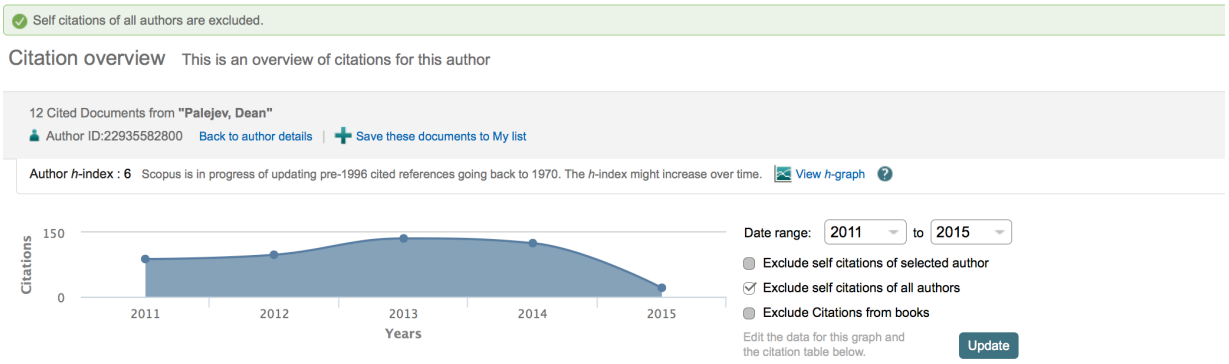
Деян Палежев

Списък на цитиранията

Цитиранията по-долу са към 27.03.2015 г. според SCOPUS, като не са включени самоцитирания (на всички съавтори). Въпреки старанието на SCOPUS е възможно някои самоцитирания да са включени в списъка, както и някои цитирания да са пропуснати.



Documents

Citations

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			<2011	2011	2012	2013	2014	2015	Subtotal	>2015	Total
		Total	**304**	**88**	**98**	**136**	**125**	**21**	**468**	**0**	**772**
1	Transcriptome Analysis of the Human Striatum in Tourette Syn...	2014						2	**2**		**2**
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5	Somatic copy number mosaicism in human skin revealed by indu...	2012				37	35	9	**81**		**81**
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10	EBNA1 regulates cellular gene expression by binding cellular...	2009		5	5	5	6	7			**23**		**28**
11	A procedure for highly specific, sensitive, and unbiased who...	2008		6	6	8	6	10			**30**		**36**
12	Paired-end mapping reveals extensive structural variation in...	2007		293	77	84	58	41	4		**264**		**557**

12. Lennington J, Coppola G, Kataoka-Sasaki Y, Fernandez T, **Palejev D**, Li Y, Huttner A, Pletikos M, Sestan N, Leckman JF, Vaccarino F (2014) Transcriptome analysis of the human striatum in Tourette syndrome. **Biol. Psychiatry**. doi: 10.1016/j.biopsych.2014.07.018. IF (2013): 9.472

Цитирания:

Pogorelov, V., Xu, M., Smith, H.R., Buchanan, G.F., Pittenger, C.
Corticostriatal interactions in the generation of tic-like behaviors after local striatal disinhibition
(2015) Experimental Neurology, 265, pp. 122-128.

DOI: 10.1016/j.expneurol.2015.01.001
DOCUMENT TYPE: Article

Robertson, M.M.
A personal 35 year perspective on Gilles de la Tourette syndrome: Assessment, investigations, and management
(2015) The Lancet Psychiatry, 2 (1), pp. 88-104.

DOI: 10.1016/S2215-0366(14)00133-3
DOCUMENT TYPE: Review

---

8. Abyzov A, Mariani J*, **Palejev D***, Zhang Y*, Haney M*, Tomasini L*, Ferrandino A, Rosenberg Belmaker L, Szekely A, Wilson M, Kocabas A, Calixto N , Grigorenko EL, Huttner A, Chawarska K, Weissman SM, Urban AE, Gerstein M, Vaccarino F (2012). Somatic copy-number mosaicism in human skin revealed by induced pluripotent stem cells. **Nature** 492, 438–442. doi:10.1038/nature11629. PMID:23160490. IF (2013): 42.351

Цитирания:

Ben-David, U.
Genomic instability, driver genes and cell selection: PROJECTIONS from cancer to stem cells
(2015) Biochimica et Biophysica Acta - Gene Regulatory Mechanisms, 1849 (4), pp. 427-435.

DOI: 10.1016/j.bbagrm.2014.08.005
DOCUMENT TYPE: Review

Gao, S., Zheng, C., Chang, G., Liu, W., Kou, X., Tan, K., Tao, L., Xu, K., Wang, H., Cai, J., Tian, J., Gao, S.
Unique features of mutations revealed by sequentially reprogrammed induced pluripotent stem cells
(2015) Nature Communications, 6, art. no. 6318, .

DOI: 10.1038/ncomms7318
DOCUMENT TYPE: Article

Yue, Y.J., Liu, J.B., Yang, M., Han, J.L., Guo, T.T., Guo, J., Feng, R.L., Yang, B.H.
De novo assembly and characterization of skin transcriptome using RNAseq in sheep (Ovis aries)
(2015) Genetics and Molecular Research, 14 (1), art. no. A146, pp. 1371-1384.

DOCUMENT TYPE: Article

Huch, M., Gehart, H., Van Boxtel, R., Hamer, K., Blokzijl, F., Verstegen, M.M.A., Ellis, E., Van Wenum, M., Fuchs, S.A., De Ligt, J., Van De Wetering, M., Sasaki, N., Boers, S.J., Kemperman, H., De Jonge, J., Ijzermans, J.N.M., Nieuwenhuis, E.E.S., Hoekstra, R., Strom, S., Vries, R.R.G., Van Der Laan, L.J.W., Cuppen, E., Clevers, H.
Long-term culture of genome-stable bipotent stem cells from adult human liver
(2015) Cell, 160 (1-2), pp. 299-312.

DOI: 10.1016/j.cell.2014.11.050
DOCUMENT TYPE: Article

Payne, N.L., Sylvain, A., O'Brien, C., Herszfeld, D., Sun, G., Bernard, C.C.A.
Application of human induced pluripotent stem cells for modeling and treating neurodegenerative diseases
(2015) New Biotechnology, 32 (1), pp. 212-228.

DOI: 10.1016/j.nbt.2014.05.001
DOCUMENT TYPE: Article

Isobe, K.-I., Cheng, Z., Nishio, N., Suganya, T., Tanaka, Y., Ito, S.
Reprint of "iPSCs, aging and age-related diseases"
(2015) New Biotechnology, 32 (1), pp. 169-179.

DOI: 10.1016/j.nbt.2014.11.002
DOCUMENT TYPE: Review

Anderson, G.M.
Autism Biomarkers: Challenges, Pitfalls and Possibilities
(2015) Journal of Autism and Developmental Disorders, 45 (4), pp. 1103-1113.

DOI: 10.1007/s10803-014-2225-4
DOCUMENT TYPE: Review

Steichen, C., Maluenda, J., Tosca, L., Luce, E., Pineau, D., Dianat, N., Hannoun, Z., Tachdjian, G., Melki, J., Dubart-Kupperschmitt, A.
An atypical human induced pluripotent stem cell line with a complex, stable, and balanced genomic rearrangement including a large de novo 1q uniparental disomy
(2015) Stem Cells Translational Medicine, 4 (3), pp. 224-229.

DOI: 10.5966/sctm.2014-0186
DOCUMENT TYPE: Article

Giri, S., Bader, A.
A low-cost, high-quality new drug discovery process using patient-derived induced pluripotent stem cells
(2015) Drug Discovery Today, 20 (1), pp. 37-49.

DOI: 10.1016/j.drudis.2014.10.011
DOCUMENT TYPE: Review

Lu, J., Li, H., Hu, M., Sasaki, T., Baccei, A., Gilbert, D.M., Liu, J.S., Collins, J.J., Lerou, P.H.
The distribution of genomic variations in human iPSCs is related to replication-timing reorganization during reprogramming
(2014) Cell Reports, 7 (1), pp. 70-78. Cited 3 times.

DOI: 10.1016/j.celrep.2014.03.007
DOCUMENT TYPE: Article

Isobe, K.-I., Cheng, Z., Nishio, N., Suganya, T., Tanaka, Y., Ito, S.
IPSCs, aging and age-related diseases
(2014) New Biotechnology, 31 (5), pp. 411-421. Cited 1 time.

DOI: 10.1016/j.nbt.2014.04.004
DOCUMENT TYPE: Review

Eshel, O., Shirak, A., Dor, L., Band, M., Zak, T., Markovich-Gordon, M., Chalifa-Caspi, V., Feldmesser, E., Weller, J.I., Seroussi, E., Hulata, G., Ron, M.
Identification of male-specific amh duplication, sexually differentially expressed genes and microRNAs at early embryonic development of Nile tilapia (Oreochromis niloticus)
(2014) BMC Genomics, 15 (1), art. no. 774, .

DOI: 10.1186/1471-2164-15-774
DOCUMENT TYPE: Article

Cai, J., Miao, X., Li, Y., Smith, C., Tsang, K., Cheng, L., Wang, Q.-F.
Whole-genome sequencing identifies genetic variances in culture-expanded human mesenchymal stem cells
(2014) Stem Cell Reports, 3 (2), pp. 227-233. Cited 1 time.

DOI: 10.1016/j.stemcr.2014.05.019
DOCUMENT TYPE: Article

Marques, F.Z., Prestes, P.R., Pinheiro, L.B., Scurrah, K., Emslie, K.R., Tomaszewski, M., Harrap, S.B., Charchar, F.J.
Measurement of absolute copy number variation reveals association with essential hypertension
(2014) BMC Medical Genomics, 7 (1), art. no. 44, .

DOI: 10.1186/1755-8794-7-44
DOCUMENT TYPE: Article

Ross, K.A.
Coherent somatic mutation in autoimmune disease
(2014) PLoS ONE, 9 (7), art. no. e101093, .

DOI: 10.1371/journal.pone.0101093
DOCUMENT TYPE: Review

Jacobs, K., Mertzanidou, A., Geens, M., Thi Nguyen, H., Staessen, C., Spits, C.
Low-grade chromosomal mosaicism in human somatic and embryonic stem cell populations
(2014) Nature Communications, 5, art. no. 4227, . Cited 2 times.

DOI: 10.1038/ncomms5227
DOCUMENT TYPE: Article

Frank, S.A.
Somatic mosaicism and disease
(2014) Current Biology, 24 (12), pp. R577-R581.

DOI: 10.1016/j.cub.2014.05.021
DOCUMENT TYPE: Review

Firas, J., Liu, X., Nefzger, C.M., Polo, J.M.
GM-CSF and MEF-conditioned media support feeder-free reprogramming of mouse granulocytes to iPS cells
(2014) Differentiation, 87 (5), pp. 193-199.

DOI: 10.1016/j.diff.2014.05.003
DOCUMENT TYPE: Article

Aghili, L., Foo, J., DeGregori, J., De, S.
Patterns of somatically acquired amplifications and deletions in apparently normal tissues of ovarian cancer patients
(2014) Cell Reports, 7 (4), pp. 1310-1319. Cited 2 times.

DOI: 10.1016/j.celrep.2014.03.071
DOCUMENT TYPE: Article

Li, Z., Lu, H., Yang, W., Yong, J., Zhang, Z.-N., Zhang, K., Deng, H., Xu, Y.
Mouse SCNT ESCs have lower somatic mutation load than syngeneic iPSCs
(2014) Stem Cell Reports, 2 (4), pp. 399-405. Cited 2 times.

DOI: 10.1016/j.stemcr.2014.02.005
DOCUMENT TYPE: Article

Dotto, G.P.
Multifocal epithelial tumors and field cancerization: Stroma as a primary determinant
(2014) Journal of Clinical Investigation, 124 (4), pp. 1446-1453. Cited 2 times.

DOI: 10.1172/JCI72589
DOCUMENT TYPE: Review

Vijg, J.
Aging genomes: A necessary evil in the logic of life
(2014) BioEssays, 36 (3), pp. 282-292. Cited 1 time.

DOI: 10.1002/bies.201300127
DOCUMENT TYPE: Article

Engle, S.J., Vincent, F.
Small molecule screening in human induced pluripotent stem cell-derived terminal cell types
(2014) Journal of Biological Chemistry, 289 (8), pp. 4562-4570. Cited 3 times.

DOI: 10.1074/jbc.R113.529156
DOCUMENT TYPE: Review

Peterson, S.E., Loring, J.F.
Genomic instability in pluripotent stem cells: Implications for clinical applications
(2014) Journal of Biological Chemistry, 289 (8), pp. 4578-4584. Cited 3 times.

DOI: 10.1074/jbc.R113.516419
DOCUMENT TYPE: Review

Ogbogu, U., Burningham, S., Ollenberger, A., Calder, K., Du, L., El Emam, K., Hyde-Lay, R., Isasi, R., Joly, Y., Kerr, I., Malin, B., McDonald, M., Penney, S., Piat, G., Roy, D.-C., Sugarman, J., Vercauteren, S., Verhenneman, G., West, L., Caulfield, T.
Policy recommendations for addressing privacy challenges associated with cell-based research and interventions
(2014) BMC Medical Ethics, 15 (1), art. no. 7, .

DOI: 10.1186/1472-6939-15-7
DOCUMENT TYPE: Article

Insel, T.R.
Brain somatic mutations: The dark matter of psychiatric genetics
(2014) Molecular Psychiatry, 19 (2), pp. 156-158. Cited 3 times.

DOI: 10.1038/mp.2013.168
DOCUMENT TYPE: Review

Nagata, N., Yamanaka, S.
Perspectives for induced pluripotent stem cell technology: New insights into human physiology involved in somatic mosaicism
(2014) Circulation Research, 114 (3), pp. 505-510. Cited 2 times.

DOI: 10.1161/CIRCRESAHA.114.303043
DOCUMENT TYPE: Review

Chen, Y., Guo, L., Chen, J., Zhao, X., Zhou, W., Zhang, C., Wang, J., Jin, L., Pei, D., Zhang, F.
Genome-wide CNV analysis in mouse induced pluripotent stem cells reveals dosage effect of pluripotent factors on genome integrity
(2014) BMC Genomics, 15 (1), art. no. 79, . Cited 2 times.

DOI: 10.1186/1471-2164-15-79
DOCUMENT TYPE: Article

Kim, J., Shin, J.-Y., Kim, J.-I., Seo, J.-S., Webster, M.J., Lee, D., Kim, S.
Somatic deletions implicated in functional diversity of brain cells of individuals with schizophrenia and unaffected controls
(2014) Scientific Reports, 4, art. no. 3807, .

DOI: 10.1038/srep03807
DOCUMENT TYPE: Article

Weissbein, U., Benvenisty, N., Ben-David, U.
Genome maintenance in pluripotent stem cells
(2014) Journal of Cell Biology, 204 (2), pp. 153-163. Cited 4 times.

DOI: 10.1083/jcb.201310135
DOCUMENT TYPE: Review

Liu, P., Kaplan, A., Yuan, B., Hanna, J.H., Lupski, J.R., Reiner, O.
Passage number is a major contributor to genomic structural variations in mouse iPSCs
(2014) Stem Cells, 32 (10), pp. 2657-2667.

DOI: 10.1002/stem.1779
DOCUMENT TYPE: Article

Hirschi, K.K., Li, S., Roy, K.
Induced pluripotent stem cells for regenerative medicine
(2014) Annual Review of Biomedical Engineering, 16, pp. 277-294.

DOI: 10.1146/annurev-bioeng-071813-105108
DOCUMENT TYPE: Review

Daughtry, B., Mitalipov, S.
Concise review: Parthenote stem cells for regenerative medicine: Genetic, epigenetic, and developmental features
(2014) Stem Cells Translational Medicine, 3 (3), pp. 290-298. Cited 3 times.

DOI: 10.5966/sctm.2013-0127
DOCUMENT TYPE: Article

Etzrodt, M., Endele, M., Schroeder, T.
Quantitative single-cell approaches to stem cell research
(2014) Cell Stem Cell, 15 (5), pp. 546-558.

DOI: 10.1016/j.stem.2014.10.015
DOCUMENT TYPE: Article

Macaulay, I.C., Voet, T.
Single Cell Genomics: Advances and Future Perspectives
(2014) PLoS Genetics, 10 (1), art. no. e1004126, . Cited 22 times.

DOI: 10.1371/journal.pgen.1004126
DOCUMENT TYPE: Review

McCole, R.B., Fonseka, C.Y., Koren, A., Wu, C.-T.
Abnormal Dosage of Ultraconserved Elements Is Highly Disfavored in Healthy Cells but Not Cancer Cells
(2014) PLoS Genetics, 10 (10), 17 p.

DOI: 10.1371/journal.pgen.1004646
DOCUMENT TYPE: Article

Hannibal, R.L., Chuong, E.B., Rivera-Mulia, J.C., Gilbert, D.M., Valouev, A., Baker, J.C.
Copy Number Variation Is a Fundamental Aspect of the Placental Genome
(2014) PLoS Genetics, 10 (5), art. no. e1004290, . Cited 2 times.

DOI: 10.1371/journal.pgen.1004290
DOCUMENT TYPE: Article

Chao, T.-H., Chen, I.-C., Tseng, S.-Y., Li, Y.-H.
Pluripotent stem cell therapy in ischemic cardiovascular disease
(2014) Acta Cardiologica Sinica, 30 (5), pp. 365-374.

DOCUMENT TYPE: Review

Avila, J., Gómez-Ramos, A., Soriano, E.
Variations in brain DNA
(2014) Frontiers in Aging Neuroscience, 6 (NOV), art. no. 323, .

DOI: 10.3389/fnagi.2014.00323
DOCUMENT TYPE: Review

Cai, X., Evrony, G.D., Lehmann, H.S., Elhosary, P.C., Mehta, B.K., Poduri, A., Walsh, C.A.
Single-Cell, Genome-wide Sequencing Identifies Clonal Somatic Copy-Number Variation in the Human Brain
(2014) Cell Reports, 8 (5), pp. 1280-1289.

DOI: 10.1016/j.celrep.2014.07.043
DOCUMENT TYPE: Article

Mourier, T., Nielsen, L.P., Hansen, A.J., Willerslev, E.
Transposable elements in cancer as a by-product of stress-induced evolvability
(2014) Frontiers in Genetics, 5 (MAY), art. no. 156, .

DOI: 10.3389/fgene.2014.00156
DOCUMENT TYPE: Article

Weissbein, U., Benvenisty, N., Ben-David, U.
Genome maintenance in pluripotent stem cells
(2014) Journal of Animal and Veterinary Advances, 12 (7), pp. 153-163.

DOI: 10.1083/jcb.201310135
DOCUMENT TYPE: Review

Grieshammer, U., Shepard, K.A.
Proceedings: Consideration of genetics in the design of induced pluripotent stem cell-based models of complex disease
(2014) Stem Cells Translational Medicine, 3 (11), pp. 1253-1259.

DOI: 10.5966/sctm.2014-0191
DOCUMENT TYPE: Article

Proukakis, C., Shoaee, M., Morris, J., Brier, T., Kara, E., Sheerin, U.-M., Charlesworth, G., Tolosa, E., Houlden, H., Wood, N.W., Schapira, A.H.
Analysis of Parkinson's disease brain-derived DNA for alpha-synuclein coding somatic mutations
(2014) Movement Disorders, 29 (8), pp. 1060-1064.

DOI: 10.1002/mds.25883
DOCUMENT TYPE: Article

Bellin, M., Casini, S., Davis, R.P., D'Aniello, C., Haas, J., Ward-Van Oostwaard, D., Tertoolen, L.G.J., Jung, C.B., Elliott, D.A., Welling, A., Laugwitz, K.-L., Moretti, A., Mummery, C.L.
Isogenic human pluripotent stem cell pairs reveal the role of a KCNH2 mutation in long-QT syndrome
(2013) EMBO Journal, 32 (24), pp. 3161-3175. Cited 11 times.

DOI: 10.1038/emboj.2013.240
DOCUMENT TYPE: Article

Napoletani, D., Signore, M., Struppa, D.C.
Cancer quasispecies and stem-like adaptive aneuploidy
(2013) F1000Research, 2, art. no. 2-268.v1, .

DOI: 10.12688/f1000research.2-268.v1
DOCUMENT TYPE: Article

McDermott, G.P., Do, D., Litterst, C.M., Maar, D., Hindson, C.M., Steenblock, E.R., Legler, T.C., Jouvenot, Y., Marrs, S.H., Bemis, A., Shah, P., Wong, J., Wang, S., Sally, D., Javier, L., Dinio, T., Han, C., Brackbill, T.P., Hodges, S.P., Ling, Y., Klitgord, N., Carman, G.J., Berman, J.R., Koehler, R.T., Hiddessen, A.L., Walse, P., Bousse, L., Tzonev, S., Hefner, E., Hindson, B.J., Cauly, T.H., Hamby, K., Patel, V.P., Regan, J.F., Wyatt, P.W., Karlin-Neumann, G.A., Stumbo, D.P., Lowe, A.J.
Multiplexed target detection using DNA-binding dye chemistry in droplet digital PCR
(2013) Analytical Chemistry, 85 (23), pp. 11619-11627. Cited 8 times.

DOI: 10.1021/ac403061n
DOCUMENT TYPE: Article

Wu, Y.-L., Pandian, G.N., Ding, Y.-P., Zhang, W., Tanaka, Y., Sugiyama, H.
Clinical grade iPS cells: Need for versatile small molecules and optimal cell sources
(2013) Chemistry and Biology, 20 (11), pp. 1311-1322. Cited 6 times.

DOI: 10.1016/j.chembiol.2013.09.016
DOCUMENT TYPE: Review

Maguire, G.
Using a systems-based approach to overcome reductionist strategies in the development of diagnostics
(2013) Expert Review of Molecular Diagnostics, 13 (8), pp. 895-905. Cited 1 time.

DOI: 10.1586/14737159.2013.846828
DOCUMENT TYPE: Review

Wang, J., Gu, Q., Hao, J., Bai, D., Liu, L., Zhao, X., Liu, Z., Wang, L., Zhou, Q.
Generation of Induced Pluripotent Stem Cells with High Efficiency from Human Umbilical Cord Blood Mononuclear Cells
(2013) Genomics, Proteomics and Bioinformatics, 11 (5), pp. 304-311. Cited 2 times.

DOI: 10.1016/j.gpb.2013.08.002
DOCUMENT TYPE: Article

Campbell, C.D., Eichler, E.E.
Properties and rates of germline mutations in humans
(2013) Trends in Genetics, 29 (10), pp. 575-584. Cited 20 times.

DOI: 10.1016/j.tig.2013.04.005
DOCUMENT TYPE: Review

Trounson, A., Mitalipov, S.
Foreword to the Second Edition
(2013) Principles of Cloning: Second Edition, pp. xi-xii.

DOI: 10.1016/B978-0-12-386541-0.00049-7
DOCUMENT TYPE: Editorial

Zhang, X.-B.
Cellular Reprogramming of Human Peripheral Blood Cells
(2013) Genomics, Proteomics and Bioinformatics, 11 (5), pp. 264-274. Cited 2 times.

DOI: 10.1016/j.gpb.2013.09.001
DOCUMENT TYPE: Review

Almeida, S., Gascon, E., Tran, H., Chou, H.J., Gendron, T.F., Degroot, S., Tapper, A.R., Sellier, C., Charlet-Berguerand, N., Karydas, A., Seeley, W.W., Boxer, A.L., Petrucelli, L., Miller, B.L., Gao, F.-B.
Modeling key pathological features of frontotemporal dementia with C9ORF72 repeat expansion in iPSC-derived human neurons
(2013) Acta Neuropathologica, 126 (3), pp. 385-399. Cited 38 times.

DOI: 10.1007/s00401-013-1149-y
DOCUMENT TYPE: Article

Gallegos, T.F., Sancho-Martinez, I., Izpisua Belmonte, J.C.
Advances in cellular reprogramming: Moving toward a reprieve from immunogenicity
(2013) Immunology Letters, 155 (1-2), pp. 14-17.

DOI: 10.1016/j.imlet.2013.09.019
DOCUMENT TYPE: Review

Filippich, C., Wolvetang, E.J., Mowry, B.J.
Will brain cells derived from induced pluripotent stem cells or directly converted from somatic cells (iNs) be useful for schizophrenia research?
(2013) Schizophrenia Bulletin, 39 (5), pp. 948-954. Cited 1 time.

DOI: 10.1093/schbul/sbt103
DOCUMENT TYPE: Article

Speicher, M.R.
Single-cell analysis: Toward the clinic
(2013) Genome Medicine, 5 (8), art. no. 74, . Cited 3 times.

DOI: 10.1186/gm478
DOCUMENT TYPE: Editorial

Polanco, J.C., Ho, M.S.H., Wang, B., Zhou, Q., Wolvetang, E., Mason, E., Wells, C.A., Kolle, G., Grimmond, S.M., Bertoncello, I., O'brien, C., Laslett, A.L.
Identification of unsafe human induced pluripotent stem cell lines using a robust surrogate assay for pluripotency
(2013) Stem Cells, 31 (8), pp. 1498-1510. Cited 4 times.

DOI: 10.1002/stem.1425
DOCUMENT TYPE: Article

Liang, G., Zhang, Y.
Genetic and epigenetic variations in iPSCs: Potential causes and implications for application
(2013) Cell Stem Cell, 13 (2), pp. 149-159. Cited 24 times.

DOI: 10.1016/j.stem.2013.07.001
DOCUMENT TYPE: Review

Silveira, S.M., Villacis, R.A.R., Marchi, F.A., de Barros Filho, M.C., Drigo, S.A., Neto, C.S., Lopes, A., da Cunha, I.W., Rogatto, S.R.
Genomic Signatures Predict Poor Outcome in Undifferentiated Pleomorphic Sarcomas and Leiomyosarcomas
(2013) PLoS ONE, 8 (6), art. no. e67643, . Cited 3 times.

DOI: 10.1371/journal.pone.0067643
DOCUMENT TYPE: Article

Miyake, K., Yang, C., Minakuchi, Y., Ohori, K., Soutome, M., Hirasawa, T., Kazuki, Y., Adachi, N., Suzuki, S., Itoh, M., Goto, Y.-I., Andoh, T., Kurosawa, H., Oshimura, M., Sasaki, M., Toyoda, A., Kubota, T.
Comparison of Genomic and Epigenomic Expression in Monozygotic Twins Discordant for Rett Syndrome
(2013) PLoS ONE, 8 (6), art. no. e66729, . Cited 13 times.

DOI: 10.1371/journal.pone.0066729
DOCUMENT TYPE: Article

Buganim, Y., Faddah, D.A., Jaenisch, R.
Mechanisms and models of somatic cell reprogramming
(2013) Nature Reviews Genetics, 14 (6), pp. 427-439. Cited 48 times.

DOI: 10.1038/nrg3473
DOCUMENT TYPE: Review

Cahan, P., Daley, G.Q.
Origins and implications of pluripotent stem cell variability and heterogeneity
(2013) Nature Reviews Molecular Cell Biology, 14 (6), pp. 357-368. Cited 44 times.

DOI: 10.1038/nrm3584
DOCUMENT TYPE: Review

Proukakis, C., Houlden, H., Schapira, A.H.
Somatic alpha-synuclein mutations in Parkinson's disease: Hypothesis and preliminary data
(2013) Movement Disorders, 28 (6), pp. 705-712. Cited 7 times.

DOI: 10.1002/mds.25502
DOCUMENT TYPE: Article

Su, R.-J., Baylink, D.J., Neises, A., Kiroyan, J.B., Meng, X., Payne, K.J., Tschudy-Seney, B., Duan, Y., Appleby, N., Kearns-Jonker, M., Gridley, D.S., Wang, J., Lau, K.-H.W., Zhang, X.-B.
Efficient Generation of Integration-Free iPS Cells from Human Adult Peripheral Blood Using BCL-XL Together with Yamanaka Factors
(2013) PLoS ONE, 8 (5), art. no. e64496, . Cited 10 times.

DOI: 10.1371/journal.pone.0064496
DOCUMENT TYPE: Article

Pryzhkova, M.V.
Concise review: Carbon nanotechnology: Perspectives in stem cell research
(2013) Stem Cells Translational Medicine, 2 (5), pp. 376-383. Cited 2 times.

DOI: 10.5966/sctm.2012-0151
DOCUMENT TYPE: Review

Thanasegaran, S., Cheng, Z., Ito, S., Nishio, N., Isobe, K.-I.
No immunogenicity of IPS cells in syngeneic host studied by in vivo injection and 3D scaffold experiments
(2013) BioMed Research International, 2013, art. no. 378207, . Cited 4 times.

DOI: 10.1155/2013/378207
DOCUMENT TYPE: Article

Biesecker, L.G., Spinner, N.B.
A genomic view of mosaicism and human disease
(2013) Nature Reviews Genetics, 14 (5), pp. 307-320. Cited 50 times.

DOI: 10.1038/nrg3424
DOCUMENT TYPE: Review

Bai, Q., Desprat, R., Klein, B., Lemaître, J.-M., de Vos, J.
Embryonic stem cells or induced Pluripotent stem cells? A DNA integrity perspective
(2013) Current Gene Therapy, 13 (2), pp. 93-98. Cited 3 times.

DOI: 10.2174/1566523211313020003
DOCUMENT TYPE: Article

Dianat, N., Steichen, C., Vallier, L., Weber, A., Dubart-Kupperschmitt, A.
Human pluripotent stem cells for modelling human liver diseases and cell therapy
(2013) Current Gene Therapy, 13 (2), pp. 120-132. Cited 9 times.

DOI: 10.2174/1566523211313020006
DOCUMENT TYPE: Review

Bayart, E., Cohen-Haguenauer, O.
Technological overview of iPS induction from human adult somatic cells
(2013) Current Gene Therapy, 13 (2), pp. 73-92. Cited 18 times.

DOI: 10.2174/1566523211313020002
DOCUMENT TYPE: Review

Ben-David, U., Mayshar, Y., Benvenisty, N.
Virtual karyotyping of pluripotent stem cells on the basis of their global gene expression profiles
(2013) Nature Protocols, 8 (5), pp. 989-997. Cited 9 times.

DOI: 10.1038/nprot.2013.051
DOCUMENT TYPE: Article

Su, R.-J., Yang, Y., Neises, A., Payne, K.J., Wang, J., Viswanathan, K., Wakeland, E.K., Fang, X., Zhang, X.-B.
Few Single Nucleotide Variations in Exomes of Human Cord Blood Induced Pluripotent Stem Cells
(2013) PLoS ONE, 8 (4), art. no. e59908, . Cited 2 times.

DOI: 10.1371/journal.pone.0059908
DOCUMENT TYPE: Article

Katsouras, C.S., Baltogiannis, G.G., Naka, K.K., Roukos, D.H., Michalis, L.K.
Decoding coronary artery disease: Somatic mosaicism and genomics for personal and population risk prediction
(2013) Biomarkers in Medicine, 7 (2), pp. 189-192.

DOI: 10.2217/bmm.13.4
DOCUMENT TYPE: Review

Panchision, D.M.
Meeting report: Using stem cells for biological and therapeutics discovery in mental illness, April 2012
(2013) Stem Cells Translational Medicine, 2 (3), pp. 217-222. Cited 2 times.

DOI: 10.5966/sctm.2012-0149
DOCUMENT TYPE: Article

Ess, K.C.
Patient heal thyself: Modeling and treating neurological disorders using patient-derived stem cells
(2013) Experimental Biology and Medicine, 238 (3), pp. 308-314. Cited 3 times.

DOI: 10.1177/1535370213480713
DOCUMENT TYPE: Review

Pryzhkova, M.V.
Stem cells: Will they ever be the same?
(2013) Regenerative Medicine, 8 (2), pp. 97-99. Cited 2 times.

DOI: 10.2217/rme.13.9
DOCUMENT TYPE: Editorial

De Almeida, P.E., Ransohoff, J.D., Nahid, A., Wu, J.C.
Immunogenicity of pluripotent stem cells and their derivatives
(2013) Circulation Research, 112 (3), pp. 549-561. Cited 19 times.

DOI: 10.1161/CIRCRESAHA.111.249243
DOCUMENT TYPE: Review

Lupski, J.R.
Genome mosaicism - One human, multiple genomes
(2013) Science, 341 (6144), pp. 358-359. Cited 28 times.

DOI: 10.1126/science.1239503
DOCUMENT TYPE: Short Survey

Macosko, E.Z., McCarroll, S.A.
Our fallen genomes
(2013) Science, 342 (6158), pp. 564-565. Cited 1 time.

DOI: 10.1126/science.1246942
DOCUMENT TYPE: Short Survey

McConnell, M.J., Lindberg, M.R., Brennand, K.J., Piper, J.C., Voet, T., Cowing-Zitron, C., Shumilina, S., Lasken, R.S., Vermeesch, J.R., Hall, I.M., Gage, F.H.
Mosaic copy number variation in human neurons
(2013) Science, 342 (6158), pp. 632-637. Cited 53 times.

DOI: 10.1126/science.1243472
DOCUMENT TYPE: Article

---

7. Naumova OY, **Palejev D**, Vlasova NV, Lee M, Rychkov SY, Babich ON, Vaccarino F, Grigorenko EL (2012) Age-related changes of gene expression in the neocortex: Preliminary data on RNA-seq of the transcriptome in three functionally distinct cortical areas. **Dev. Psychopathol.** 24 (2012), 1427–1442. doi:10.1017/S0954579412000818. PMID: 23062308. IF (2013): 4.40

Цитирания:

Prasad, H., Rao, R.
The Na+/H+ exchanger NHE6 modulates endosomal pH to control processing of amyloid precursor protein in a cell culture model of alzheimer disease
(2015) Journal of Biological Chemistry, 290 (9), pp. 5311-5327.

DOI: 10.1074/jbc.M114.602219

DOCUMENT TYPE: Article

Plomin, R., Simpson, M.A.
The future of genomics for developmentalists
(2013) Development and Psychopathology, 25 (4 PART 2), pp. 1263-1278. Cited 9 times.

DOI: 10.1017/S0954579413000606
DOCUMENT TYPE: Article

---

6. Mariani J, Simonini MV, **Palejev D**, Tomasini L, Coppola G, Szekely A, Horvath T, Vaccarino F. (2012). Modeling Human Cortical Development in vitro using induced Pluripotent Stem Cells. **Proc. Natl. Acad. Sci. U. S. A.** 109(31):12770-5. doi: 10.1073/pnas.1202944109. PMCID: PMC3411972. IF (2013): 9.809

Цитирания:

Topol, A., Tran, N.N., Brennand, K.J.
A guide to generating and using hiPSC derived NPCs for the study of neurological diseases
(2015) Journal of Visualized Experiments, (96), art. no. e52495, .

DOI: 10.3791/52495
DOCUMENT TYPE: Article

Brennand, K., Savas, J.N., Kim, Y., Tran, N., Simone, A., Hashimoto-Torii, K., Beaumont, K.G., Kim, H.J., Topol, A., Ladran, I., Abdelrahim, M., Matikainen-Ankney, B., Chao, S.-H., Mrksich, M., Rakic, P., Fang, G., Zhang, B., Yates, J.R., Gage, F.H.
Phenotypic differences in hiPSC NPCs derived from patients with schizophrenia
(2015) Molecular Psychiatry, 20 (3), pp. 361-368.

DOI: 10.1038/mp.2014.22
DOCUMENT TYPE: Article

Brändl, B., Schneider, S.A., Loring, J.F., Hardy, J., Gribbon, P., Müller, F.-J.
Stem cell reprogramming: Basic implications and future perspective for movement disorders
(2015) Movement Disorders, 30 (3), pp. 301-312.

DOI: 10.1002/mds.26113
DOCUMENT TYPE: Review

Schmidt, D., Cho, Y.K.
Natural photoreceptors and their application to synthetic biology
(2015) Trends in Biotechnology, 33 (2), pp. 80-91.

DOI: 10.1016/j.tibtech.2014.10.007
DOCUMENT TYPE: Review

Tang-Schomer, M.D., White, J.D., Tien, L.W., Schmitt, L.I., Valentin, T.M., Graziano, D.J., Hopkins, A.M., Omenetto, F.G., Haydon, P.G., Kaplan, D.L.
Bioengineered functional brain-like cortical tissue

(2014) Proceedings of the National Academy of Sciences of the United States of America, 111 (38), pp. 13811-13816. Cited 4 times.

DOI: 10.1073/pnas.1324214111
DOCUMENT TYPE: Article

Klincumhom, N., Tharasanit, T., Thongkittidilok, C., Tiptanavattana, N., Rungarunlert, S., Dinnyés, A., Techakumphu, M.
Selective TGF-β1/ALK inhibitor improves neuronal differentiation of mouse embryonic stem cells
(2014) Neuroscience Letters, 578, pp. 1-6.

DOI: 10.1016/j.neulet.2014.06.001
DOCUMENT TYPE: Article

Gualda, E.J., Simão, D., Pinto, C., Alves, P.M., Brito, C.
Imaging of human differentiated 3D neural aggregates using light sheet fluorescence microscopy
(2014) Frontiers in Cellular Neuroscience, 8 (AUG), art. no. 221, .

DOI: 10.3389/fncel.2014.00221
DOCUMENT TYPE: Article

Stein, J.L., de la Torre-Ubieta, L., Tian, Y., Parikshak, N.N., Hernández, I.A., Marchetto, M.C., Baker, D.K., Lu, D., Hinman, C.R., Lowe, J.K., Wexler, E.M., Muotri, A.R., Gage, F.H., Kosik, K.S., Geschwind, D.H.
A quantitative framework to evaluate modeling of cortical development by neural stem cells
(2014) Neuron, 83 (1), pp. 69-86. Cited 2 times.

DOI: 10.1016/j.neuron.2014.05.035
DOCUMENT TYPE: Article

Haggarty, S.J., Perlis, R.H.
Translation: Screening for novel therapeutics with disease-relevant cell types derived from human stem cell models
(2014) Biological Psychiatry, 75 (12), pp. 952-960. Cited 3 times.

DOI: 10.1016/j.biopsych.2013.05.028
DOCUMENT TYPE: Review

Brennand, K.J., Landek-Salgado, M.A., Sawa, A.
Modeling heterogeneous patients with a clinical diagnosis of schizophrenia with induced pluripotent stem cells
(2014) Biological Psychiatry, 75 (12), pp. 936-944. Cited 5 times.

DOI: 10.1016/j.biopsych.2013.10.025
DOCUMENT TYPE: Review

Lin, M., Zhao, D., Hrabovsky, A., Pedrosa, E., Zheng, D., Lachman, H.M.
Heat shock alters the expression of schizophrenia and autism candidate genes in an induced pluripotent stem cell model of the human telencephalon
(2014) PLoS ONE, 9 (4), art. no. e94968, . Cited 1 time.

DOI: 10.1371/journal.pone.0094968
DOCUMENT TYPE: Article

Kim, D.-S., Joel Ross, P., Zaslavsky, K., Ellis, J.
Optimizing neuronal differentiation from induced pluripotent stem cells to model ASD
(2014) Frontiers in Cellular Neuroscience, 8 (1 APR), art. no. 109, . Cited 1 time.

DOI: 10.3389/fncel.2014.00109
DOCUMENT TYPE: Review

Li, Y., Xu, C., Ma, T.
In vitro organogenesis from pluripotent stem cells
(2014) Organogenesis, 10 (2), pp. 159-163.

DOI: 10.4161/org.28918
DOCUMENT TYPE: Review

Dage, J.L., Colvin, E.M., Fouillet, A., Langron, E., Roell, W.C., Li, J., Mathur, S.X., Mogg, A.J., Schmitt, M.G., Felder, C.C., Merchant, K.M., Isaac, J., Broad, L.M., Sher, E., Ursu, D.
Pharmacological characterisation of ligand- and voltage-gated ion channels expressed in human iPSC-derived forebrain neurons
(2014) Psychopharmacology, 231 (6), pp. 1105-1124. Cited 4 times.

DOI: 10.1007/s00213-013-3384-2
DOCUMENT TYPE: Article

Prilutsky, D., Palmer, N.P., Smedemark-Margulies, N., Schlaeger, T.M., Margulies, D.M., Kohane, I.S.
IPSC-derived neurons as a higher-throughput readout for autism: Promises and pitfalls
(2014) Trends in Molecular Medicine, 20 (2), pp. 91-104.

DOI: 10.1016/j.molmed.2013.11.004
DOCUMENT TYPE: Review

Bilican, B., Livesey, M.R., Haghi, G., Qiu, J., Burr, K., Siller, R., Hardingham, G.E., Wyllie, D.J.A., Chandran, S.
Physiological normoxia and absence of EGF is required for the long-term propagation of anterior neural precursors from human pluripotent cells
(2014) PLoS ONE, 9 (1), art. no. e85932, . Cited 4 times.

DOI: 10.1371/journal.pone.0085932
DOCUMENT TYPE: Article

Patterson, M., Gaeta, X., Loo, K., Edwards, M., Smale, S., Cinkornpumin, J., Xie, Y., Listgarten, J., Azghadi, S., Douglass, S.M., Pellegrini, M., Lowry, W.E.
Let-7 miRNAs can act through NOTCH to regulate human gliogenesis
(2014) Stem Cell Reports, 3 (5), pp. 758-773.

DOI: 10.1016/j.stemcr.2014.08.015
DOCUMENT TYPE: Article

Campos, P.B., Paulsen, B.S., Rehen, S.K.
Accelerating neuronal aging in in vitro model brain disorders: A focus on reactive oxygen species
(2014) Frontiers in Aging Neuroscience, 6 (OCT), art. no. 292, .

DOI: 10.3389/fnagi.2014.00292
DOCUMENT TYPE: Review

Pamies, D., Hartung, T., Hogberg, H.T.
Biological and medical applications of a brain-on-a-chip
(2014) Experimental Biology and Medicine, 239 (9), pp. 1096-1107. Cited 3 times.

DOI: 10.1177/1535370214537738

DOCUMENT TYPE: Article

Balmer, N.V., Klima, S., Rempel, E., Ivanova, V.N., Kolde, R., Weng, M.K., Meganathan, K., Henry, M., Sachinidis, A., Berthold, M.R., Hengstler, J.G., Rahnenführer, J., Waldmann, T., Leist, M.
From transient transcriptome responses to disturbed neurodevelopment: Role of histone acetylation and methylation as epigenetic switch between reversible and irreversible drug effects
(2014) Archives of Toxicology, 88 (7), pp. 1451-1468. Cited 5 times.

DOI: 10.1007/s00204-014-1279-6
DOCUMENT TYPE: Article

Anderson, S., Vanderhaeghen, P.
Cortical neurogenesis from pluripotent stem cells: Complexity emerging from simplicity
(2014) Current Opinion in Neurobiology, 27, pp. 151-157. Cited 3 times.

DOI: 10.1016/j.conb.2014.03.012
DOCUMENT TYPE: Review

Srikanth, P., Young-Pearse, T.L.
Stem cells on the brain: Modeling neurodevelopmental and neurodegenerative diseases using human induced pluripotent stem cells
(2014) Journal of Neurogenetics, 28 (1-2), pp. 5-29. Cited 2 times.

DOI: 10.3109/01677063.2014.881358
DOCUMENT TYPE: Review

Schadt, E.E., Buchanan, S., Brennand, K.J., Merchant, K.M.
Evolving toward a human-cell based and multiscale approach to drug discovery for CNS disorders
(2014) Frontiers in Pharmacology, 5 (DEC), art. no. 252, .

DOI: 10.3389/fphar.2014.00252
DOCUMENT TYPE: Review

Ader, M., Tanaka, E.M.
Modeling human development in 3D culture
(2014) Current Opinion in Cell Biology, 31 (1), pp. 23-28. Cited 1 time.

DOI: 10.1016/j.ceb.2014.06.013
DOCUMENT TYPE: Review

Van den Ameele, J., Tiberi, L., Vanderhaeghen, P., Espuny-Camacho, I.
Thinking out of the dish: What to learn about cortical development using pluripotent stem cells
(2014) Trends in Neurosciences, 37 (6), pp. 334-342. Cited 2 times.

DOI: 10.1016/j.tins.2014.03.005
DOCUMENT TYPE: Review

Karus, M., Blaess, S., Brüstle, O.
Self-organization of neural tissue architectures from pluripotent stem cells
(2014) Journal of Comparative Neurology, 522 (12), pp. 2831-2844.

DOI: 10.1002/cne.23608
DOCUMENT TYPE: Review

Kim, T.-G., Yao, R., Monnell, T., Cho, J.-H., Vasudevan, A., Koh, A., Peeyush, K.T., Moon, M., Datta, D., Bolshakov, V.Y., Kim, K.-S., Chung, S.
Efficient specification of interneurons from human pluripotent stem cells by dorsoventral and rostrocaudal modulation
(2014) Stem Cells, 32 (7), pp. 1789-1804. Cited 4 times.

DOI: 10.1002/stem.1704
DOCUMENT TYPE: Article

Sadegh, C., Macklis, J.D.
Established monolayer differentiation of mouse embryonic stem cells generates heterogeneous neocortical-like neurons stalled at a stage equivalent to midcorticogenesis
(2014) Journal of Comparative Neurology, 522 (12), pp. 2691-2706.

DOI: 10.1002/cne.23576
DOCUMENT TYPE: Article

Radonjić, N.V., Ayoub, A.E., Memi, F., Yu, X., Maroof, A., Jakovcevski, I., Anderson, S.A., Rakic, P., Zecevic, N.
Diversity of Cortical Interneurons in Primates: The Role of the Dorsal Proliferative Niche
(2014) Cell Reports, 9 (6), pp. 2139-2151. Cited 1 time.

DOI: 10.1016/j.celrep.2014.11.026
DOCUMENT TYPE: Article

Kadoshima, T., Sakaguchi, H., Nakano, T., Soen, M., Ando, S., Eiraku, M., Sasai, Y.
Self-organization of axial polarity, inside-out layer pattern, and species-specific progenitor dynamics in human ES cell-derived neocortex
(2013) Proceedings of the National Academy of Sciences of the United States of America, 110 (50), pp. 20284-20289. Cited 11 times.

DOI: 10.1073/pnas.1315710110
DOCUMENT TYPE: Article

Brennand, K.J.
Inducing cellular aging: Enabling neurodegeneration-in-a-dish
(2013) Cell Stem Cell, 13 (6), pp. 635-636. Cited 1 time.

DOI: 10.1016/j.stem.2013.11.017
DOCUMENT TYPE: Article

Klefenz, F., Williamson, A.
Modeling the formation process of grouping stimuli sets through cortical columns and microcircuits to feature neurons
(2013) Computational Intelligence and Neuroscience, 2013, art. no. 290358, .

DOI: 10.1155/2013/290358
DOCUMENT TYPE: Review

Tornero, D., Wattananit, S., Madsen, M.G., Koch, P., Wood, J., Tatarishvili, J., Mine, Y., Ge, R., Monni, E., Devaraju, K., Hevner, R.F., Brüstle, O., Lindvall, O., Kokaia, Z.
Human induced pluripotent stem cell-derived cortical neurons integrate in stroke-injured cortex and improve functional recovery
(2013) Brain, 136 (12), pp. 3561-3577. Cited 14 times.

