

## 8 Reference

- Akaike, H.: Fitting autoregressive models for prediction. *Annals of the institute of statistical mathematics* 21 (1969) 243–
- Abeyasinghe, S.S., N. Chuzhanova, M. Krawczak, E.V. Ball, and D.N. Cooper. 2003. Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. *Hum Mutat.* 22:229-44.
- and Analysis ConsortiumThe Chimpanzee, S. 2005. Initial sequence of the chimpanzee genome and comparison with the human genome. 437:69.
- Autio, R., S. Hautaniemi, P. Kauraniemi, O. Yli-Harja, J. Astola, M. Wolf, and A. Kallioniemi. 2003. CGH-Plotter: MATLAB toolbox for CGH-data analysis. *Bioinformatics.* 19:1714-5.
- Bailey, J.A., R. Baertsch, W.J. Kent, D. Haussler, and E.E. Eichler. 2004. Hotspots of mammalian chromosomal evolution. *Genome Biol.* 5:R23.
- Bailey, J.A., Z. Gu, R.A. Clark, K. Reinert, R.V. Samonte, S. Schwartz, M.D. Adams, E.W. Myers, P.W. Li, and E.E. Eichler. 2002. Recent segmental duplications in the human genome. *Science.* 297:1003-7.
- Bailey, J.A., A.M. Yavor, H.F. Massa, B.J. Trask, and E.E. Eichler. 2001. Segmental duplications: organization and impact within the current human genome project assembly. *Genome Res.* 11:1005-17.
- Bailey, J.A., A.M. Yavor, L. Viggiano, D. Misceo, J.E. Horvath, N. Archidiacono, S. Schwartz, M. Rocchi, and E.E. Eichler. 2002. Human-specific duplication and mosaic transcripts: the recent paralogous structure of chromosome 22. *Am J Hum Genet.* 70:83-100.
- Barrett, M.T., A. Scheffer, A. Ben-Dor, N. Sampas, D. Lipson, R. Kincaid, P. Tsang, B. Curry, K. Baird, P.S. Meltzer, Z. Yakhini, L. Bruhn, and S. Laderman. 2004. Comparative genomic hybridization using oligonucleotide microarrays and total genomic DNA. *Proc Natl Acad Sci U S A.* 101:17765-70.
- Bignell, G.R., J. Huang, J. Greshock, S. Watt, A. Butler, S. West, M. Grigorova, K.W. Jones, W. Wei, M.R. Stratton, P.A. Futreal, B. Weber, M.H. Shaper, and R. Wooster. 2004. High-resolution analysis of DNA copy number using oligonucleotide microarrays. *Genome Res.* 14:287-95.
- Bredel, M., C. Bredel, D. Juric, G.R. Harsh, H. Vogel, L.D. Recht, and B.I. Sikic. 2005. High-resolution genome-wide mapping of genetic alterations in human glial brain tumors. *Cancer Res.* 65:4088-96.
- Brennan, C., Y. Zhang, C. Leo, B. Feng, C. Cauwels, A.J. Aguirre, M. Kim, A. Protopopov, and L. Chin. 2004. High-resolution global profiling of genomic alterations with long oligonucleotide microarray. *Cancer Res.* 64:4744-8.
- Bugge, M., G. Bruun-Petersen, K. Brondum-Nielsen, U. Friedrich, J. Hansen, G. Jensen, P.K. Jensen, U. Kristoffersson, C. Lundsteen, E. Niebuhr, K.R. Rasmussen, K. Rasmussen, and N. Tommerup. 2000. Disease associated balanced chromosome rearrangements: a resource for large scale genotype-phenotype delineation in man. *J Med Genet.* 37:858-65.
- Carter, N.P., M.A. Ferguson-Smith, M.T. Perryman, H. Telenius, A.H. Pelmear, M.A. Leversha, M.T. Glancy, S.L. Wood, K. Cook, H.M. Dyson, and et al. 1992. Reverse chromosome painting: a method for the rapid analysis of aberrant chromosomes in clinical cytogenetics. *J Med Genet.* 29:299-307.

