

Chapter 8

References

- Abagyan, R. A., Totrov, M.M., and Kuznetsov, D.N. ICM - a new method for protein modeling and design. Applications to docking and structure prediction from the distorted native conformation. (1994) *J Comp Chem*, **15**, 488 - 506.
- Adachi, N. and Lieber, M. R. Bidirectional gene organization: a common architectural feature of the human genome. (2002) *Cell*, **109**, 807-9.
- Adams, M. D., Celniker, S. E., Holt, R. A., Evans, C. A., Gocayne, J. D., Amanatides, P. G., et al. The genome sequence of *Drosophila melanogaster*. (2000) *Science*, **287**, 2185-95.
- Adams, M. D., Kelley, J. M., Gocayne, J. D., Dubnick, M., Polymeropoulos, M. H., Xiao, H., et al. Complementary DNA sequencing: expressed sequence tags and human genome project. (1991) *Science*, **252**, 1651-6.
- Adams, M. D., Soares, M. B., Kerlavage, A. R., Fields, C. and Venter, J. C. Rapid cDNA sequencing (expressed sequence tags) from a directionally cloned human infant brain cDNA library. (1993) *Nat Genet*, **4**, 373-80.
- Alexandrie, A. K., Rannug, A., Juronen, E., Tasa, G. and Warholm, M. Detection and characterization of a novel functional polymorphism in the GSTT1 gene. (2002) *Pharmacogenetics*, **12**, 613-9.
- Allan, J. M., Wild, C. P., Rollinson, S., Willett, E. V., Moorman, A. V., Dovey, G. J., et al. Polymorphism in glutathione S-transferase P1 is associated with susceptibility to chemotherapy-induced leukemia. (2001) *Proc Natl Acad Sci U S A*, **98**, 11592-7.
- Altschul, S. F., Gish, W., Miller, W., Myers, E. W. and Lipman, D. J. Basic local alignment search tool. (1990) *J Mol Biol*, **215**, 403-10.
- Altschul, S. F., Madden, T. L., Schaffer, A. A., Zhang, J., Zhang, Z., Miller, W., et al. Gapped BLAST and PSI-BLAST: a new generation of protein database search programs. (1997) *Nucleic Acids Res*, **25**, 3389-402.

- Altshuler, D., Pollara, V. J., Cowles, C. R., Van Etten, W. J., Baldwin, J., Linton, L., et al. An SNP map of the human genome generated by reduced representation shotgun sequencing. (2000) *Nature*, **407**, 513-6.
- Anderson, S. Shotgun DNA sequencing using cloned DNase I-generated fragments. (1981) *Nucleic Acids Res*, **9**, 3015-27.
- Anderson, S., Bankier, A. T., Barrell, B. G., de Bruijn, M. H., Coulson, A. R., Drouin, J., et al. Sequence and organization of the human mitochondrial genome. (1981) *Nature*, **290**, 457-65.
- Angelisova, P., Vlcek, C., Stefanova, I., Lipoldova, M. and Horejsi, V. The human leucocyte surface antigen CD53 is a protein structurally similar to the CD37 and MRC OX-44 antigens. (1990) *Immunogenetics*, **32**, 281-5.
- Ansari-Lari, M. A., Oeltjen, J. C., Schwartz, S., Zhang, Z., Muzny, D. M., Lu, J., et al. Comparative sequence analysis of a gene-rich cluster at human chromosome 12p13 and its syntenic region in mouse chromosome 6. (1998) *Genome Res*, **8**, 29-40.
- Antequera, F. and Bird, A. Number of CpG islands and genes in human and mouse. (1993) *Proc Natl Acad Sci U S A*, **90**, 11995-9.
- Arabidopsis Genome Initiative, T. Analysis of the genome sequence of the flowering plant *Arabidopsis thaliana*. (2000) *Nature*, **408**, 796-815.
- Attali, B., Romey, G., Honore, E., Schmid-Alliana, A., Mattei, M. G., Lesage, F., et al. Cloning, functional expression, and regulation of two K⁺ channels in human T lymphocytes. (1992) *J Biol Chem*, **267**, 8650-7.
- Bailey, J. A., Yavor, A. M., Viggiano, L., Misceo, D., Horvath, J. E., Archidiacono, N., et al. Human-specific duplication and mosaic transcripts: the recent paralogous structure of chromosome 22. (2002) *Am J Hum Genet*, **70**, 83-100.
- Bankier, A. T., Weston, K. M. and Barrell, B. G. Random cloning and sequencing by the M13/dideoxynucleotide chain termination method. (1987) *Methods Enzymol*, **155**, 51-93.