DOI: 10.1093/brain/awt278
DOCUMENT TYPE: Article

Ladran, I., Tran, N., Topol, A., Brennand, K.J.
Neural stem and progenitor cells in health and disease
(2013) Wiley Interdisciplinary Reviews: Systems Biology and Medicine, 5 (6), pp. 701-715.

DOI: 10.1002/wsbm.1239
DOCUMENT TYPE: Review

Veenstra-Vanderweele, J.
Here/in this issue and there/abstract thinking: Human brain development in a dish
(2013) Journal of the American Academy of Child and Adolescent Psychiatry, 52 (10), pp. 993-994.

DOI: 10.1016/j.jaac.2013.07.015
DOCUMENT TYPE: Editorial

Bershteyn, M., Kriegstein, A.R.
XCerebral organoids in a dish: Progress and prospects
(2013) Cell, 155 (1), pp. X19-20. Cited 5 times.

DOI: 10.1016/j.cell.2013.09.010
DOCUMENT TYPE: Review

DeBoer, E.M., Kraushar, M.L., Hart, R.P., Rasin, M.-R.
Post-transcriptional regulatory elements and spatiotemporal specification of neocortical stem cells and projection neurons
(2013) Neuroscience, 248, pp. 499-528. Cited 2 times.

DOI: 10.1016/j.neuroscience.2013.05.042
DOCUMENT TYPE: Review

Lancaster, M.A., Renner, M., Martin, C.-A., Wenzel, D., Bicknell, L.S., Hurles, M.E., Homfray, T., Penninger, J.M., Jackson, A.P., Knoblich, J.A.
Cerebral organoids model human brain development and microcephaly
(2013) Nature, 501 (7467), pp. 373-379. Cited 129 times.

DOI: 10.1038/nature12517
DOCUMENT TYPE: Article

Brüstle, O.
Developmental neuroscience: Miniature human brains
(2013) Nature, 501 (7467), pp. 319-320. Cited 4 times.

DOI: 10.1038/nature12552
DOCUMENT TYPE: Note

Boissart, C., Poulet, A., Georges, P., Darville, H., Julita, E., Delorme, R., Bourgeron, T., Peschanski, M., Benchoua, A.
Differentiation from human pluripotent stem cells of cortical neurons of the superficial layers amenable to psychiatric disease modeling and high-throughput drug screening
(2013) Translational Psychiatry, 3, art. no. e294, . Cited 3 times.

DOI: 10.1038/tp.2013.71
DOCUMENT TYPE: Article

Hjelm, B.E., Salhia, B., Kurdoglu, A., Szelinger, S., Reiman, R.A., Sue, L.I., Beach, T.G., Huentelman, M.J., Craig, D.W.

In vitro-differentiated neural cell cultures progress towards donor-identical brain tissue
(2013) Human Molecular Genetics, 22 (17), art. no. ddt208, pp. 3534-3546. Cited 3 times.

DOI: 10.1093/hmg/ddt208
DOCUMENT TYPE: Article

Nicoleau, C., Varela, C., Bonnefond, C., Maury, Y., Bugi, A., Aubry, L., Viegas, P., Bourgois-Rocha, F., Peschanski, M., Perrier, A.L.
Embryonic stem cells neural differentiation qualifies the role of Wnt/β-Catenin signals in human telencephalic specification and regionalization
(2013) Stem Cells, 31 (9), pp. 1763-1774. Cited 6 times.

DOI: 10.1002/stem.1462
DOCUMENT TYPE: Article

Filippich, C., Wolvetang, E.J., Mowry, B.J.
Will brain cells derived from induced pluripotent stem cells or directly converted from somatic cells (iNs) be useful for schizophrenia research?
(2013) Schizophrenia Bulletin, 39 (5), pp. 948-954. Cited 1 time.

DOI: 10.1093/schbul/sbt103
DOCUMENT TYPE: Article

Nestor, M.W., Noggle, S.A.
Standardization of human stem cell pluripotency using bioinformatics
(2013) Stem Cell Research and Therapy, 4 (2), art. no. 37, . Cited 4 times.

DOI: 10.1186/scrt185
DOCUMENT TYPE: Review

Millan, M.J.
An epigenetic framework for neurodevelopmental disorders: From pathogenesis to potential therapy
(2013) Neuropharmacology, 68, pp. 2-82. Cited 38 times.

DOI: 10.1016/j.neuropharm.2012.11.015
DOCUMENT TYPE: Review

Hick, A., Wattenhofer-Donzé, M., Chintawar, S., Tropel, P., Simard, J.P., Vaucamps, N., Gall, D., Lambot, L., André, C., Reutenauer, L., Rai, M., Teletin, M., Messaddeq, N., Schiffmann, S.N., Viville, S., Pearson, C.E., Pandolfo, M., Puccio, H.
Neurons and cardiomyocytes derived from induced pluripotent stem cells as a model for mitochondrial defects in Friedreich's ataxia
(2013) DMM Disease Models and Mechanisms, 6 (3), pp. 608-621. Cited 24 times.

DOI: 10.1242/dmm.010900
DOCUMENT TYPE: Article

Zhang, K., Yi, F., Liu, G.-H., Izpisua Belmonte, J.C.
New march towards the regeneration of sensation and cognition: Hear more, see more and learn more
(2013) Journal of Molecular Cell Biology, 5 (2), pp. 151-153. Cited 1 time.

DOI: 10.1093/jmcb/mjs060
DOCUMENT TYPE: Article

Müller, W.

Induced Stem Cells and Tissue Engineering [Ersatzgewebe aus Bioreaktoren: Stammzellen III: Regeneration außerhalb des Körpers]
(2013) Biologie in Unserer Zeit, 43 (2), pp. 104-110.

DOI: 10.1002/biuz.201310503
DOCUMENT TYPE: Article

Panchision, D.M.
Meeting report: Using stem cells for biological and therapeutics discovery in mental illness, April 2012
(2013) Stem Cells Translational Medicine, 2 (3), pp. 217-222. Cited 2 times.

DOI: 10.5966/sctm.2012-0149
DOCUMENT TYPE: Article

Sasai, Y.
Cytosystems dynamics in self-organization of tissue architecture
(2013) Nature, 493 (7432), pp. 318-326. Cited 35 times.

DOI: 10.1038/nature11859
DOCUMENT TYPE: Review

Tran, N.N., Ladran, I.G., Brennand, K.J.
Modeling schizophrenia using induced pluripotent stem cell-derived and fibroblast-induced neurons
(2013) Schizophrenia Bulletin, 39 (1), pp. 4-10. Cited 5 times.

DOI: 10.1093/schbul/sbs127
DOCUMENT TYPE: Article

---

5. Vaccarino FM, Stevens HE, Kocabas A, **Palejev D**, Szekely A, Grigorenko EL, Weissman S. (2011) Induced pluripotent stem cells: a new tool to confront the challenge of neuropsychiatric disorders. **Neuropharmacol.** 60(7-8):1355-63. doi: 10.1016/j.neuropharm.2011.02.021. PMCID: PMC3087494. IF (2013): 4.819

Цитирания:

Duan, J.
Path from schizophrenia genomics to biology: Gene regulation and perturbation in neurons derived from induced pluripotent stem cells and genome editing
(2015) Neuroscience Bulletin, 31 (1), pp. 113-127. Cited 1 time.

DOI: 10.1007/s12264-014-1488-2
DOCUMENT TYPE: Review

Liu, G., Rustom, N., Litteljohn, D., Bobyn, J., Rudyk, C., Anisman, H., Hayley, S.
Use of induced pluripotent stem cell derived neurons engineered to express BDNF for modulation of stressor related pathology
(2014) Frontiers in Cellular Neuroscience, 8 (OCT), art. no. 316, 12 p.

DOI: 10.3389/fncel.2014.00316
DOCUMENT TYPE: Article

Dage, J.L., Colvin, E.M., Fouillet, A., Langron, E., Roell, W.C., Li, J., Mathur, S.X., Mogg, A.J., Schmitt, M.G., Felder, C.C., Merchant, K.M., Isaac, J., Broad, L.M., Sher, E., Ursu, D.
Pharmacological characterisation of ligand- and voltage-gated ion channels expressed in human iPSC-derived forebrain neurons
(2014) Psychopharmacology, 231 (6), pp. 1105-1124. Cited 4 times.

DOI: 10.1007/s00213-013-3384-2
DOCUMENT TYPE: Article

Prilutsky, D., Palmer, N.P., Smedemark-Margulies, N., Schlaeger, T.M., Margulies, D.M., Kohane, I.S.
IPSC-derived neurons as a higher-throughput readout for autism: Promises and pitfalls
(2014) Trends in Molecular Medicine, 20 (2), pp. 91-104.

DOI: 10.1016/j.molmed.2013.11.004
DOCUMENT TYPE: Review

Srikanth, P., Young-Pearse, T.L.
Stem cells on the brain: Modeling neurodevelopmental and neurodegenerative diseases using human induced pluripotent stem cells
(2014) Journal of Neurogenetics, 28 (1-2), pp. 5-29. Cited 2 times.

DOI: 10.3109/01677063.2014.881358
DOCUMENT TYPE: Review

Liu, G., Anisman, H., Bobyn, J., Hayley, S.
Interaction Between Nonviral Reprogrammed Fibroblast Stem Cells and Trophic Factors for Brain Repair
(2014) Molecular Neurobiology, 50 (2), pp. 673-684.

DOI: 10.1007/s12035-014-8680-2
DOCUMENT TYPE: Article

Ratajczak, M.Z., Kucharska-Mazur, J., Samochowiec, J.
Stem cell research and its growing impact on contemporary psychiatry [Badania nad komórkami macierzystymi i ich rosnĄcy wpływ na współczesnĄ psychiatrię]
(2014) Psychiatria Polska, 48 (6), pp. 1073-1085.

DOCUMENT TYPE: Article

Langley, G.R.
Considering a new paradigm for Alzheimer's disease research
(2014) Drug Discovery Today, 19 (8), pp. 1114-1124. Cited 1 time.

DOI: 10.1016/j.drudis.2014.03.013
DOCUMENT TYPE: Review

Petrov, Y.P., Kukhareva, L.V., Krylova, T.A.
The effect of type I collagen and fibronectin on the morphology of human mesenchymal stromal cells in culture
(2013) Cell and Tissue Biology, 7 (6), pp. 545-555. Cited 1 time.

DOI: 10.1134/S1990519X13060096
DOCUMENT TYPE: Article

Qu, X., Liu, T., Song, K., Li, X., Ge, D.

Differentiation of reprogrammed human adipose mesenchymal stem cells toward neural cells with defined transcription factors
(2013) Biochemical and Biophysical Research Communications, 439 (4), pp. 552-558.

DOI: 10.1016/j.bbrc.2013.09.005
DOCUMENT TYPE: Article

Lasalle, J.M.
Epigenomic strategies at the interface of genetic and environmental risk factors for autism
(2013) Journal of Human Genetics, 58 (7), pp. 396-401. Cited 13 times.

DOI: 10.1038/jhg.2013.49
DOCUMENT TYPE: Review

Da Silveira Paulsen, B., Souza Da Silveira, M., Galina, A., Kastrup Rehen, S.
Pluripotent stem cells as a model to study oxygen metabolism in neurogenesis and neurodevelopmental disorders
(2013) Archives of Biochemistry and Biophysics, 534 (1-2), pp. 3-10. Cited 4 times.

DOI: 10.1016/j.abb.2012.10.009
DOCUMENT TYPE: Review

Millan, M.J.
An epigenetic framework for neurodevelopmental disorders: From pathogenesis to potential therapy
(2013) Neuropharmacology, 68, pp. 2-82. Cited 38 times.

DOI: 10.1016/j.neuropharm.2012.11.015
DOCUMENT TYPE: Review

Petrov, Yu.P., Kukhareva, L.V., Krylova, T.A.
Effect of type I collagen and fibronectin on cell morphology of human MSCs in vitro
(2013) Tsitologiya, 55 (7), pp. 452-462. Cited 1 time.

DOCUMENT TYPE: Article

Lowenthal, J., Lipnick, S., Rao, M., Hull, S.C.
Specimen collection for induced pluripotent stem cell research: Harmonizing the approach to informed consent
(2012) Stem Cells Translational Medicine, 1 (5), pp. 409-421. Cited 17 times.

DOI: 10.5966/sctm.2012-0029
DOCUMENT TYPE: Article

---

3. Canaan A, Haviv I, Urban AE, Schulz VP, Hartman S, Zhang Z, **Palejev D**, Deisseroth AB, Lacy J, Snyder M, Gerstein M, Weissman SM (2009) EBNA1 regulates cellular gene expression by binding cellular promoters. **Proc. Natl. Acad. Sci. U. S. A.** 106:22421-22426. doi: 10.1073/pnas.0911676106. PMCID: PMC2799695. IF (2013): 9.809

Цитирания:

Li, L., Zhang, Y., Guo, B.-B., Chan, F.K., Tao, Q.

Oncogenic induction of cellular high CpG methylation by Epstein-Barr virus in malignant epithelial cells
(2014) Chinese Journal of Cancer, 33 (12), pp. 604-608. Cited 1 time.

DOI: 10.5732/cjc.014.10191
DOCUMENT TYPE: Review

Song, C., Xie, G., Wang, L., Liu, L., Tian, G., Xiang, H.
DNA-based hybridization chain reaction for an ultrasensitive cancer marker EBNA-1 electrochemical immunosensor
(2014) Biosensors and Bioelectronics, 58, pp. 68-74. Cited 3 times.

DOI: 10.1016/j.bios.2014.02.031
DOCUMENT TYPE: Article

Lu, F., Tempera, I., Lee, H.T., Dewispelaere, K., Lieberman, P.M.
EBNA1 binding and epigenetic regulation of gastrokine tumor suppressor genes in gastric carcinoma cells
(2014) Virology Journal, 11 (1), art. no. 12, . Cited 1 time.

DOI: 10.1186/1743-422X-11-12
DOCUMENT TYPE: Article

Tempera, I., Lieberman, P.M.
Epigenetic regulation of EBV persistence and oncogenesis
(2014) Seminars in Cancer Biology, 26, pp. 22-29. Cited 4 times.

DOI: 10.1016/j.semcancer.2014.01.003
DOCUMENT TYPE: Review

Niller, H.H., Szenthe, K., Minarovits, J.
Epstein-Barr virus-host cell interactions: An epigenetic dialog?
(2014) Frontiers in Genetics, 5 (OCT), art. no. 367, .

DOI: 10.3389/fgene.2014.00367
DOCUMENT TYPE: Article

Mansouri, S., Pan, Q., Blencowe, B.J., Claycomb, J.M., Frappier, L.
Epstein-Barr virus EBNA1 protein regulates viral latency through effects on let-7 microRNA and Dicer
(2014) Journal of Virology, 88 (19), pp. 11166-11177. Cited 1 time.

DOI: 10.1128/JVI.01785-14
DOCUMENT TYPE: Article

Hussain, M., Gatherer, D., Wilson, J.B.
Modelling the structure of full-length Epstein–Barr virus nuclear antigen 1
(2014) Virus Genes, 49 (3), pp. 358-372.

DOI: 10.1007/s11262-014-1101-9
DOCUMENT TYPE: Article

Bernasconi, M., Ueda, S., Krukowski, P., Bornhauser, B.C., Ladell, K., Dorner, M., Sigrist, J.A., Campidelli, C., Aslandogmus, R., Alessi, D., Berger, C., Pileri, S.A., Speck, R.F., Nadal, D.
Early gene expression changes by Epstein-Barr virus infection of B-cells indicate CDKs and survivin as therapeutic targets for post-transplant lymphoproliferative diseases
(2013) International Journal of Cancer, 133 (10), pp. 2341-2350. Cited 1 time.

DOI: 10.1002/ijc.28239
DOCUMENT TYPE: Article

Satoh, J.-I., Kawana, N., Yamamoto, Y.
Molecular network of chromatin immunoprecipitation followed by deep sequencing-based (ChIP-Seq) Epstein-Barr virus nuclear antigen 1-target cellular genes supports biological implications of Epstein-Barr virus persistence in multiple sclerosis
(2013) Clinical and Experimental Neuroimmunology, 4 (2), pp. 181-192.

DOI: 10.1111/cen3.12035
DOCUMENT TYPE: Article

Ansari, M.A., Singh, V.V., Dutta, S., Veettil, M.V., Dutta, D., Chikoti, L., Lu, J., Everly, D., Chandran, B.
Constitutive interferon-inducible protein 16-inflammasome activation during epstein-barr virus latency I, II, and III in B and epithelial cells
(2013) Journal of Virology, 87 (15), pp. 8606-8623. Cited 19 times.

DOI: 10.1128/JVI.00805-13
DOCUMENT TYPE: Article

Mughal, N., Coppotelli, G., Callegari, S., Gastaldello, S., Masucci, M.G.
Interaction of Gamma-Herpesvirus Genome Maintenance Proteins with Cellular Chromatin
(2013) PLoS ONE, 8 (5), art. no. e62783, . Cited 2 times.

DOI: 10.1371/journal.pone.0062783
DOCUMENT TYPE: Article

Coppotelli, G., Mughal, N., Callegari, S., Sompallae, R., Caja, L., Luijsterburg, M.S., Dantuma, N.P., Moustakas, A., Masucci, M.G.
The Epstein-Barr virus nuclear antigen-1 reprograms transcription by mimicry of high mobility group A proteins
(2013) Nucleic Acids Research, 41 (5), pp. 2950-2962. Cited 9 times.

DOI: 10.1093/nar/gkt032
DOCUMENT TYPE: Article

Smith, D.W., Sugden, B.
Potential cellular functions of Epstein-Barr nuclear antigen 1 (EBNA1) of Epstein-Barr virus
(2013) Viruses, 5 (1), pp. 226-240. Cited 8 times.

DOI: 10.3390/v5010226
DOCUMENT TYPE: Review

Jourdan, N., Jobart-Malfait, A., Reis, G.D., Quignon, F., Piolot, T., Klein, C., Tramier, M., Coppey-Moisan, M., Marechal, V.
Live-cell imaging reveals multiple interactions between epstein-barr virus nuclear antigen 1 and cellular chromatin during interphase and mitosis
(2012) Journal of Virology, 86 (9), pp. 5314-5329. Cited 8 times.

DOI: 10.1128/JVI.06303-11
DOCUMENT TYPE: Article

Frappier, L.
Role of EBNA1 in NPC tumourigenesis
(2012) Seminars in Cancer Biology, 22 (2), pp. 154-161. Cited 19 times.

DOI: 10.1016/j.semcancer.2011.12.002
DOCUMENT TYPE: Review

Borkosky, S.S., Whitley, C., Kopp-Schneider, A., Hausen, H., de Villiers, E.-M.
Epstein-barr virus stimulates torque teno virus replication: A possible relationship to multiple sclerosis
(2012) PLoS ONE, 7 (2), art. no. e32160, . Cited 11 times.

DOI: 10.1371/journal.pone.0032160
DOCUMENT TYPE: Article

Võsa, L., Sudakov, A., Remm, M., Ustav, M., Kurg, R.
Identification and analysis of papillomavirus E2 protein binding sites in the human genome
(2012) Journal of Virology, 86 (1), pp. 348-357. Cited 6 times.

DOI: 10.1128/JVI.05606-11
DOCUMENT TYPE: Article

Cao, J.Y., Mansouri, S., Frappier, L.
Changes in the nasopharyngeal carcinoma nuclear proteome induced by the EBNA1 protein of epstein-barr virus reveal potential roles for EBNA1 in metastasis and oxidative stress responses
(2012) Journal of Virology, 86 (1), pp. 382-394. Cited 14 times.

DOI: 10.1128/JVI.05648-11
DOCUMENT TYPE: Article

Yasuda, A., Noguchi, K., Minoshima, M., Kashiwazaki, G., Kanda, T., Katayama, K., Mitsuhashi, J., Bando, T., Sugiyama, H., Sugimoto, Y.
DNA ligand designed to antagonize EBNA1 represses Epstein-Barr virus-induced immortalization
(2011) Cancer Science, 102 (12), pp. 2221-2230. Cited 3 times.

DOI: 10.1111/j.1349-7006.2011.02098.x
DOCUMENT TYPE: Article

Sofueva, S., Osman, F., Lorenz, A., Steinacher, R., Castagnetti, S., Ledesma, J., Whitby, M.C.
Ultrafine anaphase bridges, broken DNA and illegitimate recombination induced by a replication fork barrier
(2011) Nucleic Acids Research, 39 (15), pp. 6568-6584. Cited 18 times.

DOI: 10.1093/nar/gkr340
DOCUMENT TYPE: Article

Hammerschmidt, W.
What keeps the power on in lymphomas?
(2011) Blood, 117 (6), pp. 1777-1778.

DOI: 10.1182/blood-2010-12-322222
DOCUMENT TYPE: Note

Lu, J., Murakami, M., Verma, S.C., Cai, Q., Haldar, S., Kaul, R., Wasik, M.A., Middeldorp, J., Robertson, E.S.
Epstein-Barr Virus nuclear antigen 1 (EBNA1) confers resistance to apoptosis in EBV-positive B-lymphoma cells through up-regulation of survivin
(2011) Virology, 410 (1), pp. 64-75. Cited 26 times.

DOI: 10.1016/j.virol.2010.10.029
DOCUMENT TYPE: Article

Durzyńska, J., Pacholska-Bogalska, J., Kaczmarek, M., Hanć, T., Durda, M., Skrzypczak, M., Goździcka-Józefiak, A.
Multiplex PCR for identification of herpes virus infections in adolescents
(2011) Journal of Medical Virology, 83 (2), pp. 267-271.

DOI: 10.1002/jmv.21972
DOCUMENT TYPE: Article

Epeldegui, M., Vendrame, E., Martínez-Maza, O.
HIV-associated immune dysfunction and viral infection: Role in the pathogenesis of AIDS-related lymphoma
(2010) Immunologic Research, 48 (1-3), pp. 72-83. Cited 24 times.

DOI: 10.1007/s12026-010-8168-8
DOCUMENT TYPE: Review

Lu, F., Wikramasinghe, P., Norseen, J., Tsai, K., Wang, P., Showe, L., Davuluri, R.V., Lieberman, P.M.
Genome-wide analysis of host-chromosome binding sites for Epstein-Barr Virus Nuclear Antigen 1 (EBNA1)
(2010) Virology Journal, 7, art. no. 262, . Cited 22 times.

DOI: 10.1186/1743-422X-7-262
DOCUMENT TYPE: Article

Sia, K.C., Chong, W.K., Ho, I.a.W., Yulyana, Y., Endaya, B., Huynh, H., Lam, P.Y.P.
Hybrid herpes simplex virus/Epstein-Barr virus amplicon viral vectors confer enhanced transgene expression in primary human tumors and human bone marrow-derived mesenchymal stem cells
(2010) Journal of Gene Medicine, 12 (10), pp. 848-858. Cited 8 times.

DOI: 10.1002/jgm.1506
DOCUMENT TYPE: Article

d'Hérouël, A.F., Birgersdotter, A., Werner, M.
FR-like EBNA1 binding repeats in the human genome
(2010) Virology, 405 (2), pp. 524-529. Cited 4 times.

DOI: 10.1016/j.virol.2010.06.040
DOCUMENT TYPE: Article

Owen, T.J., O'Neil, J.D., Dawson, C.W., Hu, C., Chen, X., Yao, Y., Wood, V.H.J., Mitchell, L.E., White, R.J., Young, L.S., Arrand, J.R.
Epstein-Barr virus-encoded EBNA1 enhances RNA polymerase III-dependent EBER expression through induction of EBER-associated cellular transcription factors
(2010) Molecular Cancer, 9, art. no. 241, . Cited 6 times.

DOI: 10.1186/1476-4598-9-241
DOCUMENT TYPE: Article

---

2. Pan X, Urban AE, **Palejev D,** Schulz V, Grubert F, Hu Y, Snyder M, Weissman SM (2008) A procedure for highly specific, sensitive, and unbiased whole-genome amplification. **Proc. Natl. Acad. Sci. U. S. A.** 05:15499-15504. doi: 10.1073/pnas.0808028105. PMCID: PMC2563063. IF (2013): 9.809

Цитирания:

Yang, Y., Swennenhuis, J.F., Rho, H.S., Le Gac, S., Terstappen, L.W.M.M.
Parallel single cancer cell whole genome amplification using button-valve assisted mixing in nanoliter chambers
(2014) PLoS ONE, 9 (9), art. no. e107958, p. e155.

DOI: 10.1371/journal.pone.0107958
DOCUMENT TYPE: Article

Norman, A., Riber, L., Luo, W., Li, L.L., Hansen, L.H., Sørensen, S.J.
An improved method for including upper size range plasmids in metamobilomes
(2014) PLoS ONE, 9 (8), art. no. e104405, .

DOI: 10.1371/journal.pone.0104405
DOCUMENT TYPE: Article

Motley, S.T., Picuri, J.M., Crowder, C.D., Minich, J.J., Hofstadler, S.A., Eshoo, M.W.
Improved multiple displacement amplification (iMDA) and ultraclean reagents
(2014) BMC Genomics, 15 (1), art. no. 443, .

DOI: 10.1186/1471-2164-15-443
DOCUMENT TYPE: Article

Džunková, M., Garcia-Garcerà, M., Martínez-Priego, L., D'Auria, G., Calafell, F., Moya, A.
Direct squencing from the minimal number of DNA molecules needed to fill a 454 picotiterplate
(2014) PLoS ONE, 9 (6), art. no. e97379, . Cited 1 time.

DOI: 10.1371/journal.pone.0097379
DOCUMENT TYPE: Article

Ali, M.M., Li, F., Zhang, Z., Zhang, K., Kang, D.-K., Ankrum, J.A., Le, X.C., Zhao, W.
Rolling circle amplification: A versatile tool for chemical biology, materials science and medicine
(2014) Chemical Society Reviews, 43 (10), pp. 3324-3341. Cited 22 times.

DOI: 10.1039/c3cs60439j
DOCUMENT TYPE: Review

Takahashi, H., Yamazaki, H., Akanuma, S., Kanahara, H., Saito, T., Chimuro, T., Kobayashi, T., Ohtani, T., Yamamoto, K., Sugiyama, S., Kobori, T.
Preparation of Phi29 DNA polymerase free of amplifiable DNA using ethidium monoazide, an ultraviolet-free light-emitting diode lamp and trehalose
(2014) PLoS ONE, 9 (2), art. no. e82624, .

DOI: 10.1371/journal.pone.0082624
DOCUMENT TYPE: Article

Clingenpeel, S., Clum, A., Schwientek, P., Rinke, C., Woyke, T.
Reconstructing each cell's genome within complex microbial communities - dream or reality?
(2014) Frontiers in Microbiology, 5 (DEC), art. no. 771, .

DOI: 10.3389/fmicb.2014.00771
DOCUMENT TYPE: Article

Dichosa, A.E.K., Daughton, A.R., Reitenga, K.G., Fitzsimons, M.S., Han, C.S.
Capturing and cultivating single bacterial cells in gel microdroplets to obtain near-complete genomes
(2014) Nature Protocols, 9 (3), pp. 608-621. Cited 3 times.

DOI: 10.1038/nprot.2014.034
DOCUMENT TYPE: Article

Sujayanont, P., Chininmanu, K., Tassaneetrithep, B., Tangthawornchaikul, N., Malasit, P., Suriyaphol, P.
Comparison of phi29-based whole genome amplification and whole transcriptome amplification in dengue virus
(2014) Journal of Virological Methods, 195, pp. 141-147.

DOI: 10.1016/j.jviromet.2013.10.005
DOCUMENT TYPE: Article

Tsai, I.J., Hunt, M., Holroyd, N., Huckvale, T., Berriman, M., Kikuchi, T.
Summarizing specific profiles in illumina sequencing from whole-genome amplified DNA
(2014) DNA Research, 21 (3), pp. 243-254.

DOI: 10.1093/dnares/dst054
DOCUMENT TYPE: Article

Gole, J., Gore, A., Richards, A., Chiu, Y.-J., Fung, H.-L., Bushman, D., Chiang, H.-I., Chun, J., Lo, Y.-H., Zhang, K.
Massively parallel polymerase cloning and genome sequencing of single cells using nanoliter microwells
(2013) Nature Biotechnology, 31 (12), pp. 1126-1132. Cited 20 times.

DOI: 10.1038/nbt.2720
DOCUMENT TYPE: Article

Wang, Q., Zhu, X., Feng, Y., Xue, Z., Fan, G.
Single-cell genomics: An overview
(2013) Frontiers in Biology, 8 (6), pp. 569-576.

DOI: 10.1007/s11515-013-1285-8
DOCUMENT TYPE: Review

Close, D.W., Ferrara, F., Dichosa, A.E., Kumar, S., Daughton, A.R., Daligault, H.E., Reitenga, K.G., Velappan, N., Sanchez, T.C., Iyer, S., Kiss, C., Han, C.S., Bradbury, A.R.M.
Using phage display selected antibodies to dissect microbiomes for complete de novo genome sequencing of low abundance microbes
(2013) BMC Microbiology, 13 (1), art. no. 270, .

DOI: 10.1186/1471-2180-13-270
DOCUMENT TYPE: Article

Thompson, A., Bench, S., Carter, B., Zehr, J.
Coupling FACS and genomic methods for the characterization of uncultivated symbionts
(2013) Methods in Enzymology, 531, pp. 45-60. Cited 1 time.

DOI: 10.1016/B978-0-12-407863-5.00003-4
DOCUMENT TYPE: Article

Wang, Y., Lee, O.O., Yang, J.K., Li, T.G., Qian, P.Y.
Artifactual pyrosequencing reads in multiple-displacement-amplified sediment metagenomes fromthe Red Sea
(2013) PeerJ, 2013 (1), art. no. e69, . Cited 2 times.

DOI: 10.7717/peerj.69
DOCUMENT TYPE: Article

Fitzsimons, M.S., Novotny, M., Lo, C.-C., Dichosa, A.E.K., Yee-Greenbaum, J.L., Snook, J.P., Gu, W., Chertkov, O., Davenport, K.W., McMurry, K., Reitenga, K.G., Daughton, A.R., He, J., Johnson, S.L., Gleasner, C.D., Wills, P.L., Parson-Quintana, B., Chain, P.S., Detter, J.C., Lasken, R.S., Han, C.S.
Nearly finished genomes produced using gel microdroplet culturing reveal substantial intraspecies genomic diversity within the human microbiome
(2013) Genome Research, 23 (5), pp. 878-888. Cited 15 times.

DOI: 10.1101/gr.142208.112
DOCUMENT TYPE: Article

Särkinen, T., Staats, M., Richardson, J.E., Cowan, R.S., Bakker, F.T.
How to Open the Treasure Chest? Optimising DNA Extraction from Herbarium Specimens
(2012) PLoS ONE, 7 (8), art. no. e43808, . Cited 15 times.

DOI: 10.1371/journal.pone.0043808
DOCUMENT TYPE: Article

Kamke, J., Bayer, K., Woyke, T., Hentschel, U.
Exploring symbioses by single-cell genomics
(2012) Biological Bulletin, 223 (1), pp. 30-43. Cited 7 times.

DOCUMENT TYPE: Review

Parks, M., Cronn, R., Liston, A.
Separating the wheat from the chaff: Mitigating the effects of noise in a plastome phylogenomic data set from Pinus L. (Pinaceae)
(2012) BMC Evolutionary Biology, 12 (1), art. no. 100, . Cited 20 times.

DOI: 10.1186/1471-2148-12-100
DOCUMENT TYPE: Article

Yilmaz, S., Singh, A.K.
Single cell genome sequencing
(2012) Current Opinion in Biotechnology, 23 (3), pp. 437-443. Cited 17 times.

DOI: 10.1016/j.copbio.2011.11.018
DOCUMENT TYPE: Review

Dichosa, A.E.K., Fitzsimons, M.S., Lo, C.-C., Weston, L.L., Preteska, L.G., Snook, J.P., Zhang, X., Gu, W., McMurry, K., Green, L.D., Chain, P.S., Detter, J.C., Han, C.S.
Artificial polyploidy improves bacterial single cell genome recovery
(2012) PLoS ONE, 7 (5), art. no. e37387, . Cited 11 times.

DOI: 10.1371/journal.pone.0037387
DOCUMENT TYPE: Article

Tate, C.M., Nuñez, A.N., Goldstein, C.A., Gomes, I., Robertson, J.M., Kavlick, M.F., Budowle, B.
Evaluation of circular DNA substrates for whole genome amplification prior to forensic analysis
(2012) Forensic Science International: Genetics, 6 (2), pp. 185-190. Cited 7 times.

DOI: 10.1016/j.fsigen.2011.04.011
DOCUMENT TYPE: Article

Malecki, M., Szybalski, W.
Isolation of single, intact chromosomes from single, selected ovarian cancer cells for in situ hybridization and sequencing
(2012) Gene, 493 (1), pp. 132-139. Cited 2 times.

DOI: 10.1016/j.gene.2011.11.044
DOCUMENT TYPE: Article

Hansen, A.K., Vorburger, C., Moran, N.A.
Genomic basis of endosymbiont-conferred protection against an insect parasitoid
(2012) Genome Research, 22 (1), pp. 106-114. Cited 16 times.

DOI: 10.1101/gr.125351.111
DOCUMENT TYPE: Article

LaTuga, M.S., Ellis, J.C., Cotton, C.M., Goldberg, R.N., Wynn, J.L., Jackson, R.B., Seed, P.C.
Beyond bacteria: A study of the enteric microbial consortium in extremely low birth weight infants
(2011) PLoS ONE, 6 (12), art. no. e27858, . Cited 30 times.

DOI: 10.1371/journal.pone.0027858
DOCUMENT TYPE: Article

Kalisky, T., Blainey, P., Quake, S.R.
Genomic analysis at the single-cell level
(2011) Annual Review of Genetics, 45, pp. 431-445. Cited 54 times.

DOI: 10.1146/annurev-genet-102209-163607
DOCUMENT TYPE: Article

Burke, G.R., Moran, N.A.
Massive genomic decay in Serratia symbiotica, a recently evolved symbiont of aphids
(2011) Genome Biology and Evolution, 3 (1), pp. 195-208. Cited 51 times.

DOI: 10.1093/gbe/evr002
DOCUMENT TYPE: Article

Akasaka, E., Ozawa, A., Mori, H., Mizobe, Y., Yoshida, M., Miyoshi, K., Sato, M.
Whole-genome amplification-based GenomiPhi for multiple genomic analysis of individual early porcine embryos
(2011) Theriogenology, 75 (8), pp. 1543-1549. Cited 2 times.

DOI: 10.1016/j.theriogenology.2010.12.018
DOCUMENT TYPE: Article

Blainey, P.C., Quake, S.R.
Digital MDA for enumeration of total nucleic acid contamination
(2011) Nucleic Acids Research, 39 (4), . Cited 38 times.

DOI: 10.1093/nar/gkq1074

DOCUMENT TYPE: Article

Kim, J., Easley, C.J.
Isothermal DNA amplification in bioanalysis: Strategies and applications
(2011) Bioanalysis, 3 (2), pp. 227-239. Cited 43 times.

DOI: 10.4155/bio.10.172
DOCUMENT TYPE: Review

van Oorschot, R.A.H., Ballantyne, K.N., Mitchell, R.J.
Forensic trace DNA: A review
(2010) Investigative Genetics, 1 (1), art. no. 14, . Cited 44 times.

DOI: 10.1186/2041-2223-1-14
DOCUMENT TYPE: Review

Durstewitz, G., Polley, A., Plieske, J., Luerssen, H., Graner, E.M., Wieseke, R., Ganal, M.W.
SNP discovery by amplicon sequencing and multiplex SNP genotyping in the allopolyploid species Brassica napus
(2010) Genome, 53 (11), pp. 948-956. Cited 24 times.

DOI: 10.1139/G10-079
DOCUMENT TYPE: Conference Paper

Cuàys, E., Olano-Martn, E., Khymenets, O., Hernández, L., Jofre-Monseny, L., Grandoso, L., Tejedor, D., Martnez, A., Farré, M., De La Torre, R.
Errors and reproducibility of DNA array-based detection of allelic variants in ADME genes: PHARMAchip™
(2010) Pharmacogenomics, 11 (2), pp. 257-266. Cited 14 times.

DOI: 10.2217/pgs.09.165
DOCUMENT TYPE: Article

Sánchez, C.C., Smith, T.P.L., Wiedmann, R.T., Vallejo, R.L., Salem, M., Yao, J., Rexroad III, C.E.
Single nucleotide polymorphism discovery in rainbow trout by deep sequencing of a reduced representation library
(2009) BMC Genomics, 10, art. no. 559, . Cited 79 times.

DOI: 10.1186/1471-2164-10-559
DOCUMENT TYPE: Article

Rodrigue, S., Malmstrom, R.R., Berlin, A.M., Birren, B.W., Henn, M.R., Chisholm, S.W.
Whole genome amplification and de novo assembly of single bacterial cells
(2009) PLoS ONE, 4 (9), art. no. e6864, . Cited 101 times.

DOI: 10.1371/journal.pone.0006864
DOCUMENT TYPE: Article

Zhulin, I.B.
It is computation time for bacteriology!
(2009) Journal of Bacteriology, 91 (1), pp. 20-22. Cited 4 times.

DOI: 10.1128/JB.01491-08
DOCUMENT TYPE: Note

1. Korbel JO, Urban AE, Affourtit JP, Godwin B, Grubert F, Simons JF, Kim PM, **Palejev D**, Carriero NJ, Du L, Taillon BE, Chen Z, Tanzer A, Saunders AC, Chi J, Yang F, Carter NP, Hurles ME, Weissman SM, Harkins TT, Gerstein MB, Egholm M, Snyder M (2007) Paired-end mapping reveals extensive structural variation in the human genome. **Science** 318:420-426. doi: 10.1126/science.1149504. PMCID: PMC2674581. IF (2013): 31.48

Цитирания:

Schlick-Steiner, B.C., Arthofer, W., Moder, K., Steiner, F.M.
Recent insertion/deletion (reINDEL) mutations: Increasing awareness to boost molecular-based research in ecology and evolution
(2015) Ecology and Evolution, 5 (1), pp. 24-35.

DOI: 10.1002/ece3.1330
DOCUMENT TYPE: Article

Gelernter, J.
Genetics of complex traits in psychiatry
(2015) Biological Psychiatry, 77 (1), pp. 36-42. Cited 1 time.

DOI: 10.1016/j.biopsych.2014.08.005
DOCUMENT TYPE: Review

Tang, J., Fang, F., Miller, D.F., Pilrose, J.M., Matei, D., Huang, T.H.-M., Nephew, K.P.
Global DNA methylation profiling technologies and the ovarian cancer methylome
(2015) Methods in Molecular Biology, 1238, pp. 653-675.

DOI: 10.1007/978-1-4939-1804-1_34
DOCUMENT TYPE: Article

Hancarova, M., Puchmajerova, A., Drabova, J., Karaskova, E., Vlckova, M., Sedlacek, Z.
Deletions of 9q21.3 including NTRK2 are associated with severe phenotype
(2015) American Journal of Medical Genetics, Part A, 167 (1), pp. 264-267.

DOI: 10.1002/ajmg.a.36797
DOCUMENT TYPE: Letter

Sante, T., Vergult, S., Volders, P.-J., Kloosterman, W.P., Trooskens, G., De Preter, K., Dheedene, A., Speleman, F., De Meyer, T., Menten, B.
ViVar: A comprehensive platform for the analysis and visualization of structural genomic variation
(2014) PLoS ONE, 9 (12), art. no. e113800, .

DOI: 10.1371/journal.pone.0113800
DOCUMENT TYPE: Article

Snow, A.N., Stence, A.A., Pruessner, J.A., Bossler, A.D., Ma, D.

A simple and cost-effective method of DNA extraction from small formalin-fixed paraffin-embedded tissue for molecular oncologic testing
(2014) BMC Clinical Pathology, 14 (1), art. no. 30, .

DOI: 10.1186/1472-6890-14-30
DOCUMENT TYPE: Article

Hommelsheim, C.M., Frantzeskakis, L., Huang, M., Ülker, B.
PCR amplification of repetitive DNA: A limitation to genome editing technologies and many other applications
(2014) Scientific Reports, 4, art. no. 5052, .

DOI: 10.1038/srep05052
DOCUMENT TYPE: Article

Ordulu, Z., Wong, K.E., Currall, B.B., Ivanov, A.R., Pereira, S., Althari, S., Gusella, J.F., Talkowski, M.E., Morton, C.C.
Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature
(2014) American Journal of Human Genetics, 94 (5), pp. 695-709. Cited 1 time.

DOI: 10.1016/j.ajhg.2014.03.020
DOCUMENT TYPE: Article

Looso, M.
Opening the genetic toolbox of niche model organisms with high throughput techniques: Novel proteins in regeneration as a case study
(2014) BioEssays, 36 (4), pp. 407-418. Cited 3 times.

DOI: 10.1002/bies.201300093
DOCUMENT TYPE: Article

Blake, J., Riddell, A., Theiss, S., Gonzalez, A.P., Haase, B., Jauch, A., Janssen, J.W.G., Ibberson, D., Pavlinic, D., Moog, U., Benes, V., Runz, H.
Sequencing of a patient with balanced chromosome abnormalities and neurodevelopmental disease identifies disruption of multiple high risk loci by structural variation
(2014) PLoS ONE, 9 (3), art. no. e90894, . Cited 1 time.

DOI: 10.1371/journal.pone.0090894
DOCUMENT TYPE: Article

Utami, K.H., Hillmer, A.M., Aksoy, I., Chew, E.G.Y., Teo, A.S.M., Zhang, Z., Lee, C.W.H., Chen, P.J., Seng, C.C., Ariyaratne, P.N., Rouam, S.L., Soo, L.S., Yousoof, S., Prokudin, I., Peters, G., Collins, F., Wilson, M., Kakakios, A., Haddad, G., Menuet, A., Perche, O., Tay, S.K.H., Sung, K.W.K., Ruan, X., Ruan, Y., Liu, E.T., Briault, S., Jamieson, R.V., Davila, S., Cacheux, V.
Detection of chromosomal breakpoints in patients with developmental delay and speech disorders
(2014) PLoS ONE, 9 (3), art. no. e90852, . Cited 1 time.

DOI: 10.1371/journal.pone.0090852
DOCUMENT TYPE: Article

Mosen-Ansorena, D., Telleria, N., Veganzones, S., la Orden, V.D., Maestro, M.L., Aransay, A.M.
SeqCNA: An R package for DNA copy number analysis in cancer using high-throughput sequencing
(2014) BMC Genomics, 15 (1), art. no. 178, .

DOI: 10.1186/1471-2164-15-178
DOCUMENT TYPE: Article

Yavaş, G., Koyutürk, M., Gould, M.P., McMahon, S., LaFramboise, T.
DB2: A probabilistic approach for accurate detection of tandem duplication breakpoints using paired-end reads
(2014) BMC Genomics, 15 (1), art. no. 175, .

DOI: 10.1186/1471-2164-15-175
DOCUMENT TYPE: Article

Duan, J., Deng, H.-W., Wang, Y.-P.
Common copy number variation detection from multiple sequenced samples
(2014) IEEE Transactions on Biomedical Engineering, 61 (3), art. no. 6675802, pp. 928-937.

DOI: 10.1109/TBME.2013.2292588
DOCUMENT TYPE: Article

Yang, Z.-H., Zheng, R., Gao, Y., Zhang, Q., Zhang, H.
Abnormal gene expression and gene fusion in lung adenocarcinoma with high-throughput RNA sequencing
(2014) Cancer Gene Therapy, 21 (2), pp. 74-82. Cited 1 time.

DOI: 10.1038/cgt.2013.86
DOCUMENT TYPE: Article

Chen, K., Chen, L., Fan, X., Wallis, J., Ding, L., Weinstock, G.
TIGRA: A targeted iterative graph routing assembler for breakpoint assembly
(2014) Genome Research, 24 (2), pp. 310-317. Cited 6 times.

DOI: 10.1101/gr.162883.113
DOCUMENT TYPE: Article

Flagel, L.E., Willis, J.H., Vision, T.J.
The standing pool of genomic structural variation in a natural population of Mimulus guttatus
(2014) Genome Biology and Evolution, 6 (1), pp. 53-64.

DOI: 10.1093/gbe/evt199
DOCUMENT TYPE: Article

Ma, S., Wang, X., Liu, Y., Gao, J., Zhang, S., Shi, R., Chang, J., Zhao, P., Xia, Q.
Multiplex genomic structure variation mediated by TALEN and ssODN
(2014) BMC Genomics, 15 (1), art. no. 41, . Cited 5 times.

DOI: 10.1186/1471-2164-15-41
DOCUMENT TYPE: Article

Li, W., Freudenberg, J., Miramontes, P.
Diminishing return for increased Mappability with longer sequencing reads: Implications of the k-mer distributions in the human genome
(2014) BMC Bioinformatics, 15 (1), art. no. 2, . Cited 5 times.

DOI: 10.1186/1471-2105-15-2
DOCUMENT TYPE: Article

Bolon, Y.-T., Stec, A.O., Michno, J.-M., Roessler, J., Bhaskar, P.B., Ries, L., Dobbels, A.A., Campbell, B.W., Young, N.P., Anderson, J.E., Grant, D.M., Orf, J.H., Naeve, S.L., Muehlbauer, G.J., Vance, C.P., Stupar, R.M.

Genome resilience and prevalence of segmental duplications following fast neutron irradiation of soybean
(2014) Genetics, 198 (3), pp. 967-981. Cited 1 time.

DOI: 10.1534/genetics.114.170340/-/DC1
DOCUMENT TYPE: Article

Vergult, S., Van Binsbergen, E., Sante, T., Nowak, S., Vanakker, O., Claes, K., Poppe, B., Van Der Aa, N., Van Roosmalen, M.J., Duran, K., Tavakoli-Yaraki, M., Swinkels, M., Van Den Boogaard, M.-J., Van Haelst, M., Roelens, F., Speleman, F., Cuppen, E., Mortier, G., Kloosterman, W.P., Menten, B.
Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations
(2014) European Journal of Human Genetics, 22 (5), pp. 652-659. Cited 2 times.

DOI: 10.1038/ejhg.2013.220
DOCUMENT TYPE: Article

Fernandez-Banet, J., Lee, N.P., Chan, K.T., Gao, H., Liu, X., Sung, W.-K., Tan, W., Fan, S.T., Poon, R.T., Li, S., Ching, K., Rejto, P.A., Mao, M., Kan, Z.
Decoding complex patterns of genomic rearrangement in hepatocellular carcinoma
(2014) Genomics, 103 (2-3), pp. 189-203.

DOI: 10.1016/j.ygeno.2014.01.003
DOCUMENT TYPE: Article

Hanscom, C., Talkowski, M.
Design of large-insert jumping libraries for structural variant detection using Illumina sequencing
(2014) Current Protocols in Human Genetics, (SUPPL.80), art. no. 7.22, . Cited 1 time.