- Carvalho, B., E. Ouwerkerk, G.A. Meijer, and B. Ylstra. 2004. High resolution microarray comparative genomic hybridisation analysis using spotted oligonucleotides. *J Clin Pathol.* 57:644-6.
- Cheung, J., X. Estivill, R. Khaja, J.R. MacDonald, K. Lau, L.C. Tsui, and S.W. Scherer. 2003. Genome-wide detection of segmental duplications and potential assembly errors in the human genome sequence. *Genome Biol.* 4:R25.
- Cheung, V.G., N. Nowak, W. Jang, I.R. Kirsch, S. Zhao, X.N. Chen, T.S. Furey, U.J. Kim, W.L. Kuo, M. Olivier, J. Conroy, A. Kasprzyk, H. Massa, R. Yonescu, S. Sait, C. Thoreen, A. Snijders, E. Lemyre, J.A. Bailey, A. Bruzel, W.D. Burrill, S.M. Clegg, S. Collins, P. Dhimi, C. Friedman, C.S. Han, S. Herrick, J. Lee, A.H. Ligon, S. Lowry, M. Morley, S. Narasimhan, K. Osoegawa, Z. Peng, I. Plajzer-Frick, B.J. Quade, D. Scott, K. Sirotkin, A.A. Thorpe, J.W. Gray, J. Hudson, D. Pinkel, T. Ried, L. Rowen, G.L. Shen-Ong, R.L. Strausberg, E. Birney, D.F. Callen, J.F. Cheng, D.R. Cox, N.A. Doggett, N.P. Carter, E.E. Eichler, D. Haussler, J.R. Korenberg, C.C. Morton, D. Albertson, G. Schuler, P.J. de Jong, and B.J. Trask. 2001. Integration of cytogenetic landmarks into the draft sequence of the human genome. *Nature.* 409:953-8.
- Chi, B., R.J. DeLeeuw, B.P. Coe, C. MacAulay, and W.L. Lam. 2004. SeeGH--a software tool for visualization of whole genome array comparative genomic hybridization data. *BMC Bioinformatics.* 5:13.
- Daruwala, R.S., A. Rudra, H. Ostrer, R. Lucito, M. Wigler, and B. Mishra. 2004. A versatile statistical analysis algorithm to detect genome copy number variation. *Proc Natl Acad Sci U S A.* 101:16292-7.
- de Vries, B.B., R. Pfundt, M. Leisink, D.A. Koolen, L.E. Vissers, I.M. Janssen, S. Reijmersdal, W.M. Nillesen, E.H. Huys, N. Leeuw, D. Smeets, E.A. Sistermans, T. Feuth, C.M. van Ravenswaaij-Arts, A.G. van Kessel, E.F. Schoenmakers, H.G. Brunner, and J.A. Veltman. 2005. Diagnostic genome profiling in mental retardation. *Am J Hum Genet.* 77:606-16.
- Deininger, P.L., and M.A. Batzer. 1999. Alu repeats and human disease. *Mol Genet Metab.* 67:183-93.
- Dhimi, P., A.J. Coffey, S. Abbs, J.R. Vermeesch, J.P. Dumanski, K.J. Woodward, R.M. Andrews, C. Langford, and D. Vetrie. 2005. Exon array CGH: detection of copy-number changes at the resolution of individual exons in the human genome. *Am J Hum Genet.* 76:750-62.
- du Manoir, S., M.R. Speicher, S. Joos, E. Schrock, S. Popp, H. Dohner, G. Kovacs, M. Robert-Nicoud, P. Lichter, and T. Cremer. 1993. Detection of complete and partial chromosome gains and losses by comparative genomic in situ hybridization. *Hum Genet.* 90:590-610.
- Edelmann, L., E. Spiteri, K. Koren, V. Pulijaal, M.G. Bialer, A. Shanske, R. Goldberg, and B.E. Morrow. 2001. AT-rich palindromes mediate the constitutional t(11;22) translocation. *Am J Hum Genet.* 68:1-13.