- Bates, G. P., Valdes, J., Hummerich, H., Baxendale, S., Le Paslier, D. L., Monaco, A. P., et al. Characterization of a yeast artificial chromosome contig spanning the Huntington's disease gene candidate region. (1992) *Nat Genet*, **1**, 180-7.
- Beaudoin, E., Freier, S., Wyatt, J. R., Claverie, J. M. and Gautheret, D. Patterns of variant polyadenylation signal usage in human genes. (2000) *Genome Res*, **10**, 1001-10.
- Bentley, D. R. The Human Genome Project--an overview. (2000) *Med Res Rev*, **20**, 189-96.
- Bentley, D. R., Deloukas, P., Dunham, A., French, L., Gregory, S. G., Humphray, S. J., et al. The physical maps for sequencing human chromosomes 1, 6, 9, 10, 13, 20 and X. (2001) *Nature*, **409**, 942-3.
- Bernaola-Galvan, P., Roman-Roldan, R. and Oliver, J. L. Compositional segmentation and long-range fractal correlations in DNA sequences. (1996) *Physical Review. E. Statistical Physics, Plasmas, Fluids, and Related Interdisciplinary Topics*, **53**, 5181-5189.
- Bernardi, G., Olofsson, B., Filipski, J., Zerial, M., Salinas, J., Cuny, G., et al. The mosaic genome of warm-blooded vertebrates. (1985) *Science*, **228**, 953-8.
- Bickmore, W. A. and Sumner, A. T. Mammalian chromosome banding--an expression of genome organization. (1989) *Trends Genet*, **5**, 144-8.
- Bird, A. P. CpG-rich islands and the function of DNA methylation. (1986) *Nature*, **321**, 209-13.
- Birnboim, H. C. and Doly, J. A rapid alkaline extraction procedure for screening recombinant plasmid DNA. (1979) *Nucleic Acids Res*, **7**, 1513-23.
- Blattner, F. R., Plunkett, G., 3rd, Bloch, C. A., Perna, N. T., Burland, V., Riley, M., et al. The complete genome sequence of Escherichia coli K-12. (1997) *Science*, **277**, 1453-74.
- Board, P., Coggan, M., Johnston, P., Ross, V., Suzuki, T. and Webb, G. Genetic heterogeneity of the human glutathione transferases: a complex of gene families. (1990) *Pharmacol Ther*, **48**, 357-69.
- Boguski, M. S. and Schuler, G. D. ESTablishing a human transcript map. (1995) *Nat Genet*, **10**, 369-71.