DOI: 10.1002/0471142905.hg0722s80
DOCUMENT TYPE: Article

Martínez-Fundichely, A., Casillas, S., Egea, R., Ràmia, M., Barbadilla, A., Pantano, L., Puig, M., Cáceres, M.
InvFEST, a database integrating information of polymorphic inversions in the human genome
(2014) Nucleic Acids Research, 42 (D1), pp. D1027-D1032. Cited 4 times.

DOI: 10.1093/nar/gkt1122
DOCUMENT TYPE: Article

Chain, F.J.J., Feulner, P.G.D., Panchal, M., Eizaguirre, C., Samonte, I.E., Kalbe, M., Lenz, T.L., Stoll, M., Bornberg-Bauer, E., Milinski, M., Reusch, T.B.H.
Extensive Copy-Number Variation of Young Genes across Stickleback Populations
(2014) PLoS Genetics, 10 (12), 18 p. Cited 1 time.

DOI: 10.1371/journal.pgen.1004830
DOCUMENT TYPE: Article

Bickhart, D.M., Liu, G.E.
The challenges and importance of structural variation detection in livestock
(2014) Frontiers in Genetics, 5 (FEB), art. no. Article 37, . Cited 3 times.

DOI: 10.3389/fgene.2014.00037
DOCUMENT TYPE: Review

Steinberg, K.M., Schneider, V.A., Graves-Lindsay, T.A., Fulton, R.S., Agarwala, R., Huddleston, J., Shiryev, S.A., Morgulis, A., Surti, U., Warren, W.C., Church, D.M., Eichler, E.E., Wilson, R.K.
Single haplotype assembly of the human genome from a hydatidiform mole
(2014) Genome Research, 24 (12), pp. 2066-2076. Cited 1 time.

DOI: 10.1101/gr.180893.114
DOCUMENT TYPE: Article

Gao, J., Guan, R., Qi, F.
Strategies for improving accuracy of structural variation prediction using read pairs
(2014) Proceedings - 2013 International Conference on Information Science and Cloud Computing Companion, ISCC-C 2013, art. no. 6973636, pp. 463-468.

DOI: 10.1109/ISCC-C.2013.127
DOCUMENT TYPE: Conference Paper

Baik, J.Y., Lee, K.H.
Toward product attribute control: Developments from genome sequencing
(2014) Current Opinion in Biotechnology, 30, pp. 40-44. Cited 2 times.

DOI: 10.1016/j.copbio.2014.05.001
DOCUMENT TYPE: Review

Nuttle, X., Itsara, A., Shendure, J., Eichler, E.E.
Resolving genomic disorder-associated breakpoints within segmental DNA duplications using massively parallel sequencing
(2014) Nature Protocols, 9 (6), pp. 1496-1513. Cited 1 time.

DOI: 10.1038/nprot.2014.096
DOCUMENT TYPE: Article

Schwarz, R.F., Trinh, A., Sipos, B., Brenton, J.D., Goldman, N., Markowetz, F.
Phylogenetic Quantification of Intra-tumour Heterogeneity
(2014) PLoS Computational Biology, 10 (4), art. no. e1003535, . Cited 4 times.

DOI: 10.1371/journal.pcbi.1003535
DOCUMENT TYPE: Article

Zhang, L.-M., Luo, H., Liu, Z.-Q., Zhao, Y., Luo, J.-C., Hao, D.-Y., Jing, H.-C.
Genome-wide patterns of large-size presence/absence variants in sorghum
(2014) Journal of Integrative Plant Biology, 56 (1), pp. 24-37. Cited 2 times.

DOI: 10.1111/jipb.12121
DOCUMENT TYPE: Article

Liu, T., Xie, L., Ye, J., He, X.
Family-based analysis identified CD2 as a susceptibility gene for primary open angle glaucoma in Chinese Han population
(2014) Journal of Cellular and Molecular Medicine, 18 (4), pp. 600-609.

DOI: 10.1111/jcmm.12201
DOCUMENT TYPE: Article

Wilson-Sánchez, D., Rubio-Díaz, S., Muñoz-Viana, R., Pérez-Pérez, J.M., Jover-Gil, S., Ponce, M.R., Micol, J.L.
Leaf phenomics: A systematic reverse genetic screen for Arabidopsis leaf mutants

(2014) Plant Journal, 79 (5), pp. 878-891. Cited 1 time.

DOI: 10.1111/tpj.12595
DOCUMENT TYPE: Article

Ritz, A., Bashir, A., Sindi, S., Hsu, D., Hajirasouliha, I., Raphael, B.J.
Characterization of Structural variants with single molecule and hybrid sequencing approaches
(2014) Bioinformatics, 30 (24), pp. 3458-3466.

DOI: 10.1093/bioinformatics/btu714
DOCUMENT TYPE: Article

Muñoz-Minjares, J., Cabal-Aragón, J., Shmaliy, Y.S.
Confidence masks for genome DNA copy number variations in applications to HR-CGH array measurements
(2014) Biomedical Signal Processing and Control, 13 (1), pp. 337-344.

DOI: 10.1016/j.bspc.2014.06.006
DOCUMENT TYPE: Article

Rogers, R.L., Cridland, J.M., Shao, L., Hu, T.T., Andolfatto, P., Thornton, K.R.
Landscape of standing variation for tandem duplications in drosophila yakuba and drosophila simulans
(2014) Molecular Biology and Evolution, 31 (7), pp. 1750-1766.

DOI: 10.1093/molbev/msu124
DOCUMENT TYPE: Article

Li, J., Kannan, M., Trivett, A.L., Liao, H., Wu, X., Akagi, K., Symer, D.E.
An antisense promoter in mouse L1 retrotransposon open reading frame-1 initiates expression of diverse fusion transcripts and limits retrotransposition
(2014) Nucleic Acids Research, 42 (7), pp. 4546-4562. Cited 3 times.

DOI: 10.1093/nar/gku091
DOCUMENT TYPE: Article

Ma, J., Xiong, M., You, M., Lozano, G., Amos, C.I.
Genome-wide association tests of inversions with application to psoriasis
(2014) Human Genetics, 133 (8), pp. 967-974.

DOI: 10.1007/s00439-014-1437-1
DOCUMENT TYPE: Article

Kitchen, R.R., Rozowsky, J.S., Gerstein, M.B., Nairn, A.C.
Decoding neuroproteomics: Integrating the genome, translatome and functional anatomy
(2014) Nature Neuroscience, 17 (11), pp. 1491-1499.

DOI: 10.1038/nn.3829
DOCUMENT TYPE: Review

Keane, T.M., Wong, K., Adams, D.J., Flint, J., Reymond, A., Yalcin, B.
Identification of structural variation in mouse genomes
(2014) Frontiers in Genetics, 5 (JUL), art. no. Article 192, . Cited 1 time.

DOI: 10.3389/fgene.2014.00192
DOCUMENT TYPE: Review

Aguado, C., Gayà-Vidal, M., Villatoro, S., Oliva, M., Izquierdo, D., Giner-Delgado, C., Montalvo, V., García-González, J., Martínez-Fundichely, A., Capilla, L., Ruiz-Herrera, A., Estivill, X., Puig, M., Cáceres, M.
Validation and Genotyping of Multiple Human Polymorphic Inversions Mediated by Inverted Repeats Reveals a High Degree of Recurrence
(2014) PLoS Genetics, 10 (3), art. no. e1004208, .

DOI: 10.1371/journal.pgen.1004208
DOCUMENT TYPE: Article

Talkowski, M.E., Minikel, E.V., Gusella, J.F.
Autism spectrum disorder genetics: Diverse genes with diverse clinical outcomes
(2014) Harvard Review of Psychiatry, 22 (2), pp. 65-75. Cited 1 time.

DOI: 10.1097/HRP.0000000000000002
DOCUMENT TYPE: Review

Adam-Blondon, A.-F.
Grapevine genome update and beyond
(2014) Acta Horticulturae, 1046, pp. 311-318. Cited 1 time.

DOCUMENT TYPE: Article

Gillespie, R.L., O'Sullivan, J., Ashworth, J., Bhaskar, S., Williams, S., Biswas, S., Kehdi, E., Ramsden, S.C., Clayton-Smith, J., Black, G.C., Lloyd, I.C.
Personalized diagnosis and management of congenital cataract by next-generation sequencing
(2014) Ophthalmology, 121 (11), pp. 2124-2137.e2. Cited 1 time.

DOI: 10.1016/j.ophtha.2014.06.006
DOCUMENT TYPE: Article

Flynn, T.J., Phipps-Green, A., Hollis-Moffatt, J.E., Merriman, M.E., Topless, R., Montgomery, G., Chapman, B., Stamp, L.K., Dalbeth, N., Merriman, T.R.
Association analysis of the SLC22A11 (organic anion transporter 4) and SLC22A12 (urate transporter 1) urate transporter locus with gout in New Zealand case-control sample sets reveals multiple ancestral-specific effects
(2013) Arthritis Research and Therapy, 15 (6), art. no. R220, . Cited 1 time.

DOI: 10.1186/ar4417
DOCUMENT TYPE: Article

Zhang, C.-Z., Leibowitz, M.L., Pellman, D.
Chromothripsis and beyond: Rapid genome evolution from complex chromosomal rearrangements
(2013) Genes and Development, 27 (23), pp. 2513-2530. Cited 10 times.

DOI: 10.1101/gad.229559.113
DOCUMENT TYPE: Review

Kasaian, K., Li, Y.Y., Jones, S.J.M.
Bioinformatics for Cancer Genomics
(2013) Cancer Genomics: From Bench to Personalized Medicine, pp. 133-152.

DOI: 10.1016/B978-0-12-396967-5.00009-8
DOCUMENT TYPE: Book Chapter

Gao, J., Guan, R., Qi, F.
Methods for detecting genome structural variation based on PEM
(2013) Beijing Jiaotong Daxue Xuebao/Journal of Beijing Jiaotong University, 37 (6), pp. 8-12.

DOCUMENT TYPE: Article

Duan, J., Wan, M., Deng, H.-W., Wang, Y.-P.
Modeling exome sequencing data with generalized Gaussian distribution with application to copy number variation detection
(2013) Proceedings - 2013 IEEE International Conference on Bioinformatics and Biomedicine, IEEE BIBM 2013, art. no. 6732619, pp. 1-7.

DOI: 10.1109/BIBM.2013.6732619
DOCUMENT TYPE: Conference Paper

Livnat, A.
Interaction-based evolution: How natural selection and nonrandom mutation work together
(2013) Biology Direct, 8 (1), art. no. 24, . Cited 2 times.

DOI: 10.1186/1745-6150-8-24
DOCUMENT TYPE: Article

Plass, C., Pfister, S.M., Lindroth, A.M., Bogatyrova, O., Claus, R., Lichter, P.
Mutations in regulators of the epigenome and their connections to global chromatin patterns in cancer
(2013) Nature Reviews Genetics, 14 (11), pp. 765-780. Cited 40 times.

DOI: 10.1038/nrg3554
DOCUMENT TYPE: Review

Liu, B., Morrison, C.D., Johnson, C.S., Trump, D.L., Qin, M., Conroy, J.C., Wang, J., Liu, S.
Computational methods for detecting copy number variations in cancer genome using next generation sequencing: Principles and challenges
(2013) Oncotarget, 4 (11), pp. 1868-1881. Cited 3 times.

DOCUMENT TYPE: Article

Haraksingh, R.R., Snyder, M.P.
Impacts of variation in the human genome on gene regulation
(2013) Journal of Molecular Biology, 425 (21), pp. 3970-3977. Cited 10 times.

DOI: 10.1016/j.jmb.2013.07.015
DOCUMENT TYPE: Review

Ratnapriya, R., Swaroop, A.
Genetic architecture of retinal and macular degenerative diseases: The promise and challenges of next-generation sequencing
(2013) Genome Medicine, 5 (9), art. no. 84, . Cited 1 time.

DOI: 10.1186/gm488
DOCUMENT TYPE: Review

Sankaranarayanan, K., Taleei, R., Rahmanian, S., Nikjoo, H.

Ionizing radiation and genetic risks. XVII. Formation mechanisms underlying naturally occurring DNA deletions in the human genome and their potential relevance for bridging the gap between induced DNA double-strand breaks and deletions in irradiated germ cells
(2013) Mutation Research - Reviews in Mutation Research, 753 (2), pp. 114-130. Cited 5 times.

DOI: 10.1016/j.mrrev.2013.07.003
DOCUMENT TYPE: Review

Dong, Z.C., Chen, Y.
Transcriptomics: Advances and approaches
(2013) Science China Life Sciences, 56 (10), pp. 960-967.

DOI: 10.1007/s11427-013-4557-2
DOCUMENT TYPE: Review

Zhao, M., Wang, Q., Wang, Q., Jia, P., Zhao, Z.
Computational tools for copy number variation (CNV) detection using next-generation sequencing data: Features and perspectives
(2013) BMC Bioinformatics, 14 (SUPPL11), art. no. S1, . Cited 7 times.

DOI: 10.1186/1471-2105-14-S11-S1
DOCUMENT TYPE: Article

Yegnasubramanian, S.
Explanatory chapter: Next generation sequencing
(2013) Methods in Enzymology, 529, pp. 201-208.

DOI: 10.1016/B978-0-12-418687-3.00016-1
DOCUMENT TYPE: Article

Valsesia, A., Macé, A., Jacquemont, S., Beckmann, J.S., Kutalik, Z.
The growing importance of CNVs: New insights for detection and clinical interpretation
(2013) Frontiers in Genetics, 4 (MAY), art. no. Article 92, . Cited 6 times.

DOI: 10.3389/fgene.2013.00092
DOCUMENT TYPE: Review

Lehrach, H.
DNA sequencing methods in human genetics and disease research
(2013) F1000Prime Reports, 5, art. no. 34, .

DOI: 10.12703/P5-34
DOCUMENT TYPE: Article

Morey, M., Fernández-Marmiesse, A., Castiñeiras, D., Fraga, J.M., Couce, M.L., Cocho, J.A.
A glimpse into past, present, and future DNA sequencing
(2013) Molecular Genetics and Metabolism, 110 (1-2), pp. 3-24. Cited 11 times.

DOI: 10.1016/j.ymgme.2013.04.024
DOCUMENT TYPE: Review

Sakai, R., Moisse, M., Reumers, J., Aerts, J.
Pipit: Visualizing functional impacts of structural variations
(2013) Bioinformatics, 29 (17), pp. 2206-2207.

DOI: 10.1093/bioinformatics/btt367
DOCUMENT TYPE: Article

Geurts Van Kessel, A., Venkatachalam, R., Kuiper, R.P.
Colorectal Cancer
(2013) Genomic and Personalized Medicine, 2, pp. 722-732.

DOI: 10.1016/B978-0-12-382227-7.00062-8
DOCUMENT TYPE: Book Chapter

Lee, C.
Structural Genomic Variation in the Human Genome
(2013) Genomic and Personalized Medicine, 1, pp. 123-132.

DOI: 10.1016/B978-0-12-382227-7.00010-0
DOCUMENT TYPE: Book Chapter

Guffanti, G., Torri, F., Rasmussen, J., Clark, A.P., Lakatos, A., Turner, J.A., Fallon, J.H., Saykin, A.J., Weiner, M., Vawter, M.P., Knowles, J.A., Potkin, S.G., Macciardi, F.
Increased CNV-Region deletions in mild cognitive impairment (MCI) and Alzheimer's disease (AD) subjects in the ADNI sample
(2013) Genomics, 102 (2), pp. 112-122. Cited 1 time.

DOI: 10.1016/j.ygeno.2013.04.004
DOCUMENT TYPE: Review

Chen, G., Wang, C., Shi, L., Tong, W., Qu, X., Chen, J., Yang, J., Shi, C., Chen, L., Zhou, P., Lu, B., Shi, T.
Comprehensively identifying and characterizing the missing gene sequences in human reference genome with integrated analytic approaches
(2013) Human Genetics, 132 (8), pp. 899-911. Cited 1 time.

DOI: 10.1007/s00439-013-1300-9
DOCUMENT TYPE: Article

Guo, X., Brenner, M., Zhang, X., Laragione, T., Tai, S., Li, Y., Bu, J., Yin, Y., Shah, A.A., Kwan, K., Li, Y., Jun, W., Gulko, P.S.
Whole-genome sequences of DA and F344 rats with different susceptibilities to arthritis, autoimmunity, inflammation and cancer
(2013) Genetics, 194 (4), pp. 1017-1028. Cited 2 times.

DOI: 10.1534/genetics.113.153049
DOCUMENT TYPE: Article

Wellensiek, B.P., Larsen, A.C., Stephens, B., Kukurba, K., Waern, K., Briones, N., Liu, L., Snyder, M., Jacobs, B.L., Kumar, S., Chaput, J.C.
Genome-wide profiling of human cap-independent translation-enhancing elements
(2013) Nature Methods, 10 (8), pp. 747-750. Cited 1 time.

DOI: 10.1038/nmeth.2522
DOCUMENT TYPE: Article

Paudel, Y., Madsen, O., Megens, H.-J., Frantz, L.A.F., Bosse, M., Bastiaansen, J.W.M., Crooijmans, R.P.M.A., Groenen, M.A.M.

Evolutionary dynamics of copy number variation in pig genomes in the context of adaptation and domestication
(2013) BMC Genomics, 14 (1), art. no. 449, . Cited 11 times.

DOI: 10.1186/1471-2164-14-449
DOCUMENT TYPE: Article

El-Sayed Moustafa, J.S., Froguel, P.
From obesity genetics to the future of personalized obesity therapy
(2013) Nature Reviews Endocrinology, 9 (7), pp. 402-413. Cited 17 times.

DOI: 10.1038/nrendo.2013.57
DOCUMENT TYPE: Review

Kim, S., Medvedev, P., Paton, T.A., Bafna, V.
Reprever: Resolving low-copy duplicated sequences using template driven assembly
(2013) Nucleic Acids Research, 41 (12), . Cited 2 times.

DOI: 10.1093/nar/gkt339
DOCUMENT TYPE: Article

Zhou, W., Zhang, F., Chen, X., Shen, Y., Lupski, J.R., Jin, L.
Increased genome instability in human DNA segments with self-chains: Homology-induced structural variations via replicative mechanisms
(2013) Human Molecular Genetics, 22 (13), pp. 2642-2651. Cited 2 times.

DOI: 10.1093/hmg/ddt113
DOCUMENT TYPE: Article

Brunham, L.R., Hayden, M.R.
Hunting human disease genes: Lessons from the past, challenges for the future
(2013) Human Genetics, 132 (6), pp. 603-617. Cited 4 times.

DOI: 10.1007/s00439-013-1286-3
DOCUMENT TYPE: Review

Mardis, E.R.
Next-generation sequencing platforms
(2013) Annual Review of Analytical Chemistry, 6, pp. 287-303. Cited 49 times.

DOI: 10.1146/annurev-anchem-062012-092628
DOCUMENT TYPE: Article

Escaramís, G., Tornador, C., Bassaganyas, L., Rabionet, R., Tubio, J.M.C., Martínez-Fundichely, A., Cáceres, M., Gut, M., Ossowski, S., Estivill, X.
PeSV-Fisher: Identification of Somatic and Non-Somatic Structural Variants Using Next Generation Sequencing Data
(2013) PLoS ONE, 8 (5), art. no. e63377, . Cited 3 times.

DOI: 10.1371/journal.pone.0063377
DOCUMENT TYPE: Article

Ezawa, K., Landan, G., Graur, D.
Detecting negative selection on recurrent mutations using gene genealogy
(2013) BMC Genetics, 14, art. no. 37, .

DOI: 10.1186/1471-2156-14-37
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.
CNV-TV: A robust method to discover copy number variation from short sequencing reads
(2013) BMC Bioinformatics, 14 (1), art. no. 150, .

DOI: 10.1186/1471-2105-14-150
DOCUMENT TYPE: Article

Mwenifumbo, J.C., Marra, M.A.
Cancer genome-sequencing study design
(2013) Nature Reviews Genetics, 14 (5), pp. 321-332. Cited 19 times.

DOI: 10.1038/nrg3445
DOCUMENT TYPE: Review

Lucas Lledó, J.I., Cáceres, M.
On the Power and the Systematic Biases of the Detection of Chromosomal Inversions by Paired-End Genome Sequencing
(2013) PLoS ONE, 8 (4), art. no. e61292, . Cited 3 times.

DOI: 10.1371/journal.pone.0061292
DOCUMENT TYPE: Article

Bassaganyas, L., Riveira-Muñoz, E., García-Aragonés, M., González, J.R., Cáceres, M., Armengol, L., Estivill, X.
Worldwide population distribution of the common LCE3C-LCE3B deletion associated with psoriasis and other autoimmune disorders
(2013) BMC Genomics, 14 (1), art. no. 261, . Cited 2 times.

DOI: 10.1186/1471-2164-14-261
DOCUMENT TYPE: Article

Gahan, P.B.
Circulating nucleic acids in plasma and serum: Applications in diagnostic techniques for noninvasive prenatal diagnosis
(2013) International Journal of Women's Health, 5 (1), pp. 177-186. Cited 2 times.

DOI: 10.2147/IJWH.S34442
DOCUMENT TYPE: Review

Verdin, H., D'haene, B., Beysen, D., Novikova, Y., Menten, B., Sante, T., Lapunzina, P., Nevado, J., Carvalho, C.M.B., Lupski, J.R., de Baere, E.
Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain
(2013) PLoS Genetics, 9 (3), art. no. e1003358, . Cited 10 times.

DOI: 10.1371/journal.pgen.1003358
DOCUMENT TYPE: Article

Walter, V., Nobel, A.B., Hayes, D.N., Wright, F.A.
Identification of Recurrent DNA Copy Number Aberrations in Tumors
(2013) Statistical Diagnostics for Cancer: Analyzing High-Dimensional Data, 3, pp. 239-260.

DOI: 10.1002/9783527665471.ch13
DOCUMENT TYPE: Book Chapter

Ray, F.A., Zimmerman, E., Robinson, B., Cornforth, M.N., Bedford, J.S., Goodwin, E.H., Bailey, S.M.
Directional genomic hybridization for chromosomal inversion discovery and detection
(2013) Chromosome Research, 21 (2), pp. 165-174. Cited 2 times.

DOI: 10.1007/s10577-013-9345-0
DOCUMENT TYPE: Article

Van Den Bossche, M.J., Strazisar, M., Cammaerts, S., Liekens, A.M., Vandeweyer, G., Depreeuw, V., Mattheijssens, M., Lenaerts, A..-S., De Zutter, S., De Rijk, P., Sabbe, B., Del-Favero, J.
Identification of rare copy number variants in high burden schizophrenia families
(2013) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 162 (3), pp. 273-282. Cited 4 times.

DOI: 10.1002/ajmg.b.32146
DOCUMENT TYPE: Article

Lundin, S., Gruselius, J., Nystedt, B., Lexow, P., Käller, M., Lundeberg, J.
Hierarchical molecular tagging to resolve long continuous sequences by massively parallel sequencing
(2013) Scientific Reports, 3, art. no. 1186, .

DOI: 10.1038/srep01186
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.
Comparative Studies of Copy Number Variation Detection Methods for Next-Generation Sequencing Technologies
(2013) PLoS ONE, 8 (3), art. no. e59128, . Cited 24 times.

DOI: 10.1371/journal.pone.0059128
DOCUMENT TYPE: Article

Ragheb, M.N., Ford, C.B., Chase, M.R., Lin, P.L., Flynn, J.L., Fortune, S.M.
The mutation rate of mycobacterial repetitive unit loci in strains of M. tuberculosis from cynomolgus macaque infection
(2013) BMC Genomics, 14 (1), art. no. 145, . Cited 3 times.

DOI: 10.1186/1471-2164-14-145
DOCUMENT TYPE: Article

Singer, M.A.
Are chronic degenerative diseases part of the ageing process? Insights from comparative biology
(2013) Are Chronic Degenerative Diseases Part of the Ageing Process? Insights from Comparative Biology, pp. 1-217.

DOCUMENT TYPE: Book

Coonrod, E.M., Durtschi, J.D., Margraf, R.L., Voelkerding, K.V.
Developing genome and exome sequencing for candidate gene identification in inherited disorders: An integrated technical and bioinformatics approach
(2013) Archives of Pathology and Laboratory Medicine, 137 (3), pp. 415-433. Cited 11 times.

DOI: 10.5858/arpa.2012-0107-RA
DOCUMENT TYPE: Review

Grimm, D., Hagmann, J., Koenig, D., Weigel, D., Borgwardt, K.
Accurate indel prediction using paired-end short reads

(2013) BMC Genomics, 14 (1), art. no. 132, . Cited 10 times.

DOI: 10.1186/1471-2164-14-132
DOCUMENT TYPE: Article

Soon, W.W., Hariharan, M., Snyder, M.P.
High-throughput sequencing for biology and medicine
(2013) Molecular Systems Biology, 9, art. no. 640, . Cited 40 times.

DOI: 10.1038/msb.2012.61
DOCUMENT TYPE: Review

Hong, S.G., Dunbar, C.E., Winkler, T.
Assessing the risks of genotoxicity in the therapeutic development of induced pluripotent stem cells
(2013) Molecular Therapy, 21 (2), pp. 272-281. Cited 7 times.

DOI: 10.1038/mt.2012.255
DOCUMENT TYPE: Review

Haasl, R.J., Payseur, B.A.
Microsatellites as targets of natural selection
(2013) Molecular Biology and Evolution, 30 (2), pp. 285-298. Cited 8 times.

DOI: 10.1093/molbev/mss247
DOCUMENT TYPE: Article

Feulner, P.G.D., Chain, F.J.J., Panchal, M., Eizaguirre, C., Kalbe, M., Lenz, T.L., Mundry, M., Samonte, I.E., Stoll, M., Milinski, M., Reusch, T.B.H., Bornberg-Bauer, E.
Genome-wide patterns of standing genetic variation in a marine population of three-spined sticklebacks
(2013) Molecular Ecology, 22 (3), pp. 635-649. Cited 21 times.

DOI: 10.1111/j.1365-294X.2012.05680.x
DOCUMENT TYPE: Conference Paper

Gong, Q., Tao, Y., Yang, J.-R., Cai, J., Yuan, Y., Ruan, J., Yang, J., Liu, H., Li, W., Lu, X., Zhuang, S.-M., Wang, S.M., Wu, C.-I.
Identification of medium-sized genomic deletions with low coverage, mate-paired restricted tags
(2013) BMC Genomics, 14 (1), art. no. 51, .

DOI: 10.1186/1471-2164-14-51
DOCUMENT TYPE: Article

Schluth-Bolard, C., Labalme, A., Cordier, M.-P., Till, M., Nadeau, G., Tevissen, H., Lesca, G., Boutry-Kryza, N., Rossignol, S., Rocas, D., Dubruc, E., Edery, P., Sanlaville, D.
Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations
(2013) Journal of Medical Genetics, 50 (3), pp. 144-150. Cited 14 times.

DOI: 10.1136/jmedgenet-2012-101351
DOCUMENT TYPE: Article

Li, W., Olivier, M.
Current analysis platforms and methods for detecting copy number variation
(2013) Physiological Genomics, 45 (1), pp. 1-6. Cited 6 times.

DOI: 10.1152/physiolgenomics.00082.2012
DOCUMENT TYPE: Article

Li, H.
Systems genetics in "-omics" era: Current and future development
(2013) Theory in Biosciences, 132 (1), pp. 1-16. Cited 2 times.

DOI: 10.1007/s12064-012-0168-x
DOCUMENT TYPE: Review

Solieri, L., Dakal, T.C., Giudici, P.
Next-generation sequencing and its potential impact on food microbial genomics
(2013) Annals of Microbiology, 63 (1), pp. 21-37. Cited 6 times.

DOI: 10.1007/s13213-012-0478-8
DOCUMENT TYPE: Review

Kunz, M., Dannemann, M., Kelso, J.
High-throughput sequencing of the melanoma genome
(2013) Experimental Dermatology, 22 (1), pp. 10-17. Cited 8 times.

DOI: 10.1111/exd.12054
DOCUMENT TYPE: Article

Wittler, R.
Unraveling overlapping deletions by agglomerative clustering
(2013) BMC Genomics, 14, art. no. S12, . Cited 1 time.

DOI: 10.1186/1471-2164-14-S1-S12
DOCUMENT TYPE: Article

Schulte, I., Batty, E.M., Pole, J.C.M., Blood, K.A., Mo, S., Cooke, S.L., Ng, C., Howe, K.L., Chin, S.-F., Brenton, J.D., Caldas, C., Howarth, K.D., Edwards, P.A.W.
Structural analysis of the genome of breast cancer cell line ZR-75-30 identifies twelve expressed fusion genes
(2012) BMC Genomics, 13 (1), art. no. 719, . Cited 2 times.

DOI: 10.1186/1471-2164-13-719
DOCUMENT TYPE: Article

Xu, H., Luo, X., Qian, J., Pang, X., Song, J., Qian, G., Chen, J., Chen, S.
FastUniq: A Fast De Novo Duplicates Removal Tool for Paired Short Reads
(2012) PLoS ONE, 7 (12), art. no. e52249, . Cited 4 times.

DOI: 10.1371/journal.pone.0052249
DOCUMENT TYPE: Article

Zhou, S., Fu, Y., Li, J., He, L., Cai, X., Yan, Q., Rao, X., Huang, S., Li, G., Wang, Y., Xu, A.
HTS-PEG: A Method for High Throughput Sequencing of the Paired-Ends of Genomic Libraries
(2012) PLoS ONE, 7 (12), art. no. e52257, .

DOI: 10.1371/journal.pone.0052257
DOCUMENT TYPE: Article

Wu, X., Zhang, D., Li, G.
Insights into the regulation of human CNV-miRNAs from the view of their target genes
(2012) BMC Genomics, 13 (1), art. no. 707, . Cited 5 times.

DOI: 10.1186/1471-2164-13-707
DOCUMENT TYPE: Article

Talkowski, M.E., Ordulu, Z., Pillalamarri, V., Benson, C.B., Blumenthal, I., Connolly, S., Hanscom, C., Hussain, N., Pereira, S., Picker, J., Rosenfeld, J.A., Shaffer, L.G., Wilkins-Haug, L.E., Gusella, J.F., Morton, C.C.
Clinical diagnosis by whole-genome sequencing of a prenatal sample
(2012) New England Journal of Medicine, 367 (23), pp. 2226-2232. Cited 30 times.

DOI: 10.1056/NEJMoa1208594
DOCUMENT TYPE: Article

Alves, J.M., Lopes, A.M., Chikhi, L., Amorim, A.
On the structural plasticity of the human genome: Chromosomal inversions revisited
(2012) Current Genomics, 13 (8), pp. 623-632. Cited 4 times.

DOI: 10.2174/138920212803759703
DOCUMENT TYPE: Review

Pan, S., Caleshu, C.A., Dunn, K.E., Ashley, E.A.
Cardiac structural and sarcomere genes associated with cardiomyopathy exhibit marked intolerance of genetic variation
(2012) Circulation: Cardiovascular Genetics, 5 (6), pp. 602-610. Cited 15 times.

DOI: 10.1161/CIRCGENETICS.112.963421
DOCUMENT TYPE: Article

Chin, B.L., Ryan, O., Lewitter, F., Boone, C., Fink, G.R.
Genetic variation in Saccharomyces cerevisiae: Circuit diversification in a signal transduction network
(2012) Genetics, 192 (4), pp. 1523-1532. Cited 5 times.

DOI: 10.1534/genetics.112.145573
DOCUMENT TYPE: Article

Raphael, B.J.
Chapter 6: Structural Variation and Medical Genomics
(2012) PLoS Computational Biology, 8 (12), art. no. e1002821, . Cited 3 times.

DOI: 10.1371/journal.pcbi.1002821
DOCUMENT TYPE: Article

Milward, E.A., Daneshi, N., Johnstone, D.M.
Emerging real-time technologies in molecular medicine and the evolution of integrated 'pharmacomics' approaches to personalized medicine and drug discovery
(2012) Pharmacology and Therapeutics, 136 (3), pp. 295-304. Cited 2 times.

DOI: 10.1016/j.pharmthera.2012.08.008
DOCUMENT TYPE: Review

dela Paz, J.S., Stronghill, P.E., Douglas, S.J., Saravia, S., Hasenkampf, C.A., Riggs, C.D.

Chromosome Fragile Sites in Arabidopsis Harbor Matrix Attachment Regions That May Be Associated with Ancestral Chromosome Rearrangement Events
(2012) PLoS Genetics, 8 (12), art. no. e1003136, . Cited 1 time.

DOI: 10.1371/journal.pgen.1003136
DOCUMENT TYPE: Article

Hardiman, G.
Application of Ultra-High Throughput Sequencing and Microarray Technologies in Pharmacogenomics Testing
(2012) Therapeutic Drug Monitoring, pp. 143-159. Cited 1 time.

DOI: 10.1016/B978-0-12-385467-4.00007-5
DOCUMENT TYPE: Book Chapter

Elbaidouri, M., Panaud, O.
Genome-wide analysis of transposition using next generation sequencing technologies
(2012) Topics in Current Genetics, 24, pp. 59-70. Cited 2 times.

DOI: 10.1007/978-3-642-31842-9-4
DOCUMENT TYPE: Article

Gahan, P.B.
Biology of circulating nucleic acids and possible roles in diagnosis and treatment in diabetes and cancer
(2012) Infectious Disorders - Drug Targets, 12 (5), pp. 360-370. Cited 4 times.

DOI: 10.2174/187152612804142224
DOCUMENT TYPE: Article

Jones, M.J.K., Jallepalli, P.V.
Chromothripsis: Chromosomes in Crisis
(2012) Developmental Cell, 23 (5), pp. 908-917. Cited 22 times.

DOI: 10.1016/j.devcel.2012.10.010
DOCUMENT TYPE: Review

Schwartz, J.J., Lee, C., Hiatt, J.B., Adey, A., Shendure, J.
Capturing native long-range contiguity by in situ library construction and optical sequencing
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (46), pp. 18749-18754. Cited 3 times.

DOI: 10.1073/pnas.1202680109
DOCUMENT TYPE: Article

Falconer, E., Hills, M., Naumann, U., Poon, S.S.S., Chavez, E.A., Sanders, A.D., Zhao, Y., Hirst, M., Lansdorp, P.M.
DNA template strand sequencing of single-cells maps genomic rearrangements at high resolution
(2012) Nature Methods, 9 (11), pp. 1107-1112. Cited 17 times.

DOI: 10.1038/nmeth.2206
DOCUMENT TYPE: Article

Marian, A.J.
Challenges in Medical Applications of Whole Exome/Genome Sequencing Discoveries
(2012) Trends in Cardiovascular Medicine, 22 (8), pp. 219-223. Cited 8 times.

DOI: 10.1016/j.tcm.2012.08.001
DOCUMENT TYPE: Review

Han, J.S., Shao, S.
Circular retrotransposition products generated by a LINE retrotransposon
(2012) Nucleic Acids Research, 40 (21), pp. 10866-10877. Cited 3 times.

DOI: 10.1093/nar/gks859
DOCUMENT TYPE: Article

Liu, G.E., Bickhart, D.M.
Copy number variation in the cattle genome
(2012) Functional and Integrative Genomics, 12 (4), pp. 609-624. Cited 10 times.

DOI: 10.1007/s10142-012-0289-9
DOCUMENT TYPE: Review

Coughlin II, C.R., Scharer, G.H., Shaikh, T.H.
Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns
(2012) Genome Medicine, 4 (10), art. no. 80, . Cited 1 time.

DOI: 10.1186/gm381
DOCUMENT TYPE: Review

Mijušković, M., Brown, S.M., Tang, Z., Lindsay, C.R., Efstathiadis, E., Deriano, L., Roth, D.B.
A Streamlined Method for Detecting Structural Variants in Cancer Genomes by Short Read Paired-End Sequencing
(2012) PLoS ONE, 7 (10), art. no. e48314, . Cited 6 times.

DOI: 10.1371/journal.pone.0048314
DOCUMENT TYPE: Article

Hunter, R.G., Murakami, G., Dewell, S., Seligsohn, M., Baker, M.E.R., Datson, N.A., McEwen, B.S., Pfaff, D.W.
Acute stress and hippocampal histone H3 lysine 9 trimethylation, a retrotransposon silencing response
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (43), pp. 17657-17662. Cited 31 times.

DOI: 10.1073/pnas.1215810109
DOCUMENT TYPE: Article

Shibata, T.
Cancer genomics and pathology: All Together Now
(2012) Pathology International, 62 (10), pp. 647-659. Cited 3 times.

DOI: 10.1111/j.1440-1827.2012.02855.x
DOCUMENT TYPE: Review

Langley, C.H., Stevens, K., Cardeno, C., Lee, Y.C.G., Schrider, D.R., Pool, J.E., Langley, S.A., Suarez, C., Corbett-Detig, R.B., Kolaczkowski, B., Fang, S., Nista, P.M., Holloway, A.K., Kern, A.D., Dewey, C.N., Song, Y.S., Hahn, M.W., Begun, D.J.
Genomic variation in natural populations of Drosophila melanogaster
(2012) Genetics, 192 (2), pp. 533-598. Cited 65 times.

DOI: 10.1534/genetics.112.142018

DOCUMENT TYPE: Article

Simon, M.M., Mallon, A.-M., Howell, G.R., Reinholdt, L.G.
High throughput sequencing approaches to mutation discovery in the mouse
(2012) Mammalian Genome, 23 (9-10), pp. 499-513.

DOI: 10.1007/s00335-012-9424-0
DOCUMENT TYPE: Article

Ehli, E.A., Abdellaoui, A., Hu, Y., Hottenga, J.J., Kattenberg, M., Van Beijsterveldt, T., Bartels, M., Althoff, R.R., Xiao, X., Scheet, P., De Geus, E.J., Hudziak, J.J., Boomsma, D.I., Davies, G.E.
De novo and inherited CNVs in MZ twin pairs selected for discordance and concordance on Attention Problems
(2012) European Journal of Human Genetics, 20 (10), pp. 1037-1043. Cited 14 times.

DOI: 10.1038/ejhg.2012.49
DOCUMENT TYPE: Article

Charney, E.
Humans, fruit flies, and automatons
(2012) Behavioral and Brain Sciences, 35 (5), pp. 381-410.

DOI: 10.1017/S0140525X12001501
DOCUMENT TYPE: Article

Zhang, N.R., Siegmund, D.O.
Model selection for high-dimensional, multi-sequence change-point problems
(2012) Statistica Sinica, 22 (4), pp. 1507-1538. Cited 1 time.

DOI: 10.5705/ss.2010.257
DOCUMENT TYPE: Article

Yao, F., Ariyaratne, P.N., Hillmer, A.M., Lee, W.H., Li, G., Teo, A.S.M., Woo, X.Y., Zhang, Z., Chen, J.P., Poh, W.T., Zawack, K.F.B., Chan, C.S., Leong, S.T., Neo, S.C., Choi, P.S.D., Gao, S., Nagarajan, N., Thoreau, H., Shahab, A., Ruan, X., Cacheux-Rataboul, V., Wei, C.-L., Bourque, G., Sung, W.-K., Liu, E.T., Ruan, Y.
Long Span DNA Paired-End-Tag (DNA-PET) Sequencing Strategy for the Interrogation of Genomic Structural Mutations and Fusion-Point-Guided Reconstruction of Amplicons
(2012) PLoS ONE, 7 (9), art. no. e46152, . Cited 4 times.

DOI: 10.1371/journal.pone.0046152
DOCUMENT TYPE: Article

Asan, Geng, C., Chen, Y., Wu, K., Cai, Q., Wang, Y., Lang, Y., Cao, H., Yang, H., Wang, J., Zhang, X.
Paired-End Sequencing of Long-Range DNA Fragments for De Novo Assembly of Large, Complex Mammalian Genomes by Direct Intra-Molecule Ligation
(2012) PLoS ONE, 7 (9), art. no. e46211, . Cited 4 times.

DOI: 10.1371/journal.pone.0046211
DOCUMENT TYPE: Article

Soemedi, R., Wilson, I.J., Bentham, J., Darlay, R., Töpf, A., Zelenika, D., Cosgrove, C., Setchfield, K., Thornborough, C., Granados-Riveron, J., Blue, G.M., Breckpot, J., Hellens, S., Zwolinkski, S., Glen, E., Mamasoula, C., Rahman, T.J., Hall, D., Rauch, A., Devriendt, K., Gewillig, M., O'sullivan, J., Winlaw, D.S., Bu'lock, F., Brook, J.D., Bhattacharya, S., Lathrop, M., Santibanez-Koref, M., Cordell, H.J., Goodship, J.A., Keavney, B.D.
Contribution of global rare copy-number variants to the risk of sporadic congenital heart disease

(2012) American Journal of Human Genetics, 91 (3), pp. 489-501. Cited 43 times.

DOI: 10.1016/j.ajhg.2012.08.003
DOCUMENT TYPE: Article

Rolfe, P.A., Bernstein, D.A., Grisafi, P., Fink, G.R., Gifford, D.K.
Ruler arrays reveal haploid genomic structural variation
(2012) PLoS ONE, 7 (8), art. no. e43210, .

DOI: 10.1371/journal.pone.0043210
DOCUMENT TYPE: Article

Sun, S., Ke, R., Hughes, D., Nilsson, M., Andersson, D.I.
Genome-wide detection of spontaneous chromosomal rearrangements in bacteria
(2012) PLoS ONE, 7 (8), art. no. e42639, . Cited 7 times.

DOI: 10.1371/journal.pone.0042639
DOCUMENT TYPE: Article

Du, R., Lu, C., Jiang, Z., Li, S., Ma, R., An, H., Xu, M., An, Y., Xia, Y., Jin, L., Wang, X., Zhang, F.
Efficient typing of copy number variations in a segmental duplication-mediated rearrangement hotspot using multiplex competitive amplification
(2012) Journal of Human Genetics, 57 (8), pp. 545-551. Cited 12 times.

DOI: 10.1038/jhg.2012.66
DOCUMENT TYPE: Article

Jiang, H., Zeng, X., He, N.
An integrated view of current progress in copy number variations analysis of genome
(2012) Advanced Science Letters, 7, pp. 1-8. Cited 5 times.

DOI: 10.1166/asl.2012.3315
DOCUMENT TYPE: Review

Valencia, A., Hidalgo, M.
Getting personalized cancer genome analysis into the clinic: the challenges in bioinformatics
(2012) Genome Medicine, 4 (8), art. no. 61, . Cited 11 times.

DOI: 10.1186/gm362
DOCUMENT TYPE: Review

Casals, F., Idaghdour, Y., Hussin, J., Awadalla, P.
Next-generation sequencing approaches for genetic mapping of complex diseases
(2012) Journal of Neuroimmunology, 248 (1-2), pp. 10-22. Cited 9 times.

DOI: 10.1016/j.jneuroim.2011.12.017
DOCUMENT TYPE: Review

Belfield, E.J., Gan, X., Mithani, A., Brown, C., Jiang, C., Franklin, K., Alvey, E., Wibowo, A., Jung, M., Bailey, K., Kalwani, S., Ragoussis, J., Mott, R., Harberd, N.P.
Genome-wide analysis of mutations in mutant lineages selected following fast-neutron irradiation mutagenesis of Arabidopsis thaliana
(2012) Genome Research, 22 (7), pp. 1306-1315. Cited 12 times.

DOI: 10.1101/gr.131474.111
DOCUMENT TYPE: Article

Xing, M.-N., Zhang, X.-Z., Huang, H.
Application of metagenomic techniques in mining enzymes from microbial communities for biofuel synthesis
(2012) Biotechnology Advances, 30 (4), pp. 920-929. Cited 14 times.

DOI: 10.1016/j.biotechadv.2012.01.021
DOCUMENT TYPE: Review

Kim, S., Millard, S.P., Yu, C.-E., Leong, L., Radant, A., Dobie, D., Tsuang, D.W., Wijsman, E.M.
Inheritance Model Introduces Differential Bias in CNV Calls Between Parents and Offspring
(2012) Genetic Epidemiology, 36 (5), pp. 488-498. Cited 1 time.

DOI: 10.1002/gepi.21643
DOCUMENT TYPE: Article

González, J.R., Abellán, C., Abellán, J.J.
Bayesian model to detect phenotype-specific genes for copy number data
(2012) BMC Bioinformatics, 13 (1), art. no. 130, .

DOI: 10.1186/1471-2105-13-130
DOCUMENT TYPE: Article

Iskow, R.C., Gokcumen, O., Lee, C.
Exploring the role of copy number variants in human adaptation
(2012) Trends in Genetics, 28 (6), pp. 245-257. Cited 21 times.

DOI: 10.1016/j.tig.2012.03.002
DOCUMENT TYPE: Review

Van Den Bossche, M.J., Strazisar, M., De Bruyne, S., Bervoets, C., Lenaerts, A.-S., De Zutter, S., Nordin, A., Norrback, K.-F., Goossens, D., De Rijk, P., Green, E.K., Grozeva, D., Mendlewicz, J., Craddock, N., Sabbe, B.G., Adolfsson, R., Souery, D., Del-Favero, J.
Identification of a CACNA2D4 deletion in late onset bipolar disorder patients and implications for the involvement of voltage-dependent calcium channels in psychiatric disorders
(2012) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 159 B (4), pp. 465-475. Cited 3 times.

DOI: 10.1002/ajmg.b.32053
DOCUMENT TYPE: Article

Arlt, M.F., Wilson, T.E., Glover, T.W.
Replication stress and mechanisms of CNV formation
(2012) Current Opinion in Genetics and Development, 22 (3), pp. 204-210. Cited 23 times.

DOI: 10.1016/j.gde.2012.01.009
DOCUMENT TYPE: Review

Lai, B., Ding, R., Li, Y., Duan, L., Zhu, H.
A de novo metagenomic assembly program for shotgun DNA reads
(2012) Bioinformatics, 28 (11), art. no. bts162, pp. 1455-1462. Cited 14 times.

DOI: 10.1093/bioinformatics/bts162
DOCUMENT TYPE: Article

Doan, R., Cohen, N., Harrington, J., Veazy, K., Juras, R., Cothran, G., McCue, M.E., Skow, L., Dindot, S.V.
Identification of copy number variants in horses
(2012) Genome Research, 22 (5), pp. 899-907. Cited 13 times.

DOI: 10.1101/gr.128991.111
DOCUMENT TYPE: Article

Li, J., Harris, R.A., Cheung, S.W., Coarfa, C., Jeong, M., Goodell, M.A., White, L.D., Patel, A., Kang, S.-H., Shaw, C., Chinault, A.C., Gambin, T., Gambin, A., Lupski, J.R., Milosavljevic, A.
Genomic hypomethylation in the human germline associates with selective structural mutability in the human genome
(2012) PLoS Genetics, 8 (5), art. no. e1002692, . Cited 19 times.