- Eichler, E.E. 2001. Recent duplication, domain accretion and the dynamic mutation of the human genome. *Trends Genet.* 17:661-9.
- Eichler, E.E., and D. Sankoff. 2003. Structural dynamics of eukaryotic chromosome evolution. *Science.* 301:793-7.
- Fiegler, H., P. Carr, E.J. Douglas, D.C. Burford, S. Hunt, C.E. Scott, J. Smith, D. Vetrie, P. Gorman, I.P. Tomlinson, and N.P. Carter. 2003. DNA microarrays for comparative genomic hybridization based on DOP-PCR

- amplification of BAC and PAC clones. *Genes Chromosomes Cancer*. 36:361-74.
- Fiegler, H., S.M. Gribble, D.C. Burford, P. Carr, E. Prigmore, K.M. Porter, S. Clegg, J.A. Crolla, N.R. Dennis, P. Jacobs, and N.P. Carter. 2003. Array painting: a method for the rapid analysis of aberrant chromosomes using DNA microarrays. *J Med Genet*. 40:664-70.
- Fortna, A., Y. Kim, E. MacLaren, K. Marshall, G. Hahn, L. Meltesen, M. Brenton, R. Hink, S. Burgers, T. Hernandez-Boussard, A. Karimpour-Fard, D. Glueck, L. McGavran, R. Berry, J. Pollack, and J.M. Sikela. 2004. Lineage-specific gene duplication and loss in human and great ape evolution. *PLoS Biol*. 2:E207.
- Fridlyand, J., A.M. Snijders, D. Pinkel, D.G. Albertson, and A.N.A.N. Jain. 2004. Hidden Markov models approach to the analysis of array CGH data. 90:132.
- Fujiyama, A., H. Watanabe, A. Toyoda, T.D. Taylor, T. Itoh, S.F. Tsai, H.S. Park, M.L. Yaspo, H. Lehrach, Z. Chen, G. Fu, N. Saitou, K. Osoegawa, P.J. de Jong, Y. Suto, M. Hattori, and Y. Sakaki. 2002. Construction and analysis of a human-chimpanzee comparative clone map. *Science*. 295:131-4.
- Gotter, A.L., T.H. Shaikh, M.L. Budarf, C.H. Rhodes, and B.S. Emanuel. 2004. A palindrome-mediated mechanism distinguishes translocations involving LCR-B of chromosome 22q11.2. *Hum Mol Genet*. 13:103-15.
- Gribble, S.M., H. Fiegler, D.C. Burford, E. Prigmore, F. Yang, P. Carr, B.L. Ng, T. Sun, E.S. Kamberov, V.L. Makarov, J.P. Langmore, and N.P. Carter. 2004. Applications of combined DNA microarray and chromosome sorting technologies. *Chromosome Res*. 12:35-43.
- Hodgson, G., J.H. Hager, S. Volik, S. Hariono, M. Wernick, D. Moore, N. Nowak, D.G. Albertson, D. Pinkel, C. Collins, D. Hanahan, and J.W. Gray. 2001. Genome scanning with array CGH delineates regional alterations in mouse islet carcinomas. *Nat Genet*. 29:459-64.
- Horvath, J.E., J.A. Bailey, D.P. Locke, and E.E. Eichler. 2001. Lessons from the human genome: transitions between euchromatin and heterochromatin. *Hum Mol Genet*. 10:2215-23.
- Hupe, P., N. Stransky, J.P. Thiery, F. Radvanyi, and E. Barillot. 2004. Analysis of array CGH data: from signal ratio to gain and loss of DNA regions. *Bioinformatics*. 20:3413-22.
- Inoue, K., and J.R. Lupski. 2002. Molecular mechanisms for genomic disorders. *Annu Rev Genomics Hum Genet*. 3:199-242.
- Inoue, K., H. Osaka, V.C. Thurston, J.T. Clarke, A. Yoneyama, L. Rosenbarker, T.D. Bird, M.E. Hodes, L.G. Shaffer, and J.R. Lupski. 2002. Genomic rearrangements resulting in PLP1 deletion occur by nonhomologous end joining and cause different dysmyelinating phenotypes in males and females. *Am J Hum Genet*. 71:838-53.