- Bonfield, J. K., Smith, K. and Staden, R. A new DNA sequence assembly program. (1995) *Nucleic Acids Res*, **23**, 4992-9.
- Booth, J., Boyland, E., and Sims, P. An enzyme from rat liver catalyzing conjugation with glutathione. (1961) *Biochem J*, **79**, 516 - 524.
- Botstein, D., White, R. L., Skolnick, M. and Davis, R. W. Construction of a genetic linkage map in man using restriction fragment length polymorphisms. (1980) *Am J Hum Genet*, **32**, 314-31.
- Bouffard, G. G., Idol, J. R., Braden, V. V., Iyer, L. M., Cunningham, A. F., Weintraub, L. A., et al. A physical map of human chromosome 7: an integrated YAC contig map with average STS spacing of 79 kb. (1997) *Genome Res*, **7**, 673-92.
- Brenner, M., Lampel, K., Nakatani, Y., Mill, J., Banner, C., Mearow, K., et al. Characterization of human cDNA and genomic clones for glial fibrillary acidic protein. (1990) *Brain Res Mol Brain Res*, **7**, 277-86.
- Brenner, S. E., Chothia, C. and Hubbard, T. J. Assessing sequence comparison methods with reliable structurally identified distant evolutionary relationships. (1998) *Proc Natl Acad Sci U S A*, **95**, 6073-8.
- Brintnell, B., Hey, Y., Jones, D., Hoggard, N., James, L. and Varley, J. M. Generation of a contig comprising YACs and BACs within chromosome region 1p13.1. (1997) *Somat Cell Mol Genet*, **23**, 153-7.
- Brown, L., Espinosa, R., 3rd, Le Beau, M. M., Siciliano, M. J. and Baer, R. HEN1 and HEN2: a subgroup of basic helix-loop-helix genes that are coexpressed in a human neuroblastoma. (1992) *Proc Natl Acad Sci U S A*, **89**, 8492-6.
- Burge, C. and Karlin, S. Prediction of complete gene structures in human genomic DNA. (1997) *J Mol Biol*, **268**, 78-94.
- Burke, D. T., Carle, G. F. and Olson, M. V. Cloning of large segments of exogenous DNA into yeast by means of artificial chromosome vectors. (1987) *Science*, **236**, 806-12.
- C. elegans Sequencing Consortium, T. Genome sequence of the nematode *C. elegans*: a platform for investigating biology. The *C. elegans* Sequencing Consortium. (1998) *Science*, **282**, 2012-8.

- Carpten, J., Nupponen, N., Isaacs, S., Sood, R., Robbins, C., Xu, J., et al. Germline mutations in the ribonuclease L gene in families showing linkage with HPC1. (2002) *Nat Genet*, **30**, 181-4.
- Carrano, A. V., Lamerdin, J., Ashworth, L. K., Watkins, B., Branscomb, E., Slezak, T., et al. A high-resolution, fluorescence-based, semiautomated method for DNA fingerprinting. (1989) *Genomics*, **4**, 129-36.
- Carrier, A., Rosier, M. F., Guillemot, F., Goguel, A. F., Pulcini, F., Bernheim, A., et al. Integrated physical, genetic, and genic map covering 3 Mb around the human NGF gene (NGFB) at 1p13. (1996) *Genomics*, **31**, 80-9.
- Charroux, B., Pellizzoni, L., Perkinson, R. A., Shevchenko, A., Mann, M. and Dreyfuss, G. Gemin3: A novel DEAD box protein that interacts with SMN, the spinal muscular atrophy gene product, and is a component of gems. (1999) *J Cell Biol*, **147**, 1181-94.
- Chasman, D. and Adams, R. M. Predicting the functional consequences of non-synonymous single nucleotide polymorphisms: structure-based assessment of amino acid variation. (2001) *J Mol Biol*, **307**, 683-706.
- Chelly, J., Tumer, Z., Tonnesen, T., Petterson, A., Ishikawa-Brush, Y., Tommerup, N., et al. Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. (1993) *Nat Genet*, **3**, 14-9.
- Chen, C. L., Liu, Q. and Relling, M. V. Simultaneous characterization of glutathione S-transferase M1 and T1 polymorphisms by polymerase chain reaction in American whites and blacks. (1996) *Pharmacogenetics*, **6**, 187-91.
- Choo, K. H., Gould, K. G., Rees, D. J. and Brownlee, G. G. Molecular cloning of the gene for human anti-haemophilic factor IX. (1982) *Nature*, **299**, 178-80.
- Chumakov, I. M., Le Gall, I., Billault, A., Ougen, P., Soularue, P., Guillou, S., et al. Isolation of chromosome 21-specific yeast artificial chromosomes from a total human genome library. (1992) *Nat Genet*, **1**, 222-5.
- Chumakov, I. M., Rigault, P., Le Gall, I., Bellanne-Chantelot, C., Billault, A., Guillou, S., et al. A YAC contig map of the human genome. (1995) *Nature*, **377**, 175-297.