DOI: 10.1371/journal.pgen.1002692
DOCUMENT TYPE: Article

Rossetti, S., Hopp, K., Sikkink, R.A., Sundsbak, J.L., Lee, Y.K., Kubly, V., Eckloff, B.W., Ward, C.J., Winearls, C.G., Torres, V.E., Harris, P.C.
Identification of gene mutations in autosomal dominant polycystic kidney disease through targeted resequencing
(2012) Journal of the American Society of Nephrology, 23 (5), pp. 915-933. Cited 27 times.

DOI: 10.1681/ASN.2011101032
DOCUMENT TYPE: Article

Furney, S.J., Gundem, G., Lopez-Bigas, N.
Oncogenomics methods and resources
(2012) Cold Spring Harbor Protocols, 7 (5), pp. 546-564. Cited 3 times.

DOI: 10.1101/pdb.top069229
DOCUMENT TYPE: Review

Weise, A., Mrasek, K., Klein, E., Mulatinho, M., Llerena Jr., J.C., Hardekopf, D., Pekova, S., Bhatt, S., Kosyakova, N., Liehr, T.
Microdeletion and Microduplication Syndromes
(2012) Journal of Histochemistry and Cytochemistry, 60 (5), pp. 346-358. Cited 21 times.

DOI: 10.1369/0022155412440001
DOCUMENT TYPE: Article

Jain, K.K.
Integration of Biotechnologies for the Development of Personalized Medicine
(2012) Pharmaceutical Biotechnology: Drug Discovery and Clinical Applications, pp. 553-580.

DOI: 10.1002/9783527632909.ch21
DOCUMENT TYPE: Book Chapter

Adelson, D.L.
Bovine Genome Architecture
(2012) Bovine Genomics, pp. 123-143.

DOI: 10.1002/9781118301739.ch10
DOCUMENT TYPE: Book Chapter

Itsara, A., Vissers, L.E.L.M., Steinberg, K.M., Meyer, K.J., Zody, M.C., Koolen, D.A., De Ligt, J., Cuppen, E., Baker, C., Lee, C., Graves, T.A., Wilson, R.K., Jenkins, R.B., Veltman, J.A., Eichler, E.E.
Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing
(2012) American Journal of Human Genetics, 90 (4), pp. 599-613. Cited 8 times.

DOI: 10.1016/j.ajhg.2012.02.013
DOCUMENT TYPE: Article

Rodríguez-Santiago, B., Armengol, L.
Next generation sequencing technology in pre- and postnatal genetic diagnosis [Tecnologías de secuenciación de nueva generación en diagnóstico genético pre- y postnatal]
(2012) Diagnostico Prenatal, 23 (2), pp. 56-66.

DOI: 10.1016/j.diapre.2012.02.001
DOCUMENT TYPE: Article

Chiang, C., Jacobsen, J.C., Ernst, C., Hanscom, C., Heilbut, A., Blumenthal, I., Mills, R.E., Kirby, A., Lindgren, A.M., Rudiger, S.R., McLaughlan, C.J., Bawden, C.S., Reid, S.J., Faull, R.L.M., Snell, R.G., Hall, I.M., Shen, Y., Ohsumi, T.K., Borowsky, M.L., Daly, M.J., Lee, C., Morton, C.C., MacDonald, M.E., Gusella, J.F., Talkowski, M.E.
Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration
(2012) Nature Genetics, 44 (4), pp. 390-397. Cited 61 times.

DOI: 10.1038/ng.2202
DOCUMENT TYPE: Article

Sindi, S.S., Önal, S., Peng, L.C., Wu, H.-T., Raphael, B.J.
An integrative probabilistic model for identification of structural variation in sequencing data
(2012) Genome Biology, 13 (3), art. no. R22, . Cited 24 times.

DOI: 10.1186/gb-2012-13-3-r22
DOCUMENT TYPE: Article

Yalcin, B., Wong, K., Bhomra, A., Goodson, M., Keane, T.M., Adams, D.J., Flint, J.
The fine-scale architecture of structural variants in 17 mouse genomes
(2012) Genome Biology, 13 (3), art. no. R18, . Cited 13 times.

DOI: 10.1186/gb-2012-13-3-r18
DOCUMENT TYPE: Article

Ariyadasa, R., Stein, N.
Advances in BAC-based physical mapping and map integration strategies in plants
(2012) Journal of Biomedicine and Biotechnology, 2012, art. no. 184854, . Cited 11 times.

DOI: 10.1155/2012/184854
DOCUMENT TYPE: Review

Jensen, H., Kjeldsen, E., Hjortdal, V.E.
Could submicroscopical chromosomal imbalances cause cono-truncal malformations in twins?
(2012) Congenital Heart Disease, 7 (2), pp. 170-177.

DOI: 10.1111/j.1747-0803.2011.00544.x
DOCUMENT TYPE: Article

Lee, H.J., Kweon, J., Kim, E., Kim, S., Kim, J.-S.
Targeted chromosomal duplications and inversions in the human genome using zinc finger nucleases
(2012) Genome Research, 22 (3), pp. 539-548. Cited 50 times.

DOI: 10.1101/gr.129635.111
DOCUMENT TYPE: Article

Dewey, F.E., Pan, S., Wheeler, M.T., Quake, S.R., Ashley, E.A.
DNA sequencing clinical applications of new DNA sequencing technologies
(2012) Circulation, 125 (7), pp. 931-944. Cited 15 times.

DOI: 10.1161/CIRCULATIONAHA.110.972828
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Veltman, J.A.
Impact of Genomewide Structural Variation on Gene Discovery
(2012) Gene Discovery for Disease Models, pp. 443-470.

DOI: 10.1002/9780470933947.ch21
DOCUMENT TYPE: Book Chapter

Cáceres, A., Sindi, S.S., Raphael, B.J., Cáceres, M., González, J.R.
Identification of polymorphic inversions from genotypes
(2012) BMC Bioinformatics, 13 (1), art. no. 28, . Cited 5 times.

DOI: 10.1186/1471-2105-13-28
DOCUMENT TYPE: Article

Koboldt, D.C., Larson, D.E., Chen, K., Ding, L., Wilson, R.K.
Massively parallel sequencing approaches for characterization of structural variation
(2012) Methods in Molecular Biology, 838, pp. 369-384. Cited 15 times.

DOI: 10.1007/978-1-61779-507-7_18
DOCUMENT TYPE: Article

Nowrousian, M., Teichert, I., Masloff, S., Kück, U.
Whole-genome sequencing of sordaria macrospora mutants identifies developmental genes
(2012) G3: Genes, Genomes, Genetics, 2 (2), pp. 261-270. Cited 17 times.

DOI: 10.1534/g3.111.001479
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Stankiewicz, P.
Microdeletion and microduplication syndromes
(2012) Methods in Molecular Biology, 838, pp. 29-75. Cited 20 times.

DOI: 10.1007/978-1-61779-507-7_2
DOCUMENT TYPE: Article

Pfundt, R., Veltman, J.A.
Structural genomic variation in intellectual disability
(2012) Methods in Molecular Biology, 838, pp. 77-95. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_3

DOCUMENT TYPE: Article

Sneddon, T.P., Church, D.M.
Online resources for genomic structural variation
(2012) Methods in Molecular Biology, 838, pp. 273-289. Cited 2 times.

DOI: 10.1007/978-1-61779-507-7_13
DOCUMENT TYPE: Article

Johansson, A.C.V., Feuk, L.
Characterizing and interpreting genetic variation from personal genome sequencing
(2012) Methods in Molecular Biology, 838, pp. 343-367. Cited 3 times.

DOI: 10.1007/978-1-61779-507-7_17
DOCUMENT TYPE: Article

Hollox, E.J.
The challenges of studying complex and dynamic regions of the human genome
(2012) Methods in Molecular Biology, 838, pp. 187-207. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_9
DOCUMENT TYPE: Article

Jiang, J., Li, J., Kwan, H., Au, C., Wan Law, P., Li, L., Kam, K., Lun Ling, J., Leung, F.C.
A cost-effective and universal strategy for complete prokaryotic genomic sequencing proposed by computer simulation
(2012) BMC Research Notes, 5, art. no. 80, . Cited 2 times.

DOI: 10.1186/1756-0500-5-80
DOCUMENT TYPE: Article

Hall, I.M., Quinlan, A.R.
Detection and interpretation of genomic structural variation in mammals
(2012) Methods in Molecular Biology, 838, pp. 225-248. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_11
DOCUMENT TYPE: Article

Iqbal, Z., Caccamo, M., Turner, I., Flicek, P., McVean, G.
De novo assembly and genotyping of variants using colored de Bruijn graphs
(2012) Nature Genetics, 44 (2), pp. 226-232. Cited 73 times.

DOI: 10.1038/ng.1028
DOCUMENT TYPE: Article

Dumanski, J.P., Piotrowski, A.
Structural genetic variation in the context of somatic mosaicism
(2012) Methods in Molecular Biology, 838, pp. 249-272. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_12
DOCUMENT TYPE: Article

Simmons, A.D., Carvalho, C.M.B., Lupski, J.R.
What have studies of genomic disorders taught us about our genome?

(2012) Methods in Molecular Biology, 838, pp. 1-27. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_1
DOCUMENT TYPE: Article

Dewal, N., Hu, Y., Freedman, M.L., LaFramboise, T., Pe'Er, I.
Calling amplified haplotypes in next generation tumor sequence data
(2012) Genome Research, 22 (2), pp. 362-374. Cited 4 times.

DOI: 10.1101/gr.122564.111
DOCUMENT TYPE: Article

Ceulemans, S., Van Der Ven, K., Del-Favero, J.
Targeted screening and validation of copy number variations
(2012) Methods in Molecular Biology, 838, pp. 311-328. Cited 6 times.

DOI: 10.1007/978-1-61779-507-7_15
DOCUMENT TYPE: Article

Chen, J., Kim, Y.C., Wang, S.M.
DGS (Ditag Genome Scanning) - A Restriction-Based Paired-End Sequencing Approach for Genome Structural Analysis
(2012) Tag-Based Next Generation Sequencing, pp. 277-285.

DOI: 10.1002/9783527644582.ch16
DOCUMENT TYPE: Book Chapter

Gogol-Döring, A., Chen, W.
An overview of the analysis of next generation sequencing data
(2012) Methods in Molecular Biology, 802, pp. 249-257. Cited 12 times.

DOI: 10.1007/978-1-61779-400-1_16
DOCUMENT TYPE: Article

Le Scouarnec, S., Gribble, S.M.
Characterising chromosome rearrangements: Recent technical advances in molecular cytogenetics
(2012) Heredity, 108 (1), pp. 75-85. Cited 22 times.

DOI: 10.1038/hdy.2011.100
DOCUMENT TYPE: Review

Xu, X., Liu, X., Ge, S., Jensen, J.D., Hu, F., Li, X., Dong, Y., Gutenkunst, R.N., Fang, L., Huang, L., Li, J., He, W., Zhang, G., Zheng, X., Zhang, F., Li, Y., Yu, C., Kristiansen, K., Zhang, X., Wang, J., Wright, M., McCouch, S., Nielsen, R., Wang, J., Wang, W.
Resequencing 50 accessions of cultivated and wild rice yields markers for identifying agronomically important genes
(2012) Nature Biotechnology, 30 (1), pp. 105-111. Cited 145 times.

DOI: 10.1038/nbt.2050
DOCUMENT TYPE: Article

Abeel, T., Van Parys, T., Saeys, Y., Galagan, J., Van De Peer, Y.
GenomeView: A next-generation genome browser
(2012) Nucleic Acids Research, 40 (2), . Cited 25 times.

DOI: 10.1093/nar/gkr995

DOCUMENT TYPE: Article

Severson, D.W., Behura, S.K.
Mosquito genomics: Progress and challenges
(2012) Annual Review of Entomology, 57, pp. 143-166. Cited 27 times.

DOI: 10.1146/annurev-ento-120710-100651
DOCUMENT TYPE: Article

Brooks, M.J., Rajasimha, H.K., Roger, J.E., Swaroop, A.
Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl -/- retinal transcriptomes
(2011) Molecular Vision, 17, pp. 3034-3054. Cited 27 times.

DOCUMENT TYPE: Article

Liu, Y.
Cancer and signaling pathway deregulation
(2011) Handbook of Research on Computational and Systems Biology: Interdisciplinary Applications, pp. 369-379.

DOI: 10.4018/978-1-60960-491-2.ch017
DOCUMENT TYPE: Book Chapter

Zhao, Z., Nguyen, T.C., Deng, N., Johnson, K.M., Zhu, D.
SPATA: A seeding and patching algorithm for de novo transcriptome assembly
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112351, pp. 26-33. Cited 1 time.

DOI: 10.1109/BIBMW.2011.6112351
DOCUMENT TYPE: Conference Paper

Zhang, J., Gao, Y., Zhao, X., Guan, M., Zhang, W., Wan, J., Yu, B.
Investigation of copy-number variations of C8orf4 in hematological malignancies
(2011) Medical Oncology, 28 (SUPPL. 1), pp. S647-S652. Cited 3 times.

DOI: 10.1007/s12032-010-9698-6
DOCUMENT TYPE: Article

Lai, A.G., Denton-Giles, M., Mueller-Roeber, B., Schippers, J.H.M., Dijkwel, P.P.
Positional information resolves structural variations and uncovers an evolutionarily divergent genetic locus in accessions of Arabidopsis thaliana
(2011) Genome Biology and Evolution, 3 (1), pp. 627-640. Cited 2 times.

DOI: 10.1093/gbe/evr038
DOCUMENT TYPE: Article

Schrider, D.R., Stevens, K., Cardeño, C.M., Langley, C.H., Hahn, M.W.
Genome-wide analysis of retrogene polymorphisms in Drosophila melanogaster
(2011) Genome Research, 21 (12), pp. 2087-2095. Cited 21 times.

DOI: 10.1101/gr.116434.110
DOCUMENT TYPE: Article

Hochstenbach, R., Buizer-Voskamp, J.E., Vorstman, J.A.S., Ophoff, R.A.

Genome arrays for the detection of copy number variations in idiopathic mental retardation, idiopathic generalized epilepsy and neuropsychiatric disorders: Lessons for diagnostic workflow and research
(2011) Cytogenetic and Genome Research, 135 (3-4), pp. 174-202. Cited 45 times.

DOI: 10.1159/000332928
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Lefante, J., Deng, H.-W., Wang, Y.-P.
Detection of copy number variation from next generation sequencing data with total variation penalized least square optimization
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112348, pp. 3-12. Cited 5 times.

DOI: 10.1109/BIBMW.2011.6112348
DOCUMENT TYPE: Conference Paper

Liu, X., Choi, S.-W., Wong, T.K.F., Lam, T.-W., Yiu, S.-M.
Detection of novel tandem duplication with next-generation sequencing
(2011) 2011 ACM Conference on Bioinformatics, Computational Biology and Biomedicine, BCB 2011, pp. 415-419.

DOI: 10.1145/2147805.2147861
DOCUMENT TYPE: Conference Paper

Marian, A.J.
Heart Failure as a Consequence of Restrictive Cardiomyopathy
(2011) Heart Failure, pp. 395-407.

DOI: 10.1016/B978-1-4160-5895-3.10025-7
DOCUMENT TYPE: Book Chapter

Girirajan, S., Campbell, C.D., Eichler, E.E.
Human copy number variation and complex genetic disease
(2011) Annual Review of Genetics, 45, pp. 203-226. Cited 70 times.

DOI: 10.1146/annurev-genet-102209-163544
DOCUMENT TYPE: Article

Kaganovich, M., Snyder, M.
Deciphering DNA Sequence Information
(2011) Genome Organization and Function in the Cell Nucleus, pp. 1-20.

DOI: 10.1002/9783527639991.ch1
DOCUMENT TYPE: Book Chapter

Greisman, H.A., Hoffman, N.G., Yi, H.S.
Rapid high-resolution mapping of balanced chromosomal rearrangements on tiling CGH arrays
(2011) Journal of Molecular Diagnostics, 13 (6), pp. 621-633. Cited 11 times.

DOI: 10.1016/j.jmoldx.2011.07.005
DOCUMENT TYPE: Article

Schlick-Steiner, B.C., Arthofer, W., Moder, K., Steiner, F.M.
Recent insertion/deletion (reINDEL) mutations: Increasing awareness to boost molecular-based research in ecology and evolution

(2015) Ecology and Evolution, 5 (1), pp. 24-35.

DOI: 10.1002/ece3.1330
DOCUMENT TYPE: Article

Gelernter, J.
Genetics of complex traits in psychiatry
(2015) Biological Psychiatry, 77 (1), pp. 36-42. Cited 1 time.

DOI: 10.1016/j.biopsych.2014.08.005
DOCUMENT TYPE: Review

Tang, J., Fang, F., Miller, D.F., Pilrose, J.M., Matei, D., Huang, T.H.-M., Nephew, K.P.
Global DNA methylation profiling technologies and the ovarian cancer methylome
(2015) Methods in Molecular Biology, 1238, pp. 653-675.

DOI: 10.1007/978-1-4939-1804-1_34
DOCUMENT TYPE: Article

Hancarova, M., Puchmajerova, A., Drabova, J., Karaskova, E., Vlckova, M., Sedlacek, Z.
Deletions of 9q21.3 including NTRK2 are associated with severe phenotype
(2015) American Journal of Medical Genetics, Part A, 167 (1), pp. 264-267.

DOI: 10.1002/ajmg.a.36797
DOCUMENT TYPE: Letter

Sante, T., Vergult, S., Volders, P.-J., Kloosterman, W.P., Trooskens, G., De Preter, K., Dheedene, A., Speleman, F., De Meyer, T., Menten, B.
ViVar: A comprehensive platform for the analysis and visualization of structural genomic variation
(2014) PLoS ONE, 9 (12), art. no. e113800, .

DOI: 10.1371/journal.pone.0113800
DOCUMENT TYPE: Article

Snow, A.N., Stence, A.A., Pruessner, J.A., Bossler, A.D., Ma, D.
A simple and cost-effective method of DNA extraction from small formalin-fixed paraffin-embedded tissue for molecular oncologic testing
(2014) BMC Clinical Pathology, 14 (1), art. no. 30, .

DOI: 10.1186/1472-6890-14-30
DOCUMENT TYPE: Article

Hommelsheim, C.M., Frantzeskakis, L., Huang, M., Ülker, B.
PCR amplification of repetitive DNA: A limitation to genome editing technologies and many other applications
(2014) Scientific Reports, 4, art. no. 5052, .

DOI: 10.1038/srep05052
DOCUMENT TYPE: Article

Ordulu, Z., Wong, K.E., Currall, B.B., Ivanov, A.R., Pereira, S., Althari, S., Gusella, J.F., Talkowski, M.E., Morton, C.C.
Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature
(2014) American Journal of Human Genetics, 94 (5), pp. 695-709. Cited 1 time.

DOI: 10.1016/j.ajhg.2014.03.020
DOCUMENT TYPE: Article

Looso, M.
Opening the genetic toolbox of niche model organisms with high throughput techniques: Novel proteins in regeneration as a case study
(2014) BioEssays, 36 (4), pp. 407-418. Cited 3 times.

DOI: 10.1002/bies.201300093
DOCUMENT TYPE: Article

Blake, J., Riddell, A., Theiss, S., Gonzalez, A.P., Haase, B., Jauch, A., Janssen, J.W.G., Ibberson, D., Pavlinic, D., Moog, U., Benes, V., Runz, H.
Sequencing of a patient with balanced chromosome abnormalities and neurodevelopmental disease identifies disruption of multiple high risk loci by structural variation
(2014) PLoS ONE, 9 (3), art. no. e90894, . Cited 1 time.

DOI: 10.1371/journal.pone.0090894
DOCUMENT TYPE: Article

Utami, K.H., Hillmer, A.M., Aksoy, I., Chew, E.G.Y., Teo, A.S.M., Zhang, Z., Lee, C.W.H., Chen, P.J., Seng, C.C., Ariyaratne, P.N., Rouam, S.L., Soo, L.S., Yousoof, S., Prokudin, I., Peters, G., Collins, F., Wilson, M., Kakakios, A., Haddad, G., Menuet, A., Perche, O., Tay, S.K.H., Sung, K.W.K., Ruan, X., Ruan, Y., Liu, E.T., Briault, S., Jamieson, R.V., Davila, S., Cacheux, V.
Detection of chromosomal breakpoints in patients with developmental delay and speech disorders
(2014) PLoS ONE, 9 (3), art. no. e90852, . Cited 1 time.

DOI: 10.1371/journal.pone.0090852
DOCUMENT TYPE: Article

Mosen-Ansorena, D., Telleria, N., Veganzones, S., la Orden, V.D., Maestro, M.L., Aransay, A.M.
SeqCNA: An R package for DNA copy number analysis in cancer using high-throughput sequencing
(2014) BMC Genomics, 15 (1), art. no. 178, .

DOI: 10.1186/1471-2164-15-178
DOCUMENT TYPE: Article

Yavaş, G., Koyutürk, M., Gould, M.P., McMahon, S., LaFramboise, T.
DB2: A probabilistic approach for accurate detection of tandem duplication breakpoints using paired-end reads
(2014) BMC Genomics, 15 (1), art. no. 175, .

DOI: 10.1186/1471-2164-15-175
DOCUMENT TYPE: Article

Duan, J., Deng, H.-W., Wang, Y.-P.
Common copy number variation detection from multiple sequenced samples
(2014) IEEE Transactions on Biomedical Engineering, 61 (3), art. no. 6675802, pp. 928-937.

DOI: 10.1109/TBME.2013.2292588
DOCUMENT TYPE: Article

Yang, Z.-H., Zheng, R., Gao, Y., Zhang, Q., Zhang, H.

Abnormal gene expression and gene fusion in lung adenocarcinoma with high-throughput RNA sequencing
(2014) Cancer Gene Therapy, 21 (2), pp. 74-82. Cited 1 time.

DOI: 10.1038/cgt.2013.86
DOCUMENT TYPE: Article

Chen, K., Chen, L., Fan, X., Wallis, J., Ding, L., Weinstock, G.
TIGRA: A targeted iterative graph routing assembler for breakpoint assembly
(2014) Genome Research, 24 (2), pp. 310-317. Cited 6 times.

DOI: 10.1101/gr.162883.113
DOCUMENT TYPE: Article

Flagel, L.E., Willis, J.H., Vision, T.J.
The standing pool of genomic structural variation in a natural population of Mimulus guttatus
(2014) Genome Biology and Evolution, 6 (1), pp. 53-64.

DOI: 10.1093/gbe/evt199
DOCUMENT TYPE: Article

Ma, S., Wang, X., Liu, Y., Gao, J., Zhang, S., Shi, R., Chang, J., Zhao, P., Xia, Q.
Multiplex genomic structure variation mediated by TALEN and ssODN
(2014) BMC Genomics, 15 (1), art. no. 41, . Cited 5 times.

DOI: 10.1186/1471-2164-15-41
DOCUMENT TYPE: Article

Li, W., Freudenberg, J., Miramontes, P.
Diminishing return for increased Mappability with longer sequencing reads: Implications of the k-mer distributions in the human genome
(2014) BMC Bioinformatics, 15 (1), art. no. 2, . Cited 5 times.

DOI: 10.1186/1471-2105-15-2
DOCUMENT TYPE: Article

Bolon, Y.-T., Stec, A.O., Michno, J.-M., Roessler, J., Bhaskar, P.B., Ries, L., Dobbels, A.A., Campbell, B.W., Young, N.P., Anderson, J.E., Grant, D.M., Orf, J.H., Naeve, S.L., Muehlbauer, G.J., Vance, C.P., Stupar, R.M.
Genome resilience and prevalence of segmental duplications following fast neutron irradiation of soybean
(2014) Genetics, 198 (3), pp. 967-981. Cited 1 time.

DOI: 10.1534/genetics.114.170340/-/DC1
DOCUMENT TYPE: Article

Vergult, S., Van Binsbergen, E., Sante, T., Nowak, S., Vanakker, O., Claes, K., Poppe, B., Van Der Aa, N., Van Roosmalen, M.J., Duran, K., Tavakoli-Yaraki, M., Swinkels, M., Van Den Boogaard, M.-J., Van Haelst, M., Roelens, F., Speleman, F., Cuppen, E., Mortier, G., Kloosterman, W.P., Menten, B.
Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations
(2014) European Journal of Human Genetics, 22 (5), pp. 652-659. Cited 2 times.

DOI: 10.1038/ejhg.2013.220
DOCUMENT TYPE: Article

Fernandez-Banet, J., Lee, N.P., Chan, K.T., Gao, H., Liu, X., Sung, W.-K., Tan, W., Fan, S.T., Poon, R.T., Li, S., Ching, K., Rejto, P.A., Mao, M., Kan, Z.
Decoding complex patterns of genomic rearrangement in hepatocellular carcinoma
(2014) Genomics, 103 (2-3), pp. 189-203.

DOI: 10.1016/j.ygeno.2014.01.003
DOCUMENT TYPE: Article

Hanscom, C., Talkowski, M.
Design of large-insert jumping libraries for structural variant detection using Illumina sequencing
(2014) Current Protocols in Human Genetics, (SUPPL.80), art. no. 7.22, . Cited 1 time.

DOI: 10.1002/0471142905.hg0722s80
DOCUMENT TYPE: Article

Martínez-Fundichely, A., Casillas, S., Egea, R., Ràmia, M., Barbadilla, A., Pantano, L., Puig, M., Cáceres, M.
InvFEST, a database integrating information of polymorphic inversions in the human genome
(2014) Nucleic Acids Research, 42 (D1), pp. D1027-D1032. Cited 4 times.

DOI: 10.1093/nar/gkt1122
DOCUMENT TYPE: Article

Chain, F.J.J., Feulner, P.G.D., Panchal, M., Eizaguirre, C., Samonte, I.E., Kalbe, M., Lenz, T.L., Stoll, M., Bornberg-Bauer, E., Milinski, M., Reusch, T.B.H.
Extensive Copy-Number Variation of Young Genes across Stickleback Populations
(2014) PLoS Genetics, 10 (12), 18 p. Cited 1 time.

DOI: 10.1371/journal.pgen.1004830
DOCUMENT TYPE: Article

Bickhart, D.M., Liu, G.E.
The challenges and importance of structural variation detection in livestock
(2014) Frontiers in Genetics, 5 (FEB), art. no. Article 37, . Cited 3 times.

DOI: 10.3389/fgene.2014.00037
DOCUMENT TYPE: Review

Steinberg, K.M., Schneider, V.A., Graves-Lindsay, T.A., Fulton, R.S., Agarwala, R., Huddleston, J., Shiryev, S.A., Morgulis, A., Surti, U., Warren, W.C., Church, D.M., Eichler, E.E., Wilson, R.K.
Single haplotype assembly of the human genome from a hydatidiform mole
(2014) Genome Research, 24 (12), pp. 2066-2076. Cited 1 time.

DOI: 10.1101/gr.180893.114
DOCUMENT TYPE: Article

Gao, J., Guan, R., Qi, F.
Strategies for improving accuracy of structural variation prediction using read pairs
(2014) Proceedings - 2013 International Conference on Information Science and Cloud Computing Companion, ISCC-C 2013, art. no. 6973636, pp. 463-468.

DOI: 10.1109/ISCC-C.2013.127
DOCUMENT TYPE: Conference Paper

Baik, J.Y., Lee, K.H.

Toward product attribute control: Developments from genome sequencing
(2014) Current Opinion in Biotechnology, 30, pp. 40-44. Cited 2 times.

DOI: 10.1016/j.copbio.2014.05.001
DOCUMENT TYPE: Review

Nuttle, X., Itsara, A., Shendure, J., Eichler, E.E.
Resolving genomic disorder-associated breakpoints within segmental DNA duplications using massively parallel sequencing
(2014) Nature Protocols, 9 (6), pp. 1496-1513. Cited 1 time.

DOI: 10.1038/nprot.2014.096
DOCUMENT TYPE: Article

Schwarz, R.F., Trinh, A., Sipos, B., Brenton, J.D., Goldman, N., Markowetz, F.
Phylogenetic Quantification of Intra-tumour Heterogeneity
(2014) PLoS Computational Biology, 10 (4), art. no. e1003535, . Cited 4 times.

DOI: 10.1371/journal.pcbi.1003535
DOCUMENT TYPE: Article

Zhang, L.-M., Luo, H., Liu, Z.-Q., Zhao, Y., Luo, J.-C., Hao, D.-Y., Jing, H.-C.
Genome-wide patterns of large-size presence/absence variants in sorghum
(2014) Journal of Integrative Plant Biology, 56 (1), pp. 24-37. Cited 2 times.

DOI: 10.1111/jipb.12121
DOCUMENT TYPE: Article

Liu, T., Xie, L., Ye, J., He, X.
Family-based analysis identified CD2 as a susceptibility gene for primary open angle glaucoma in Chinese Han population
(2014) Journal of Cellular and Molecular Medicine, 18 (4), pp. 600-609.

DOI: 10.1111/jcmm.12201
DOCUMENT TYPE: Article

Wilson-Sánchez, D., Rubio-Díaz, S., Muñoz-Viana, R., Pérez-Pérez, J.M., Jover-Gil, S., Ponce, M.R., Micol, J.L.
Leaf phenomics: A systematic reverse genetic screen for Arabidopsis leaf mutants
(2014) Plant Journal, 79 (5), pp. 878-891. Cited 1 time.

DOI: 10.1111/tpj.12595
DOCUMENT TYPE: Article

Ritz, A., Bashir, A., Sindi, S., Hsu, D., Hajirasouliha, I., Raphael, B.J.
Characterization of Structural variants with single molecule and hybrid sequencing approaches
(2014) Bioinformatics, 30 (24), pp. 3458-3466.

DOI: 10.1093/bioinformatics/btu714
DOCUMENT TYPE: Article

Muñoz-Minjares, J., Cabal-Aragón, J., Shmaliy, Y.S.
Confidence masks for genome DNA copy number variations in applications to HR-CGH array measurements
(2014) Biomedical Signal Processing and Control, 13 (1), pp. 337-344.

DOI: 10.1016/j.bspc.2014.06.006

DOCUMENT TYPE: Article

Rogers, R.L., Cridland, J.M., Shao, L., Hu, T.T., Andolfatto, P., Thornton, K.R.
Landscape of standing variation for tandem duplications in drosophila yakuba and drosophila simulans
(2014) Molecular Biology and Evolution, 31 (7), pp. 1750-1766.

DOI: 10.1093/molbev/msu124
DOCUMENT TYPE: Article

Li, J., Kannan, M., Trivett, A.L., Liao, H., Wu, X., Akagi, K., Symer, D.E.
An antisense promoter in mouse L1 retrotransposon open reading frame-1 initiates expression of diverse fusion transcripts and limits retrotransposition
(2014) Nucleic Acids Research, 42 (7), pp. 4546-4562. Cited 3 times.

DOI: 10.1093/nar/gku091
DOCUMENT TYPE: Article

Ma, J., Xiong, M., You, M., Lozano, G., Amos, C.I.
Genome-wide association tests of inversions with application to psoriasis
(2014) Human Genetics, 133 (8), pp. 967-974.

DOI: 10.1007/s00439-014-1437-1
DOCUMENT TYPE: Article

Kitchen, R.R., Rozowsky, J.S., Gerstein, M.B., Nairn, A.C.
Decoding neuroproteomics: Integrating the genome, translatome and functional anatomy
(2014) Nature Neuroscience, 17 (11), pp. 1491-1499.

DOI: 10.1038/nn.3829
DOCUMENT TYPE: Review

Keane, T.M., Wong, K., Adams, D.J., Flint, J., Reymond, A., Yalcin, B.
Identification of structural variation in mouse genomes
(2014) Frontiers in Genetics, 5 (JUL), art. no. Article 192, . Cited 1 time.

DOI: 10.3389/fgene.2014.00192
DOCUMENT TYPE: Review

Aguado, C., Gayà-Vidal, M., Villatoro, S., Oliva, M., Izquierdo, D., Giner-Delgado, C., Montalvo, V., García-González, J., Martínez-Fundichely, A., Capilla, L., Ruiz-Herrera, A., Estivill, X., Puig, M., Cáceres, M.
Validation and Genotyping of Multiple Human Polymorphic Inversions Mediated by Inverted Repeats Reveals a High Degree of Recurrence
(2014) PLoS Genetics, 10 (3), art. no. e1004208, .

DOI: 10.1371/journal.pgen.1004208
DOCUMENT TYPE: Article

Talkowski, M.E., Minikel, E.V., Gusella, J.F.
Autism spectrum disorder genetics: Diverse genes with diverse clinical outcomes
(2014) Harvard Review of Psychiatry, 22 (2), pp. 65-75. Cited 1 time.

DOI: 10.1097/HRP.0000000000000002
DOCUMENT TYPE: Review

Adam-Blondon, A.-F.
Grapevine genome update and beyond
(2014) Acta Horticulturae, 1046, pp. 311-318. Cited 1 time.

DOCUMENT TYPE: Article

Gillespie, R.L., O'Sullivan, J., Ashworth, J., Bhaskar, S., Williams, S., Biswas, S., Kehdi, E., Ramsden, S.C., Clayton-Smith, J., Black, G.C., Lloyd, I.C.
Personalized diagnosis and management of congenital cataract by next-generation sequencing
(2014) Ophthalmology, 121 (11), pp. 2124-2137.e2. Cited 1 time.

DOI: 10.1016/j.ophtha.2014.06.006
DOCUMENT TYPE: Article

Flynn, T.J., Phipps-Green, A., Hollis-Moffatt, J.E., Merriman, M.E., Topless, R., Montgomery, G., Chapman, B., Stamp, L.K., Dalbeth, N., Merriman, T.R.
Association analysis of the SLC22A11 (organic anion transporter 4) and SLC22A12 (urate transporter 1) urate transporter locus with gout in New Zealand case-control sample sets reveals multiple ancestral-specific effects
(2013) Arthritis Research and Therapy, 15 (6), art. no. R220, . Cited 1 time.

DOI: 10.1186/ar4417
DOCUMENT TYPE: Article

Zhang, C.-Z., Leibowitz, M.L., Pellman, D.
Chromothripsis and beyond: Rapid genome evolution from complex chromosomal rearrangements
(2013) Genes and Development, 27 (23), pp. 2513-2530. Cited 10 times.

DOI: 10.1101/gad.229559.113
DOCUMENT TYPE: Review

Kasaian, K., Li, Y.Y., Jones, S.J.M.
Bioinformatics for Cancer Genomics
(2013) Cancer Genomics: From Bench to Personalized Medicine, pp. 133-152.

DOI: 10.1016/B978-0-12-396967-5.00009-8
DOCUMENT TYPE: Book Chapter

Gao, J., Guan, R., Qi, F.
Methods for detecting genome structural variation based on PEM
(2013) Beijing Jiaotong Daxue Xuebao/Journal of Beijing Jiaotong University, 37 (6), pp. 8-12.

DOCUMENT TYPE: Article

Duan, J., Wan, M., Deng, H.-W., Wang, Y.-P.
Modeling exome sequencing data with generalized Gaussian distribution with application to copy number variation detection
(2013) Proceedings - 2013 IEEE International Conference on Bioinformatics and Biomedicine, IEEE BIBM 2013, art. no. 6732619, pp. 1-7.

DOI: 10.1109/BIBM.2013.6732619
DOCUMENT TYPE: Conference Paper

Livnat, A.

Interaction-based evolution: How natural selection and nonrandom mutation work together
(2013) Biology Direct, 8 (1), art. no. 24, . Cited 2 times.

DOI: 10.1186/1745-6150-8-24
DOCUMENT TYPE: Article

Plass, C., Pfister, S.M., Lindroth, A.M., Bogatyrova, O., Claus, R., Lichter, P.
Mutations in regulators of the epigenome and their connections to global chromatin patterns in cancer
(2013) Nature Reviews Genetics, 14 (11), pp. 765-780. Cited 40 times.

DOI: 10.1038/nrg3554
DOCUMENT TYPE: Review

Liu, B., Morrison, C.D., Johnson, C.S., Trump, D.L., Qin, M., Conroy, J.C., Wang, J., Liu, S.
Computational methods for detecting copy number variations in cancer genome using next generation sequencing: Principles and challenges
(2013) Oncotarget, 4 (11), pp. 1868-1881. Cited 3 times.

DOCUMENT TYPE: Article

Haraksingh, R.R., Snyder, M.P.
Impacts of variation in the human genome on gene regulation
(2013) Journal of Molecular Biology, 425 (21), pp. 3970-3977. Cited 10 times.

DOI: 10.1016/j.jmb.2013.07.015
DOCUMENT TYPE: Review

Ratnapriya, R., Swaroop, A.
Genetic architecture of retinal and macular degenerative diseases: The promise and challenges of next-generation sequencing
(2013) Genome Medicine, 5 (9), art. no. 84, . Cited 1 time.

DOI: 10.1186/gm488
DOCUMENT TYPE: Review

Sankaranarayanan, K., Taleei, R., Rahmanian, S., Nikjoo, H.
Ionizing radiation and genetic risks. XVII. Formation mechanisms underlying naturally occurring DNA deletions in the human genome and their potential relevance for bridging the gap between induced DNA double-strand breaks and deletions in irradiated germ cells
(2013) Mutation Research - Reviews in Mutation Research, 753 (2), pp. 114-130. Cited 5 times.

DOI: 10.1016/j.mrrev.2013.07.003
DOCUMENT TYPE: Review

Dong, Z.C., Chen, Y.
Transcriptomics: Advances and approaches
(2013) Science China Life Sciences, 56 (10), pp. 960-967.

DOI: 10.1007/s11427-013-4557-2
DOCUMENT TYPE: Review

Zhao, M., Wang, Q., Wang, Q., Jia, P., Zhao, Z.
Computational tools for copy number variation (CNV) detection using next-generation sequencing data: Features and perspectives

(2013) BMC Bioinformatics, 14 (SUPPL11), art. no. S1, . Cited 7 times.

DOI: 10.1186/1471-2105-14-S11-S1
DOCUMENT TYPE: Article

Yegnasubramanian, S.
Explanatory chapter: Next generation sequencing
(2013) Methods in Enzymology, 529, pp. 201-208.

DOI: 10.1016/B978-0-12-418687-3.00016-1
DOCUMENT TYPE: Article

Valsesia, A., Macé, A., Jacquemont, S., Beckmann, J.S., Kutalik, Z.
The growing importance of CNVs: New insights for detection and clinical interpretation
(2013) Frontiers in Genetics, 4 (MAY), art. no. Article 92, . Cited 6 times.

DOI: 10.3389/fgene.2013.00092
DOCUMENT TYPE: Review

Lehrach, H.
DNA sequencing methods in human genetics and disease research
(2013) F1000Prime Reports, 5, art. no. 34, .

DOI: 10.12703/P5-34
DOCUMENT TYPE: Article

Morey, M., Fernández-Marmiesse, A., Castiñeiras, D., Fraga, J.M., Couce, M.L., Cocho, J.A.
A glimpse into past, present, and future DNA sequencing
(2013) Molecular Genetics and Metabolism, 110 (1-2), pp. 3-24. Cited 11 times.

DOI: 10.1016/j.ymgme.2013.04.024
DOCUMENT TYPE: Review

Sakai, R., Moisse, M., Reumers, J., Aerts, J.
Pipit: Visualizing functional impacts of structural variations
(2013) Bioinformatics, 29 (17), pp. 2206-2207.

DOI: 10.1093/bioinformatics/btt367
DOCUMENT TYPE: Article

Geurts Van Kessel, A., Venkatachalam, R., Kuiper, R.P.
Colorectal Cancer
(2013) Genomic and Personalized Medicine, 2, pp. 722-732.

DOI: 10.1016/B978-0-12-382227-7.00062-8
DOCUMENT TYPE: Book Chapter

Lee, C.
Structural Genomic Variation in the Human Genome
(2013) Genomic and Personalized Medicine, 1, pp. 123-132.

DOI: 10.1016/B978-0-12-382227-7.00010-0
DOCUMENT TYPE: Book Chapter

Guffanti, G., Torri, F., Rasmussen, J., Clark, A.P., Lakatos, A., Turner, J.A., Fallon, J.H., Saykin, A.J., Weiner, M., Vawter, M.P., Knowles, J.A., Potkin, S.G., Macciardi, F.
Increased CNV-Region deletions in mild cognitive impairment (MCI) and Alzheimer's disease (AD) subjects in the ADNI sample
(2013) Genomics, 102 (2), pp. 112-122. Cited 1 time.

DOI: 10.1016/j.ygeno.2013.04.004
DOCUMENT TYPE: Review

Chen, G., Wang, C., Shi, L., Tong, W., Qu, X., Chen, J., Yang, J., Shi, C., Chen, L., Zhou, P., Lu, B., Shi, T.
Comprehensively identifying and characterizing the missing gene sequences in human reference genome with integrated analytic approaches
(2013) Human Genetics, 132 (8), pp. 899-911. Cited 1 time.

DOI: 10.1007/s00439-013-1300-9
DOCUMENT TYPE: Article

Guo, X., Brenner, M., Zhang, X., Laragione, T., Tai, S., Li, Y., Bu, J., Yin, Y., Shah, A.A., Kwan, K., Li, Y., Jun, W., Gulko, P.S.
Whole-genome sequences of DA and F344 rats with different susceptibilities to arthritis, autoimmunity, inflammation and cancer
(2013) Genetics, 194 (4), pp. 1017-1028. Cited 2 times.

DOI: 10.1534/genetics.113.153049
DOCUMENT TYPE: Article

Wellensiek, B.P., Larsen, A.C., Stephens, B., Kukurba, K., Waern, K., Briones, N., Liu, L., Snyder, M., Jacobs, B.L., Kumar, S., Chaput, J.C.
Genome-wide profiling of human cap-independent translation-enhancing elements
(2013) Nature Methods, 10 (8), pp. 747-750. Cited 1 time.

DOI: 10.1038/nmeth.2522
DOCUMENT TYPE: Article

Paudel, Y., Madsen, O., Megens, H.-J., Frantz, L.A.F., Bosse, M., Bastiaansen, J.W.M., Crooijmans, R.P.M.A., Groenen, M.A.M.
Evolutionary dynamics of copy number variation in pig genomes in the context of adaptation and domestication
(2013) BMC Genomics, 14 (1), art. no. 449, . Cited 11 times.

DOI: 10.1186/1471-2164-14-449
DOCUMENT TYPE: Article

El-Sayed Moustafa, J.S., Froguel, P.
From obesity genetics to the future of personalized obesity therapy
(2013) Nature Reviews Endocrinology, 9 (7), pp. 402-413. Cited 17 times.

DOI: 10.1038/nrendo.2013.57
DOCUMENT TYPE: Review

Kim, S., Medvedev, P., Paton, T.A., Bafna, V.
Reprever: Resolving low-copy duplicated sequences using template driven assembly
(2013) Nucleic Acids Research, 41 (12), . Cited 2 times.

DOI: 10.1093/nar/gkt339

DOCUMENT TYPE: Article

Zhou, W., Zhang, F., Chen, X., Shen, Y., Lupski, J.R., Jin, L.
Increased genome instability in human DNA segments with self-chains: Homology-induced structural variations via replicative mechanisms
(2013) Human Molecular Genetics, 22 (13), pp. 2642-2651. Cited 2 times.

DOI: 10.1093/hmg/ddt113
DOCUMENT TYPE: Article

Brunham, L.R., Hayden, M.R.
Hunting human disease genes: Lessons from the past, challenges for the future
(2013) Human Genetics, 132 (6), pp. 603-617. Cited 4 times.

DOI: 10.1007/s00439-013-1286-3
DOCUMENT TYPE: Review

Mardis, E.R.
Next-generation sequencing platforms
(2013) Annual Review of Analytical Chemistry, 6, pp. 287-303. Cited 49 times.

DOI: 10.1146/annurev-anchem-062012-092628
DOCUMENT TYPE: Article

Escaramís, G., Tornador, C., Bassaganyas, L., Rabionet, R., Tubio, J.M.C., Martínez-Fundichely, A., Cáceres, M., Gut, M., Ossowski, S., Estivill, X.
PeSV-Fisher: Identification of Somatic and Non-Somatic Structural Variants Using Next Generation Sequencing Data
(2013) PLoS ONE, 8 (5), art. no. e63377, . Cited 3 times.

DOI: 10.1371/journal.pone.0063377
DOCUMENT TYPE: Article

Ezawa, K., Landan, G., Graur, D.
Detecting negative selection on recurrent mutations using gene genealogy
(2013) BMC Genetics, 14, art. no. 37, .

DOI: 10.1186/1471-2156-14-37
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.
CNV-TV: A robust method to discover copy number variation from short sequencing reads
(2013) BMC Bioinformatics, 14 (1), art. no. 150, .

DOI: 10.1186/1471-2105-14-150
DOCUMENT TYPE: Article

Mwenifumbo, J.C., Marra, M.A.
Cancer genome-sequencing study design
(2013) Nature Reviews Genetics, 14 (5), pp. 321-332. Cited 19 times.

DOI: 10.1038/nrg3445
DOCUMENT TYPE: Review

Lucas Lledó, J.I., Cáceres, M.
On the Power and the Systematic Biases of the Detection of Chromosomal Inversions by Paired-End Genome Sequencing
(2013) PLoS ONE, 8 (4), art. no. e61292, . Cited 3 times.

DOI: 10.1371/journal.pone.0061292
DOCUMENT TYPE: Article

Bassaganyas, L., Riveira-Muñoz, E., García-Aragonés, M., González, J.R., Cáceres, M., Armengol, L., Estivill, X.
Worldwide population distribution of the common LCE3C-LCE3B deletion associated with psoriasis and other autoimmune disorders
(2013) BMC Genomics, 14 (1), art. no. 261, . Cited 2 times.

DOI: 10.1186/1471-2164-14-261
DOCUMENT TYPE: Article

Gahan, P.B.
Circulating nucleic acids in plasma and serum: Applications in diagnostic techniques for noninvasive prenatal diagnosis
(2013) International Journal of Women's Health, 5 (1), pp. 177-186. Cited 2 times.

DOI: 10.2147/IJWH.S34442
DOCUMENT TYPE: Review

Verdin, H., D'haene, B., Beysen, D., Novikova, Y., Menten, B., Sante, T., Lapunzina, P., Nevado, J., Carvalho, C.M.B., Lupski, J.R., de Baere, E.
Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain
(2013) PLoS Genetics, 9 (3), art. no. e1003358, . Cited 10 times.

DOI: 10.1371/journal.pgen.1003358
DOCUMENT TYPE: Article

Walter, V., Nobel, A.B., Hayes, D.N., Wright, F.A.
Identification of Recurrent DNA Copy Number Aberrations in Tumors
(2013) Statistical Diagnostics for Cancer: Analyzing High-Dimensional Data, 3, pp. 239-260.

DOI: 10.1002/9783527665471.ch13
DOCUMENT TYPE: Book Chapter

Ray, F.A., Zimmerman, E., Robinson, B., Cornforth, M.N., Bedford, J.S., Goodwin, E.H., Bailey, S.M.
Directional genomic hybridization for chromosomal inversion discovery and detection
(2013) Chromosome Research, 21 (2), pp. 165-174. Cited 2 times.