- Ishkanian, A.S., C.A. Malloff, S.K. Watson, R.J. DeLeeuw, B. Chi, B.P. Coe, A. Snijders, D.G. Albertson, D. Pinkel, M.A. Marra, V. Ling, C. MacAulay, and W.L. Lam. 2004. A tiling resolution DNA microarray with complete coverage of the human genome. *Nat Genet*. 36:299-303.
- Kallioniemi, A., O.P. Kallioniemi, D. Sudar, D. Rutovitz, J.W. Gray, F. Waldman, and D. Pinkel. 1992. Comparative genomic hybridization for molecular cytogenetic analysis of solid tumors. *Science*. 258:818-21.

- Kennedy, G.C., H. Matsuzaki, S. Dong, W.M. Liu, J. Huang, G. Liu, X. Su, M. Cao, W. Chen, J. Zhang, W. Liu, G. Yang, X. Di, T. Ryder, Z. He, U. Surti, M.S. Phillips, M.T. Boyce-Jacino, S.P. Fodor, and K.W. Jones. 2003. Large-scale genotyping of complex DNA. *Nat Biotechnol.* 21:1233-7.
- Kirchhoff, M., T. Gerdes, J. Maahr, H. Rose, M. Bentz, H. Dohner, and C. Lundsteen. 1999. Deletions below 10 megabasepairs are detected in comparative genomic hybridization by standard reference intervals. *Genes Chromosomes Cancer.* 25:410-3.
- Knuutila, S., A.M. Bjorkqvist, K. Autio, M. Tarkkanen, M. Wolf, O. Monni, J. Szymanska, M.L. Larramendy, J. Tapper, H. Pere, W. El-Rifai, S. Hemmer, V.M. Wasenius, V. Vidgren, and Y. Zhu. 1998. DNA copy number amplifications in human neoplasms: review of comparative genomic hybridization studies. *Am J Pathol.* 152:1107-23.
- Krzywinski, M., I. Bosdet, D. Smailus, R. Chiu, C. Mathewson, N. Wye, S. Barber, M. Brown-John, S. Chan, S. Chand, A. Cloutier, N. Girn, D. Lee, A. Masson, M. Mayo, T. Olson, P. Pandoh, A.L. Prabhu, E. Schoenmakers, M. Tsai, D. Albertson, W. Lam, C.O. Choy, K. Osoegawa, S. Zhao, P.J. de Jong, J. Schein, S. Jones, and M.A. Marra. 2004. A set of BAC clones spanning the human genome. *Nucleic Acids Res.* 32:3651-60.
- Kurahashi, H., H. Inagaki, K. Yamada, T. Ohye, M. Taniguchi, B.S. Emanuel, and T. Toda. 2004. Cruciform DNA structure underlies the etiology for palindrome-mediated human chromosomal translocations. *J Biol Chem.* 279:35377-83.
- Kurahashi, H., T. Shaikh, M. Takata, T. Toda, and B.S. Emanuel. 2003. The constitutional t(17;22): another translocation mediated by palindromic AT-rich repeats. *Am J Hum Genet.* 72:733-8.
- Kurahashi, H., T.H. Shaikh, P. Hu, B.A. Roe, B.S. Emanuel, and M.L. Budarf. 2000. Regions of genomic instability on 22q11 and 11q23 as the etiology for the recurrent constitutional t(11;22). *Hum Mol Genet.* 9:1665-70.
- Kurahashi, H., T.H. Shaikh, E.H. Zackai, L. Celle, D.A. Driscoll, M.L. Budarf, and B.S. Emanuel. 2000. Tightly clustered 11q23 and 22q11 breakpoints permit PCR-based detection of the recurrent constitutional t(11;22). *Am J Hum Genet.* 67:763-8.
- Li, J., T. Jiang, J.H. Mao, A. Balmain, L. Peterson, C. Harris, P.H. Rao, P. Havlak, R. Gibbs, and W.W. Cai. 2004. Genomic segmental polymorphisms in inbred mouse strains. *Nat Genet.* 36:952-4.