- Claudio, J. O., Liew, C. C., Ma, J., Heng, H. H., Stewart, A. K. and Hawley, R. G. Cloning and expression analysis of a novel WD repeat gene, WDR3, mapping to 1p12-p13. (1999) *Genomics*, **59**, 85-9.
- Coffey, A. J., Roberts, R. G., Green, E. D., Cole, C. G., Butler, R., Anand, R., et al. Construction of a 2.6-Mb contig in yeast artificial chromosomes spanning the human dystrophin gene using an STS-based approach. (1992) *Genomics*, **12**, 474-84.
- Cohen, D., Chumakov, I. and Weissenbach, J. A first-generation physical map of the human genome. (1993) *Nature*, **366**, 698-701.
- Collins, F. S., Brooks, L. D. and Chakravarti, A. A DNA polymorphism discovery resource for research on human genetic variation. (1998) *Genome Res*, **8**, 1229-31.
- Collins, J. E., Cole, C. G., Smink, L. J., Garrett, C. L., Levensha, M. A., Soderlund, C. A., et al. A high-density YAC contig map of human chromosome 22. (1995) *Nature*, **377**, 367-79.
- Collins, J. E., Goward, M. E., Cole, C. G., Smink, L. J., Huckle, E. J., Knowles, S., et al. Reevaluating human gene annotation: a second-generation analysis of chromosome 22. (2003) *Genome Res*, **13**, 27-36.
- Connelly, M. A., Zhang, H., Kieleczawa, J. and Anderson, C. W. The promoters for human DNA-PKcs (PRKDC) and MCM4: divergently transcribed genes located at chromosome 8 band q11. (1998) *Genomics*, **47**, 71-83.
- Coulondre, C., Miller, J. H., Farabaugh, P. J. and Gilbert, W. Molecular basis of base substitution hotspots in *Escherichia coli*. (1978) *Nature*, **274**, 775-80.
- Coulson, A., Huynh, C., Kozono, Y. and Shownkeen, R. The physical map of the *Caenorhabditis elegans* genome. (1995) *Methods Cell Biol*, **48**, 533-50.
- Coulson, A., Sulston, J., Brenner, S., Karn, J. Toward a physical map of the genome of the nematode *Caenorhabditis elegans*. (1986) *Proc Natl Acad Sci U S A*, **83**, 7821-7825.
- Couzin, J. Human genome. HapMap launched with pledges of \$100 million. (2002) *Science*, **298**, 941-2.

- Cox, D. R., Burmeister, M., Price, E. R., Kim, S. and Myers, R. M. Radiation hybrid mapping: a somatic cell genetic method for constructing high-resolution maps of mammalian chromosomes. (1990) *Science*, **250**, 245-50.
- Dauwerse, J. G., Kievits, T., Beverstock, G. C., van der Keur, D., Smit, E., Wessels, H. W., et al. Rapid detection of chromosome 16 inversion in acute nonlymphocytic leukemia, subtype M4: regional localization of the breakpoint in 16p. (1990) *Cytogenet Cell Genet*, **53**, 126-8.
- Dawson, E., Chen, Y., Hunt, S., Smink, L. J., Hunt, A., Rice, K., et al. A SNP resource for human chromosome 22: extracting dense clusters of SNPs from the genomic sequence. (2001) *Genome Res*, **11**, 170-8.
- Deininger, P. L. a. D., G.R. The recent evolution of DNA repetitive elements. (1986) *Trends Genet*, **2**, 76-80.
- Dekker, J. W., Budhia, S., Angel, N. Z., Cooper, B. J., Clark, G. J., Hart, D. N., et al. Identification of an S-adenosylhomocysteine hydrolase-like transcript induced during dendritic cell differentiation. (2002) *Immunogenetics*, **53**, 993-1001.
- Dell'Angelica, E. C., Mullins, C. and Bonifacino, J. S. AP-4, a novel protein complex related to clathrin adaptors. (1999) *J Biol Chem*, **274**, 7278-85.
- Deloukas, P., Matthews, L. H., Ashurst, J., Burton, J., Gilbert, J. G., Jones, M., et al. The DNA sequence and comparative analysis of human chromosome 20. (2001) *Nature*, **414**, 865-71.
- Deloukas, P., Schuler, G. D., Gyapay, G., Beasley, E. M., Soderlund, C., Rodriguez-Tome, P., et al. A physical map of 30,000 human genes. (1998) *Science*, **282**, 744-6.
- DeMartino, G. N., Orth, K., McCullough, M. L., Lee, L. W., Munn, T. Z., Moomaw, C. R., et al. The primary structures of four subunits of the human, high-molecular-weight proteinase, macropain (proteasome), are distinct but homologous. (1991) *Biochim Biophys Acta*, **1079**, 29-38.
- Deutsch, S., Iseli, C., Bucher, P., Antonarakis, S. E. and Scott, H. S. A cSNP map and database for human chromosome 21. (2001) *Genome Res*, **11**, 300-7.