DOI: 10.1007/s10577-013-9345-0
DOCUMENT TYPE: Article

Van Den Bossche, M.J., Strazisar, M., Cammaerts, S., Liekens, A.M., Vandeweyer, G., Depreeuw, V., Mattheijssens, M., Lenaerts, A..-S., De Zutter, S., De Rijk, P., Sabbe, B., Del-Favero, J.
Identification of rare copy number variants in high burden schizophrenia families
(2013) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 162 (3), pp. 273-282. Cited 4 times.

DOI: 10.1002/ajmg.b.32146
DOCUMENT TYPE: Article

Lundin, S., Gruselius, J., Nystedt, B., Lexow, P., Käller, M., Lundeberg, J.

Hierarchical molecular tagging to resolve long continuous sequences by massively parallel sequencing
(2013) Scientific Reports, 3, art. no. 1186, .

DOI: 10.1038/srep01186
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.
Comparative Studies of Copy Number Variation Detection Methods for Next-Generation Sequencing Technologies
(2013) PLoS ONE, 8 (3), art. no. e59128, . Cited 24 times.

DOI: 10.1371/journal.pone.0059128
DOCUMENT TYPE: Article

Ragheb, M.N., Ford, C.B., Chase, M.R., Lin, P.L., Flynn, J.L., Fortune, S.M.
The mutation rate of mycobacterial repetitive unit loci in strains of M. tuberculosis from cynomolgus macaque infection
(2013) BMC Genomics, 14 (1), art. no. 145, . Cited 3 times.

DOI: 10.1186/1471-2164-14-145
DOCUMENT TYPE: Article

Singer, M.A.
Are chronic degenerative diseases part of the ageing process? Insights from comparative biology
(2013) Are Chronic Degenerative Diseases Part of the Ageing Process? Insights from Comparative Biology, pp. 1-217.

DOCUMENT TYPE: Book

Coonrod, E.M., Durtschi, J.D., Margraf, R.L., Voelkerding, K.V.
Developing genome and exome sequencing for candidate gene identification in inherited disorders: An integrated technical and bioinformatics approach
(2013) Archives of Pathology and Laboratory Medicine, 137 (3), pp. 415-433. Cited 11 times.

DOI: 10.5858/arpa.2012-0107-RA
DOCUMENT TYPE: Review

Grimm, D., Hagmann, J., Koenig, D., Weigel, D., Borgwardt, K.
Accurate indel prediction using paired-end short reads
(2013) BMC Genomics, 14 (1), art. no. 132, . Cited 10 times.

DOI: 10.1186/1471-2164-14-132
DOCUMENT TYPE: Article

Soon, W.W., Hariharan, M., Snyder, M.P.
High-throughput sequencing for biology and medicine
(2013) Molecular Systems Biology, 9, art. no. 640, . Cited 40 times.

DOI: 10.1038/msb.2012.61
DOCUMENT TYPE: Review

Hong, S.G., Dunbar, C.E., Winkler, T.
Assessing the risks of genotoxicity in the therapeutic development of induced pluripotent stem cells
(2013) Molecular Therapy, 21 (2), pp. 272-281. Cited 7 times.

DOI: 10.1038/mt.2012.255
DOCUMENT TYPE: Review

Haasl, R.J., Payseur, B.A.
Microsatellites as targets of natural selection
(2013) Molecular Biology and Evolution, 30 (2), pp. 285-298. Cited 8 times.

DOI: 10.1093/molbev/mss247
DOCUMENT TYPE: Article

Feulner, P.G.D., Chain, F.J.J., Panchal, M., Eizaguirre, C., Kalbe, M., Lenz, T.L., Mundry, M., Samonte, I.E., Stoll, M., Milinski, M., Reusch, T.B.H., Bornberg-Bauer, E.
Genome-wide patterns of standing genetic variation in a marine population of three-spined sticklebacks
(2013) Molecular Ecology, 22 (3), pp. 635-649. Cited 21 times.

DOI: 10.1111/j.1365-294X.2012.05680.x
DOCUMENT TYPE: Conference Paper

Gong, Q., Tao, Y., Yang, J.-R., Cai, J., Yuan, Y., Ruan, J., Yang, J., Liu, H., Li, W., Lu, X., Zhuang, S.-M., Wang, S.M., Wu, C.-I.
Identification of medium-sized genomic deletions with low coverage, mate-paired restricted tags
(2013) BMC Genomics, 14 (1), art. no. 51, .

DOI: 10.1186/1471-2164-14-51
DOCUMENT TYPE: Article

Schluth-Bolard, C., Labalme, A., Cordier, M.-P., Till, M., Nadeau, G., Tevissen, H., Lesca, G., Boutry-Kryza, N., Rossignol, S., Rocas, D., Dubruc, E., Edery, P., Sanlaville, D.
Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations
(2013) Journal of Medical Genetics, 50 (3), pp. 144-150. Cited 14 times.

DOI: 10.1136/jmedgenet-2012-101351
DOCUMENT TYPE: Article

Li, W., Olivier, M.
Current analysis platforms and methods for detecting copy number variation
(2013) Physiological Genomics, 45 (1), pp. 1-6. Cited 6 times.

DOI: 10.1152/physiolgenomics.00082.2012
DOCUMENT TYPE: Article

Li, H.
Systems genetics in "-omics" era: Current and future development
(2013) Theory in Biosciences, 132 (1), pp. 1-16. Cited 2 times.

DOI: 10.1007/s12064-012-0168-x
DOCUMENT TYPE: Review

Solieri, L., Dakal, T.C., Giudici, P.
Next-generation sequencing and its potential impact on food microbial genomics
(2013) Annals of Microbiology, 63 (1), pp. 21-37. Cited 6 times.

DOI: 10.1007/s13213-012-0478-8
DOCUMENT TYPE: Review

Kunz, M., Dannemann, M., Kelso, J.
High-throughput sequencing of the melanoma genome
(2013) Experimental Dermatology, 22 (1), pp. 10-17. Cited 8 times.

DOI: 10.1111/exd.12054
DOCUMENT TYPE: Article

Wittler, R.
Unraveling overlapping deletions by agglomerative clustering
(2013) BMC Genomics, 14, art. no. S12, . Cited 1 time.

DOI: 10.1186/1471-2164-14-S1-S12
DOCUMENT TYPE: Article

Schulte, I., Batty, E.M., Pole, J.C.M., Blood, K.A., Mo, S., Cooke, S.L., Ng, C., Howe, K.L., Chin, S.-F., Brenton, J.D., Caldas, C., Howarth, K.D., Edwards, P.A.W.
Structural analysis of the genome of breast cancer cell line ZR-75-30 identifies twelve expressed fusion genes
(2012) BMC Genomics, 13 (1), art. no. 719, . Cited 2 times.

DOI: 10.1186/1471-2164-13-719
DOCUMENT TYPE: Article

Xu, H., Luo, X., Qian, J., Pang, X., Song, J., Qian, G., Chen, J., Chen, S.
FastUniq: A Fast De Novo Duplicates Removal Tool for Paired Short Reads
(2012) PLoS ONE, 7 (12), art. no. e52249, . Cited 4 times.

DOI: 10.1371/journal.pone.0052249
DOCUMENT TYPE: Article

Zhou, S., Fu, Y., Li, J., He, L., Cai, X., Yan, Q., Rao, X., Huang, S., Li, G., Wang, Y., Xu, A.
HTS-PEG: A Method for High Throughput Sequencing of the Paired-Ends of Genomic Libraries
(2012) PLoS ONE, 7 (12), art. no. e52257, .

DOI: 10.1371/journal.pone.0052257
DOCUMENT TYPE: Article

Wu, X., Zhang, D., Li, G.
Insights into the regulation of human CNV-miRNAs from the view of their target genes
(2012) BMC Genomics, 13 (1), art. no. 707, . Cited 5 times.

DOI: 10.1186/1471-2164-13-707
DOCUMENT TYPE: Article

Talkowski, M.E., Ordulu, Z., Pillalamarri, V., Benson, C.B., Blumenthal, I., Connolly, S., Hanscom, C., Hussain, N., Pereira, S., Picker, J., Rosenfeld, J.A., Shaffer, L.G., Wilkins-Haug, L.E., Gusella, J.F., Morton, C.C.
Clinical diagnosis by whole-genome sequencing of a prenatal sample
(2012) New England Journal of Medicine, 367 (23), pp. 2226-2232. Cited 30 times.

DOI: 10.1056/NEJMoa1208594
DOCUMENT TYPE: Article

Alves, J.M., Lopes, A.M., Chikhi, L., Amorim, A.
On the structural plasticity of the human genome: Chromosomal inversions revisited

(2012) Current Genomics, 13 (8), pp. 623-632. Cited 4 times.

DOI: 10.2174/138920212803759703
DOCUMENT TYPE: Review

Pan, S., Caleshu, C.A., Dunn, K.E., Ashley, E.A.
Cardiac structural and sarcomere genes associated with cardiomyopathy exhibit marked intolerance of genetic variation
(2012) Circulation: Cardiovascular Genetics, 5 (6), pp. 602-610. Cited 15 times.

DOI: 10.1161/CIRCGENETICS.112.963421
DOCUMENT TYPE: Article

Chin, B.L., Ryan, O., Lewitter, F., Boone, C., Fink, G.R.
Genetic variation in Saccharomyces cerevisiae: Circuit diversification in a signal transduction network
(2012) Genetics, 192 (4), pp. 1523-1532. Cited 5 times.

DOI: 10.1534/genetics.112.145573
DOCUMENT TYPE: Article

Raphael, B.J.
Chapter 6: Structural Variation and Medical Genomics
(2012) PLoS Computational Biology, 8 (12), art. no. e1002821, . Cited 3 times.

DOI: 10.1371/journal.pcbi.1002821
DOCUMENT TYPE: Article

Milward, E.A., Daneshi, N., Johnstone, D.M.
Emerging real-time technologies in molecular medicine and the evolution of integrated 'pharmacomics' approaches to personalized medicine and drug discovery
(2012) Pharmacology and Therapeutics, 136 (3), pp. 295-304. Cited 2 times.

DOI: 10.1016/j.pharmthera.2012.08.008
DOCUMENT TYPE: Review

dela Paz, J.S., Stronghill, P.E., Douglas, S.J., Saravia, S., Hasenkampf, C.A., Riggs, C.D.
Chromosome Fragile Sites in Arabidopsis Harbor Matrix Attachment Regions That May Be Associated with Ancestral Chromosome Rearrangement Events
(2012) PLoS Genetics, 8 (12), art. no. e1003136, . Cited 1 time.

DOI: 10.1371/journal.pgen.1003136
DOCUMENT TYPE: Article

Hardiman, G.
Application of Ultra-High Throughput Sequencing and Microarray Technologies in Pharmacogenomics Testing
(2012) Therapeutic Drug Monitoring, pp. 143-159. Cited 1 time.

DOI: 10.1016/B978-0-12-385467-4.00007-5
DOCUMENT TYPE: Book Chapter

Elbaidouri, M., Panaud, O.
Genome-wide analysis of transposition using next generation sequencing technologies
(2012) Topics in Current Genetics, 24, pp. 59-70. Cited 2 times.

DOI: 10.1007/978-3-642-31842-9-4

DOCUMENT TYPE: Article

Gahan, P.B.
Biology of circulating nucleic acids and possible roles in diagnosis and treatment in diabetes and cancer
(2012) Infectious Disorders - Drug Targets, 12 (5), pp. 360-370. Cited 4 times.

DOI: 10.2174/187152612804142224
DOCUMENT TYPE: Article

Jones, M.J.K., Jallepalli, P.V.
Chromothripsis: Chromosomes in Crisis
(2012) Developmental Cell, 23 (5), pp. 908-917. Cited 22 times.

DOI: 10.1016/j.devcel.2012.10.010
DOCUMENT TYPE: Review

Schwartz, J.J., Lee, C., Hiatt, J.B., Adey, A., Shendure, J.
Capturing native long-range contiguity by in situ library construction and optical sequencing
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (46), pp. 18749-18754. Cited 3 times.

DOI: 10.1073/pnas.1202680109
DOCUMENT TYPE: Article

Falconer, E., Hills, M., Naumann, U., Poon, S.S.S., Chavez, E.A., Sanders, A.D., Zhao, Y., Hirst, M., Lansdorp, P.M.
DNA template strand sequencing of single-cells maps genomic rearrangements at high resolution
(2012) Nature Methods, 9 (11), pp. 1107-1112. Cited 17 times.

DOI: 10.1038/nmeth.2206
DOCUMENT TYPE: Article

Marian, A.J.
Challenges in Medical Applications of Whole Exome/Genome Sequencing Discoveries
(2012) Trends in Cardiovascular Medicine, 22 (8), pp. 219-223. Cited 8 times.

DOI: 10.1016/j.tcm.2012.08.001
DOCUMENT TYPE: Review

Han, J.S., Shao, S.
Circular retrotransposition products generated by a LINE retrotransposon
(2012) Nucleic Acids Research, 40 (21), pp. 10866-10877. Cited 3 times.

DOI: 10.1093/nar/gks859
DOCUMENT TYPE: Article

Liu, G.E., Bickhart, D.M.
Copy number variation in the cattle genome
(2012) Functional and Integrative Genomics, 12 (4), pp. 609-624. Cited 10 times.

DOI: 10.1007/s10142-012-0289-9
DOCUMENT TYPE: Review

Coughlin II, C.R., Scharer, G.H., Shaikh, T.H.

Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns
(2012) Genome Medicine, 4 (10), art. no. 80, . Cited 1 time.

DOI: 10.1186/gm381
DOCUMENT TYPE: Review

Mijušković, M., Brown, S.M., Tang, Z., Lindsay, C.R., Efstathiadis, E., Deriano, L., Roth, D.B.
A Streamlined Method for Detecting Structural Variants in Cancer Genomes by Short Read Paired-End Sequencing
(2012) PLoS ONE, 7 (10), art. no. e48314, . Cited 6 times.

DOI: 10.1371/journal.pone.0048314
DOCUMENT TYPE: Article

Hunter, R.G., Murakami, G., Dewell, S., Seligsohn, M., Baker, M.E.R., Datson, N.A., McEwen, B.S., Pfaff, D.W.
Acute stress and hippocampal histone H3 lysine 9 trimethylation, a retrotransposon silencing response
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (43), pp. 17657-17662. Cited 31 times.

DOI: 10.1073/pnas.1215810109
DOCUMENT TYPE: Article

Shibata, T.
Cancer genomics and pathology: All Together Now
(2012) Pathology International, 62 (10), pp. 647-659. Cited 3 times.

DOI: 10.1111/j.1440-1827.2012.02855.x
DOCUMENT TYPE: Review

Langley, C.H., Stevens, K., Cardeno, C., Lee, Y.C.G., Schrider, D.R., Pool, J.E., Langley, S.A., Suarez, C., Corbett-Detig, R.B., Kolaczkowski, B., Fang, S., Nista, P.M., Holloway, A.K., Kern, A.D., Dewey, C.N., Song, Y.S., Hahn, M.W., Begun, D.J.
Genomic variation in natural populations of Drosophila melanogaster
(2012) Genetics, 192 (2), pp. 533-598. Cited 65 times.

DOI: 10.1534/genetics.112.142018
DOCUMENT TYPE: Article

Simon, M.M., Mallon, A.-M., Howell, G.R., Reinholdt, L.G.
High throughput sequencing approaches to mutation discovery in the mouse
(2012) Mammalian Genome, 23 (9-10), pp. 499-513.

DOI: 10.1007/s00335-012-9424-0
DOCUMENT TYPE: Article

Ehli, E.A., Abdellaoui, A., Hu, Y., Hottenga, J.J., Kattenberg, M., Van Beijsterveldt, T., Bartels, M., Althoff, R.R., Xiao, X., Scheet, P., De Geus, E.J., Hudziak, J.J., Boomsma, D.I., Davies, G.E.
De novo and inherited CNVs in MZ twin pairs selected for discordance and concordance on Attention Problems
(2012) European Journal of Human Genetics, 20 (10), pp. 1037-1043. Cited 14 times.

DOI: 10.1038/ejhg.2012.49
DOCUMENT TYPE: Article

Charney, E.

Humans, fruit flies, and automatons
(2012) Behavioral and Brain Sciences, 35 (5), pp. 381-410.

DOI: 10.1017/S0140525X12001501
DOCUMENT TYPE: Article

Zhang, N.R., Siegmund, D.O.
Model selection for high-dimensional, multi-sequence change-point problems
(2012) Statistica Sinica, 22 (4), pp. 1507-1538. Cited 1 time.

DOI: 10.5705/ss.2010.257
DOCUMENT TYPE: Article

Yao, F., Ariyaratne, P.N., Hillmer, A.M., Lee, W.H., Li, G., Teo, A.S.M., Woo, X.Y., Zhang, Z., Chen, J.P., Poh, W.T., Zawack, K.F.B., Chan, C.S., Leong, S.T., Neo, S.C., Choi, P.S.D., Gao, S., Nagarajan, N., Thoreau, H., Shahab, A., Ruan, X., Cacheux-Rataboul, V., Wei, C.-L., Bourque, G., Sung, W.-K., Liu, E.T., Ruan, Y.
Long Span DNA Paired-End-Tag (DNA-PET) Sequencing Strategy for the Interrogation of Genomic Structural Mutations and Fusion-Point-Guided Reconstruction of Amplicons
(2012) PLoS ONE, 7 (9), art. no. e46152, . Cited 4 times.

DOI: 10.1371/journal.pone.0046152
DOCUMENT TYPE: Article

Asan, Geng, C., Chen, Y., Wu, K., Cai, Q., Wang, Y., Lang, Y., Cao, H., Yang, H., Wang, J., Zhang, X.
Paired-End Sequencing of Long-Range DNA Fragments for De Novo Assembly of Large, Complex Mammalian Genomes by Direct Intra-Molecule Ligation
(2012) PLoS ONE, 7 (9), art. no. e46211, . Cited 4 times.

DOI: 10.1371/journal.pone.0046211
DOCUMENT TYPE: Article

Soemedi, R., Wilson, I.J., Bentham, J., Darlay, R., Töpf, A., Zelenika, D., Cosgrove, C., Setchfield, K., Thornborough, C., Granados-Riveron, J., Blue, G.M., Breckpot, J., Hellens, S., Zwolinkski, S., Glen, E., Mamasoula, C., Rahman, T.J., Hall, D., Rauch, A., Devriendt, K., Gewillig, M., O'sullivan, J., Winlaw, D.S., Bu'lock, F., Brook, J.D., Bhattacharya, S., Lathrop, M., Santibanez-Koref, M., Cordell, H.J., Goodship, J.A., Keavney, B.D.
Contribution of global rare copy-number variants to the risk of sporadic congenital heart disease
(2012) American Journal of Human Genetics, 91 (3), pp. 489-501. Cited 43 times.

DOI: 10.1016/j.ajhg.2012.08.003
DOCUMENT TYPE: Article

Rolfe, P.A., Bernstein, D.A., Grisafi, P., Fink, G.R., Gifford, D.K.
Ruler arrays reveal haploid genomic structural variation
(2012) PLoS ONE, 7 (8), art. no. e43210, .

DOI: 10.1371/journal.pone.0043210
DOCUMENT TYPE: Article

Sun, S., Ke, R., Hughes, D., Nilsson, M., Andersson, D.I.
Genome-wide detection of spontaneous chromosomal rearrangements in bacteria
(2012) PLoS ONE, 7 (8), art. no. e42639, . Cited 7 times.

DOI: 10.1371/journal.pone.0042639
DOCUMENT TYPE: Article

Du, R., Lu, C., Jiang, Z., Li, S., Ma, R., An, H., Xu, M., An, Y., Xia, Y., Jin, L., Wang, X., Zhang, F.
Efficient typing of copy number variations in a segmental duplication-mediated rearrangement hotspot using multiplex competitive amplification
(2012) Journal of Human Genetics, 57 (8), pp. 545-551. Cited 12 times.

DOI: 10.1038/jhg.2012.66
DOCUMENT TYPE: Article

Jiang, H., Zeng, X., He, N.
An integrated view of current progress in copy number variations analysis of genome
(2012) Advanced Science Letters, 7, pp. 1-8. Cited 5 times.

DOI: 10.1166/asl.2012.3315
DOCUMENT TYPE: Review

Valencia, A., Hidalgo, M.
Getting personalized cancer genome analysis into the clinic: the challenges in bioinformatics
(2012) Genome Medicine, 4 (8), art. no. 61, . Cited 11 times.

DOI: 10.1186/gm362
DOCUMENT TYPE: Review

Casals, F., Idaghdour, Y., Hussin, J., Awadalla, P.
Next-generation sequencing approaches for genetic mapping of complex diseases
(2012) Journal of Neuroimmunology, 248 (1-2), pp. 10-22. Cited 9 times.

DOI: 10.1016/j.jneuroim.2011.12.017
DOCUMENT TYPE: Review

Belfield, E.J., Gan, X., Mithani, A., Brown, C., Jiang, C., Franklin, K., Alvey, E., Wibowo, A., Jung, M., Bailey, K., Kalwani, S., Ragoussis, J., Mott, R., Harberd, N.P.
Genome-wide analysis of mutations in mutant lineages selected following fast-neutron irradiation mutagenesis of Arabidopsis thaliana
(2012) Genome Research, 22 (7), pp. 1306-1315. Cited 12 times.

DOI: 10.1101/gr.131474.111
DOCUMENT TYPE: Article

Xing, M.-N., Zhang, X.-Z., Huang, H.
Application of metagenomic techniques in mining enzymes from microbial communities for biofuel synthesis
(2012) Biotechnology Advances, 30 (4), pp. 920-929. Cited 14 times.

DOI: 10.1016/j.biotechadv.2012.01.021
DOCUMENT TYPE: Review

Kim, S., Millard, S.P., Yu, C.-E., Leong, L., Radant, A., Dobie, D., Tsuang, D.W., Wijsman, E.M.
Inheritance Model Introduces Differential Bias in CNV Calls Between Parents and Offspring
(2012) Genetic Epidemiology, 36 (5), pp. 488-498. Cited 1 time.

DOI: 10.1002/gepi.21643
DOCUMENT TYPE: Article

González, J.R., Abellán, C., Abellán, J.J.

Bayesian model to detect phenotype-specific genes for copy number data
(2012) BMC Bioinformatics, 13 (1), art. no. 130, .

DOI: 10.1186/1471-2105-13-130
DOCUMENT TYPE: Article

Iskow, R.C., Gokcumen, O., Lee, C.
Exploring the role of copy number variants in human adaptation
(2012) Trends in Genetics, 28 (6), pp. 245-257. Cited 21 times.

DOI: 10.1016/j.tig.2012.03.002
DOCUMENT TYPE: Review

Van Den Bossche, M.J., Strazisar, M., De Bruyne, S., Bervoets, C., Lenaerts, A.-S., De Zutter, S., Nordin, A., Norrback, K.-F., Goossens, D., De Rijk, P., Green, E.K., Grozeva, D., Mendlewicz, J., Craddock, N., Sabbe, B.G., Adolfsson, R., Souery, D., Del-Favero, J.
Identification of a CACNA2D4 deletion in late onset bipolar disorder patients and implications for the involvement of voltage-dependent calcium channels in psychiatric disorders
(2012) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 159 B (4), pp. 465-475. Cited 3 times.

DOI: 10.1002/ajmg.b.32053
DOCUMENT TYPE: Article

Arlt, M.F., Wilson, T.E., Glover, T.W.
Replication stress and mechanisms of CNV formation
(2012) Current Opinion in Genetics and Development, 22 (3), pp. 204-210. Cited 23 times.

DOI: 10.1016/j.gde.2012.01.009
DOCUMENT TYPE: Review

Lai, B., Ding, R., Li, Y., Duan, L., Zhu, H.
A de novo metagenomic assembly program for shotgun DNA reads
(2012) Bioinformatics, 28 (11), art. no. bts162, pp. 1455-1462. Cited 14 times.

DOI: 10.1093/bioinformatics/bts162
DOCUMENT TYPE: Article

Doan, R., Cohen, N., Harrington, J., Veazy, K., Juras, R., Cothran, G., McCue, M.E., Skow, L., Dindot, S.V.
Identification of copy number variants in horses
(2012) Genome Research, 22 (5), pp. 899-907. Cited 13 times.

DOI: 10.1101/gr.128991.111
DOCUMENT TYPE: Article

Li, J., Harris, R.A., Cheung, S.W., Coarfa, C., Jeong, M., Goodell, M.A., White, L.D., Patel, A., Kang, S.-H., Shaw, C., Chinault, A.C., Gambin, T., Gambin, A., Lupski, J.R., Milosavljevic, A.
Genomic hypomethylation in the human germline associates with selective structural mutability in the human genome
(2012) PLoS Genetics, 8 (5), art. no. e1002692, . Cited 19 times.

DOI: 10.1371/journal.pgen.1002692
DOCUMENT TYPE: Article

Rossetti, S., Hopp, K., Sikkink, R.A., Sundsbak, J.L., Lee, Y.K., Kubly, V., Eckloff, B.W., Ward, C.J., Winearls, C.G., Torres, V.E., Harris, P.C.

Identification of gene mutations in autosomal dominant polycystic kidney disease through targeted resequencing
(2012) Journal of the American Society of Nephrology, 23 (5), pp. 915-933. Cited 27 times.

DOI: 10.1681/ASN.2011101032
DOCUMENT TYPE: Article

Furney, S.J., Gundem, G., Lopez-Bigas, N.
Oncogenomics methods and resources
(2012) Cold Spring Harbor Protocols, 7 (5), pp. 546-564. Cited 3 times.

DOI: 10.1101/pdb.top069229
DOCUMENT TYPE: Review

Weise, A., Mrasek, K., Klein, E., Mulatinho, M., Llerena Jr., J.C., Hardekopf, D., Pekova, S., Bhatt, S., Kosyakova, N., Liehr, T.
Microdeletion and Microduplication Syndromes
(2012) Journal of Histochemistry and Cytochemistry, 60 (5), pp. 346-358. Cited 21 times.

DOI: 10.1369/0022155412440001
DOCUMENT TYPE: Article

Jain, K.K.
Integration of Biotechnologies for the Development of Personalized Medicine
(2012) Pharmaceutical Biotechnology: Drug Discovery and Clinical Applications, pp. 553-580.

DOI: 10.1002/9783527632909.ch21
DOCUMENT TYPE: Book Chapter

Adelson, D.L.
Bovine Genome Architecture
(2012) Bovine Genomics, pp. 123-143.

DOI: 10.1002/9781118301739.ch10
DOCUMENT TYPE: Book Chapter

Itsara, A., Vissers, L.E.L.M., Steinberg, K.M., Meyer, K.J., Zody, M.C., Koolen, D.A., De Ligt, J., Cuppen, E., Baker, C., Lee, C., Graves, T.A., Wilson, R.K., Jenkins, R.B., Veltman, J.A., Eichler, E.E.
Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing
(2012) American Journal of Human Genetics, 90 (4), pp. 599-613. Cited 8 times.

DOI: 10.1016/j.ajhg.2012.02.013
DOCUMENT TYPE: Article

Rodríguez-Santiago, B., Armengol, L.
Next generation sequencing technology in pre- and postnatal genetic diagnosis [Tecnologías de secuenciación de nueva generación en diagnóstico genético pre- y postnatal]
(2012) Diagnostico Prenatal, 23 (2), pp. 56-66.

DOI: 10.1016/j.diapre.2012.02.001
DOCUMENT TYPE: Article

Chiang, C., Jacobsen, J.C., Ernst, C., Hanscom, C., Heilbut, A., Blumenthal, I., Mills, R.E., Kirby, A., Lindgren, A.M., Rudiger, S.R., McLaughlan, C.J., Bawden, C.S., Reid, S.J., Faull, R.L.M., Snell, R.G., Hall, I.M., Shen, Y., Ohsumi, T.K., Borowsky, M.L., Daly, M.J., Lee, C., Morton, C.C., MacDonald, M.E., Gusella, J.F., Talkowski, M.E.

Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration
(2012) Nature Genetics, 44 (4), pp. 390-397. Cited 61 times.

DOI: 10.1038/ng.2202
DOCUMENT TYPE: Article

Sindi, S.S., Önal, S., Peng, L.C., Wu, H.-T., Raphael, B.J.
An integrative probabilistic model for identification of structural variation in sequencing data
(2012) Genome Biology, 13 (3), art. no. R22, . Cited 24 times.

DOI: 10.1186/gb-2012-13-3-r22
DOCUMENT TYPE: Article

Yalcin, B., Wong, K., Bhomra, A., Goodson, M., Keane, T.M., Adams, D.J., Flint, J.
The fine-scale architecture of structural variants in 17 mouse genomes
(2012) Genome Biology, 13 (3), art. no. R18, . Cited 13 times.

DOI: 10.1186/gb-2012-13-3-r18
DOCUMENT TYPE: Article

Ariyadasa, R., Stein, N.
Advances in BAC-based physical mapping and map integration strategies in plants
(2012) Journal of Biomedicine and Biotechnology, 2012, art. no. 184854, . Cited 11 times.

DOI: 10.1155/2012/184854
DOCUMENT TYPE: Review

Jensen, H., Kjeldsen, E., Hjortdal, V.E.
Could submicroscopical chromosomal imbalances cause cono-truncal malformations in twins?
(2012) Congenital Heart Disease, 7 (2), pp. 170-177.

DOI: 10.1111/j.1747-0803.2011.00544.x
DOCUMENT TYPE: Article

Lee, H.J., Kweon, J., Kim, E., Kim, S., Kim, J.-S.
Targeted chromosomal duplications and inversions in the human genome using zinc finger nucleases
(2012) Genome Research, 22 (3), pp. 539-548. Cited 50 times.

DOI: 10.1101/gr.129635.111
DOCUMENT TYPE: Article

Dewey, F.E., Pan, S., Wheeler, M.T., Quake, S.R., Ashley, E.A.
DNA sequencing clinical applications of new DNA sequencing technologies
(2012) Circulation, 125 (7), pp. 931-944. Cited 15 times.

DOI: 10.1161/CIRCULATIONAHA.110.972828
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Veltman, J.A.
Impact of Genomewide Structural Variation on Gene Discovery
(2012) Gene Discovery for Disease Models, pp. 443-470.

DOI: 10.1002/9780470933947.ch21

DOCUMENT TYPE: Book Chapter

Cáceres, A., Sindi, S.S., Raphael, B.J., Cáceres, M., González, J.R.
Identification of polymorphic inversions from genotypes
(2012) BMC Bioinformatics, 13 (1), art. no. 28, . Cited 5 times.

DOI: 10.1186/1471-2105-13-28
DOCUMENT TYPE: Article

Koboldt, D.C., Larson, D.E., Chen, K., Ding, L., Wilson, R.K.
Massively parallel sequencing approaches for characterization of structural variation
(2012) Methods in Molecular Biology, 838, pp. 369-384. Cited 15 times.

DOI: 10.1007/978-1-61779-507-7_18
DOCUMENT TYPE: Article

Nowrousian, M., Teichert, I., Masloff, S., Kück, U.
Whole-genome sequencing of sordaria macrospora mutants identifies developmental genes
(2012) G3: Genes, Genomes, Genetics, 2 (2), pp. 261-270. Cited 17 times.

DOI: 10.1534/g3.111.001479
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Stankiewicz, P.
Microdeletion and microduplication syndromes
(2012) Methods in Molecular Biology, 838, pp. 29-75. Cited 20 times.

DOI: 10.1007/978-1-61779-507-7_2
DOCUMENT TYPE: Article

Pfundt, R., Veltman, J.A.
Structural genomic variation in intellectual disability
(2012) Methods in Molecular Biology, 838, pp. 77-95. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_3
DOCUMENT TYPE: Article

Sneddon, T.P., Church, D.M.
Online resources for genomic structural variation
(2012) Methods in Molecular Biology, 838, pp. 273-289. Cited 2 times.

DOI: 10.1007/978-1-61779-507-7_13
DOCUMENT TYPE: Article

Johansson, A.C.V., Feuk, L.
Characterizing and interpreting genetic variation from personal genome sequencing
(2012) Methods in Molecular Biology, 838, pp. 343-367. Cited 3 times.

DOI: 10.1007/978-1-61779-507-7_17
DOCUMENT TYPE: Article

Hollox, E.J.
The challenges of studying complex and dynamic regions of the human genome

(2012) Methods in Molecular Biology, 838, pp. 187-207. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_9
DOCUMENT TYPE: Article

Jiang, J., Li, J., Kwan, H., Au, C., Wan Law, P., Li, L., Kam, K., Lun Ling, J., Leung, F.C.
A cost-effective and universal strategy for complete prokaryotic genomic sequencing proposed by computer simulation
(2012) BMC Research Notes, 5, art. no. 80, . Cited 2 times.

DOI: 10.1186/1756-0500-5-80
DOCUMENT TYPE: Article

Hall, I.M., Quinlan, A.R.
Detection and interpretation of genomic structural variation in mammals
(2012) Methods in Molecular Biology, 838, pp. 225-248. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_11
DOCUMENT TYPE: Article

Iqbal, Z., Caccamo, M., Turner, I., Flicek, P., McVean, G.
De novo assembly and genotyping of variants using colored de Bruijn graphs
(2012) Nature Genetics, 44 (2), pp. 226-232. Cited 73 times.

DOI: 10.1038/ng.1028
DOCUMENT TYPE: Article

Dumanski, J.P., Piotrowski, A.
Structural genetic variation in the context of somatic mosaicism
(2012) Methods in Molecular Biology, 838, pp. 249-272. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_12
DOCUMENT TYPE: Article

Simmons, A.D., Carvalho, C.M.B., Lupski, J.R.
What have studies of genomic disorders taught us about our genome?
(2012) Methods in Molecular Biology, 838, pp. 1-27. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_1
DOCUMENT TYPE: Article

Dewal, N., Hu, Y., Freedman, M.L., LaFramboise, T., Pe'Er, I.
Calling amplified haplotypes in next generation tumor sequence data
(2012) Genome Research, 22 (2), pp. 362-374. Cited 4 times.

DOI: 10.1101/gr.122564.111
DOCUMENT TYPE: Article

Ceulemans, S., Van Der Ven, K., Del-Favero, J.
Targeted screening and validation of copy number variations
(2012) Methods in Molecular Biology, 838, pp. 311-328. Cited 6 times.

DOI: 10.1007/978-1-61779-507-7_15
DOCUMENT TYPE: Article

Chen, J., Kim, Y.C., Wang, S.M.
DGS (Ditag Genome Scanning) - A Restriction-Based Paired-End Sequencing Approach for Genome Structural Analysis
(2012) Tag-Based Next Generation Sequencing, pp. 277-285.

DOI: 10.1002/9783527644582.ch16
DOCUMENT TYPE: Book Chapter

Gogol-Döring, A., Chen, W.
An overview of the analysis of next generation sequencing data
(2012) Methods in Molecular Biology, 802, pp. 249-257. Cited 12 times.

DOI: 10.1007/978-1-61779-400-1_16
DOCUMENT TYPE: Article

Le Scouarnec, S., Gribble, S.M.
Characterising chromosome rearrangements: Recent technical advances in molecular cytogenetics
(2012) Heredity, 108 (1), pp. 75-85. Cited 22 times.

DOI: 10.1038/hdy.2011.100
DOCUMENT TYPE: Review

Xu, X., Liu, X., Ge, S., Jensen, J.D., Hu, F., Li, X., Dong, Y., Gutenkunst, R.N., Fang, L., Huang, L., Li, J., He, W., Zhang, G., Zheng, X., Zhang, F., Li, Y., Yu, C., Kristiansen, K., Zhang, X., Wang, J., Wright, M., McCouch, S., Nielsen, R., Wang, J., Wang, W.
Resequencing 50 accessions of cultivated and wild rice yields markers for identifying agronomically important genes
(2012) Nature Biotechnology, 30 (1), pp. 105-111. Cited 145 times.

DOI: 10.1038/nbt.2050
DOCUMENT TYPE: Article

Abeel, T., Van Parys, T., Saeys, Y., Galagan, J., Van De Peer, Y.
GenomeView: A next-generation genome browser
(2012) Nucleic Acids Research, 40 (2), . Cited 25 times.

DOI: 10.1093/nar/gkr995
DOCUMENT TYPE: Article

Severson, D.W., Behura, S.K.
Mosquito genomics: Progress and challenges
(2012) Annual Review of Entomology, 57, pp. 143-166. Cited 27 times.

DOI: 10.1146/annurev-ento-120710-100651
DOCUMENT TYPE: Article

Brooks, M.J., Rajasimha, H.K., Roger, J.E., Swaroop, A.
Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl -/- retinal transcriptomes
(2011) Molecular Vision, 17, pp. 3034-3054. Cited 27 times.

DOCUMENT TYPE: Article

Liu, Y.
Cancer and signaling pathway deregulation
(2011) Handbook of Research on Computational and Systems Biology: Interdisciplinary Applications, pp. 369-379.

DOI: 10.4018/978-1-60960-491-2.ch017
DOCUMENT TYPE: Book Chapter

Zhao, Z., Nguyen, T.C., Deng, N., Johnson, K.M., Zhu, D.
SPATA: A seeding and patching algorithm for de novo transcriptome assembly
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112351, pp. 26-33. Cited 1 time.

DOI: 10.1109/BIBMW.2011.6112351
DOCUMENT TYPE: Conference Paper

Zhang, J., Gao, Y., Zhao, X., Guan, M., Zhang, W., Wan, J., Yu, B.
Investigation of copy-number variations of C8orf4 in hematological malignancies
(2011) Medical Oncology, 28 (SUPPL. 1), pp. S647-S652. Cited 3 times.

DOI: 10.1007/s12032-010-9698-6
DOCUMENT TYPE: Article

Lai, A.G., Denton-Giles, M., Mueller-Roeber, B., Schippers, J.H.M., Dijkwel, P.P.
Positional information resolves structural variations and uncovers an evolutionarily divergent genetic locus in accessions of Arabidopsis thaliana
(2011) Genome Biology and Evolution, 3 (1), pp. 627-640. Cited 2 times.

DOI: 10.1093/gbe/evr038
DOCUMENT TYPE: Article

Schrider, D.R., Stevens, K., Cardeño, C.M., Langley, C.H., Hahn, M.W.
Genome-wide analysis of retrogene polymorphisms in Drosophila melanogaster
(2011) Genome Research, 21 (12), pp. 2087-2095. Cited 21 times.

DOI: 10.1101/gr.116434.110
DOCUMENT TYPE: Article

Hochstenbach, R., Buizer-Voskamp, J.E., Vorstman, J.A.S., Ophoff, R.A.
Genome arrays for the detection of copy number variations in idiopathic mental retardation, idiopathic generalized epilepsy and neuropsychiatric disorders: Lessons for diagnostic workflow and research
(2011) Cytogenetic and Genome Research, 135 (3-4), pp. 174-202. Cited 45 times.

DOI: 10.1159/000332928
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Lefante, J., Deng, H.-W., Wang, Y.-P.
Detection of copy number variation from next generation sequencing data with total variation penalized least square optimization
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112348, pp. 3-12. Cited 5 times.

DOI: 10.1109/BIBMW.2011.6112348
DOCUMENT TYPE: Conference Paper

Liu, X., Choi, S.-W., Wong, T.K.F., Lam, T.-W., Yiu, S.-M.
Detection of novel tandem duplication with next-generation sequencing
(2011) 2011 ACM Conference on Bioinformatics, Computational Biology and Biomedicine, BCB 2011, pp. 415-419.

DOI: 10.1145/2147805.2147861
DOCUMENT TYPE: Conference Paper

Marian, A.J.
Heart Failure as a Consequence of Restrictive Cardiomyopathy
(2011) Heart Failure, pp. 395-407.

DOI: 10.1016/B978-1-4160-5895-3.10025-7
DOCUMENT TYPE: Book Chapter

Girirajan, S., Campbell, C.D., Eichler, E.E.
Human copy number variation and complex genetic disease
(2011) Annual Review of Genetics, 45, pp. 203-226. Cited 70 times.

DOI: 10.1146/annurev-genet-102209-163544
DOCUMENT TYPE: Article

Kaganovich, M., Snyder, M.
Deciphering DNA Sequence Information
(2011) Genome Organization and Function in the Cell Nucleus, pp. 1-20.

DOI: 10.1002/9783527639991.ch1
DOCUMENT TYPE: Book Chapter

Greisman, H.A., Hoffman, N.G., Yi, H.S.
Rapid high-resolution mapping of balanced chromosomal rearrangements on tiling CGH arrays
(2011) Journal of Molecular Diagnostics, 13 (6), pp. 621-633. Cited 11 times.

DOI: 10.1016/j.jmoldx.2011.07.005
DOCUMENT TYPE: Article

Moon, S., Kim, Y.J., Hong, C.B., Kim, D.-J., Lee, J.-Y., Kim, B.-J.
Data-driven approach to detect common copy-number variations and frequency profiles in a population-based Korean cohort
(2011) European Journal of Human Genetics, 19 (11), pp. 1167-1172. Cited 6 times.

DOI: 10.1038/ejhg.2011.103
DOCUMENT TYPE: Article

Arlt, M.F., Ozdemir, A.C., Birkeland, S.R., Wilson, T.E., Glover, T.W.
Hydroxyurea induces de novo copy number variants in human cells
(2011) Proceedings of the National Academy of Sciences of the United States of America, 108 (42), pp. 17360-17365. Cited 28 times.

DOI: 10.1073/pnas.1109272108
DOCUMENT TYPE: Article

Wittler, R., Chauve, C.
Consistency-based detection of potential tumor-specific deletions in matched normal/tumor genomes
(2011) BMC Bioinformatics, 12 (SUPPL. 9), art. no. S21, . Cited 2 times.

DOI: 10.1186/1471-2105-12-S9-S21
DOCUMENT TYPE: Article

Gijsbers, A.C.J., Ruivenkamp, C.A.L.
Molecular karyotyping: From microscope to SNP arrays
(2011) Hormone Research in Paediatrics, 76 (3), pp. 208-213. Cited 5 times.

DOI: 10.1159/000330406
DOCUMENT TYPE: Article

Nelson, F.K., Snyder, M., Gardner, A.F., Hendrickson, C.L., Shendure, J.A., Porreca, G.J., Church, G.M., Ausubel, F.M., Ju, J., Kieleczawa, J., Slatko, B.E.
Introduction and historical overview of DNA sequencing
(2011) Current Protocols in Molecular Biology, (SUPPL.96), art. no. 7.0, .

DOI: 10.1002/0471142727.mb0700s96
DOCUMENT TYPE: Article

Torrezan, G.T., da Silva, F.C.C., Krepischi, A.C.V., Santos, T.M.M., de O Ferreira, F., Rossi, B.M., Carraro, D.M.
Breakpoint characterization of a novel large intragenic deletion of MUTYH detected in a MAP patient: Case report
(2011) BMC Medical Genetics, 12, art. no. 128, . Cited 6 times.

DOI: 10.1186/1471-2350-12-128
DOCUMENT TYPE: Article

Baetens, M., Van Laer, L., De Leeneer, K., Hellemans, J., De Schrijver, J., Van De Voorde, H., Renard, M., Dietz, H., Lacro, R.V., Menten, B., Van Criekinge, W., De Backer, J., De Paepe, A., Loeys, B., Coucke, P.J.
Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes
(2011) Human Mutation, 32 (9), pp. 1053-1062. Cited 14 times.

DOI: 10.1002/humu.21525
DOCUMENT TYPE: Article

Levin, H.L., Moran, J.V.
Dynamic interactions between transposable elements and their hosts
(2011) Nature Reviews Genetics, 12 (9), pp. 615-627. Cited 97 times.

DOI: 10.1038/nrg3030
DOCUMENT TYPE: Review

Saxena, A., Carninci, P.
Whole transcriptome analysis: What are we still missing?
(2011) Wiley Interdisciplinary Reviews: Systems Biology and Medicine, 3 (5), pp. 527-543. Cited 12 times.

DOI: 10.1002/wsbm.135
DOCUMENT TYPE: Review

Jiang, C., Mithani, A., Gan, X., Belfield, E.J., Klingler, J.P., Zhu, J.-K., Ragoussis, J., Mott, R., Harberd, N.P.
Regenerant arabidopsis lineages display a distinct genome-wide spectrum of mutations conferring variant phenotypes
(2011) Current Biology, 21 (16), pp. 1385-1390. Cited 16 times.

DOI: 10.1016/j.cub.2011.07.002
DOCUMENT TYPE: Article

Klein, J.D., Ossowski, S., Schneeberger, K., Weigel, D., Huson, D.H.

Locas - A low coverage assembly tool for resequencing projects
(2011) PLoS ONE, 6 (8), art. no. e23455, . Cited 8 times.

DOI: 10.1371/journal.pone.0023455
DOCUMENT TYPE: Article

Robinson, T., Campino, S.G., Auburn, S., Assefa, S.A., Polley, S.D., Manske, M., MacInnis, B., Rockett, K.A., Maslen, G.L., Sanders, M., Quail, M.A., Chiodini, P.L., Kwiatkowski, D.P., Clark, T.G., Sutherland, C.J.
Drug-resistant genotypes and multi-clonality in Plasmodium falciparum analysed by direct genome sequencing from peripheral blood of malaria patients
(2011) PLoS ONE, 6 (8), art. no. e23204, . Cited 22 times.

DOI: 10.1371/journal.pone.0023204
DOCUMENT TYPE: Article

Wang, J., Mullighan, C.G., Easton, J., Roberts, S., Heatley, S.L., Ma, J., Rusch, M.C., Chen, K., Harris, C.C., Ding, L., Holmfeldt, L., Payne-Turner, D., Fan, X., Wei, L., Zhao, D., Obenauer, J.C., Naeve, C., Mardis, E.R., Wilson, R.K., Downing, J.R., Zhang, J.
CREST maps somatic structural variation in cancer genomes with base-pair resolution
(2011) Nature Methods, 8 (8), pp. 652-654. Cited 99 times.

DOI: 10.1038/nmeth.1628
DOCUMENT TYPE: Article

Hampton, O.A., Miller, C.A., Koriabine, M., Li, J., Den Hollander, P., Carbone, L., Nefedov, M., Ten Hallers, B.F.H., Lee, A.V., De Jong, P.J., Milosavljevic, A.
Long-range massively parallel mate pair sequencing detects distinct mutations and similar patterns of structural mutability in two breast cancer cell lines
(2011) Cancer Genetics, 204 (8), pp. 447-457. Cited 6 times.

DOI: 10.1016/j.cancergen.2011.07.009
DOCUMENT TYPE: Article

Ku, C.-S., Teo, S.-M., Naidoo, N., Sim, X., Teo, Y.-Y., Pawitan, Y., Seielstad, M., Chia, K.-S., Salim, A.
Copy number polymorphisms in new HapMap III and Singapore populations
(2011) Journal of Human Genetics, 56 (8), pp. 552-560.