- Locke, D.P., R. Segraves, L. Carbone, N. Archidiacono, D.G. Albertson, D. Pinkel, and E.E. Eichler. 2003. Large-scale variation among human and great ape genomes determined by array comparative genomic hybridization. *Genome Res.* 13:347-57.
- Locke, D.P., R. Segraves, R.D. Nicholls, S. Schwartz, D. Pinkel, D.G. Albertson, and E.E. Eichler. 2004. BAC microarray analysis of 15q11-q13 rearrangements and the impact of segmental duplications. *J Med Genet.* 41:175-82.
- Lucito, R., J. Healy, J. Alexander, A. Reiner, D. Esposito, M. Chi, L. Rodgers, A. Brady, J. Sebat, J. Troge, J.A. West, S. Rostan, K.C. Nguyen, S. Powers, K.Q. Ye, A. Olshen, E. Venkatraman, L. Norton, and M. Wigler. 2003. Representational oligonucleotide microarray analysis: a high-resolution

- method to detect genome copy number variation. *Genome Res.* 13:2291-305.
- Lupski, J.R. 1998. Genomic disorders: structural features of the genome can lead to DNA rearrangements and human disease traits. *Trends Genet.* 14:417-22.
- McNeil, N., and T. Ried. 2000. Novel molecular cytogenetic techniques for identifying complex chromosomal rearrangements: technology and applications in molecular medicine. *Expert Rev Mol Med.* 2000:1-14.
- Menten, B., F. Pattyn, K. De Preter, P. Robbrecht, E. Michels, K. Buysse, G. Mortier, A. De Paepe, S. van Vooren, J. Vermeesch, Y. Moreau, B. De Moor, S. Vermeulen, F. Speleman, and J. Vandesompele. 2005. arrayCGHbase: an analysis platform for comparative genomic hybridization microarrays. *BMC Bioinformatics.* 6:124.
- Newman, T.L., E. Tuzun, V.A. Morrison, K.E. Hayden, M. Ventura, S.D. McGrath, M. Rocchi, and E.E. Eichler. 2005. A genome-wide survey of structural variation between human and chimpanzee. *Genome Res.* 15:1344-56.
- Nimmakayalu, M.A., A.L. Gotter, T.H. Shaikh, and B.S. Emanuel. 2003. A novel sequence-based approach to localize translocation breakpoints identifies the molecular basis of a t(4;22). *Hum Mol Genet.* 12:2817-25.
- Nobile, C., L. Toffolatti, F. Rizzi, B. Simionati, V. Nigro, B. Cardazzo, T. Patarnello, G. Valle, and G.A. Danieli. 2002. Analysis of 22 deletion breakpoints in dystrophin intron 49. *Hum Genet.* 110:418-21.
- Olshen, A.B., E.S. Venkatraman, R. Lucito, and M. Wigler. 2004. Circular binary segmentation for the analysis of array-based DNA copy number data. *Biostatistics.* 5:557-72.
- Oostlander, A.E., G.A. Meijer, and B. Ylstra. 2004. Microarray-based comparative genomic hybridization and its applications in human genetics. *Clin Genet.* 66:488-95.
- Osoegawa, K., A.G. Mammoser, C. Wu, E. Frengen, C. Zeng, J.J. Catanese, and P.J. de Jong. 2001. A bacterial artificial chromosome library for sequencing the complete human genome. *Genome Res.* 11:483-96.
- Pinkel, D., R. Seagraves, D. Sudar, S. Clark, I. Poole, D. Kowbel, C. Collins, W.L. Kuo, C. Chen, Y. Zhai, S.H. Dairkee, B.M. Ljung, J.W. Gray, and D.G. Albertson. 1998. High resolution analysis of DNA copy number variation using comparative genomic hybridization to microarrays. *Nat Genet.* 20:207-11.
- Pollack, J.R., C.M. Perou, A.A. Alizadeh, M.B. Eisen, A. Pergamenschikov, C.F. Williams, S.S. Jeffrey, D. Botstein, and P.O. Brown. 1999. Genome-wide analysis of DNA copy-number changes using cDNA microarrays. *Nat Genet.* 23:41-6.