- Dib, C., Faure, S., Fizames, C., Samson, D., Drouot, N., Vignal, A., et al. A comprehensive genetic map of the human genome based on 5,264 microsatellites. (1996) *Nature*, **380**, 152-4.
- Ding, Y., Johnson, M. D., Chen, W. Q., Wong, D., Chen, Y. J., Benson, S. C., et al. Five-color-based high-information-content fingerprinting of bacterial artificial chromosome clones using type IIS restriction endonucleases. (2001) *Genomics*, **74**, 142-54.
- Doggett, N. A., Goodwin, L. A., Tesmer, J. G., Meincke, L. J., Bruce, D. C., Clark, L. M., et al. An integrated physical map of human chromosome 16. (1995) *Nature*, **377**, 335-65.
- Donis-Keller, H., Green, P., Helms, C., Cartinhour, S., Weiffenbach, B., Stephens, K., et al. A genetic linkage map of the human genome. (1987) *Cell*, **51**, 319-37.
- Down, T. A. and Hubbard, T. J. Computational detection and location of transcription start sites in mammalian genomic DNA. (2002) *Genome Res*, **12**, 458-61.
- Dracopoli, N. C., Rettig, W. J., Whitfield, G. K., Darlington, G. J., Spengler, B. A., Biedler, J. L., et al. Assignment of the gene for the beta subunit of thyroid-stimulating hormone to the short arm of human chromosome 1. (1986) *Proc Natl Acad Sci U S A*, **83**, 1822-6.
- Dumont, M., Luu-The, V., Dupont, E., Pelletier, G. and Labrie, F. Characterization, expression, and immunohistochemical localization of 3 beta-hydroxysteroid dehydrogenase/delta 5-delta 4 isomerase in human skin. (1992) *J Invest Dermatol*, **99**, 415-21.
- Dunham, I., Shimizu, N., Roe, B. A., Chissoe, S., Hunt, A. R., Collins, J. E., et al. The DNA sequence of human chromosome 22. (1999) *Nature*, **402**, 489-95.
- El-Rifai, W., Sarlomo-Rikala, M., Andersson, L. C., Knuutila, S. and Miettinen, M. DNA sequence copy number changes in gastrointestinal stromal tumors: tumor progression and prognostic significance. (2000) *Cancer Res*, **60**, 3899-903.
- Evans, G. A. and Lewis, K. A. Physical mapping of complex genomes by cosmid multiplex analysis. (1989) *Proc Natl Acad Sci U S A*, **86**, 5030-4.
- Ewing, B. and Green, P. Analysis of expressed sequence tags indicates 35,000 human genes. (2000) *Nat Genet*, **25**, 232-4.