DOI: 10.1038/jhg.2011.54
DOCUMENT TYPE: Article

Li, Y., Zheng, H., Luo, R., Wu, H., Zhu, H., Li, R., Cao, H., Wu, B., Huang, S., Shao, H., Ma, H., Zhang, F., Feng, S., Zhang, W., Du, H., Tian, G., Li, J., Zhang, X., Li, S., Bolund, L., Kristiansen, K., De Smith, A.J., Blakemore, A.I.F., Coin, L.J.M., Yang, H., Wang, J., Wang, J.
Structural variation in two human genomes mapped at single-nucleotide resolution by whole genome de novo assembly
(2011) Nature Biotechnology, 29 (8), pp. 723-730. Cited 55 times.

DOI: 10.1038/nbt.1904
DOCUMENT TYPE: Article

Parrish, N., Hormozdiari, F., Eskin, E.
Assembly of non-unique insertion content using next-generation sequencing
(2011) BMC Bioinformatics, 12 (SUPPL.6), art. no. S3, . Cited 3 times.

DOI: 10.1186/1471-2105-12-S6-S3
DOCUMENT TYPE: Article

Yu, P., Wang, C., Xu, Q., Feng, Y., Yuan, X., Yu, H., Wang, Y., Tang, S., Wei, X.
Detection of copy number variations in rice using array-based comparative genomic hybridization
(2011) BMC Genomics, 12, art. no. 372, . Cited 15 times.

DOI: 10.1186/1471-2164-12-372
DOCUMENT TYPE: Article

Beck, C.R., Garcia-Perez, J.L., Badge, R.M., Moran, J.V.
LINE-1 elements in structural variation and disease
(2011) Annual Review of Genomics and Human Genetics, 12, pp. 187-215. Cited 75 times.

DOI: 10.1146/annurev-genom-082509-141802
DOCUMENT TYPE: Review

Pole, J.C.M., McCaughan, F., Newman, S., Howarth, K.D., Dear, P.H., Edwards, P.A.W.
Single-molecule analysis of genome rearrangements in cancer
(2011) Nucleic Acids Research, 39 (13), . Cited 1 time.

DOI: 10.1093/nar/gkr227
DOCUMENT TYPE: Article

Casbon, J.A., Osborne, R.J., Brenner, S., Lichtenstein, C.P.
A method for counting PCR template molecules with application to next-generation sequencing
(2011) Nucleic Acids Research, 39 (12), . Cited 30 times.

DOI: 10.1093/nar/gkr217
DOCUMENT TYPE: Article

Novais, R.C., Thorstenson, Y.R.
The evolution of Pyrosequencing® for microbiology: From genes to genomes
(2011) Journal of Microbiological Methods, 86 (1), pp. 1-7. Cited 15 times.

DOI: 10.1016/j.mimet.2011.04.006
DOCUMENT TYPE: Review

Teo, S.-M., Ku, C.-S., Naidoo, N., Hall, P., Chia, K.-S., Salim, A., Pawitan, Y.
A population-based study of copy number variants and regions of homozygosity in healthy Swedish individuals
(2011) Journal of Human Genetics, 56 (7), pp. 524-533. Cited 6 times.

DOI: 10.1038/jhg.2011.52
DOCUMENT TYPE: Article

Qi, J., Zhao, F.
InGAP-sv: A novel scheme to identify and visualize structural variation from paired end mapping data
(2011) Nucleic Acids Research, 39 (SUPPL. 2), pp. W567-W575. Cited 23 times.

DOI: 10.1093/nar/gkr506
DOCUMENT TYPE: Article

Alverson, A.J., Rice, D.W., Dickinson, S., Barry, K., Palmer, J.D.
Origins and recombination of the bacterial-sized multichromosomal mitochondrial genome of cucumber
(2011) Plant Cell, 23 (7), pp. 2499-2513. Cited 40 times.

DOI: 10.1105/tpc.111.087189
DOCUMENT TYPE: Article

Schneeberger, K., Ossowski, S., Ott, F., Klein, J.D., Wang, X., Lanz, C., Smith, L.M., Cao, J., Fitz, J., Warthmann, N., Henz, S.R., Huson, D.H., Weigel, D.
Reference-guided assembly of four diverse Arabidopsis thaliana genomes
(2011) Proceedings of the National Academy of Sciences of the United States of America, 108 (25), pp. 10249-10254. Cited 74 times.

DOI: 10.1073/pnas.1107739108
DOCUMENT TYPE: Article

Dong, H., Chen, Y., Shen, Y., Wang, S., Zhao, G., Jin, W.
Artificial duplicate reads in sequencing data of 454 genome sequencer FLX system
(2011) Acta Biochimica et Biophysica Sinica, 43 (6), pp. 496-500. Cited 10 times.

DOI: 10.1093/abbs/gmr030
DOCUMENT TYPE: Article

Mills, R.E., Pittard, W.S., Mullaney, J.M., Farooq, U., Creasy, T.H., Mahurkar, A.A., Kemeza, D.M., Strassler, D.S., Ponting, C.P., Webber, C., Devine, S.E.
Natural genetic variation caused by small insertions and deletions in the human genome
(2011) Genome Research, 21 (6), pp. 830-839. Cited 68 times.

DOI: 10.1101/gr.115907.110
DOCUMENT TYPE: Article

Richter, D.C., Ott, F., Auch, A.F., Schmid, R., Huson, D.H.
MetaSim: A Sequencing Simulator for Genomics and Metagenomics
(2011) Handbook of Molecular Microbial Ecology I: Metagenomics and Complementary Approaches, pp. 417-421.

DOI: 10.1002/9781118010518.ch48
DOCUMENT TYPE: Book Chapter

Kloosterman, W.P., Guryev, V., van Roosmalen, M., Duran, K.J., de Bruijn, E., Bakker, S.C.M., Letteboer, T., van Nesselrooij, B., Hochstenbach, R., Poot, M., Cuppen, E.
Chromothripsis as a mechanism driving complex de novo structural rearrangements in the germline
(2011) Human Molecular Genetics, 20 (10), art. no. ddr073, pp. 1916-1924. Cited 65 times.

DOI: 10.1093/hmg/ddr073
DOCUMENT TYPE: Article

Bolon, Y.-T., Haun, W.J., Xu, W.W., Grant, D., Stacey, M.G., Nelson, R.T., Gerhardt, D.J., Jeddeloh, J.A., Stacey, G., Muehlbauer, G.J., Orf, J.H., Naeve, S.L., Stupar, R.M., Vance, C.P.
Phenotypic and genomic analyses of a fast neutron mutant population resource in soybean
(2011) Plant Physiology, 156 (1), pp. 240-253. Cited 36 times.

DOI: 10.1104/pp.110.170811
DOCUMENT TYPE: Article

Inaki, K., Hillmer, A.M., Ukil, L., Yao, F., Woo, X.Y., Vardy, L.A., Zawack, K.F.B., Lee, C.W.H., Ariyaratne, P.N., Chan, Y.S., Desai, K.V., Bergh, J., Hall, P., Putti, T.C., Ong, W.L., Shahab, A., Cacheux-Rataboul, V., Karuturi, R.K.M., Sung, W.-K., Ruan, X., Bourque, G., Ruan, Y., Liu, E.T.

Transcriptional consequences of genomic structural aberrations in breast cancer
(2011) Genome Research, 21 (5), pp. 676-687. Cited 31 times.

DOI: 10.1101/gr.113225.110
DOCUMENT TYPE: Article

Fang, Z., Cui, X.
Design and validation issues in RNA-seq experiments
(2011) Briefings in Bioinformatics, 12 (3), art. no. bbr004, pp. 280-287. Cited 39 times.

DOI: 10.1093/bib/bbr004
DOCUMENT TYPE: Article

Alkan, C., Coe, B.P., Eichler, E.E.
Genome structural variation discovery and genotyping
(2011) Nature Reviews Genetics, 12 (5), pp. 363-376. Cited 257 times.

DOI: 10.1038/nrg2958
DOCUMENT TYPE: Review

Hu, T.T., Pattyn, P., Bakker, E.G., Cao, J., Cheng, J.-F., Clark, R.M., Fahlgren, N., Fawcett, J.A., Grimwood, J., Gundlach, H., Haberer, G., Hollister, J.D., Ossowski, S., Ottilar, R.P., Salamov, A.A., Schneeberger, K., Spannagl, M., Wang, X., Yang, L., Nasrallah, M.E., Bergelson, J., Carrington, J.C., Gaut, B.S., Schmutz, J., Mayer, K.F.X., Van De Peer, Y., Grigoriev, I.V., Nordborg, M., Weigel, D., Guo, Y.-L.
The Arabidopsis lyrata genome sequence and the basis of rapid genome size change
(2011) Nature Genetics, 43 (5), pp. 476-483. Cited 222 times.

DOI: 10.1038/ng.807
DOCUMENT TYPE: Article

Hillmer, A.M., Yao, F., Inaki, K., Lee, W.H., Ariyaratne, P.N., Teo, A.S.M., Woo, X.Y., Zhang, Z., Zhao, H., Ukil, L., Chen, J.P., Zhu, F., So, J.B.Y., Salto-Tellez, M., Poh, W.T., Zawack, K.F.B., Nagarajan, N., Gao, S., Li, G., Kumar, V., Lim, H.P.J., Sia, Y.Y., Chan, C.S., Leong, S.T., Neo, S.C., Choi, P.S.D., Thoreau, H., Tan, P.B.O., Shahab, A., Ruan, X., Bergh, J., Hall, P., Cacheux-Rataboul, V., Wei, C.-L., Yeoh, K.G., Sung, W.-K., Bourque, G., Liu, E.T., Ruan, Y.
Comprehensive long-span paired-end-tag mapping reveals characteristic patterns of structural variations in epithelial cancer genomes
(2011) Genome Research, 21 (5), pp. 665-675. Cited 36 times.

DOI: 10.1101/gr.113555.110
DOCUMENT TYPE: Article

Pyon, Y.S., Hayes, M., Li, J.
Model based clustering approach for identifying structural variation using next generation sequencing data
(2011) 2011 IEEE 1st International Conference on Computational Advances in Bio and Medical Sciences, ICCABS 2011, art. no. 5729871, pp. 153-158.

DOI: 10.1109/ICCABS.2011.5729871
DOCUMENT TYPE: Conference Paper

Nord, A.S., Lee, M., King, M.-C., Walsh, T.
Accurate and exact CNV identification from targeted high-throughput sequence data
(2011) BMC Genomics, 12, art. no. 184, . Cited 40 times.

DOI: 10.1186/1471-2164-12-184
DOCUMENT TYPE: Article

Talkowski, M.E., Ernst, C., Heilbut, A., Chiang, C., Hanscom, C., Lindgren, A., Kirby, A., Liu, S., Muddukrishna, B., Ohsumi, T.K., Shen, Y., Borowsky, M., Daly, M.J., Morton, C.C., Gusella, J.F.
Next-generation sequencing strategies enable routine detection of balanced chromosome rearrangements for clinical diagnostics and genetic research
(2011) American Journal of Human Genetics, 88 (4), pp. 469-481. Cited 49 times.

DOI: 10.1016/j.ajhg.2011.03.013
DOCUMENT TYPE: Article

Kitada, K., Taima, A., Ogasawara, K., Metsugi, S., Aikawa, S.
Chromosome-specific segmentation revealed by structural analysis of individually isolated chromosomes
(2011) Genes Chromosomes and Cancer, 50 (4), pp. 217-227. Cited 10 times.

DOI: 10.1002/gcc.20847
DOCUMENT TYPE: Article

Sabot, F., Picault, N., El-Baidouri, M., Llauro, C., Chaparro, C., Piegu, B., Roulin, A., Guiderdoni, E., Delabastide, M., McCombie, R., Panaud, O.
Transpositional landscape of the rice genome revealed by paired-end mapping of high-throughput re-sequencing data
(2011) Plant Journal, 66 (2), pp. 241-246. Cited 25 times.

DOI: 10.1111/j.1365-313X.2011.04492.x
DOCUMENT TYPE: Article

Ku, C.-S., Naidoo, N., Pawitan, Y.
Revisiting Mendelian disorders through exome sequencing
(2011) Human Genetics, 129 (4), pp. 351-370. Cited 109 times.

DOI: 10.1007/s00439-011-0964-2
DOCUMENT TYPE: Review

Koike, A., Nishida, N., Yamashita, D., Tokunaga, K.
Comparative analysis of copy number variation detection methods and database construction
(2011) BMC Genetics, 12, art. no. 29, . Cited 6 times.

DOI: 10.1186/1471-2156-12-29
DOCUMENT TYPE: Article

Fode, P., Jespersgaard, C., Hardwick, R.J., Bogle, H., Theisen, M., Dodoo, D., Lenicek, M., Vitek, L., Vieira, A., Freitas, J., Andersen, P.S., Hollox, E.J.
Determination of beta-defensin genomic copy number in different populations: A comparison of three methods
(2011) PLoS ONE, 6 (2), art. no. e16768, . Cited 22 times.

DOI: 10.1371/journal.pone.0016768
DOCUMENT TYPE: Article

Helyar, S.J., Hemmer-Hansen, J., Bekkevold, D., Taylor, M.I., Ogden, R., Limborg, M.T., Cariani, A., Maes, G.E., Diopere, E., Carvalho, G.R., Nielsen, E.E.
Application of SNPs for population genetics of nonmodel organisms: New opportunities and challenges
(2011) Molecular Ecology Resources, 11 (SUPPL. 1), pp. 123-136. Cited 97 times.

DOI: 10.1111/j.1755-0998.2010.02943.x
DOCUMENT TYPE: Article

De Leeneer, K., Hellemans, J., De Schrijver, J., Baetens, M., Poppe, B., Van Criekinge, W., De Paepe, A., Coucke, P., Claes, K.
Massive parallel amplicon sequencing of the breast cancer genes BRCA1 and BRCA2: Opportunities, challenges, and limitations
(2011) Human Mutation, 32 (3), pp. 335-344. Cited 30 times.

DOI: 10.1002/humu.21428
DOCUMENT TYPE: Article

Handsaker, R.E., Korn, J.M., Nemesh, J., McCarroll, S.A.
Discovery and genotyping of genome structural polymorphism by sequencing on a population scale
(2011) Nature Genetics, 43 (3), pp. 269-276. Cited 94 times.

DOI: 10.1038/ng.768
DOCUMENT TYPE: Article

Männik, K., Parkel, S., Palta, P., Žilina, O., Puusepp, H., Esko, T., Mägi, R., Nõukas, M., Veidenberg, A., Nelis, M., Metspalu, A., Remm, M., Õunap, K., Kurg, A.
A parallel SNP array study of genomic aberrations associated with mental retardation in patients and general population in Estonia
(2011) European Journal of Medical Genetics, 54 (2), pp. 136-143. Cited 5 times.

DOI: 10.1016/j.ejmg.2010.11.005
DOCUMENT TYPE: Article

Mardis, E.R.
A decade's perspective on DNA sequencing technology
(2011) Nature, 470 (7333), pp. 198-203. Cited 238 times.

DOI: 10.1038/nature09796
DOCUMENT TYPE: Review

Kerstens, H.H.D., Crooijmans, R.P.M.A., Dibbits, B.W., Vereijken, A., Okimoto, R., Groenen, M.A.M.
Structural variation in the chicken genome identified by paired-end next-generation DNA sequencing of reduced representation libraries
(2011) BMC Genomics, 12, art. no. 94, . Cited 10 times.

DOI: 10.1186/1471-2164-12-94
DOCUMENT TYPE: Article

Wijchers, P.J., de Laat, W.
Genome organization influences partner selection for chromosomal rearrangements
(2011) Trends in Genetics, 27 (2), pp. 63-71. Cited 21 times.

DOI: 10.1016/j.tig.2010.11.001
DOCUMENT TYPE: Review

Roth, L., Marschalek, R., Oldenburg, J., Oyen, F., Schneppenheim, R.
Characterisation of two novel large F8 deletions in patients with severe haemophilia A and factor VIII inhibitors
(2011) Thrombosis and Haemostasis, 105 (2), pp. 279-284. Cited 2 times.

DOI: 10.1160/TH10-09-0570
DOCUMENT TYPE: Article

Ozsolak, F., Milos, P.M.
RNA sequencing: Advances, challenges and opportunities
(2011) Nature Reviews Genetics, 12 (2), pp. 87-98. Cited 404 times.

DOI: 10.1038/nrg2934
DOCUMENT TYPE: Review

Guan, M., Liu, L., Zhao, X., Wu, Q., Yu, B., Shao, Y., Yang, H., Fu, X., Wan, J., Zhang, W.
Copy number variations of EphA3 are associated with multiple types of hematologic malignancies
(2011) Clinical Lymphoma, Myeloma and Leukemia, 11 (1), pp. 50-53. Cited 11 times.

DOI: 10.3816/CLML.2011.n.006
DOCUMENT TYPE: Article

Guo, X., Freyer, L., Morrow, B., Zheng, D.
Characterization of the past and current duplication activities in the human 22q11.2 region
(2011) BMC Genomics, 12, art. no. 71, . Cited 5 times.

DOI: 10.1186/1471-2164-12-71
DOCUMENT TYPE: Article

Balasubramanian, S., Habegger, L., Frankish, A., MacArthur, D.G., Harte, R., Tyler-Smith, C., Harrow, J., Gerstein, M.
Gene inactivation and its implications for annotation in the era of personal genomics
(2011) Genes and Development, 25 (1), pp. 1-10. Cited 14 times.

DOI: 10.1101/gad.1968411
DOCUMENT TYPE: Review

Wan, J., Gao, Y., Zhao, X., Wu, Q., Fu, X., Shao, Y., Yang, H., Guan, M., Yu, B., Zhang, W.
The association between the copy-number variations of ZMAT4 and hematological malignancy
(2011) Hematology, 16 (1), pp. 20-23.

DOI: 10.1179/102453311X12902908411751
DOCUMENT TYPE: Article

Hong, D., Park, S.-S., Ju, Y.S., Kim, S., Shin, J.-Y., Kim, S., Yu, S.-B., Lee, W.-C., Lee, S., Park, H., Kim, J.-I., Seo, J.-S.
TIARA: A database for accurate analysis of multiple personal genomes based on cross-technology
(2011) Nucleic Acids Research, 39 (SUPPL. 1), pp. D883-D888. Cited 12 times.

DOI: 10.1093/nar/gkq1101
DOCUMENT TYPE: Article

Naidoo, N., Pawitan, Y., Soong, R., Cooper, D.N., Ku, C.-S.
Human genetics and genomics a decade after the release of the draft sequence of the human genome
(2011) Human Genomics, 5 (6), pp. 577-622. Cited 6 times.

DOI: 10.1186/1479-7364-5-6-577
DOCUMENT TYPE: Review

Ku, C.S., Naidoo, N., Teo, S.M., Pawitan, Y.
Regions of homozygosity and their impact on complex diseases and traits
(2011) Human Genetics, 129 (1), pp. 1-15. Cited 22 times.

DOI: 10.1007/s00439-010-0920-6
DOCUMENT TYPE: Review

Kitzman, J.O., MacKenzie, A.P., Adey, A., Hiatt, J.B., Patwardhan, R.P., Sudmant, P.H., Ng, S.B., Alkan, C., Qiu, R., Eichler, E.E., Shendure, J.
Haplotype-resolved genome sequencing of a Gujarati Indian individual
(2011) Nature Biotechnology, 29 (1), pp. 59-64. Cited 97 times.

DOI: 10.1038/nbt.1740
DOCUMENT TYPE: Article

Mihály, Z., Gyorffy, B.
Next generation sequencing technologies (NGST) development and applications [Következo generációs szekvenálási technológiák kifejlodése és alkalmazásai]
(2011) Orvosi Hetilap, 152 (2), pp. 55-62.

DOI: 10.1556/OH.2011.29007
DOCUMENT TYPE: Article

Kang, S., Hong, S.
Prediction of personalized drugs based on genetic variations provided by DNA sequencing technologies
(2011) Genes and Genomics, 33 (6), pp. 591-603. Cited 1 time.

DOI: 10.1007/s13258-011-0124-z
DOCUMENT TYPE: Review

Lee, S., Xing, E., Brudno, M.
MoGUL: Detecting common insertions and deletions in a population
(2010) Lecture Notes in Computer Science (including subseries Lecture Notes in Artificial Intelligence and Lecture Notes in Bioinformatics), 6044 LNBI, pp. 357-368. Cited 5 times.

DOI: 10.1007/978-3-642-12683-3_23
DOCUMENT TYPE: Conference Paper

Hérault, Y., Duchon, A., Maréchal, D., Raveau, M., Pereira, P.L., Dalloneau, E., Brault, V.
Controlled somatic and germline copy number variation in the mouse model
(2010) Current Genomics, 11 (6), pp. 470-480.

DOCUMENT TYPE: Article

Mkrtchyan, H., Gross, M., Hinreiner, S., Polytiko, A., Manvelyan, M., Mrasek, K., Kosyakova, N., Ewers, E., Nelle, H., Liehr, T., Bhatt, S., Thoma, K., Gebhart, E., Wilhelm, S., Fahsold, R., Volleth, M., Weise, A.
The human genome puzzle - the role of copy number variation in somatic mosaicism
(2010) Current Genomics, 11 (6), pp. 426-431. Cited 11 times.

DOCUMENT TYPE: Article

He, D., Furlotte, N., Eskin, E.
Detection and reconstruction of tandemly organized de novo copy number variations
(2010) BMC Bioinformatics, 11 (SUPPL. 11), art. no. S12, . Cited 4 times.

DOI: 10.1186/1471-2105-11-S11-S12
DOCUMENT TYPE: Article

Yavaş, G., Koyutürk, M., Özsoyoğlu, M., Gould, M.P., Laframboise, T.
Çokgen: A software for the identification of rare copy number variation from SNP microarrays
(2010) Pacific Symposium on Biocomputing 2010, PSB 2010, pp. 371-382. Cited 3 times.

DOCUMENT TYPE: Conference Paper

Zhang, Q., Davis, J.C., Dove, C.G., Su, H.C.
Genetic, clinical, and laboratory markers for DOCK8 immunodeficiency syndrome
(2010) Disease Markers, 29 (3-4), pp. 131-139. Cited 26 times.

DOI: 10.3233/DMA-2010-0737
DOCUMENT TYPE: Article

Guan, B., Wang, T.-L., Shih, I.-M.
Recent advances in cancer genomics and cancer-associated genes discovery
(2010) An Omics Perspective on Cancer Research, pp. 11-29.

DOI: 10.1007/978-90-481-2675-0_2
DOCUMENT TYPE: Book Chapter

Myllykangas, S., Ji, H.P.
Targeted deep resequencing of the human cancer genome using next-generation technologies
(2010) Biotechnology and Genetic Engineering Reviews, 27, pp. 135-158. Cited 2 times.

DOCUMENT TYPE: Review

Hang, D., He, Z.-H.
The progress in cancer genome sequencing
(2010) Tumor, 30 (12), pp. 1077-1080.

DOI: 10.3781/j.issn.1000-7431.2010.12.017
DOCUMENT TYPE: Review

Lee, C., Hyland, C., Lee, A.S., Hislop, S., Ihm, C.
Copy Number Variation and Human Health
(2010) Essentials of Genomic and Personalized Medicine, pp. 46-59.

DOI: 10.1016/B978-0-12-374934-5.00005-2
DOCUMENT TYPE: Book Chapter

Xi, R., Kim, T.-M., Park, P.J.
Detecting structural variations in the human genome using next generation sequencing
(2010) Briefings in Functional Genomics, 9 (5-6), pp. 405-415. Cited 22 times.

DOI: 10.1093/bfgp/elq025
DOCUMENT TYPE: Article

Tyson, J., Majerus, T.M.O., Walker, S., Armour, J.A.L.
Screening for common copy-number variants in cancer genes
(2010) Cancer Genetics and Cytogenetics, 203 (2), pp. 316-323. Cited 2 times.

DOI: 10.1016/j.cancergencyto.2010.08.008

DOCUMENT TYPE: Article

O'Donnell, K.A., Burns, K.H.
Mobilizing diversity: Transposable element insertions in genetic variation and disease
(2010) Mobile DNA, 1 (1), art. no. 21, . Cited 18 times.

DOI: 10.1186/1759-8753-1-21
DOCUMENT TYPE: Review

Birkeland, S.R., Jin, N., Ozdemir, A.C., Lyons Jr., R.H., Weisman, L.S., Wilson, T.E.
Discovery of mutations in Saccharomyces cerevisiae by pooled linkage analysis and whole-genome sequencing
(2010) Genetics, 186 (4), pp. 1127-1137. Cited 22 times.

DOI: 10.1534/genetics.110.123232
DOCUMENT TYPE: Article

Hoang, M.L., Tan, F.J., Lai, D.C., Celniker, S.E., Hoskins, R.A., Dunham, M.J., Zheng, Y., Koshland, D.
Competitive repair by naturally dispersed repetitive DNA during non-allelic homologous recombination
(2010) PLoS Genetics, 6 (12), art. no. e1001228, pp. 1-18. Cited 17 times.

DOI: 10.1371/journal.pgen.1001228
DOCUMENT TYPE: Article

Willard, H.F.
Organization, Variation and Expression of the Human Genome
(2010) Essentials of Genomic and Personalized Medicine, pp. 13-26.

DOI: 10.1016/B978-0-12-374934-5.00002-7
DOCUMENT TYPE: Book Chapter

Vucic, E.A., Thu, K.L., Williams, A.C., Lam, W.L., Coe, B.P.
Copy number variations in the human genome and strategies for analysis
(2010) Methods in Molecular Biology, 628, pp. 103-117.

DOI: 10.1007/978-1-60327-367-1-6
DOCUMENT TYPE: Review

Guzman, N.V., Confalonieri, V.A.
The evolution of South American populations of Trimerotropis pallidipennis (Oedipodinae: Acrididae) revisited: Dispersion routes and origin of chromosomal inversion clines
(2010) Journal of Orthoptera Research, 19 (2), pp. 253-260. Cited 4 times.

DOI: 10.1665/034.019.0211
DOCUMENT TYPE: Article

Mani, R.-S., Chinnaiyan, A.M.
Triggers for genomic rearrangements: Insights into genomic, cellular and environmental influences
(2010) Nature Reviews Genetics, 11 (12), pp. 819-829. Cited 45 times.

DOI: 10.1038/nrg2883
DOCUMENT TYPE: Review

Purdy, K.J., Hurd, P.J., Moya-Laraño, J., Trimmer, M., Oakley, B.B., Woodward, G.

Systems biology for ecology. From molecules to ecosystems
(2010) Advances in Ecological Research, 43 (C), pp. 87-149. Cited 26 times.

DOI: 10.1016/B978-0-12-385005-8.00003-4
DOCUMENT TYPE: Book

Vajarova, R., Brandinova, I., Savov, A., Bichev, S., Singerska, I., Ivanova, M., Kremenski, I.
Current tools for genetic diagnosis in idiopathic mental retardation
(2010) Pediatriya, 50 (SUPPL. 1), pp. 30-35.

DOCUMENT TYPE: Article

Dahl, A., Mertes, F., Timmermann, B., Lehrach, H.
The application of massively parallel sequencing technologies in diagnostics
(2010) F1000 Biology Reports, 2 (1), art. no. 59, . Cited 3 times.

DOI: 10.3410/B2-59
DOCUMENT TYPE: Review

Kidd, J.M., Graves, T., Newman, T.L., Fulton, R., Hayden, H.S., Malig, M., Kallicki, J., Kaul, R., Wilson, R.K., Eichler, E.E.
A human genome structural variation sequencing resource reveals insights into mutational mechanisms
(2010) Cell, 143 (5), pp. 837-847. Cited 98 times.

DOI: 10.1016/j.cell.2010.10.027
DOCUMENT TYPE: Article

Fontanesi, L., Martelli, P.L., Beretti, F., Riggio, V., Dall'Olio, S., Colombo, M., Casadio, R., Russo, V., Portolano, B.
An initial comparative map of copy number variations in the goat (Capra hircus) genome
(2010) BMC Genomics, 11 (1), art. no. 639, . Cited 28 times.

DOI: 10.1186/1471-2164-11-639
DOCUMENT TYPE: Article

Schrider, D.R., Hahn, M.W.
Gene copy-number polymorphism in nature
(2010) Proceedings of the Royal Society B: Biological Sciences, 277 (1698), pp. 3213-3221. Cited 41 times.

DOI: 10.1098/rspb.2010.1180
DOCUMENT TYPE: Review

Ju, Y.S., Hong, D., Kim, S., Park, S.-S., Kim, S., Lee, S., Park, H., Kim, J.-I., Seo, J.-S.
Reference-unbiased copy number variant analysis using CGH microarrays
(2010) Nucleic Acids Research, 38 (20), . Cited 15 times.

DOI: 10.1093/nar/gkq730
DOCUMENT TYPE: Article

Itsara, A., Wu, H., Smith, J.D., Nickerson, D.A., Romieu, I., London, S.J., Eichler, E.E.
De novo rates and selection of large copy number variation
(2010) Genome Research, 20 (11), pp. 1469-1481. Cited 111 times.

DOI: 10.1101/gr.107680.110
DOCUMENT TYPE: Article

Faria, R., Navarro, A.
Chromosomal speciation revisited: Rearranging theory with pieces of evidence
(2010) Trends in Ecology and Evolution, 25 (11), pp. 660-669. Cited 71 times.

DOI: 10.1016/j.tree.2010.07.008
DOCUMENT TYPE: Review

Delseny, M., Han, B., Hsing, Y.I.
High throughput DNA sequencing: The new sequencing revolution
(2010) Plant Science, 179 (5), pp. 407-422. Cited 36 times.

DOI: 10.1016/j.plantsci.2010.07.019
DOCUMENT TYPE: Review

Cody, J.D., Heard, P., Hale, D.
Identification of two novel chromosome regions associated with isolated growth hormone deficiency
(2010) Journal of Pediatric Endocrinology and Metabolism, 23 (11), pp. 1159-1164. Cited 4 times.

DOI: 10.1515/jpem.2010.181
DOCUMENT TYPE: Article

Medvedev, P., Fiume, M., Dzamba, M., Smith, T., Brudno, M.
Detecting copy number variation with mated short reads
(2010) Genome Research, 20 (11), pp. 1613-1622. Cited 62 times.

DOI: 10.1101/gr.106344.110
DOCUMENT TYPE: Article

Cridland, J.M., Thornton, K.R.
Validation of rearrangement break points identified by paired-end sequencing in natural populations of Drosophila melanogaster
(2010) Genome Biology and Evolution, 2 (1), pp. 83-101. Cited 22 times.

DOI: 10.1093/gbe/evq001
DOCUMENT TYPE: Article

Helm, S., Mitelman, F.
Nonrandom Chromosome Abnormalities in Cancer-An Overview
(2010) Cancer Cytogenetics, pp. 25-43.

DOI: 10.1002/9781118010136.ch4
DOCUMENT TYPE: Book Chapter

Yang, H., Zhang, C., Zhao, X., Wu, Q., Fu, X., Yu, B., Shao, Y., Guan, M., Zhang, W., Wan, J., Huang, X.
Analysis of copy number variations of BS69 in multiple types of hematological malignancies
(2010) Annals of Hematology, 89 (10), pp. 959-964. Cited 2 times.

DOI: 10.1007/s00277-010-0966-5
DOCUMENT TYPE: Article

Saus, E., Brunet, A., Armengol, L., Alonso, P., Crespo, J.M., Fernández-Aranda, F., Guitart, M., Martín-Santos, R., Menchón, J.M., Navinés, R., Soria, V., Torrens, M., Urretavizcaya, M., Vallès, V., Gratacòs, M., Estivill, X.

Comprehensive copy number variant (CNV) analysis of neuronal pathways genes in psychiatric disorders identifies rare variants within patients
(2010) Journal of Psychiatric Research, 44 (14), pp. 971-978. Cited 37 times.

DOI: 10.1016/j.jpsychires.2010.03.007
DOCUMENT TYPE: Article

Peng, Y., Abercrombie, L.L.G., Yuan, J.S., Riggins, C.W., Sammons, R.D., Tranel, P.J., Stewart Jr., C.N.
Characterization of the horseweed (Conyza canadensis) transcriptome using GS-FLX 454 pyrosequencing and its application for expression analysis of candidate non-target herbicide resistance genes
(2010) Pest Management Science, 66 (10), pp. 1053-1062. Cited 27 times.

DOI: 10.1002/ps.2004
DOCUMENT TYPE: Article

Koboldt, D.C., Ding, L., Mardis, E.R., Wilson, R.K.
Challenges of sequencing human genomes
(2010) Briefings in Bioinformatics, 11 (5), art. no. bbq016, pp. 484-498. Cited 63 times.

DOI: 10.1093/bib/bbq016
DOCUMENT TYPE: Review

Klinke II, D.J.
A multiscale systems perspective on cancer, immunotherapy, and Interleukin-12
(2010) Molecular Cancer, 9, art. no. 242, . Cited 3 times.

DOI: 10.1186/1476-4598-9-242
DOCUMENT TYPE: Review

Nowrousian, M.
Next-generation sequencing techniques for eukaryotic microorganisms: Sequencing-based solutions to biological problems
(2010) Eukaryotic Cell, 9 (9), pp. 1300-1310. Cited 47 times.

DOI: 10.1128/EC.00123-10
DOCUMENT TYPE: Review

Bae, J.S., Cheong, H.S., Lae Park, B., Kim, L.H., Han, C.S., Park, T.J., Kim, J.Y., Pasaje, C.F.A., Lee, J.S., Shin, H.D.
Genome-wide profiling of structural genomic variations in Korean hapmap individuals
(2010) PLoS ONE, 5 (7), art. no. e11417, . Cited 3 times.

DOI: 10.1371/journal.pone.0011417
DOCUMENT TYPE: Article

Zhang, C., Xing, D.
Single-molecule DNA amplification and analysis using microfluidics
(2010) Chemical Reviews, 110 (8), pp. 4910-4947. Cited 57 times.

DOI: 10.1021/cr900081z
DOCUMENT TYPE: Article

Das, S.K., Austin, M.D., Akana, M.C., Deshpande, P., Cao, H., Xiao, M.
Single molecule linear analysis of DNA in nano-channel labeled with sequence specific fluorescent probes
(2010) Nucleic Acids Research, 38 (18), art. no. gkq673, pp. e177-e177. Cited 4 times.

DOI: 10.1093/nar/gkq673
DOCUMENT TYPE: Article

McNulty, S.N., Foster, J.M., Mitreva, M., Hotopp, J.C.D., Martin, J., Fischer, K., Wu, B., Davis, P.J., Kumar, S., Brattig, N.W., Slatko, B.E., Weil, G.J., Fischer, P.U.
Endosymbiont DNA in endobacteria-free filarial nematodes indicates ancient horizontal genetic transfer
(2010) PLoS ONE, 5 (6), art. no. e11029, . Cited 49 times.

DOI: 10.1371/journal.pone.0011029
DOCUMENT TYPE: Article

Voelkerding, K.V., Dames, S.A., Durtschi, J.D.
Next-generation sequencing: From basic research to diagnostics ["Next-generation sequencing": Dalla ricerca di base alla diagnostica]
(2010) Biochimica Clinica, 34 (4), pp. 287-303. Cited 1 time.

DOCUMENT TYPE: Review

Bergboer, J.G.M., Zeeuwen, P.L.J.M., Irvine, A.D., Weidinger, S., Giardina, E., Novelli, G., Heijer, M.D., Rodriguez, E., Illig, T., Riveira-Munoz, E., Campbell, L.E., Tyson, J., Dannhauser, E.N., O'Regan, G.M., Galli, E., Klopp, N., Koppelman, G.H., Novak, N., Estivill, X., McLean, W.H.I., Postma, D.S., Armour, J.A.L., Schalkwijk, J.
Deletion of late cornified envelope 3B and 3C genes is not associated with atopic dermatitis
(2010) Journal of Investigative Dermatology, 130 (8), pp. 2057-2061. Cited 22 times.

DOI: 10.1038/jid.2010.88
DOCUMENT TYPE: Article

Goldmann, R., Tichý, L., Freiberger, T., Zapletalová, P., Letocha, O., Soška, V., Fajkus, J., Fajkusová, L.
Genomic characterization of large rearrangements of the LDLR gene in Czech patients with familial hypercholesterolemia
(2010) BMC Medical Genetics, 11 (1), art. no. 115, . Cited 7 times.

DOI: 10.1186/1471-2350-11-115
DOCUMENT TYPE: Article

Zeitouni, B., Boeva, V., Janoueix-Lerosey, I., Loeillet, S., Legoix-né, P., Nicolas, A., Delattre, O., Barillot, E.
SVDetect: A tool to identify genomic structural variations from paired-end and mate-pair sequencing data
(2010) Bioinformatics, 26 (15), art. no. btq293, pp. 1895-1896. Cited 50 times.

DOI: 10.1093/bioinformatics/btq293
DOCUMENT TYPE: Article

Mitsui, J., Takahashi, Y., Goto, J., Tomiyama, H., Ishikawa, S., Yoshino, H., Minami, N., Smith, D.I., Lesage, S., Aburatani, H., Nishino, I., Brice, A., Hattori, N., Tsuji, S.
Mechanisms of genomic instabilities underlying two common fragile-site-associated Loci, PARK2 and DMD, in germ cell and cancer cell lines
(2010) American Journal of Human Genetics, 87 (1), pp. 75-89. Cited 31 times.

DOI: 10.1016/j.ajhg.2010.06.006
DOCUMENT TYPE: Article

Jain, K.K.
Innovative diagnostic technologies and their significance for personalized medicine
(2010) Molecular Diagnosis and Therapy, 14 (3), pp. 141-147. Cited 5 times.

DOI: 10.2165/11536240-000000000-00000
DOCUMENT TYPE: Note

de Magalhães, J.P., Finch, C.E., Janssens, G.
Next-generation sequencing in aging research: Emerging applications, problems, pitfalls and possible solutions
(2010) Ageing Research Reviews, 9 (3), pp. 315-323. Cited 33 times.

DOI: 10.1016/j.arr.2009.10.006
DOCUMENT TYPE: Review

Ku, C.S., Loy, E.Y., Salim, A., Pawitan, Y., Chia, K.S.
The discovery of human genetic variations and their use as disease markers: Past, present and future
(2010) Journal of Human Genetics, 55 (7), pp. 403-415. Cited 27 times.

DOI: 10.1038/jhg.2010.55
DOCUMENT TYPE: Review

Costa, F.F.
Non-coding RNAs: Meet thy masters
(2010) BioEssays, 32 (7), pp. 599-608. Cited 91 times.

DOI: 10.1002/bies.200900112
DOCUMENT TYPE: Review

Bashir, A., Bansal, V., Bafna, V.
Designing deep sequencing experiments: Detecting structural variation and estimating transcript abundance
(2010) BMC Genomics, 11 (1), art. no. 385, . Cited 13 times.

DOI: 10.1186/1471-2164-11-385
DOCUMENT TYPE: Article

Teague, B., Waterman, M.S., Goldstein, S., Potamousis, K., Zhou, S., Reslewic, S., Sarkar, D., Valouev, A., Churas, C., Kidd, J.M., Kohn, S., Runnheim, R., Lamers, C., Forrest, D., Newton, M.A., Eichler, E.E., Kent-First, M., Surti, U., Livny, M., Schwartz, D.C.
High-resolution human genome structure by single-molecule analysis
(2010) Proceedings of the National Academy of Sciences of the United States of America, 107 (24), pp. 10848-10853. Cited 51 times.

DOI: 10.1073/pnas.0914638107
DOCUMENT TYPE: Article

Leary, R.J., Kinde, I., Diehl, F., Schmidt, K., Clouser, C., Duncan, C., Antipova, A., Lee, C., McKernan, K., De La Vega, F.M., Kinzler, K.W., Vogelstein, B., Diaz Jr., L.A., Velculescu, V.E.
Development of personalized tumor biomarkers using massively parallel sequencing
(2010) Science Translational Medicine, 2 (20), . Cited 179 times.

DOI: 10.1126/scitranslmed.3000702
DOCUMENT TYPE: Article

Beck, C.R., Collier, P., Macfarlane, C., Malig, M., Kidd, J.M., Eichler, E.E., Badge, R.M., Moran, J.V.
LINE-1 retrotransposition activity in human genomes
(2010) Cell, 141 (7), pp. 1159-1170. Cited 139 times.

DOI: 10.1016/j.cell.2010.05.021

DOCUMENT TYPE: Article

Kuiper, R.P., Ligtenberg, M.J.L., Hoogerbrugge, N., Geurts van Kessel, A.
Germline copy number variation and cancer risk
(2010) Current Opinion in Genetics and Development, 20 (3), pp. 282-289. Cited 40 times.

DOI: 10.1016/j.gde.2010.03.005
DOCUMENT TYPE: Review

Huang, C.R.L., Schneider, A.M., Lu, Y., Niranjan, T., Shen, P., Robinson, M.A., Steranka, J.P., Valle, D., Civin, C.I., Wang, T., Wheelan, S.J., Ji, H., Boeke, J.D., Burns, K.H.
Mobile interspersed repeats are major structural variants in the human genome
(2010) Cell, 141 (7), pp. 1171-1182. Cited 122 times.

DOI: 10.1016/j.cell.2010.05.026
DOCUMENT TYPE: Article

Iskow, R.C., McCabe, M.T., Mills, R.E., Torene, S., Pittard, W.S., Neuwald, A.F., Van Meir, E.G., Vertino, P.M., Devine, S.E.
Natural mutagenesis of human genomes by endogenous retrotransposons
(2010) Cell, 141 (7), pp. 1253-1261. Cited 203 times.

DOI: 10.1016/j.cell.2010.05.020
DOCUMENT TYPE: Article

Anderson, M.W., Schrijver, I.
Next generation DNA sequencing and the future of genomic medicine
(2010) Genes, 1 (1), pp. 38-69. Cited 11 times.

DOI: 10.3390/genes1010038
DOCUMENT TYPE: Review

Hormozdiari, F., Hajirasouliha, I., Dao, P., Hach, F., Yorukoglu, D., Alkan, C., Eichler, E.E., Sahinalp, S.C.
Next-generation VariationHunter: Combinatorial algorithms for transposon insertion discovery
(2010) Bioinformatics, 26 (12), art. no. btq216, pp. i350-i357. Cited 73 times.

DOI: 10.1093/bioinformatics/btq216
DOCUMENT TYPE: Article

Hoffman, E.J., State, M.W.
Progress in cytogenetics: Implications for child psychopathology
(2010) Journal of the American Academy of Child and Adolescent Psychiatry, 49 (8), pp. 736-751. Cited 7 times.

DOI: 10.1016/j.jaac.2010.03.016
DOCUMENT TYPE: Review

Fadista, J., Thomsen, B., Holm, L., Bendixen, C.
Copy number variation in the bovine genome
(2010) BMC Genomics, 11 (1), art. no. 284, . Cited 53 times.

DOI: 10.1186/1471-2164-11-284
DOCUMENT TYPE: Article

Marian, A.J.
DNA sequence variants and the practice of medicine
(2010) Current Opinion in Cardiology, 25 (3), pp. 182-185.

DOI: 10.1097/HCO.0b013e3283389683
DOCUMENT TYPE: Review

Harewood, L., Schütz, F., Boyle, S., Perry, P., Delorenzi, M., Bickmore, W.A., Reymond, A.
The effect of translocation-induced nuclear reorganization on gene expression
(2010) Genome Research, 20 (5), pp. 554-564. Cited 34 times.

DOI: 10.1101/gr.103622.109
DOCUMENT TYPE: Article

Vissers, L.E.L.M., De Vries, B.B.A., Veltman, J.A.
Genomic microarrays in mental retardation: From copy number variation to gene, from research to diagnosis
(2010) Journal of Medical Genetics, 47 (5), pp. 289-297. Cited 74 times.

DOI: 10.1136/jmg.2009.072942
DOCUMENT TYPE: Review

Liu, G.E., Hou, Y., Zhu, B., Cardone, M.F., Jiang, L., Cellamare, A., Mitra, A., Alexander, L.J., Coutinho, L.L., Dell'Aquila, M.E., Gasbarre, L.C., Lacalandra, G., Li, R.W., Matukumalli, L.K., Nonneman, D., Regitano, L.C.D.A., Smith, T.P.L., Song, J., Sonstegard, T.S., Van Tassell, C.P., Ventura, M., Eichler, E.E., McDaneld, T.G., Keele, J.W.
Analysis of copy number variations among diverse cattle breeds
(2010) Genome Research, 20 (5), pp. 693-703. Cited 83 times.

DOI: 10.1101/gr.105403.110
DOCUMENT TYPE: Article

Kidd, J.M., Sampas, N., Antonacci, F., Graves, T., Fulton, R., Hayden, H.S., Alkan, C., Malig, M., Ventura, M., Giannuzzi, G., Kallicki, J., Anderson, P., Tsalenko, A., Yamada, N.A., Tsang, P., Kaul, R., Wilson, R.K., Bruhn, L., Eichler, E.E.
Characterization of missing human genome sequences and copy-number polymorphic insertions
(2010) Nature Methods, 7 (5), pp. 365-371. Cited 68 times.

DOI: 10.1038/nmeth.1451
DOCUMENT TYPE: Article

Slade, I., Stephens, P., Douglas, J., Barker, K., Stebbings, L., Abbaszadeh, F., Pritchard-Jones, K., Arbour, L., Cole, T., Sheridan, E., Price, H., Schumacher, V., Weirich, A., Royer-Pokora, B., Kingston, J., O'Meara, A., Foot, A., Dhooge, C., Gerrard, M., Dupuis, W., Levitt, G., Chompret, A., Bonaitie-Pellie, C., Tonin, P., Skeen, J., Kohler, J., Gnekow, A., Cole, R., Pizer, B., Stiller, C., Vujanic, G., Scott, R.H., Stratton, M.R., Rahman, N.
Constitutional translocation breakpoint mapping by genome-wide paired-end sequencing identifies HACE1 as a putative Wilms tumour susceptibility gene
(2010) Journal of Medical Genetics, 47 (5), pp. 342-347. Cited 17 times.

DOI: 10.1136/jmg.2009.072983
DOCUMENT TYPE: Article

Bouckenooghe, T., Flamez, D., Ortis, F., Goldman, S., Eizirik, D.L.
Identification of new pancreatic beta cell targets for In vivo imaging by a systems biology approach
(2010) Current Pharmaceutical Design, 16 (14), pp. 1609-1618. Cited 5 times.

DOI: 10.2174/138161210791164117
DOCUMENT TYPE: Article

Vlad, D., Rappaport, F., Simon, M., Loudet, O.
Gene transposition causing natural variation for growth in Arabidopsis thaliana
(2010) PLoS Genetics, 6 (5), p. 21. Cited 10 times.

DOI: 10.1371/journal.pgen.1000945
DOCUMENT TYPE: Article

Dowell, R.D., Ryan, O., Jansen, A., Cheung, D., Agarwala, S., Danford, T., Bernstein, D.A., Alexander Rolfe, P., Heisler, L.E., Chin, B., Nislow, C., Giaever, G., Phillips, P.C., Fink, G.R., Gifford, D.K., Boone, C.
Genotype to phenotype: A Complex problem
(2010) Science, 328 (5977), p. 469. Cited 120 times.