- Pollack, J.R., T. Sorlie, C.M. Perou, C.A. Rees, S.S. Jeffrey, P.E. Lonning, R. Tibshirani, D. Botstein, A.L. Borresen-Dale, and P.O. Brown. 2002. Microarray analysis reveals a major direct role of DNA copy number alteration in the transcriptional program of human breast tumors. *Proc Natl Acad Sci U S A.* 99:12963-8.
- Richardson, C., and M. Jasin. 2000. Coupled homologous and nonhomologous repair of a double-strand break preserves genomic integrity in mammalian cells. *Mol Cell Biol.* 20:9068-75.

- Roth, D.B., and J.H. Wilson. 1986. Nonhomologous recombination in mammalian cells: role for short sequence homologies in the joining reaction. *Mol Cell Biol.* 6:4295-304.
- Rozen, S., and H. Skaletsky. 2000. Primer3 on the WWW for general users and for biologist programmers. *Methods Mol Biol.* 132:365-86.
- Shaikh, T.H., H. Kurahashi, and B.S. Emanuel. 2001. Evolutionarily conserved low copy repeats (LCRs) in 22q11 mediate deletions, duplications, translocations, and genomic instability: an update and literature review. *Genet Med.* 3:6-13.
- Shaw, C.J., and J.R. Lupski. 2004. Implications of human genome architecture for rearrangement-based disorders: the genomic basis of disease. *Hum Mol Genet.* 13 Spec No 1:R57-64.
- Shaw, C.J., and J.R. Lupski. 2005. Non-recurrent 17p11.2 deletions are generated by homologous and non-homologous mechanisms. *Hum Genet.* 116:1-7.
- She, X., Z. Jiang, R.A. Clark, G. Liu, Z. Cheng, E. Tuzun, D.M. Church, G. Sutton, A.L. Halpern, and E.E. Eichler. 2004. Shotgun sequence assembly and recent segmental duplications within the human genome. *Nature.* 431:927-30.
- Singh, G.B., J.A. Kramer, and S.A. Krawetz. 1997. Mathematical model to predict regions of chromatin attachment to the nuclear matrix. *Nucleic Acids Res.* 25:1419-25.
- Smirnov, D.A., J.T. Burdick, M. Morley, and V.G. Cheung. 2004. Method for manufacturing whole-genome microarrays by rolling circle amplification. *Genes Chromosomes Cancer.* 40:72-7.
- Snijders, A.M., N. Nowak, R. Seagraves, S. Blackwood, N. Brown, J. Conroy, G. Hamilton, A.K. Hindle, B. Huey, K. Kimura, S. Law, K. Myambo, J. Palmer, B. Ylstra, J.P. Yue, J.W. Gray, A.N. Jain, D. Pinkel, and D.G. Albertson. 2001. Assembly of microarrays for genome-wide measurement of DNA copy number. *Nat Genet.* 29:263-4.
- Solinas-Toldo, S., S. Lampel, S. Stilgenbauer, J. Nickolenko, A. Benner, H. Dohner, T. Cremer, and P. Lichter. 1997. Matrix-based comparative genomic hybridization: biochips to screen for genomic imbalances. *Genes Chromosomes Cancer.* 20:399-407.
- Spiteri, E., M. Babcock, C.D. Kashork, K. Wakui, S. Gogineni, D.A. Lewis, K.M. Williams, S. Minoshima, T. Sasaki, N. Shimizu, L. Potocki, V. Pulijjal, A. Shanske, L.G. Shaffer, and B.E. Morrow. 2003. Frequent translocations occur between low copy repeats on chromosome 22q11.2 (LCR22s) and telomeric bands of partner chromosomes. *Hum Mol Genet.* 12:1823-37.
- Stankiewicz, P., and J.R. Lupski. 2002. Genome architecture, rearrangements and genomic disorders. *Trends Genet.* 18:74-82.