- Ewing, B., Hillier, L., Wendl, M. C. and Green, P. Base-calling of automated sequencer traces using phred. I. Accuracy assessment. (1998) *Genome Res*, **8**, 175-85.
- Fields, C., Adams, M. D., White, O. and Venter, J. C. How many genes in the human genome? (1994) *Nat Genet*, **7**, 345-6.
- Fleischmann, R. D., Adams, M. D., White, O., Clayton, R. A., Kirkness, E. F., Kerlavage, A. R., et al. Whole-genome random sequencing and assembly of *Haemophilus influenzae* Rd. (1995) *Science*, **269**, 496-512.
- Foote, S., Vollrath, D., Hilton, A. and Page, D. C. The human Y chromosome: overlapping DNA clones spanning the euchromatic region. (1992) *Science*, **258**, 60-6.
- Forus, A., Weghuis, D. O., Smeets, D., Fodstad, O., Myklebost, O. and van Kessel, A. G. Comparative genomic hybridization analysis of human sarcomas: I. Occurrence of genomic imbalances and identification of a novel major amplicon at 1q21-q22 in soft tissue sarcomas. (1995) *Genes Chromosomes Cancer*, **14**, 8-14.
- Francke, U. Digitized and differentially shaded human chromosome ideograms for genomic applications. (1994) *Cytogenet Cell Genet*, **65**, 206-18.
- Galgoczy, P., Rosenthal, A. and Platzer, M. Human-mouse comparative sequence analysis of the NEMO gene reveals an alternative promoter within the neighboring G6PD gene. (2001) *Gene*, **271**, 93-8.
- Gardiner, K. Base composition and gene distribution: critical patterns in mammalian genome organization. (1996) *Trends Genet*, **12**, 519-24.
- Gardiner-Garden, M. and Frommer, M. CpG islands in vertebrate genomes. (1987) *J Mol Biol*, **196**, 261-82.
- Gengyo-Ando, K. and Mitani, S. Characterization of mutations induced by ethyl methanesulfonate, UV, and trimethylpsoralen in the nematode *Caenorhabditis elegans*. (2000) *Biochem Biophys Res Commun*, **269**, 64-9.
- Gibson, T. J. and Sulston, J. E. Preparation of large numbers of plasmid DNA samples in microtiter plates by the alkaline lysis method. (1987) *Gene Anal Tech*, **4**, 41-4.

- Gilbert, W. Why genes in pieces? (1978) *Nature*, **271**, 501.
- Gilbert, W. (1992) (Ed, Kelves, D. J. a. H., L.) Harvard University Press.
- Goffeau, A., Barrell, B. G., Bussey, H., Davis, R. W., Dujon, B., Feldmann, H., et al. Life with 6000 genes. (1996) *Science*, **274**, 546, 563-7.
- Goss, S. J. and Harris, H. New method for mapping genes in human chromosomes. (1975) *Nature*, **255**, 680-4.
- Gottgens, B., Barton, L. M., Gilbert, J. G., Bench, A. J., Sanchez, M. J., Bahn, S., et al. Analysis of vertebrate SCL loci identifies conserved enhancers. (2000) *Nat Biotechnol*, **18**, 181-6.
- Green, E. D. and Chakravarti, A. The human genome sequence expedition: views from the "base camp". (2001) *Genome Res*, **11**, 645-51.
- Green, E. D. and Olson, M. V. Chromosomal region of the cystic fibrosis gene in yeast artificial chromosomes: a model for human genome mapping. (1990) *Science*, **250**, 94-8.
- Green, E. D., Riethman, H. C., Dutchik, J. E. and Olson, M. V. Detection and characterization of chimeric yeast artificial-chromosome clones. (1991) *Genomics*, **11**, 658-69.
- Green, P. Whole-genome disassembly. (2002) *Proc Natl Acad Sci U S A*, **99**, 4143-4.
- Green, P. J., Pines O, Inouye M. The role of antisense RNA in gene regulation. (1986) *Annu Rev Biochem*, **55**, 569-97.
- Gregory, S. G., Howell, G. R. and Bentley, D. R. Genome mapping by fluorescent fingerprinting. (1997) *Genome Res*, **7**, 1162-8.
- Gregory, S. G., Sekhon, M., Schein, J., Zhao, S., Osoegawa, K., Scott, C. E., et al. A physical map of the mouse genome. (2002) *Nature*, **418**, 743-50.
- Gregory, S. G., Vaudin, M., Wooster, R., Coleman, M., Mischke, D., Porter, C., et al. Report of the fourth international workshop on human chromosome 1 mapping 1998. (1998) *Cytogenet Cell Genet*, **83**, 147-75.
- Guex, N. and Peitsch, M. C. SWISS-MODEL and the Swiss-PdbViewer: an environment for comparative protein modeling. (1997) *Electrophoresis*, **18**, 2714-23.