DOI: 10.1126/science.1189015
DOCUMENT TYPE: Short Survey

Hajirasouliha, I., Hormozdiari, F., Alkan, C., Kidd, J.M., Birol, I., Eichler, E.E., Sahinalp, S.C.
Detection and characterization of novel sequence insertions using paired-end next-generation sequencing
(2010) Bioinformatics, 26 (10), art. no. btq152, pp. 1277-1283. Cited 39 times.

DOI: 10.1093/bioinformatics/btq152
DOCUMENT TYPE: Article

Ritz, A., Bashir, A., Raphael, B.J.
Structural variation analysis with strobe reads
(2010) Bioinformatics, 26 (10), art. no. btq153, pp. 1291-1298. Cited 20 times.

DOI: 10.1093/bioinformatics/btq153
DOCUMENT TYPE: Article

Bhasker, C.R., Hardiman, G.
Advances in pharmacogenomics technologies
(2010) Pharmacogenomics, 11 (4), pp. 481-485. Cited 6 times.

DOI: 10.2217/pgs.10.10
DOCUMENT TYPE: Note

Reams, A.B., Kofoid, E., Savageau, M., Roth, J.R.
Duplication frequency in a population of Salmonella enterica rapidly approaches steady state with or without recombination
(2010) Genetics, 184 (4), pp. 1077-1094. Cited 42 times.

DOI: 10.1534/genetics.109.111963
DOCUMENT TYPE: Article

Jørgensen, J.T.
Pharmacodiagnostics and personalized medicine in cancer
(2010) Molecular Diagnostics: The Key Driver in Personalized Cancer Medicine, pp. 325-344.

DOI: 10.4032/9789814241458
DOCUMENT TYPE: Book Chapter

Hehir-Kwa, J.Y., Wieskamp, N., Webber, C., Pfundt, R., Brunner, H.G., Gilissen, C., de Vries, B.B.A., Ponting, C.P., Veltman, J.A.
Accurate distinction of pathogenic from benign CNVs in mental retardation
(2010) PLoS Computational Biology, 6 (4), . Cited 31 times.

DOI: 10.1371/journal.pcbi.1000752
DOCUMENT TYPE: Article

Jenkins, S., Kahvejian, A.
True Single Molecule Sequencing (tSMS)™ by Synthesis
(2010) The Handbook of Plant Mutation Screening: Mining of Natural and Induced Alleles, pp. 287-306.

DOI: 10.1002/9783527629398.ch17
DOCUMENT TYPE: Book Chapter

Mkrtchyan, H., Gross, M., Hinreiner, S., Polytiko, A., Manvelyan, M., Mrasek, K., Kosyakova, N., Ewers, E., Nelle, H., Liehr, T., Volleth, M., Weise, A.
Early embryonic chromosome instability results in stable mosaic pattern in human tissues
(2010) PLoS ONE, 5 (3), art. no. e9591, . Cited 31 times.

DOI: 10.1371/journal.pone.0009591
DOCUMENT TYPE: Article

Ruan, Y., Wei, C.-L.
Multiplex parallel pair-end-ditag sequencing approaches in system biology
(2010) Wiley Interdisciplinary Reviews: Systems Biology and Medicine, 2 (2), pp. 224-234. Cited 4 times.

DOI: 10.1002/wsbm.40
DOCUMENT TYPE: Article

Wheat, C.W.
Rapidly developing functional genomics in ecological model systems via 454 transcriptome sequencing
(2010) Genetica, 138 (4), pp. 433-451. Cited 78 times.

DOI: 10.1007/s10709-008-9326-y
DOCUMENT TYPE: Review

Rueda, O.M., Diaz-Uriarte, R.
Finding recurrent copy number alteration regions: A review of methods
(2010) Current Bioinformatics, 5 (1), pp. 1-17. Cited 19 times.

DOI: 10.2174/157489310790596402
DOCUMENT TYPE: Review

Laird, P.W.
Principles and challenges of genome-wide DNA methylation analysis
(2010) Nature Reviews Genetics, 11 (3), pp. 191-203. Cited 508 times.

DOI: 10.1038/nrg2732
DOCUMENT TYPE: Review

Fanciulli, M., Petretto, E., Aitman, T.J.
Gene copy number variation and common human disease
(2010) Clinical Genetics, 77 (3), pp. 201-213. Cited 48 times.

DOI: 10.1111/j.1399-0004.2009.01342.x
DOCUMENT TYPE: Review

Pool, J.E., Hellmann, I., Jensen, J.D., Nielsen, R.
Population genetic inference from genomic sequence variation
(2010) Genome Research, 20 (3), pp. 291-300. Cited 88 times.

DOI: 10.1101/gr.079509.108
DOCUMENT TYPE: Review

Sindi, S.S., Raphael, B.J.
Identification and frequency estimation of inversion polymorphisms from haplotype data
(2010) Journal of Computational Biology, 17 (3), pp. 517-531. Cited 8 times.

DOI: 10.1089/cmb.2009.0185
DOCUMENT TYPE: Conference Paper

Li, G., Fullwood, M.J., Xu, H., Mulawadi, F.H., Velkov, S., Vega, V., Ariyaratne, P.N., Mohamed, Y.B., Ooi, H.-S., Tennakoon, C., Wei, C.-L., Ruan, Y., Sung, W.-K.
ChIA-PET tool for comprehensive chromatin interaction analysis with paired-end tag sequencing
(2010) Genome Biology, 11 (2), art. no. r22, . Cited 60 times.

DOI: 10.1186/gb-2010-11-2-r22
DOCUMENT TYPE: Article

Brunson, T., Wang, Q., Chambers, I., Song, Q.
A copy number variation in human NCF1 and its pseudogenes
(2010) BMC Genetics, 11, art. no. 13, . Cited 7 times.

DOI: 10.1186/1471-2156-11-13
DOCUMENT TYPE: Article

Stankiewicz, P., Lupski, J.R.
Structural variation in the human genome and its role in disease
(2010) Annual Review of Medicine, 61, pp. 437-455. Cited 363 times.

DOI: 10.1146/annurev-med-100708-204735
DOCUMENT TYPE: Review

Feuk, L.
Inversion variants in the human genome: Role in disease and genome architecture
(2010) Genome Medicine, 2 (2), art. no. 11, . Cited 16 times.

DOI: 10.1186/gm132
DOCUMENT TYPE: Review

Krawitz, P., Rödelsperger, C., Jäger, M., Jostins, L., Bauer, S., Robinson, P.N.
Microindel detection in short-read sequence data
(2010) Bioinformatics, 26 (6), art. no. btq027, pp. 722-729. Cited 50 times.

DOI: 10.1093/bioinformatics/btq027
DOCUMENT TYPE: Article

Marguerat, S., Bähler, J.
RNA-seq: From technology to biology
(2010) Cellular and Molecular Life Sciences, 67 (4), pp. 569-579. Cited 149 times.

DOI: 10.1007/s00018-009-0180-6
DOCUMENT TYPE: Review

Wilhelm, B.T., Marguerat, S., Goodhead, I., Bähler, J.
Defining transcribed regions using RNA-seq
(2010) Nature Protocols, 5 (2), pp. 255-266. Cited 39 times.

DOI: 10.1038/nprot.2009.229
DOCUMENT TYPE: Article

Carvalho, C.M.B., Zhang, F., Lupski, J.R.
Genomic disorders: A window into human gene and genome evolution
(2010) Proceedings of the National Academy of Sciences of the United States of America, 107 (SUPPL. 1), pp. 1765-1771. Cited 29 times.

DOI: 10.1073/pnas.0906222107
DOCUMENT TYPE: Conference Paper

Zepeda-Mendoza, C.J., Lemus, T., Yáñez, O., García, D., Valle-García, D., Meza-Sosa, K.F., Gutiérrez-Arcelus, M., Márquez-Ortiz, Y., Domínguez-Vidaña, R., Gonzaga-Jauregui, C., Flores, M., Palacios, R.
Identical repeated backbone of the human genome
(2010) BMC Genomics, 11 (1), art. no. 60, . Cited 2 times.

DOI: 10.1186/1471-2164-11-60
DOCUMENT TYPE: Article

Dalca, A.V., Brudno, M.
Genome variation discovery with high-throughput sequencing data
(2010) Briefings in Bioinformatics, 11 (1), art. no. bbp058, pp. 3-14. Cited 40 times.

DOI: 10.1093/bib/bbp058
DOCUMENT TYPE: Article

Metzker, M.L.
Sequencing technologies the next generation
(2010) Nature Reviews Genetics, 11 (1), pp. 31-46. Cited 2012 times.

DOI: 10.1038/nrg2626
DOCUMENT TYPE: Review

Roukos, D.H.
Systems medicine for a next-generation of biomarkers and drugs: Changing poor cancer outcomes
(2010) Gastric and Breast Cancer, 9 (1), pp. 1-23.

DOI: 10.2122/gbc.2010.0112
DOCUMENT TYPE: Review

Bell, D.W.
Our changing view of the genomic landscape of cancer

(2010) Journal of Pathology, 220 (2), pp. 231-243. Cited 48 times.

DOI: 10.1002/path.2645
DOCUMENT TYPE: Review

Rouko, D.H.
Novel clinico-genome network modeling for revolutionizing genotype-phenotype-based personalized cancer care
(2010) Expert Review of Molecular Diagnostics, 10 (1), pp. 33-48. Cited 106 times.

DOI: 10.1586/erm.09.69
DOCUMENT TYPE: Review

Edwards, P.A.W.
Fusion genes and chromosome translocations in the common epithelial cancers
(2010) Journal of Pathology, 220 (2), pp. 244-254. Cited 33 times.

DOI: 10.1002/path.2632
DOCUMENT TYPE: Review

Dermitzakis, E.T.
Population genetic principles and human populations
(2010) Vogel and Motulsky's Human Genetics: Problems and Approaches (Fourth Edition), pp. 487-506.

DOI: 10.1007/978-3-540-37654-5_18
DOCUMENT TYPE: Book Chapter

Brutsaert, T.D., Parra, E.J.
Nature versus nurture in determining athletic ability
(2009) Medicine and Sport Science, 54, pp. 11-27. Cited 2 times.

DOI: 10.1159/000235694
DOCUMENT TYPE: Article

Gibson, W.T.
Key concepts in human genetics: Understanding the complex phenotype
(2009) Medicine and Sport Science, 54, pp. 1-10.

DOI: 10.1159/000235693
DOCUMENT TYPE: Article

Kann, M.G.
Advances in translational bioinformatics: Computational approaches for the hunting of disease genes
(2009) Briefings in Bioinformatics, 11 (1), art. no. bbp048, pp. 96-110. Cited 34 times.

DOI: 10.1093/bib/bbp048
DOCUMENT TYPE: Article

De Grassi, A., Ciccarelli, F.D.
Tandem repeats modify the structure of human genes hosted in segmental duplications
(2009) Genome Biology, 10 (12), art. no. R137, . Cited 5 times.

DOI: 10.1186/gb-2009-10-12-r137
DOCUMENT TYPE: Article

Clarimón, J., Djaldetti, R., Lleó, A., Guerreiro, R.J., Molinuevo, J.L., Paisán-Ruiz, C., Gómez-Isla, T., Blesa, R., Singleton, A., Hardy, J.
Whole genome analysis in a consanguineous family with early onset Alzheimer's disease
(2009) Neurobiology of Aging, 30 (12), pp. 1986-1991. Cited 9 times.

DOI: 10.1016/j.neurobiolaging.2008.02.008
DOCUMENT TYPE: Article

Grigorenko, E.L.
Pathogenesis of autism: A patchwork of genetic causes
(2009) Future Neurology, 4 (5), pp. 591-599. Cited 5 times.

DOI: 10.2217/fnl.09.29
DOCUMENT TYPE: Review

Marian, A.J.
Nature's genetic gradients and the clinical phenotype
(2009) Circulation: Cardiovascular Genetics, 2 (6), pp. 537-539. Cited 16 times.

DOI: 10.1161/CIRCGENETICS.109.921940
DOCUMENT TYPE: Editorial

Willard, H.F.
Organization, Variation and Expression of the Human Genome as a Foundation of Genomic and Personalized Medicine
(2009) Genomic and Personalized Medicine, Two-Vol Set, pp. 1-21.

DOI: 10.1016/B978-0-12-369420-1.00001-9
DOCUMENT TYPE: Book Chapter

Sebat, J., Levy, D.L., McCarthy, S.E.
Rare structural variants in schizophrenia: one disorder, multiple mutations; one mutation, multiple disorders
(2009) Trends in Genetics, 25 (12), pp. 528-535. Cited 138 times.

DOI: 10.1016/j.tig.2009.10.004
DOCUMENT TYPE: Review

Bourque, G.
Transposable elements in gene regulation and in the evolution of vertebrate genomes
(2009) Current Opinion in Genetics and Development, 19 (6), pp. 607-612. Cited 52 times.

DOI: 10.1016/j.gde.2009.10.013
DOCUMENT TYPE: Review

Lee, C., Hyland, C., Lee, A.S., Hislop, S., Ihm, C.
Copy Number Variation and Human Health
(2009) Genomic and Personalized Medicine, Two-Vol Set, pp. 108-119.

DOI: 10.1016/B978-0-12-369420-1.00009-3
DOCUMENT TYPE: Book Chapter

Greub, G., Kebbi-Beghdadi, C., Bertelli, C., Collyn, F., Riederer, B.M., Yersin, C., Croxatto, A., Raoult, D.
High throughput sequencing and Proteomics to identify immunogenic proteins of a new pathogen: The dirty genome approach

(2009) PLoS ONE, 4 (12), art. no. e8423, . Cited 34 times.

DOI: 10.1371/journal.pone.0008423
DOCUMENT TYPE: Article

Akagi, K., Stephens, R.M., Li, J., Evdokimov, E., Kuehn, M.R., Volfovsky, N., Symer, D.E.
MouseIndelDB: A database integrating genomic indel polymorphisms that distinguish mouse strains
(2009) Nucleic Acids Research, 38 (SUPPL.1), art. no. gkp1046, pp. D600-D606. Cited 11 times.

DOI: 10.1093/nar/gkp1046
DOCUMENT TYPE: Article

Delprat, A., Negre, B., Puig, M., Ruiz, A.
The transposon Galileo generates natural chromosomal inversions in Drosophila by ectopic recombination
(2009) PLoS ONE, 4 (11), art. no. e7883, . Cited 25 times.

DOI: 10.1371/journal.pone.0007883
DOCUMENT TYPE: Article

Friedman, J.M., Adam, S., Arbour, L., Armstrong, L., Baross, A., Birch, P., Boerkoel, C., Chan, S., Chai, D., Delaney, A.D., Flibotte, S., Gibson, W.T., Langlois, S., Lemyre, E., Li, H.I., MacLeod, P., Mathers, J., Michaud, J.L., McGillivray, B.C., Patel, M.S., Qian, H., Rouleau, G.A., Van Allen, M.I., Yong, S., Zahir, F.R., Eydoux, P., Marra, M.A.
Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization
(2009) BMC Genomics, 10, art. no. 526, . Cited 14 times.

DOI: 10.1186/1471-2164-10-526
DOCUMENT TYPE: Article

Shibata, Y., Malhotra, A., Bekiranov, S., Dutta, A.
Yeast genome analysis identifies chromosomal translocation, gene conversion events and several sites of Ty element insertion
(2009) Nucleic Acids Research, 37 (19), pp. 6454-6465. Cited 5 times.

DOI: 10.1093/nar/gkp650
DOCUMENT TYPE: Article

Matsuzaki, H., Wang, P.-H., Hu, J., Rava, R., Fu, G.K.
High resolution discovery and confirmation of copy number variants in 90 Yoruba Nigerians
(2009) Genome Biology, 10 (11), art. no. 125, . Cited 42 times.

DOI: 10.1186/gb-2009-10-11-r125
DOCUMENT TYPE: Article

Shrestha, S., Tang, J., Kaslow, R.A.
Gene copy number: Learning to count past two
(2009) Nature Medicine, 15 (10), pp. 1127-1129. Cited 13 times.

DOI: 10.1038/nm1009-1127
DOCUMENT TYPE: Short Survey

Jung, Y.-C., Xu, J., Chen, J., Kim, Y.C., Winchester, D.J., Wang, S.M.
Simplified DGS procedure for large-scale genome structural study
(2009) BioTechniques, 47 (5), pp. 969-971. Cited 1 time.

DOI: 10.2144/000113294
DOCUMENT TYPE: Article

Mir, K.U.
Sequencing genomes: From individuals to populations
(2009) Briefings in Functional Genomics and Proteomics, 8 (5), pp. 367-378. Cited 10 times.

DOI: 10.1093/bfgp/elp040
DOCUMENT TYPE: Article

Winchester, L., Yau, C., Ragoussis, J.
Comparing CNV detection methods for SNP arrays
(2009) Briefings in Functional Genomics and Proteomics, 8 (5), pp. 353-366. Cited 78 times.

DOI: 10.1093/bfgp/elp017
DOCUMENT TYPE: Review

Simonis, M., Klous, P., Homminga, I., Galjaard, R.-J., Rijkers, E.-J., Grosveld, F., Meijerink, J.P.P., de Laat, W.
High-resolution identification of balanced and complex chromosomal rearrangements by 4C technology
(2009) Nature Methods, 6 (11), pp. 837-842. Cited 35 times.

DOI: 10.1038/nmeth.1391
DOCUMENT TYPE: Article

Mardis, E.R., Wilson, R.K.
Cancer genome sequencing: A review
(2009) Human Molecular Genetics, 18 (R2), pp. R163-R168. Cited 73 times.

DOI: 10.1093/hmg/ddp396
DOCUMENT TYPE: Article

Armour, J.A.L.
Human genetics: Sharp focus on the variable genome
(2009) Nature, 461 (7265), pp. 735-736. Cited 4 times.

DOI: 10.1038/461735a
DOCUMENT TYPE: Short Survey

Suzuki, G., Harper, K.M., Hiramoto, T., Funke, B., Lee, M., Kang, G., Buell, M., Geyer, M.A., Kucherlapati, R., Morrow, B., Männistö, P.T., Agatsuma, S., Hiroi, N.
Over-expression of a human chromosome 22q11.2 segment including TXNRD2, COMT and ARVCF developmentally affects incentive learning and working memory in mice
(2009) Human Molecular Genetics, 18 (20), pp. 3914-3925. Cited 18 times.

DOI: 10.1093/hmg/ddp334
DOCUMENT TYPE: Article

Wei, C.-L., Ruan, Y.
Transcriptome and genome characterization using massively parallel paired end tag (PET) sequencing analysis
(2009) Cap-Analysis Gene Expression (CAGE): The Science of Decoding Genes Transcription, pp. 41-60.

DOI: 10.4032/9789814241359
DOCUMENT TYPE: Book Chapter

Alkan, C., Kidd, J.M., Marques-Bonet, T., Aksay, G., Antonacci, F., Hormozdiari, F., Kitzman, J.O., Baker, C., Malig, M., Mutlu, O., Sahinalp, S.C., Gibbs, R.A., Eichler, E.E.
Personalized copy number and segmental duplication maps using next-generation sequencing
(2009) Nature Genetics, 41 (10), pp. 1061-1067. Cited 276 times.

DOI: 10.1038/ng.437
DOCUMENT TYPE: Article

Sieh, W., Choi, Y., Chapman, N.H., Craig, U.-K., Steinbart, E.J., Rothstein, J.H., Oyanagi, K., Garruto, R.M., Bird, T.D., Galasko, D.R., Schellenberg, G.D., Wijsman, E.M.
Identification of novel susceptibility loci for Guam neurodegenerative disease: Challenges of genome scans in genetic isolates
(2009) Human Molecular Genetics, 18 (19), pp. 3725-3738. Cited 11 times.

DOI: 10.1093/hmg/ddp300
DOCUMENT TYPE: Article

Park, P.J.
ChIP-seq: Advantages and challenges of a maturing technology
(2009) Nature Reviews Genetics, 10 (10), pp. 669-680. Cited 576 times.

DOI: 10.1038/nrg2641
DOCUMENT TYPE: Review

Brown, C.G.
The DNA sequencing renaissance and its implications for epigenomics
(2009) Epigenomics, 1 (1), pp. 5-8. Cited 3 times.

DOI: 10.2217/epi.09.7
DOCUMENT TYPE: Review

Vissers, L.E.L.M., Bhatt, S.S., Janssen, I.M., Xia, Z., Lalani, S.R., Pfundt, R., Derwinska, K., de Vries, B.B.A., Gilissen, C., Hoischen, A., Nesteruk, M., Wisniowiecka-Kowalnik, B., Smyk, M., Brunner, H.G., Cheung, S.W., van Kessel, A.G., Veltman, J.A., Stankiewicz, P.
Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture
(2009) Human Molecular Genetics, 18 (19), pp. 3579-3593. Cited 66 times.

DOI: 10.1093/hmg/ddp306
DOCUMENT TYPE: Article

Armengol, L., Villatoro, S., González, J.R., Pantano, L., García-Aragonés, M., Rabionet, R., Cáceres, M., Estivill, X.
Identification of copy number variants defining genomic differences among major human groups
(2009) PLoS ONE, 4 (9), art. no. e7230, . Cited 15 times.

DOI: 10.1371/journal.pone.0007230
DOCUMENT TYPE: Article

Tyson, J., Majerus, T.M.O., Walker, S., Armour, J.A.L.
Quadruplex MAPH: Improvement of throughput in high-resolution copy number screening
(2009) BMC Genomics, 10, art. no. 1471, p. 453. Cited 3 times.

DOI: 10.1186/1471-2164-10-453

DOCUMENT TYPE: Article

Roviello, F.
Early breast cancer: How targeted therapy, genetics and personal genomics may improve local outcomes
(2009) Gastric and Breast Cancer, 8 (2), pp. 53-69.

DOI: 10.2122/gbc.2009.0111
DOCUMENT TYPE: Review

Schlick-Steiner, B.C., Arthofer, W., Moder, K., Steiner, F.M.
Recent insertion/deletion (reINDEL) mutations: Increasing awareness to boost molecular-based research in ecology and evolution
(2015) Ecology and Evolution, 5 (1), pp. 24-35.

DOI: 10.1002/ece3.1330
DOCUMENT TYPE: Article

Gelernter, J.
Genetics of complex traits in psychiatry
(2015) Biological Psychiatry, 77 (1), pp. 36-42. Cited 1 time.

DOI: 10.1016/j.biopsych.2014.08.005
DOCUMENT TYPE: Review

Tang, J., Fang, F., Miller, D.F., Pilrose, J.M., Matei, D., Huang, T.H.-M., Nephew, K.P.
Global DNA methylation profiling technologies and the ovarian cancer methylome
(2015) Methods in Molecular Biology, 1238, pp. 653-675.

DOI: 10.1007/978-1-4939-1804-1_34
DOCUMENT TYPE: Article

Hancarova, M., Puchmajerova, A., Drabova, J., Karaskova, E., Vlckova, M., Sedlacek, Z.
Deletions of 9q21.3 including NTRK2 are associated with severe phenotype
(2015) American Journal of Medical Genetics, Part A, 167 (1), pp. 264-267.

DOI: 10.1002/ajmg.a.36797
DOCUMENT TYPE: Letter

Sante, T., Vergult, S., Volders, P.-J., Kloosterman, W.P., Trooskens, G., De Preter, K., Dheedene, A., Speleman, F., De Meyer, T., Menten, B.
ViVar: A comprehensive platform for the analysis and visualization of structural genomic variation
(2014) PLoS ONE, 9 (12), art. no. e113800, .

DOI: 10.1371/journal.pone.0113800
DOCUMENT TYPE: Article

Snow, A.N., Stence, A.A., Pruessner, J.A., Bossler, A.D., Ma, D.
A simple and cost-effective method of DNA extraction from small formalin-fixed paraffin-embedded tissue for molecular oncologic testing
(2014) BMC Clinical Pathology, 14 (1), art. no. 30, .

DOI: 10.1186/1472-6890-14-30
DOCUMENT TYPE: Article

Hommelsheim, C.M., Frantzeskakis, L., Huang, M., Ülker, B.
PCR amplification of repetitive DNA: A limitation to genome editing technologies and many other applications
(2014) Scientific Reports, 4, art. no. 5052, .

DOI: 10.1038/srep05052
DOCUMENT TYPE: Article

Ordulu, Z., Wong, K.E., Currall, B.B., Ivanov, A.R., Pereira, S., Althari, S., Gusella, J.F., Talkowski, M.E., Morton, C.C.
Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature
(2014) American Journal of Human Genetics, 94 (5), pp. 695-709. Cited 1 time.

DOI: 10.1016/j.ajhg.2014.03.020
DOCUMENT TYPE: Article

Looso, M.
Opening the genetic toolbox of niche model organisms with high throughput techniques: Novel proteins in regeneration as a case study
(2014) BioEssays, 36 (4), pp. 407-418. Cited 3 times.

DOI: 10.1002/bies.201300093
DOCUMENT TYPE: Article

Blake, J., Riddell, A., Theiss, S., Gonzalez, A.P., Haase, B., Jauch, A., Janssen, J.W.G., Ibberson, D., Pavlinic, D., Moog, U., Benes, V., Runz, H.
Sequencing of a patient with balanced chromosome abnormalities and neurodevelopmental disease identifies disruption of multiple high risk loci by structural variation
(2014) PLoS ONE, 9 (3), art. no. e90894, . Cited 1 time.

DOI: 10.1371/journal.pone.0090894
DOCUMENT TYPE: Article

Utami, K.H., Hillmer, A.M., Aksoy, I., Chew, E.G.Y., Teo, A.S.M., Zhang, Z., Lee, C.W.H., Chen, P.J., Seng, C.C., Ariyaratne, P.N., Rouam, S.L., Soo, L.S., Yousoof, S., Prokudin, I., Peters, G., Collins, F., Wilson, M., Kakakios, A., Haddad, G., Menuet, A., Perche, O., Tay, S.K.H., Sung, K.W.K., Ruan, X., Ruan, Y., Liu, E.T., Briault, S., Jamieson, R.V., Davila, S., Cacheux, V.
Detection of chromosomal breakpoints in patients with developmental delay and speech disorders
(2014) PLoS ONE, 9 (3), art. no. e90852, . Cited 1 time.

DOI: 10.1371/journal.pone.0090852
DOCUMENT TYPE: Article

Mosen-Ansorena, D., Telleria, N., Veganzones, S., la Orden, V.D., Maestro, M.L., Aransay, A.M.
SeqCNA: An R package for DNA copy number analysis in cancer using high-throughput sequencing
(2014) BMC Genomics, 15 (1), art. no. 178, .

DOI: 10.1186/1471-2164-15-178
DOCUMENT TYPE: Article

Yavaş, G., Koyutürk, M., Gould, M.P., McMahon, S., LaFramboise, T.
DB2: A probabilistic approach for accurate detection of tandem duplication breakpoints using paired-end reads

(2014) BMC Genomics, 15 (1), art. no. 175, .

DOI: 10.1186/1471-2164-15-175
DOCUMENT TYPE: Article

Duan, J., Deng, H.-W., Wang, Y.-P.
Common copy number variation detection from multiple sequenced samples
(2014) IEEE Transactions on Biomedical Engineering, 61 (3), art. no. 6675802, pp. 928-937.

DOI: 10.1109/TBME.2013.2292588
DOCUMENT TYPE: Article

Yang, Z.-H., Zheng, R., Gao, Y., Zhang, Q., Zhang, H.
Abnormal gene expression and gene fusion in lung adenocarcinoma with high-throughput RNA sequencing
(2014) Cancer Gene Therapy, 21 (2), pp. 74-82. Cited 1 time.

DOI: 10.1038/cgt.2013.86
DOCUMENT TYPE: Article

Chen, K., Chen, L., Fan, X., Wallis, J., Ding, L., Weinstock, G.
TIGRA: A targeted iterative graph routing assembler for breakpoint assembly
(2014) Genome Research, 24 (2), pp. 310-317. Cited 6 times.

DOI: 10.1101/gr.162883.113
DOCUMENT TYPE: Article

Flagel, L.E., Willis, J.H., Vision, T.J.
The standing pool of genomic structural variation in a natural population of Mimulus guttatus
(2014) Genome Biology and Evolution, 6 (1), pp. 53-64.

DOI: 10.1093/gbe/evt199
DOCUMENT TYPE: Article

Ma, S., Wang, X., Liu, Y., Gao, J., Zhang, S., Shi, R., Chang, J., Zhao, P., Xia, Q.
Multiplex genomic structure variation mediated by TALEN and ssODN
(2014) BMC Genomics, 15 (1), art. no. 41, . Cited 5 times.

DOI: 10.1186/1471-2164-15-41
DOCUMENT TYPE: Article

Li, W., Freudenberg, J., Miramontes, P.
Diminishing return for increased Mappability with longer sequencing reads: Implications of the k-mer distributions in the human genome
(2014) BMC Bioinformatics, 15 (1), art. no. 2, . Cited 5 times.

DOI: 10.1186/1471-2105-15-2
DOCUMENT TYPE: Article

Bolon, Y.-T., Stec, A.O., Michno, J.-M., Roessler, J., Bhaskar, P.B., Ries, L., Dobbels, A.A., Campbell, B.W., Young, N.P., Anderson, J.E., Grant, D.M., Orf, J.H., Naeve, S.L., Muehlbauer, G.J., Vance, C.P., Stupar, R.M.
Genome resilience and prevalence of segmental duplications following fast neutron irradiation of soybean
(2014) Genetics, 198 (3), pp. 967-981. Cited 1 time.

DOI: 10.1534/genetics.114.170340/-/DC1

DOCUMENT TYPE: Article

Vergult, S., Van Binsbergen, E., Sante, T., Nowak, S., Vanakker, O., Claes, K., Poppe, B., Van Der Aa, N., Van Roosmalen, M.J., Duran, K., Tavakoli-Yaraki, M., Swinkels, M., Van Den Boogaard, M.-J., Van Haelst, M., Roelens, F., Speleman, F., Cuppen, E., Mortier, G., Kloosterman, W.P., Menten, B.
Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations
(2014) European Journal of Human Genetics, 22 (5), pp. 652-659. Cited 2 times.

DOI: 10.1038/ejhg.2013.220
DOCUMENT TYPE: Article

Fernandez-Banet, J., Lee, N.P., Chan, K.T., Gao, H., Liu, X., Sung, W.-K., Tan, W., Fan, S.T., Poon, R.T., Li, S., Ching, K., Rejto, P.A., Mao, M., Kan, Z.
Decoding complex patterns of genomic rearrangement in hepatocellular carcinoma
(2014) Genomics, 103 (2-3), pp. 189-203.

DOI: 10.1016/j.ygeno.2014.01.003
DOCUMENT TYPE: Article

Hanscom, C., Talkowski, M.
Design of large-insert jumping libraries for structural variant detection using Illumina sequencing
(2014) Current Protocols in Human Genetics, (SUPPL.80), art. no. 7.22, . Cited 1 time.

DOI: 10.1002/0471142905.hg0722s80
DOCUMENT TYPE: Article

Martínez-Fundichely, A., Casillas, S., Egea, R., Ràmia, M., Barbadilla, A., Pantano, L., Puig, M., Cáceres, M.
InvFEST, a database integrating information of polymorphic inversions in the human genome
(2014) Nucleic Acids Research, 42 (D1), pp. D1027-D1032. Cited 4 times.

DOI: 10.1093/nar/gkt1122
DOCUMENT TYPE: Article

Chain, F.J.J., Feulner, P.G.D., Panchal, M., Eizaguirre, C., Samonte, I.E., Kalbe, M., Lenz, T.L., Stoll, M., Bornberg-Bauer, E., Milinski, M., Reusch, T.B.H.
Extensive Copy-Number Variation of Young Genes across Stickleback Populations
(2014) PLoS Genetics, 10 (12), 18 p. Cited 1 time.

DOI: 10.1371/journal.pgen.1004830
DOCUMENT TYPE: Article

Bickhart, D.M., Liu, G.E.
The challenges and importance of structural variation detection in livestock
(2014) Frontiers in Genetics, 5 (FEB), art. no. Article 37, . Cited 3 times.

DOI: 10.3389/fgene.2014.00037
DOCUMENT TYPE: Review

Steinberg, K.M., Schneider, V.A., Graves-Lindsay, T.A., Fulton, R.S., Agarwala, R., Huddleston, J., Shiryev, S.A., Morgulis, A., Surti, U., Warren, W.C., Church, D.M., Eichler, E.E., Wilson, R.K.
Single haplotype assembly of the human genome from a hydatidiform mole
(2014) Genome Research, 24 (12), pp. 2066-2076. Cited 1 time.

DOI: 10.1101/gr.180893.114
DOCUMENT TYPE: Article

Gao, J., Guan, R., Qi, F.
Strategies for improving accuracy of structural variation prediction using read pairs
(2014) Proceedings - 2013 International Conference on Information Science and Cloud Computing Companion, ISCC-C 2013, art. no. 6973636, pp. 463-468.

DOI: 10.1109/ISCC-C.2013.127
DOCUMENT TYPE: Conference Paper

Baik, J.Y., Lee, K.H.
Toward product attribute control: Developments from genome sequencing
(2014) Current Opinion in Biotechnology, 30, pp. 40-44. Cited 2 times.

DOI: 10.1016/j.copbio.2014.05.001
DOCUMENT TYPE: Review

Nuttle, X., Itsara, A., Shendure, J., Eichler, E.E.
Resolving genomic disorder-associated breakpoints within segmental DNA duplications using massively parallel sequencing
(2014) Nature Protocols, 9 (6), pp. 1496-1513. Cited 1 time.

DOI: 10.1038/nprot.2014.096
DOCUMENT TYPE: Article

Schwarz, R.F., Trinh, A., Sipos, B., Brenton, J.D., Goldman, N., Markowetz, F.
Phylogenetic Quantification of Intra-tumour Heterogeneity
(2014) PLoS Computational Biology, 10 (4), art. no. e1003535, . Cited 4 times.

DOI: 10.1371/journal.pcbi.1003535
DOCUMENT TYPE: Article

Zhang, L.-M., Luo, H., Liu, Z.-Q., Zhao, Y., Luo, J.-C., Hao, D.-Y., Jing, H.-C.
Genome-wide patterns of large-size presence/absence variants in sorghum
(2014) Journal of Integrative Plant Biology, 56 (1), pp. 24-37. Cited 2 times.

DOI: 10.1111/jipb.12121
DOCUMENT TYPE: Article

Liu, T., Xie, L., Ye, J., He, X.
Family-based analysis identified CD2 as a susceptibility gene for primary open angle glaucoma in Chinese Han population
(2014) Journal of Cellular and Molecular Medicine, 18 (4), pp. 600-609.

DOI: 10.1111/jcmm.12201
DOCUMENT TYPE: Article

Wilson-Sánchez, D., Rubio-Díaz, S., Muñoz-Viana, R., Pérez-Pérez, J.M., Jover-Gil, S., Ponce, M.R., Micol, J.L.
Leaf phenomics: A systematic reverse genetic screen for Arabidopsis leaf mutants
(2014) Plant Journal, 79 (5), pp. 878-891. Cited 1 time.

DOI: 10.1111/tpj.12595
DOCUMENT TYPE: Article

Ritz, A., Bashir, A., Sindi, S., Hsu, D., Hajirasouliha, I., Raphael, B.J.
Characterization of Structural variants with single molecule and hybrid sequencing approaches
(2014) Bioinformatics, 30 (24), pp. 3458-3466.

DOI: 10.1093/bioinformatics/btu714
DOCUMENT TYPE: Article

Muñoz-Minjares, J., Cabal-Aragón, J., Shmaliy, Y.S.
Confidence masks for genome DNA copy number variations in applications to HR-CGH array measurements
(2014) Biomedical Signal Processing and Control, 13 (1), pp. 337-344.

DOI: 10.1016/j.bspc.2014.06.006
DOCUMENT TYPE: Article

Rogers, R.L., Cridland, J.M., Shao, L., Hu, T.T., Andolfatto, P., Thornton, K.R.
Landscape of standing variation for tandem duplications in drosophila yakuba and drosophila simulans
(2014) Molecular Biology and Evolution, 31 (7), pp. 1750-1766.

DOI: 10.1093/molbev/msu124
DOCUMENT TYPE: Article

Li, J., Kannan, M., Trivett, A.L., Liao, H., Wu, X., Akagi, K., Symer, D.E.
An antisense promoter in mouse L1 retrotransposon open reading frame-1 initiates expression of diverse fusion transcripts and limits retrotransposition
(2014) Nucleic Acids Research, 42 (7), pp. 4546-4562. Cited 3 times.

DOI: 10.1093/nar/gku091
DOCUMENT TYPE: Article

Ma, J., Xiong, M., You, M., Lozano, G., Amos, C.I.
Genome-wide association tests of inversions with application to psoriasis
(2014) Human Genetics, 133 (8), pp. 967-974.

DOI: 10.1007/s00439-014-1437-1
DOCUMENT TYPE: Article

Kitchen, R.R., Rozowsky, J.S., Gerstein, M.B., Nairn, A.C.
Decoding neuroproteomics: Integrating the genome, translatome and functional anatomy
(2014) Nature Neuroscience, 17 (11), pp. 1491-1499.

DOI: 10.1038/nn.3829
DOCUMENT TYPE: Review

Keane, T.M., Wong, K., Adams, D.J., Flint, J., Reymond, A., Yalcin, B.
Identification of structural variation in mouse genomes
(2014) Frontiers in Genetics, 5 (JUL), art. no. Article 192, . Cited 1 time.

DOI: 10.3389/fgene.2014.00192
DOCUMENT TYPE: Review

Aguado, C., Gayà-Vidal, M., Villatoro, S., Oliva, M., Izquierdo, D., Giner-Delgado, C., Montalvo, V., García-González, J., Martínez-Fundichely, A., Capilla, L., Ruiz-Herrera, A., Estivill, X., Puig, M., Cáceres, M.

Validation and Genotyping of Multiple Human Polymorphic Inversions Mediated by Inverted Repeats Reveals a High Degree of Recurrence
(2014) PLoS Genetics, 10 (3), art. no. e1004208, .

DOI: 10.1371/journal.pgen.1004208
DOCUMENT TYPE: Article

Talkowski, M.E., Minikel, E.V., Gusella, J.F.
Autism spectrum disorder genetics: Diverse genes with diverse clinical outcomes
(2014) Harvard Review of Psychiatry, 22 (2), pp. 65-75. Cited 1 time.

DOI: 10.1097/HRP.0000000000000002
DOCUMENT TYPE: Review

Adam-Blondon, A.-F.
Grapevine genome update and beyond
(2014) Acta Horticulturae, 1046, pp. 311-318. Cited 1 time.

DOCUMENT TYPE: Article

Gillespie, R.L., O'Sullivan, J., Ashworth, J., Bhaskar, S., Williams, S., Biswas, S., Kehdi, E., Ramsden, S.C., Clayton-Smith, J., Black, G.C., Lloyd, I.C.
Personalized diagnosis and management of congenital cataract by next-generation sequencing
(2014) Ophthalmology, 121 (11), pp. 2124-2137.e2. Cited 1 time.

DOI: 10.1016/j.ophtha.2014.06.006
DOCUMENT TYPE: Article

Flynn, T.J., Phipps-Green, A., Hollis-Moffatt, J.E., Merriman, M.E., Topless, R., Montgomery, G., Chapman, B., Stamp, L.K., Dalbeth, N., Merriman, T.R.
Association analysis of the SLC22A11 (organic anion transporter 4) and SLC22A12 (urate transporter 1) urate transporter locus with gout in New Zealand case-control sample sets reveals multiple ancestral-specific effects
(2013) Arthritis Research and Therapy, 15 (6), art. no. R220, . Cited 1 time.

DOI: 10.1186/ar4417
DOCUMENT TYPE: Article

Zhang, C.-Z., Leibowitz, M.L., Pellman, D.
Chromothripsis and beyond: Rapid genome evolution from complex chromosomal rearrangements
(2013) Genes and Development, 27 (23), pp. 2513-2530. Cited 10 times.

DOI: 10.1101/gad.229559.113
DOCUMENT TYPE: Review

Kasaian, K., Li, Y.Y., Jones, S.J.M.
Bioinformatics for Cancer Genomics
(2013) Cancer Genomics: From Bench to Personalized Medicine, pp. 133-152.

DOI: 10.1016/B978-0-12-396967-5.00009-8
DOCUMENT TYPE: Book Chapter

Gao, J., Guan, R., Qi, F.
Methods for detecting genome structural variation based on PEM
(2013) Beijing Jiaotong Daxue Xuebao/Journal of Beijing Jiaotong University, 37 (6), pp. 8-12.

DOCUMENT TYPE: Article

Duan, J., Wan, M., Deng, H.-W., Wang, Y.-P.
Modeling exome sequencing data with generalized Gaussian distribution with application to copy number variation detection
(2013) Proceedings - 2013 IEEE International Conference on Bioinformatics and Biomedicine, IEEE BIBM 2013, art. no. 6732619, pp. 1-7.

DOI: 10.1109/BIBM.2013.6732619
DOCUMENT TYPE: Conference Paper

Livnat, A.
Interaction-based evolution: How natural selection and nonrandom mutation work together
(2013) Biology Direct, 8 (1), art. no. 24, . Cited 2 times.

DOI: 10.1186/1745-6150-8-24
DOCUMENT TYPE: Article

Plass, C., Pfister, S.M., Lindroth, A.M., Bogatyrova, O., Claus, R., Lichter, P.
Mutations in regulators of the epigenome and their connections to global chromatin patterns in cancer
(2013) Nature Reviews Genetics, 14 (11), pp. 765-780. Cited 40 times.

DOI: 10.1038/nrg3554
DOCUMENT TYPE: Review

Liu, B., Morrison, C.D., Johnson, C.S., Trump, D.L., Qin, M., Conroy, J.C., Wang, J., Liu, S.
Computational methods for detecting copy number variations in cancer genome using next generation sequencing: Principles and challenges
(2013) Oncotarget, 4 (11), pp. 1868-1881. Cited 3 times.

DOCUMENT TYPE: Article

Haraksingh, R.R., Snyder, M.P.
Impacts of variation in the human genome on gene regulation
(2013) Journal of Molecular Biology, 425 (21), pp. 3970-3977. Cited 10 times.

DOI: 10.1016/j.jmb.2013.07.015
DOCUMENT TYPE: Review

Ratnapriya, R., Swaroop, A.
Genetic architecture of retinal and macular degenerative diseases: The promise and challenges of next-generation sequencing
(2013) Genome Medicine, 5 (9), art. no. 84, . Cited 1 time.

DOI: 10.1186/gm488
DOCUMENT TYPE: Review

Sankaranarayanan, K., Taleei, R., Rahmanian, S., Nikjoo, H.
Ionizing radiation and genetic risks. XVII. Formation mechanisms underlying naturally occurring DNA deletions in the human genome and their potential relevance for bridging the gap between induced DNA double-strand breaks and deletions in irradiated germ cells
(2013) Mutation Research - Reviews in Mutation Research, 753 (2), pp. 114-130. Cited 5 times.

DOI: 10.1016/j.mrrev.2013.07.003
DOCUMENT TYPE: Review

Dong, Z.C., Chen, Y.
Transcriptomics: Advances and approaches
(2013) Science China Life Sciences, 56 (10), pp. 960-967.

DOI: 10.1007/s11427-013-4557-2
DOCUMENT TYPE: Review

Zhao, M., Wang, Q., Wang, Q., Jia, P., Zhao, Z.
Computational tools for copy number variation (CNV) detection using next-generation sequencing data: Features and perspectives
(2013) BMC Bioinformatics, 14 (SUPPL11), art. no. S1, . Cited 7 times.

DOI: 10.1186/1471-2105-14-S11-S1
DOCUMENT TYPE: Article

Yegnasubramanian, S.
Explanatory chapter: Next generation sequencing
(2013) Methods in Enzymology, 529, pp. 201-208.

DOI: 10.1016/B978-0-12-418687-3.00016-1
DOCUMENT TYPE: Article

Valsesia, A., Macé, A., Jacquemont, S., Beckmann, J.S., Kutalik, Z.
The growing importance of CNVs: New insights for detection and clinical interpretation
(2013) Frontiers in Genetics, 4 (MAY), art. no. Article 92, . Cited 6 times.

DOI: 10.3389/fgene.2013.00092
DOCUMENT TYPE: Review

Lehrach, H.
DNA sequencing methods in human genetics and disease research
(2013) F1000Prime Reports, 5, art. no. 34, .

DOI: 10.12703/P5-34
DOCUMENT TYPE: Article

Morey, M., Fernández-Marmiesse, A., Castiñeiras, D., Fraga, J.M., Couce, M.L., Cocho, J.A.
A glimpse into past, present, and future DNA sequencing
(2013) Molecular Genetics and Metabolism, 110 (1-2), pp. 3-24. Cited 11 times.

DOI: 10.1016/j.ymgme.2013.04.024
DOCUMENT TYPE: Review

Sakai, R., Moisse, M., Reumers, J., Aerts, J.
Pipit: Visualizing functional impacts of structural variations
(2013) Bioinformatics, 29 (17), pp. 2206-2207.

DOI: 10.1093/bioinformatics/btt367
DOCUMENT TYPE: Article

Geurts Van Kessel, A., Venkatachalam, R., Kuiper, R.P.
Colorectal Cancer
(2013) Genomic and Personalized Medicine, 2, pp. 722-732.

DOI: 10.1016/B978-0-12-382227-7.00062-8
DOCUMENT TYPE: Book Chapter

Lee, C.
Structural Genomic Variation in the Human Genome
(2013) Genomic and Personalized Medicine, 1, pp. 123-132.

DOI: 10.1016/B978-0-12-382227-7.00010-0
DOCUMENT TYPE: Book Chapter

Guffanti, G., Torri, F., Rasmussen, J., Clark, A.P., Lakatos, A., Turner, J.A., Fallon, J.H., Saykin, A.J., Weiner, M., Vawter, M.P., Knowles, J.A., Potkin, S.G., Macciardi, F.
Increased CNV-Region deletions in mild cognitive impairment (MCI) and Alzheimer's disease (AD) subjects in the ADNI sample
(2013) Genomics, 102 (2), pp. 112-122. Cited 1 time.

DOI: 10.1016/j.ygeno.2013.04.004
DOCUMENT TYPE: Review

Chen, G., Wang, C., Shi, L., Tong, W., Qu, X., Chen, J., Yang, J., Shi, C., Chen, L., Zhou, P., Lu, B., Shi, T.
Comprehensively identifying and characterizing the missing gene sequences in human reference genome with integrated analytic approaches
(2013) Human Genetics, 132 (8), pp. 899-911. Cited 1 time.