- Stankiewicz, P., C.J. Shaw, J.D. Dapper, K. Wakui, L.G. Shaffer, M. Withers, L. Elizondo, S.S. Park, and J.R. Lupski. 2003. Genome architecture catalyzes nonrecurrent chromosomal rearrangements. *Am J Hum Genet.* 72:1101-16.
- Telenius, H., A.H. Pelmeur, A. Tunnacliffe, N.P. Carter, A. Behmel, M.A. Ferguson-Smith, M. Nordenskjold, R. Pfragner, and B.A. Ponder. 1992. Cytogenetic analysis by chromosome painting using DOP-PCR amplified flow-sorted chromosomes. *Genes Chromosomes Cancer.* 4:257-63.
- Tjio, H.J., and A. Levan. 1956. The chromosome numbers of man. *Hereditas.* 42:1-6.

- Tuzun, E., A.J. Sharp, J.A. Bailey, R. Kaul, V.A. Morrison, L.M. Pertz, E. Haugen, H. Hayden, D. Albertson, D. Pinkel, M.V. Olson, and E.E. Eichler. 2005. Fine-scale structural variation of the human genome. *Nat Genet.* 37:727-32.
- Van Prooijen-Knegt, A.C., J.F. Van Hoek, J.G. Bauman, P. Van Duijn, I.G. Wool, and M. Van der Ploeg. 1982. In situ hybridization of DNA sequences in human metaphase chromosomes visualized by an indirect fluorescent immunocytochemical procedure. *Exp Cell Res.* 141:397-407.
- Veltman, I.M., J.A. Veltman, G. Arkesteijn, I.M. Janssen, L.E. Vissers, P.J. de Jong, A.G. van Kessel, and E.F. Schoenmakers. 2003. Chromosomal breakpoint mapping by arrayCGH using flow-sorted chromosomes. *Biotechniques.* 35:1066-70.
- Wang, P., Y. Kim, J. Pollack, B. Narasimhan, and R. Tibshirani. 2005. A method for calling gains and losses in array CGH data. *Biostatistics.* 6:45-58.
- Wilson, G.M., S. Flibotte, P.I. Missirlis, M.A. Marra, S. Jones, K. Thornton, A.G. Clark, and R.A. Holt. 2006. Identification by full-coverage array CGH of human DNA copy number increases relative to chimpanzee and gorilla. *Genome Res.* %R 10.1101/gr.4456006. 16:173-181.
- Wirth, J., H.G. Nothwang, S. van der Maarel, C. Menzel, G. Borck, I. Lopez-Pajares, K. Brondum-Nielsen, N. Tommerup, M. Bugge, H.H. Ropers, and T. Haaf. 1999. Systematic characterisation of disease associated balanced chromosome rearrangements by FISH: cytogenetically and genetically anchored YACs identify microdeletions and candidate regions for mental retardation genes. *J Med Genet.* 36:271-8.
- Woodward, K.J., M. Cundall, K. Sperle, E.A. Sistermans, M. Ross, G. Howell, S.M. Gribble, D.C. Burford, N.P. Carter, D.L. Hobson, J.Y. Garbern, J. Kamholz, H. Heng, M.E. Hodes, S. Malcolm, and G.M. Hobson. 2005. Heterogeneous duplications in patients with Pelizaeus-Merzbacher disease suggest a mechanism of coupled homologous and nonhomologous recombination. *Am J Hum Genet.* 77:966-87.
- Yang, Y.H., S. Dudoit, P. Luu, D.M. Lin, V. Peng, J. Ngai, and T.P. Speed. 2002. Normalization for cDNA microarray data: a robust composite method addressing single and multiple slide systematic variation. *Nucleic Acids Res.* 30:e15.
- Yunis, J.J., J.R. Sawyer, and K. Dunham. 1980. The striking resemblance of high-resolution G-banded chromosomes of man and chimpanzee. *Science.* 208:1145-8.
- Zhang, L., H.H. Lu, W.Y. Chung, J. Yang, and W.H. Li. 2005. Patterns of segmental duplication in the human genome. *Mol Biol Evol.* 22:135-41.