DOI: 10.1007/s00439-013-1300-9
DOCUMENT TYPE: Article

Guo, X., Brenner, M., Zhang, X., Laragione, T., Tai, S., Li, Y., Bu, J., Yin, Y., Shah, A.A., Kwan, K., Li, Y., Jun, W., Gulko, P.S.
Whole-genome sequences of DA and F344 rats with different susceptibilities to arthritis, autoimmunity, inflammation and cancer
(2013) Genetics, 194 (4), pp. 1017-1028. Cited 2 times.

DOI: 10.1534/genetics.113.153049
DOCUMENT TYPE: Article

Wellensiek, B.P., Larsen, A.C., Stephens, B., Kukurba, K., Waern, K., Briones, N., Liu, L., Snyder, M., Jacobs, B.L., Kumar, S., Chaput, J.C.
Genome-wide profiling of human cap-independent translation-enhancing elements
(2013) Nature Methods, 10 (8), pp. 747-750. Cited 1 time.

DOI: 10.1038/nmeth.2522
DOCUMENT TYPE: Article

Paudel, Y., Madsen, O., Megens, H.-J., Frantz, L.A.F., Bosse, M., Bastiaansen, J.W.M., Crooijmans, R.P.M.A., Groenen, M.A.M.
Evolutionary dynamics of copy number variation in pig genomes in the context of adaptation and domestication
(2013) BMC Genomics, 14 (1), art. no. 449, . Cited 11 times.

DOI: 10.1186/1471-2164-14-449
DOCUMENT TYPE: Article

El-Sayed Moustafa, J.S., Froguel, P.
From obesity genetics to the future of personalized obesity therapy
(2013) Nature Reviews Endocrinology, 9 (7), pp. 402-413. Cited 17 times.

DOI: 10.1038/nrendo.2013.57
DOCUMENT TYPE: Review

Kim, S., Medvedev, P., Paton, T.A., Bafna, V.
Reprever: Resolving low-copy duplicated sequences using template driven assembly
(2013) Nucleic Acids Research, 41 (12), . Cited 2 times.

DOI: 10.1093/nar/gkt339
DOCUMENT TYPE: Article

Zhou, W., Zhang, F., Chen, X., Shen, Y., Lupski, J.R., Jin, L.
Increased genome instability in human DNA segments with self-chains: Homology-induced structural variations via replicative mechanisms
(2013) Human Molecular Genetics, 22 (13), pp. 2642-2651. Cited 2 times.

DOI: 10.1093/hmg/ddt113
DOCUMENT TYPE: Article

Brunham, L.R., Hayden, M.R.
Hunting human disease genes: Lessons from the past, challenges for the future
(2013) Human Genetics, 132 (6), pp. 603-617. Cited 4 times.

DOI: 10.1007/s00439-013-1286-3
DOCUMENT TYPE: Review

Mardis, E.R.
Next-generation sequencing platforms
(2013) Annual Review of Analytical Chemistry, 6, pp. 287-303. Cited 49 times.

DOI: 10.1146/annurev-anchem-062012-092628
DOCUMENT TYPE: Article

Escaramís, G., Tornador, C., Bassaganyas, L., Rabionet, R., Tubio, J.M.C., Martínez-Fundichely, A., Cáceres, M., Gut, M., Ossowski, S., Estivill, X.
PeSV-Fisher: Identification of Somatic and Non-Somatic Structural Variants Using Next Generation Sequencing Data
(2013) PLoS ONE, 8 (5), art. no. e63377, . Cited 3 times.

DOI: 10.1371/journal.pone.0063377
DOCUMENT TYPE: Article

Ezawa, K., Landan, G., Graur, D.
Detecting negative selection on recurrent mutations using gene genealogy
(2013) BMC Genetics, 14, art. no. 37, .

DOI: 10.1186/1471-2156-14-37
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.

CNV-TV: A robust method to discover copy number variation from short sequencing reads
(2013) BMC Bioinformatics, 14 (1), art. no. 150, .

DOI: 10.1186/1471-2105-14-150
DOCUMENT TYPE: Article

Mwenifumbo, J.C., Marra, M.A.
Cancer genome-sequencing study design
(2013) Nature Reviews Genetics, 14 (5), pp. 321-332. Cited 19 times.

DOI: 10.1038/nrg3445
DOCUMENT TYPE: Review

Lucas Lledó, J.I., Cáceres, M.
On the Power and the Systematic Biases of the Detection of Chromosomal Inversions by Paired-End Genome Sequencing
(2013) PLoS ONE, 8 (4), art. no. e61292, . Cited 3 times.

DOI: 10.1371/journal.pone.0061292
DOCUMENT TYPE: Article

Bassaganyas, L., Riveira-Muñoz, E., García-Aragonés, M., González, J.R., Cáceres, M., Armengol, L., Estivill, X.
Worldwide population distribution of the common LCE3C-LCE3B deletion associated with psoriasis and other autoimmune disorders
(2013) BMC Genomics, 14 (1), art. no. 261, . Cited 2 times.

DOI: 10.1186/1471-2164-14-261
DOCUMENT TYPE: Article

Gahan, P.B.
Circulating nucleic acids in plasma and serum: Applications in diagnostic techniques for noninvasive prenatal diagnosis
(2013) International Journal of Women's Health, 5 (1), pp. 177-186. Cited 2 times.

DOI: 10.2147/IJWH.S34442
DOCUMENT TYPE: Review

Verdin, H., D'haene, B., Beysen, D., Novikova, Y., Menten, B., Sante, T., Lapunzina, P., Nevado, J., Carvalho, C.M.B., Lupski, J.R., de Baere, E.
Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain
(2013) PLoS Genetics, 9 (3), art. no. e1003358, . Cited 10 times.

DOI: 10.1371/journal.pgen.1003358
DOCUMENT TYPE: Article

Walter, V., Nobel, A.B., Hayes, D.N., Wright, F.A.
Identification of Recurrent DNA Copy Number Aberrations in Tumors
(2013) Statistical Diagnostics for Cancer: Analyzing High-Dimensional Data, 3, pp. 239-260.

DOI: 10.1002/9783527665471.ch13
DOCUMENT TYPE: Book Chapter

Ray, F.A., Zimmerman, E., Robinson, B., Cornforth, M.N., Bedford, J.S., Goodwin, E.H., Bailey, S.M.
Directional genomic hybridization for chromosomal inversion discovery and detection
(2013) Chromosome Research, 21 (2), pp. 165-174. Cited 2 times.

DOI: 10.1007/s10577-013-9345-0
DOCUMENT TYPE: Article

Van Den Bossche, M.J., Strazisar, M., Cammaerts, S., Liekens, A.M., Vandeweyer, G., Depreeuw, V., Mattheijssens, M., Lenaerts, A..-S., De Zutter, S., De Rijk, P., Sabbe, B., Del-Favero, J.
Identification of rare copy number variants in high burden schizophrenia families
(2013) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 162 (3), pp. 273-282. Cited 4 times.

DOI: 10.1002/ajmg.b.32146
DOCUMENT TYPE: Article

Lundin, S., Gruselius, J., Nystedt, B., Lexow, P., Käller, M., Lundeberg, J.
Hierarchical molecular tagging to resolve long continuous sequences by massively parallel sequencing
(2013) Scientific Reports, 3, art. no. 1186, .

DOI: 10.1038/srep01186
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Deng, H.-W., Wang, Y.-P.
Comparative Studies of Copy Number Variation Detection Methods for Next-Generation Sequencing Technologies
(2013) PLoS ONE, 8 (3), art. no. e59128, . Cited 24 times.

DOI: 10.1371/journal.pone.0059128
DOCUMENT TYPE: Article

Ragheb, M.N., Ford, C.B., Chase, M.R., Lin, P.L., Flynn, J.L., Fortune, S.M.
The mutation rate of mycobacterial repetitive unit loci in strains of M. tuberculosis from cynomolgus macaque infection
(2013) BMC Genomics, 14 (1), art. no. 145, . Cited 3 times.

DOI: 10.1186/1471-2164-14-145
DOCUMENT TYPE: Article

Singer, M.A.
Are chronic degenerative diseases part of the ageing process? Insights from comparative biology
(2013) Are Chronic Degenerative Diseases Part of the Ageing Process? Insights from Comparative Biology, pp. 1-217.

DOCUMENT TYPE: Book

Coonrod, E.M., Durtschi, J.D., Margraf, R.L., Voelkerding, K.V.
Developing genome and exome sequencing for candidate gene identification in inherited disorders: An integrated technical and bioinformatics approach
(2013) Archives of Pathology and Laboratory Medicine, 137 (3), pp. 415-433. Cited 11 times.

DOI: 10.5858/arpa.2012-0107-RA
DOCUMENT TYPE: Review

Grimm, D., Hagmann, J., Koenig, D., Weigel, D., Borgwardt, K.
Accurate indel prediction using paired-end short reads
(2013) BMC Genomics, 14 (1), art. no. 132, . Cited 10 times.

DOI: 10.1186/1471-2164-14-132
DOCUMENT TYPE: Article

Soon, W.W., Hariharan, M., Snyder, M.P.
High-throughput sequencing for biology and medicine
(2013) Molecular Systems Biology, 9, art. no. 640, . Cited 40 times.

DOI: 10.1038/msb.2012.61
DOCUMENT TYPE: Review

Hong, S.G., Dunbar, C.E., Winkler, T.
Assessing the risks of genotoxicity in the therapeutic development of induced pluripotent stem cells
(2013) Molecular Therapy, 21 (2), pp. 272-281. Cited 7 times.

DOI: 10.1038/mt.2012.255
DOCUMENT TYPE: Review

Haasl, R.J., Payseur, B.A.
Microsatellites as targets of natural selection
(2013) Molecular Biology and Evolution, 30 (2), pp. 285-298. Cited 8 times.

DOI: 10.1093/molbev/mss247
DOCUMENT TYPE: Article

Feulner, P.G.D., Chain, F.J.J., Panchal, M., Eizaguirre, C., Kalbe, M., Lenz, T.L., Mundry, M., Samonte, I.E., Stoll, M., Milinski, M., Reusch, T.B.H., Bornberg-Bauer, E.
Genome-wide patterns of standing genetic variation in a marine population of three-spined sticklebacks
(2013) Molecular Ecology, 22 (3), pp. 635-649. Cited 21 times.

DOI: 10.1111/j.1365-294X.2012.05680.x
DOCUMENT TYPE: Conference Paper

Gong, Q., Tao, Y., Yang, J.-R., Cai, J., Yuan, Y., Ruan, J., Yang, J., Liu, H., Li, W., Lu, X., Zhuang, S.-M., Wang, S.M., Wu, C.-I.
Identification of medium-sized genomic deletions with low coverage, mate-paired restricted tags
(2013) BMC Genomics, 14 (1), art. no. 51, .

DOI: 10.1186/1471-2164-14-51
DOCUMENT TYPE: Article

Schluth-Bolard, C., Labalme, A., Cordier, M.-P., Till, M., Nadeau, G., Tevissen, H., Lesca, G., Boutry-Kryza, N., Rossignol, S., Rocas, D., Dubruc, E., Edery, P., Sanlaville, D.
Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations
(2013) Journal of Medical Genetics, 50 (3), pp. 144-150. Cited 14 times.

DOI: 10.1136/jmedgenet-2012-101351
DOCUMENT TYPE: Article

Li, W., Olivier, M.
Current analysis platforms and methods for detecting copy number variation
(2013) Physiological Genomics, 45 (1), pp. 1-6. Cited 6 times.

DOI: 10.1152/physiolgenomics.00082.2012
DOCUMENT TYPE: Article

Li, H.
Systems genetics in "-omics" era: Current and future development
(2013) Theory in Biosciences, 132 (1), pp. 1-16. Cited 2 times.

DOI: 10.1007/s12064-012-0168-x
DOCUMENT TYPE: Review

Solieri, L., Dakal, T.C., Giudici, P.
Next-generation sequencing and its potential impact on food microbial genomics
(2013) Annals of Microbiology, 63 (1), pp. 21-37. Cited 6 times.

DOI: 10.1007/s13213-012-0478-8
DOCUMENT TYPE: Review

Kunz, M., Dannemann, M., Kelso, J.
High-throughput sequencing of the melanoma genome
(2013) Experimental Dermatology, 22 (1), pp. 10-17. Cited 8 times.

DOI: 10.1111/exd.12054
DOCUMENT TYPE: Article

Wittler, R.
Unraveling overlapping deletions by agglomerative clustering
(2013) BMC Genomics, 14, art. no. S12, . Cited 1 time.

DOI: 10.1186/1471-2164-14-S1-S12
DOCUMENT TYPE: Article

Schulte, I., Batty, E.M., Pole, J.C.M., Blood, K.A., Mo, S., Cooke, S.L., Ng, C., Howe, K.L., Chin, S.-F., Brenton, J.D., Caldas, C., Howarth, K.D., Edwards, P.A.W.
Structural analysis of the genome of breast cancer cell line ZR-75-30 identifies twelve expressed fusion genes
(2012) BMC Genomics, 13 (1), art. no. 719, . Cited 2 times.

DOI: 10.1186/1471-2164-13-719
DOCUMENT TYPE: Article

Xu, H., Luo, X., Qian, J., Pang, X., Song, J., Qian, G., Chen, J., Chen, S.
FastUniq: A Fast De Novo Duplicates Removal Tool for Paired Short Reads
(2012) PLoS ONE, 7 (12), art. no. e52249, . Cited 4 times.

DOI: 10.1371/journal.pone.0052249
DOCUMENT TYPE: Article

Zhou, S., Fu, Y., Li, J., He, L., Cai, X., Yan, Q., Rao, X., Huang, S., Li, G., Wang, Y., Xu, A.
HTS-PEG: A Method for High Throughput Sequencing of the Paired-Ends of Genomic Libraries
(2012) PLoS ONE, 7 (12), art. no. e52257, .

DOI: 10.1371/journal.pone.0052257
DOCUMENT TYPE: Article

Wu, X., Zhang, D., Li, G.
Insights into the regulation of human CNV-miRNAs from the view of their target genes
(2012) BMC Genomics, 13 (1), art. no. 707, . Cited 5 times.

DOI: 10.1186/1471-2164-13-707
DOCUMENT TYPE: Article

Talkowski, M.E., Ordulu, Z., Pillalamarri, V., Benson, C.B., Blumenthal, I., Connolly, S., Hanscom, C., Hussain, N., Pereira, S., Picker, J., Rosenfeld, J.A., Shaffer, L.G., Wilkins-Haug, L.E., Gusella, J.F., Morton, C.C.
Clinical diagnosis by whole-genome sequencing of a prenatal sample
(2012) New England Journal of Medicine, 367 (23), pp. 2226-2232. Cited 30 times.

DOI: 10.1056/NEJMoa1208594
DOCUMENT TYPE: Article

Alves, J.M., Lopes, A.M., Chikhi, L., Amorim, A.
On the structural plasticity of the human genome: Chromosomal inversions revisited
(2012) Current Genomics, 13 (8), pp. 623-632. Cited 4 times.

DOI: 10.2174/138920212803759703
DOCUMENT TYPE: Review

Pan, S., Caleshu, C.A., Dunn, K.E., Ashley, E.A.
Cardiac structural and sarcomere genes associated with cardiomyopathy exhibit marked intolerance of genetic variation
(2012) Circulation: Cardiovascular Genetics, 5 (6), pp. 602-610. Cited 15 times.

DOI: 10.1161/CIRCGENETICS.112.963421
DOCUMENT TYPE: Article

Chin, B.L., Ryan, O., Lewitter, F., Boone, C., Fink, G.R.
Genetic variation in Saccharomyces cerevisiae: Circuit diversification in a signal transduction network
(2012) Genetics, 192 (4), pp. 1523-1532. Cited 5 times.

DOI: 10.1534/genetics.112.145573
DOCUMENT TYPE: Article

Raphael, B.J.
Chapter 6: Structural Variation and Medical Genomics
(2012) PLoS Computational Biology, 8 (12), art. no. e1002821, . Cited 3 times.

DOI: 10.1371/journal.pcbi.1002821
DOCUMENT TYPE: Article

Milward, E.A., Daneshi, N., Johnstone, D.M.
Emerging real-time technologies in molecular medicine and the evolution of integrated 'pharmacomics' approaches to personalized medicine and drug discovery
(2012) Pharmacology and Therapeutics, 136 (3), pp. 295-304. Cited 2 times.

DOI: 10.1016/j.pharmthera.2012.08.008
DOCUMENT TYPE: Review

dela Paz, J.S., Stronghill, P.E., Douglas, S.J., Saravia, S., Hasenkampf, C.A., Riggs, C.D.
Chromosome Fragile Sites in Arabidopsis Harbor Matrix Attachment Regions That May Be Associated with Ancestral Chromosome Rearrangement Events
(2012) PLoS Genetics, 8 (12), art. no. e1003136, . Cited 1 time.

DOI: 10.1371/journal.pgen.1003136
DOCUMENT TYPE: Article

Hardiman, G.
Application of Ultra-High Throughput Sequencing and Microarray Technologies in Pharmacogenomics Testing
(2012) Therapeutic Drug Monitoring, pp. 143-159. Cited 1 time.

DOI: 10.1016/B978-0-12-385467-4.00007-5
DOCUMENT TYPE: Book Chapter

Elbaidouri, M., Panaud, O.
Genome-wide analysis of transposition using next generation sequencing technologies
(2012) Topics in Current Genetics, 24, pp. 59-70. Cited 2 times.

DOI: 10.1007/978-3-642-31842-9-4
DOCUMENT TYPE: Article

Gahan, P.B.
Biology of circulating nucleic acids and possible roles in diagnosis and treatment in diabetes and cancer
(2012) Infectious Disorders - Drug Targets, 12 (5), pp. 360-370. Cited 4 times.

DOI: 10.2174/187152612804142224
DOCUMENT TYPE: Article

Jones, M.J.K., Jallepalli, P.V.
Chromothripsis: Chromosomes in Crisis
(2012) Developmental Cell, 23 (5), pp. 908-917. Cited 22 times.

DOI: 10.1016/j.devcel.2012.10.010
DOCUMENT TYPE: Review

Schwartz, J.J., Lee, C., Hiatt, J.B., Adey, A., Shendure, J.
Capturing native long-range contiguity by in situ library construction and optical sequencing
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (46), pp. 18749-18754. Cited 3 times.

DOI: 10.1073/pnas.1202680109
DOCUMENT TYPE: Article

Falconer, E., Hills, M., Naumann, U., Poon, S.S.S., Chavez, E.A., Sanders, A.D., Zhao, Y., Hirst, M., Lansdorp, P.M.
DNA template strand sequencing of single-cells maps genomic rearrangements at high resolution
(2012) Nature Methods, 9 (11), pp. 1107-1112. Cited 17 times.

DOI: 10.1038/nmeth.2206
DOCUMENT TYPE: Article

Marian, A.J.
Challenges in Medical Applications of Whole Exome/Genome Sequencing Discoveries
(2012) Trends in Cardiovascular Medicine, 22 (8), pp. 219-223. Cited 8 times.

DOI: 10.1016/j.tcm.2012.08.001
DOCUMENT TYPE: Review

Han, J.S., Shao, S.
Circular retrotransposition products generated by a LINE retrotransposon

(2012) Nucleic Acids Research, 40 (21), pp. 10866-10877. Cited 3 times.

DOI: 10.1093/nar/gks859
DOCUMENT TYPE: Article

Liu, G.E., Bickhart, D.M.
Copy number variation in the cattle genome
(2012) Functional and Integrative Genomics, 12 (4), pp. 609-624. Cited 10 times.

DOI: 10.1007/s10142-012-0289-9
DOCUMENT TYPE: Review

Coughlin II, C.R., Scharer, G.H., Shaikh, T.H.
Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns
(2012) Genome Medicine, 4 (10), art. no. 80, . Cited 1 time.

DOI: 10.1186/gm381
DOCUMENT TYPE: Review

Mijušković, M., Brown, S.M., Tang, Z., Lindsay, C.R., Efstathiadis, E., Deriano, L., Roth, D.B.
A Streamlined Method for Detecting Structural Variants in Cancer Genomes by Short Read Paired-End Sequencing
(2012) PLoS ONE, 7 (10), art. no. e48314, . Cited 6 times.

DOI: 10.1371/journal.pone.0048314
DOCUMENT TYPE: Article

Hunter, R.G., Murakami, G., Dewell, S., Seligsohn, M., Baker, M.E.R., Datson, N.A., McEwen, B.S., Pfaff, D.W.
Acute stress and hippocampal histone H3 lysine 9 trimethylation, a retrotransposon silencing response
(2012) Proceedings of the National Academy of Sciences of the United States of America, 109 (43), pp. 17657-17662. Cited 31 times.

DOI: 10.1073/pnas.1215810109
DOCUMENT TYPE: Article

Shibata, T.
Cancer genomics and pathology: All Together Now
(2012) Pathology International, 62 (10), pp. 647-659. Cited 3 times.

DOI: 10.1111/j.1440-1827.2012.02855.x
DOCUMENT TYPE: Review

Langley, C.H., Stevens, K., Cardeno, C., Lee, Y.C.G., Schrider, D.R., Pool, J.E., Langley, S.A., Suarez, C., Corbett-Detig, R.B., Kolaczkowski, B., Fang, S., Nista, P.M., Holloway, A.K., Kern, A.D., Dewey, C.N., Song, Y.S., Hahn, M.W., Begun, D.J.
Genomic variation in natural populations of Drosophila melanogaster
(2012) Genetics, 192 (2), pp. 533-598. Cited 65 times.

DOI: 10.1534/genetics.112.142018
DOCUMENT TYPE: Article

Simon, M.M., Mallon, A.-M., Howell, G.R., Reinholdt, L.G.
High throughput sequencing approaches to mutation discovery in the mouse
(2012) Mammalian Genome, 23 (9-10), pp. 499-513.

DOI: 10.1007/s00335-012-9424-0
DOCUMENT TYPE: Article

Ehli, E.A., Abdellaoui, A., Hu, Y., Hottenga, J.J., Kattenberg, M., Van Beijsterveldt, T., Bartels, M., Althoff, R.R., Xiao, X., Scheet, P., De Geus, E.J., Hudziak, J.J., Boomsma, D.I., Davies, G.E.
De novo and inherited CNVs in MZ twin pairs selected for discordance and concordance on Attention Problems
(2012) European Journal of Human Genetics, 20 (10), pp. 1037-1043. Cited 14 times.

DOI: 10.1038/ejhg.2012.49
DOCUMENT TYPE: Article

Charney, E.
Humans, fruit flies, and automatons
(2012) Behavioral and Brain Sciences, 35 (5), pp. 381-410.

DOI: 10.1017/S0140525X12001501
DOCUMENT TYPE: Article

Zhang, N.R., Siegmund, D.O.
Model selection for high-dimensional, multi-sequence change-point problems
(2012) Statistica Sinica, 22 (4), pp. 1507-1538. Cited 1 time.

DOI: 10.5705/ss.2010.257
DOCUMENT TYPE: Article

Yao, F., Ariyaratne, P.N., Hillmer, A.M., Lee, W.H., Li, G., Teo, A.S.M., Woo, X.Y., Zhang, Z., Chen, J.P., Poh, W.T., Zawack, K.F.B., Chan, C.S., Leong, S.T., Neo, S.C., Choi, P.S.D., Gao, S., Nagarajan, N., Thoreau, H., Shahab, A., Ruan, X., Cacheux-Rataboul, V., Wei, C.-L., Bourque, G., Sung, W.-K., Liu, E.T., Ruan, Y.
Long Span DNA Paired-End-Tag (DNA-PET) Sequencing Strategy for the Interrogation of Genomic Structural Mutations and Fusion-Point-Guided Reconstruction of Amplicons
(2012) PLoS ONE, 7 (9), art. no. e46152, . Cited 4 times.

DOI: 10.1371/journal.pone.0046152
DOCUMENT TYPE: Article

Asan, Geng, C., Chen, Y., Wu, K., Cai, Q., Wang, Y., Lang, Y., Cao, H., Yang, H., Wang, J., Zhang, X.
Paired-End Sequencing of Long-Range DNA Fragments for De Novo Assembly of Large, Complex Mammalian Genomes by Direct Intra-Molecule Ligation
(2012) PLoS ONE, 7 (9), art. no. e46211, . Cited 4 times.

DOI: 10.1371/journal.pone.0046211
DOCUMENT TYPE: Article

Soemedi, R., Wilson, I.J., Bentham, J., Darlay, R., Töpf, A., Zelenika, D., Cosgrove, C., Setchfield, K., Thornborough, C., Granados-Riveron, J., Blue, G.M., Breckpot, J., Hellens, S., Zwolinkski, S., Glen, E., Mamasoula, C., Rahman, T.J., Hall, D., Rauch, A., Devriendt, K., Gewillig, M., O'sullivan, J., Winlaw, D.S., Bu'lock, F., Brook, J.D., Bhattacharya, S., Lathrop, M., Santibanez-Koref, M., Cordell, H.J., Goodship, J.A., Keavney, B.D.
Contribution of global rare copy-number variants to the risk of sporadic congenital heart disease
(2012) American Journal of Human Genetics, 91 (3), pp. 489-501. Cited 43 times.

DOI: 10.1016/j.ajhg.2012.08.003
DOCUMENT TYPE: Article

Rolfe, P.A., Bernstein, D.A., Grisafi, P., Fink, G.R., Gifford, D.K.
Ruler arrays reveal haploid genomic structural variation
(2012) PLoS ONE, 7 (8), art. no. e43210, .

DOI: 10.1371/journal.pone.0043210
DOCUMENT TYPE: Article

Sun, S., Ke, R., Hughes, D., Nilsson, M., Andersson, D.I.
Genome-wide detection of spontaneous chromosomal rearrangements in bacteria
(2012) PLoS ONE, 7 (8), art. no. e42639, . Cited 7 times.

DOI: 10.1371/journal.pone.0042639
DOCUMENT TYPE: Article

Du, R., Lu, C., Jiang, Z., Li, S., Ma, R., An, H., Xu, M., An, Y., Xia, Y., Jin, L., Wang, X., Zhang, F.
Efficient typing of copy number variations in a segmental duplication-mediated rearrangement hotspot using multiplex competitive amplification
(2012) Journal of Human Genetics, 57 (8), pp. 545-551. Cited 12 times.

DOI: 10.1038/jhg.2012.66
DOCUMENT TYPE: Article

Jiang, H., Zeng, X., He, N.
An integrated view of current progress in copy number variations analysis of genome
(2012) Advanced Science Letters, 7, pp. 1-8. Cited 5 times.

DOI: 10.1166/asl.2012.3315
DOCUMENT TYPE: Review

Valencia, A., Hidalgo, M.
Getting personalized cancer genome analysis into the clinic: the challenges in bioinformatics
(2012) Genome Medicine, 4 (8), art. no. 61, . Cited 11 times.

DOI: 10.1186/gm362
DOCUMENT TYPE: Review

Casals, F., Idaghdour, Y., Hussin, J., Awadalla, P.
Next-generation sequencing approaches for genetic mapping of complex diseases
(2012) Journal of Neuroimmunology, 248 (1-2), pp. 10-22. Cited 9 times.

DOI: 10.1016/j.jneuroim.2011.12.017
DOCUMENT TYPE: Review

Belfield, E.J., Gan, X., Mithani, A., Brown, C., Jiang, C., Franklin, K., Alvey, E., Wibowo, A., Jung, M., Bailey, K., Kalwani, S., Ragoussis, J., Mott, R., Harberd, N.P.
Genome-wide analysis of mutations in mutant lineages selected following fast-neutron irradiation mutagenesis of Arabidopsis thaliana
(2012) Genome Research, 22 (7), pp. 1306-1315. Cited 12 times.

DOI: 10.1101/gr.131474.111
DOCUMENT TYPE: Article

Xing, M.-N., Zhang, X.-Z., Huang, H.
Application of metagenomic techniques in mining enzymes from microbial communities for biofuel synthesis

(2012) Biotechnology Advances, 30 (4), pp. 920-929. Cited 14 times.

DOI: 10.1016/j.biotechadv.2012.01.021
DOCUMENT TYPE: Review

Kim, S., Millard, S.P., Yu, C.-E., Leong, L., Radant, A., Dobie, D., Tsuang, D.W., Wijsman, E.M.
Inheritance Model Introduces Differential Bias in CNV Calls Between Parents and Offspring
(2012) Genetic Epidemiology, 36 (5), pp. 488-498. Cited 1 time.

DOI: 10.1002/gepi.21643
DOCUMENT TYPE: Article

González, J.R., Abellán, C., Abellán, J.J.
Bayesian model to detect phenotype-specific genes for copy number data
(2012) BMC Bioinformatics, 13 (1), art. no. 130, .

DOI: 10.1186/1471-2105-13-130
DOCUMENT TYPE: Article

Iskow, R.C., Gokcumen, O., Lee, C.
Exploring the role of copy number variants in human adaptation
(2012) Trends in Genetics, 28 (6), pp. 245-257. Cited 21 times.

DOI: 10.1016/j.tig.2012.03.002
DOCUMENT TYPE: Review

Van Den Bossche, M.J., Strazisar, M., De Bruyne, S., Bervoets, C., Lenaerts, A.-S., De Zutter, S., Nordin, A., Norrback, K.-F., Goossens, D., De Rijk, P., Green, E.K., Grozeva, D., Mendlewicz, J., Craddock, N., Sabbe, B.G., Adolfsson, R., Souery, D., Del-Favero, J.
Identification of a CACNA2D4 deletion in late onset bipolar disorder patients and implications for the involvement of voltage-dependent calcium channels in psychiatric disorders
(2012) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 159 B (4), pp. 465-475. Cited 3 times.

DOI: 10.1002/ajmg.b.32053
DOCUMENT TYPE: Article

Arlt, M.F., Wilson, T.E., Glover, T.W.
Replication stress and mechanisms of CNV formation
(2012) Current Opinion in Genetics and Development, 22 (3), pp. 204-210. Cited 23 times.

DOI: 10.1016/j.gde.2012.01.009
DOCUMENT TYPE: Review

Lai, B., Ding, R., Li, Y., Duan, L., Zhu, H.
A de novo metagenomic assembly program for shotgun DNA reads
(2012) Bioinformatics, 28 (11), art. no. bts162, pp. 1455-1462. Cited 14 times.

DOI: 10.1093/bioinformatics/bts162
DOCUMENT TYPE: Article

Doan, R., Cohen, N., Harrington, J., Veazy, K., Juras, R., Cothran, G., McCue, M.E., Skow, L., Dindot, S.V.
Identification of copy number variants in horses
(2012) Genome Research, 22 (5), pp. 899-907. Cited 13 times.

DOI: 10.1101/gr.128991.111
DOCUMENT TYPE: Article

Li, J., Harris, R.A., Cheung, S.W., Coarfa, C., Jeong, M., Goodell, M.A., White, L.D., Patel, A., Kang, S.-H., Shaw, C., Chinault, A.C., Gambin, T., Gambin, A., Lupski, J.R., Milosavljevic, A.
Genomic hypomethylation in the human germline associates with selective structural mutability in the human genome
(2012) PLoS Genetics, 8 (5), art. no. e1002692, . Cited 19 times.

DOI: 10.1371/journal.pgen.1002692
DOCUMENT TYPE: Article

Rossetti, S., Hopp, K., Sikkink, R.A., Sundsbak, J.L., Lee, Y.K., Kubly, V., Eckloff, B.W., Ward, C.J., Winearls, C.G., Torres, V.E., Harris, P.C.
Identification of gene mutations in autosomal dominant polycystic kidney disease through targeted resequencing
(2012) Journal of the American Society of Nephrology, 23 (5), pp. 915-933. Cited 27 times.

DOI: 10.1681/ASN.2011101032
DOCUMENT TYPE: Article

Furney, S.J., Gundem, G., Lopez-Bigas, N.
Oncogenomics methods and resources
(2012) Cold Spring Harbor Protocols, 7 (5), pp. 546-564. Cited 3 times.

DOI: 10.1101/pdb.top069229
DOCUMENT TYPE: Review

Weise, A., Mrasek, K., Klein, E., Mulatinho, M., Llerena Jr., J.C., Hardekopf, D., Pekova, S., Bhatt, S., Kosyakova, N., Liehr, T.
Microdeletion and Microduplication Syndromes
(2012) Journal of Histochemistry and Cytochemistry, 60 (5), pp. 346-358. Cited 21 times.

DOI: 10.1369/0022155412440001
DOCUMENT TYPE: Article

Jain, K.K.
Integration of Biotechnologies for the Development of Personalized Medicine
(2012) Pharmaceutical Biotechnology: Drug Discovery and Clinical Applications, pp. 553-580.

DOI: 10.1002/9783527632909.ch21
DOCUMENT TYPE: Book Chapter

Adelson, D.L.
Bovine Genome Architecture
(2012) Bovine Genomics, pp. 123-143.

DOI: 10.1002/9781118301739.ch10
DOCUMENT TYPE: Book Chapter

Itsara, A., Vissers, L.E.L.M., Steinberg, K.M., Meyer, K.J., Zody, M.C., Koolen, D.A., De Ligt, J., Cuppen, E., Baker, C., Lee, C., Graves, T.A., Wilson, R.K., Jenkins, R.B., Veltman, J.A., Eichler, E.E.
Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing
(2012) American Journal of Human Genetics, 90 (4), pp. 599-613. Cited 8 times.

DOI: 10.1016/j.ajhg.2012.02.013

DOCUMENT TYPE: Article

Rodríguez-Santiago, B., Armengol, L.
Next generation sequencing technology in pre- and postnatal genetic diagnosis [Tecnologías de secuenciación de nueva generación en diagnóstico genético pre- y postnatal]
(2012) Diagnostico Prenatal, 23 (2), pp. 56-66.

DOI: 10.1016/j.diapre.2012.02.001
DOCUMENT TYPE: Article

Chiang, C., Jacobsen, J.C., Ernst, C., Hanscom, C., Heilbut, A., Blumenthal, I., Mills, R.E., Kirby, A., Lindgren, A.M., Rudiger, S.R., McLaughlan, C.J., Bawden, C.S., Reid, S.J., Faull, R.L.M., Snell, R.G., Hall, I.M., Shen, Y., Ohsumi, T.K., Borowsky, M.L., Daly, M.J., Lee, C., Morton, C.C., MacDonald, M.E., Gusella, J.F., Talkowski, M.E.
Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration
(2012) Nature Genetics, 44 (4), pp. 390-397. Cited 61 times.

DOI: 10.1038/ng.2202
DOCUMENT TYPE: Article

Sindi, S.S., Önal, S., Peng, L.C., Wu, H.-T., Raphael, B.J.
An integrative probabilistic model for identification of structural variation in sequencing data
(2012) Genome Biology, 13 (3), art. no. R22, . Cited 24 times.

DOI: 10.1186/gb-2012-13-3-r22
DOCUMENT TYPE: Article

Yalcin, B., Wong, K., Bhomra, A., Goodson, M., Keane, T.M., Adams, D.J., Flint, J.
The fine-scale architecture of structural variants in 17 mouse genomes
(2012) Genome Biology, 13 (3), art. no. R18, . Cited 13 times.

DOI: 10.1186/gb-2012-13-3-r18
DOCUMENT TYPE: Article

Ariyadasa, R., Stein, N.
Advances in BAC-based physical mapping and map integration strategies in plants
(2012) Journal of Biomedicine and Biotechnology, 2012, art. no. 184854, . Cited 11 times.

DOI: 10.1155/2012/184854
DOCUMENT TYPE: Review

Jensen, H., Kjeldsen, E., Hjortdal, V.E.
Could submicroscopical chromosomal imbalances cause cono-truncal malformations in twins?
(2012) Congenital Heart Disease, 7 (2), pp. 170-177.

DOI: 10.1111/j.1747-0803.2011.00544.x
DOCUMENT TYPE: Article

Lee, H.J., Kweon, J., Kim, E., Kim, S., Kim, J.-S.
Targeted chromosomal duplications and inversions in the human genome using zinc finger nucleases
(2012) Genome Research, 22 (3), pp. 539-548. Cited 50 times.

DOI: 10.1101/gr.129635.111
DOCUMENT TYPE: Article

Dewey, F.E., Pan, S., Wheeler, M.T., Quake, S.R., Ashley, E.A.
DNA sequencing clinical applications of new DNA sequencing technologies
(2012) Circulation, 125 (7), pp. 931-944. Cited 15 times.

DOI: 10.1161/CIRCULATIONAHA.110.972828
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Veltman, J.A.
Impact of Genomewide Structural Variation on Gene Discovery
(2012) Gene Discovery for Disease Models, pp. 443-470.

DOI: 10.1002/9780470933947.ch21
DOCUMENT TYPE: Book Chapter

Cáceres, A., Sindi, S.S., Raphael, B.J., Cáceres, M., González, J.R.
Identification of polymorphic inversions from genotypes
(2012) BMC Bioinformatics, 13 (1), art. no. 28, . Cited 5 times.

DOI: 10.1186/1471-2105-13-28
DOCUMENT TYPE: Article

Koboldt, D.C., Larson, D.E., Chen, K., Ding, L., Wilson, R.K.
Massively parallel sequencing approaches for characterization of structural variation
(2012) Methods in Molecular Biology, 838, pp. 369-384. Cited 15 times.

DOI: 10.1007/978-1-61779-507-7_18
DOCUMENT TYPE: Article

Nowrousian, M., Teichert, I., Masloff, S., Kück, U.
Whole-genome sequencing of sordaria macrospora mutants identifies developmental genes
(2012) G3: Genes, Genomes, Genetics, 2 (2), pp. 261-270. Cited 17 times.

DOI: 10.1534/g3.111.001479
DOCUMENT TYPE: Article

Vissers, L.E.L.M., Stankiewicz, P.
Microdeletion and microduplication syndromes
(2012) Methods in Molecular Biology, 838, pp. 29-75. Cited 20 times.

DOI: 10.1007/978-1-61779-507-7_2
DOCUMENT TYPE: Article

Pfundt, R., Veltman, J.A.
Structural genomic variation in intellectual disability
(2012) Methods in Molecular Biology, 838, pp. 77-95. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_3
DOCUMENT TYPE: Article

Sneddon, T.P., Church, D.M.
Online resources for genomic structural variation
(2012) Methods in Molecular Biology, 838, pp. 273-289. Cited 2 times.

DOI: 10.1007/978-1-61779-507-7_13
DOCUMENT TYPE: Article

Johansson, A.C.V., Feuk, L.
Characterizing and interpreting genetic variation from personal genome sequencing
(2012) Methods in Molecular Biology, 838, pp. 343-367. Cited 3 times.

DOI: 10.1007/978-1-61779-507-7_17
DOCUMENT TYPE: Article

Hollox, E.J.
The challenges of studying complex and dynamic regions of the human genome
(2012) Methods in Molecular Biology, 838, pp. 187-207. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_9
DOCUMENT TYPE: Article

Jiang, J., Li, J., Kwan, H., Au, C., Wan Law, P., Li, L., Kam, K., Lun Ling, J., Leung, F.C.
A cost-effective and universal strategy for complete prokaryotic genomic sequencing proposed by computer simulation
(2012) BMC Research Notes, 5, art. no. 80, . Cited 2 times.

DOI: 10.1186/1756-0500-5-80
DOCUMENT TYPE: Article

Hall, I.M., Quinlan, A.R.
Detection and interpretation of genomic structural variation in mammals
(2012) Methods in Molecular Biology, 838, pp. 225-248. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_11
DOCUMENT TYPE: Article

Iqbal, Z., Caccamo, M., Turner, I., Flicek, P., McVean, G.
De novo assembly and genotyping of variants using colored de Bruijn graphs
(2012) Nature Genetics, 44 (2), pp. 226-232. Cited 73 times.

DOI: 10.1038/ng.1028
DOCUMENT TYPE: Article

Dumanski, J.P., Piotrowski, A.
Structural genetic variation in the context of somatic mosaicism
(2012) Methods in Molecular Biology, 838, pp. 249-272. Cited 4 times.

DOI: 10.1007/978-1-61779-507-7_12
DOCUMENT TYPE: Article

Simmons, A.D., Carvalho, C.M.B., Lupski, J.R.
What have studies of genomic disorders taught us about our genome?
(2012) Methods in Molecular Biology, 838, pp. 1-27. Cited 5 times.

DOI: 10.1007/978-1-61779-507-7_1
DOCUMENT TYPE: Article

Dewal, N., Hu, Y., Freedman, M.L., LaFramboise, T., Pe'Er, I.
Calling amplified haplotypes in next generation tumor sequence data
(2012) Genome Research, 22 (2), pp. 362-374. Cited 4 times.

DOI: 10.1101/gr.122564.111
DOCUMENT TYPE: Article

Ceulemans, S., Van Der Ven, K., Del-Favero, J.
Targeted screening and validation of copy number variations
(2012) Methods in Molecular Biology, 838, pp. 311-328. Cited 6 times.

DOI: 10.1007/978-1-61779-507-7_15
DOCUMENT TYPE: Article

Chen, J., Kim, Y.C., Wang, S.M.
DGS (Ditag Genome Scanning) - A Restriction-Based Paired-End Sequencing Approach for Genome Structural Analysis
(2012) Tag-Based Next Generation Sequencing, pp. 277-285.

DOI: 10.1002/9783527644582.ch16
DOCUMENT TYPE: Book Chapter

Gogol-Döring, A., Chen, W.
An overview of the analysis of next generation sequencing data
(2012) Methods in Molecular Biology, 802, pp. 249-257. Cited 12 times.

DOI: 10.1007/978-1-61779-400-1_16
DOCUMENT TYPE: Article

Le Scouarnec, S., Gribble, S.M.
Characterising chromosome rearrangements: Recent technical advances in molecular cytogenetics
(2012) Heredity, 108 (1), pp. 75-85. Cited 22 times.

DOI: 10.1038/hdy.2011.100
DOCUMENT TYPE: Review

Xu, X., Liu, X., Ge, S., Jensen, J.D., Hu, F., Li, X., Dong, Y., Gutenkunst, R.N., Fang, L., Huang, L., Li, J., He, W., Zhang, G., Zheng, X., Zhang, F., Li, Y., Yu, C., Kristiansen, K., Zhang, X., Wang, J., Wright, M., McCouch, S., Nielsen, R., Wang, J., Wang, W.
Resequencing 50 accessions of cultivated and wild rice yields markers for identifying agronomically important genes
(2012) Nature Biotechnology, 30 (1), pp. 105-111. Cited 145 times.

DOI: 10.1038/nbt.2050
DOCUMENT TYPE: Article

Abeel, T., Van Parys, T., Saeys, Y., Galagan, J., Van De Peer, Y.
GenomeView: A next-generation genome browser
(2012) Nucleic Acids Research, 40 (2), . Cited 25 times.

DOI: 10.1093/nar/gkr995
DOCUMENT TYPE: Article

Severson, D.W., Behura, S.K.
Mosquito genomics: Progress and challenges
(2012) Annual Review of Entomology, 57, pp. 143-166. Cited 27 times.

DOI: 10.1146/annurev-ento-120710-100651
DOCUMENT TYPE: Article

Brooks, M.J., Rajasimha, H.K., Roger, J.E., Swaroop, A.
Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl -/- retinal transcriptomes
(2011) Molecular Vision, 17, pp. 3034-3054. Cited 27 times.

DOCUMENT TYPE: Article

Liu, Y.
Cancer and signaling pathway deregulation
(2011) Handbook of Research on Computational and Systems Biology: Interdisciplinary Applications, pp. 369-379.

DOI: 10.4018/978-1-60960-491-2.ch017
DOCUMENT TYPE: Book Chapter

Zhao, Z., Nguyen, T.C., Deng, N., Johnson, K.M., Zhu, D.
SPATA: A seeding and patching algorithm for de novo transcriptome assembly
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112351, pp. 26-33. Cited 1 time.

DOI: 10.1109/BIBMW.2011.6112351
DOCUMENT TYPE: Conference Paper

Zhang, J., Gao, Y., Zhao, X., Guan, M., Zhang, W., Wan, J., Yu, B.
Investigation of copy-number variations of C8orf4 in hematological malignancies
(2011) Medical Oncology, 28 (SUPPL. 1), pp. S647-S652. Cited 3 times.

DOI: 10.1007/s12032-010-9698-6
DOCUMENT TYPE: Article

Lai, A.G., Denton-Giles, M., Mueller-Roeber, B., Schippers, J.H.M., Dijkwel, P.P.
Positional information resolves structural variations and uncovers an evolutionarily divergent genetic locus in accessions of Arabidopsis thaliana
(2011) Genome Biology and Evolution, 3 (1), pp. 627-640. Cited 2 times.

DOI: 10.1093/gbe/evr038
DOCUMENT TYPE: Article

Schrider, D.R., Stevens, K., Cardeño, C.M., Langley, C.H., Hahn, M.W.
Genome-wide analysis of retrogene polymorphisms in Drosophila melanogaster
(2011) Genome Research, 21 (12), pp. 2087-2095. Cited 21 times.

DOI: 10.1101/gr.116434.110
DOCUMENT TYPE: Article

Hochstenbach, R., Buizer-Voskamp, J.E., Vorstman, J.A.S., Ophoff, R.A.
Genome arrays for the detection of copy number variations in idiopathic mental retardation, idiopathic generalized epilepsy and neuropsychiatric disorders: Lessons for diagnostic workflow and research
(2011) Cytogenetic and Genome Research, 135 (3-4), pp. 174-202. Cited 45 times.

DOI: 10.1159/000332928
DOCUMENT TYPE: Article

Duan, J., Zhang, J.-G., Lefante, J., Deng, H.-W., Wang, Y.-P.
Detection of copy number variation from next generation sequencing data with total variation penalized least square optimization
(2011) 2011 IEEE International Conference on Bioinformatics and Biomedicine Workshops, BIBMW 2011, art. no. 6112348, pp. 3-12. Cited 5 times.

DOI: 10.1109/BIBMW.2011.6112348
DOCUMENT TYPE: Conference Paper

Liu, X., Choi, S.-W., Wong, T.K.F., Lam, T.-W., Yiu, S.-M.
Detection of novel tandem duplication with next-generation sequencing
(2011) 2011 ACM Conference on Bioinformatics, Computational Biology and Biomedicine, BCB 2011, pp. 415-419.

DOI: 10.1145/2147805.2147861
DOCUMENT TYPE: Conference Paper

Marian, A.J.
Heart Failure as a Consequence of Restrictive Cardiomyopathy
(2011) Heart Failure, pp. 395-407.

DOI: 10.1016/B978-1-4160-5895-3.10025-7
DOCUMENT TYPE: Book Chapter

Girirajan, S., Campbell, C.D., Eichler, E.E.
Human copy number variation and complex genetic disease
(2011) Annual Review of Genetics, 45, pp. 203-226. Cited 70 times.

DOI: 10.1146/annurev-genet-102209-163544
DOCUMENT TYPE: Article

Greisman, H.A., Hoffman, N.G., Yi, H.S.
Rapid high-resolution mapping of balanced chromosomal rearrangements on tiling CGH arrays
(2011) Journal of Molecular Diagnostics, 13 (6), pp. 621-633. Cited 11 times.

DOI: 10.1016/j.jmoldx.2011.07.005
DOCUMENT TYPE: Article