- Guigo, R., Agarwal, P., Abril, J. F., Burset, M. and Fickett, J. W. An assessment of gene prediction accuracy in large DNA sequences. (2000) *Genome Res*, **10**, 1631-42.
- Guru, S. C., Olufemi, S. E., Manickam, P., Cummings, C., Gieser, L. M., Pike, B. L., et al. A 2.8-Mb clone contig of the multiple endocrine neoplasia type 1 (MEN1) region at 11q13. (1997) *Genomics*, **42**, 436-45.
- Gyapay, G., Morissette, J., Vignal, A., Dib, C., Fizames, C., Millasseau, P., et al. The 1993-94 Genethon human genetic linkage map. (1994) *Nat Genet*, **7**, 246-339.
- Hall, A. and Brown, R. Human N-ras: cDNA cloning and gene structure. (1985) *Nucleic Acids Res*, **13**, 5255-68.
- Halushka, M. K., Fan, J. B., Bentley, K., Hsie, L., Shen, N., Weder, A., et al. Patterns of single-nucleotide polymorphisms in candidate genes for blood-pressure homeostasis. (1999) *Nat Genet*, **22**, 239-47.
- Hardison, R., Slightom, J. L., Gumucio, D. L., Goodman, M., Stojanovic, N. and Miller, W. Locus control regions of mammalian beta-globin gene clusters: combining phylogenetic analyses and experimental results to gain functional insights. (1997) *Gene*, **205**, 73-94.
- Hardison, R., Xu, J., Jackson, J., Mansberger, J., Selifonova, O., Grotch, B., et al. Comparative analysis of the locus control region of the rabbit beta-like gene cluster: HS3 increases transient expression of an embryonic epsilon-globin gene. (1993) *Nucleic Acids Res*, **21**, 1265-72.
- Harris, R. F. Hapmap flap. (2002) *Curr Biol*, **12**, R827.
- Harrison, P. M., Hegyi, H., Balasubramanian, S., Luscombe, N. M., Bertone, P., Echols, N., et al. Molecular fossils in the human genome: identification and analysis of the pseudogenes in chromosomes 21 and 22. (2002) *Genome Res*, **12**, 272-80.
- Hattori, M., Fujiyama, A., Taylor, T. D., Watanabe, H., Yada, T., Park, H. S., et al. The DNA sequence of human chromosome 21. (2000) *Nature*, **405**, 311-9.
- Hayes, J. D. and Pulford, D. J. The glutathione S-transferase supergene family: regulation of GST and the contribution of the isoenzymes to cancer chemoprotection and drug resistance. (1995) *Crit Rev Biochem Mol Biol*, **30**, 445-600.

- Heding, I. J., Ivens, A. C., Wilson, J., Strivens, M., Gregory, S., Hoovers, J. M., et al. The generation of ordered sets of cosmid DNA clones from human chromosome region 11p. (1992) *Genomics*, **13**, 89-94.
- Heiskanen, M., Karhu, R., Hellsten, E., Peltonen, L., Kallioniemi, O. P. and Palotie, A. High resolution mapping using fluorescence in situ hybridization to extended DNA fibers prepared from agarose-embedded cells. (1994) *Biotechniques*, **17**, 928-9, 932-3.
- Henneberry, A. L. and McMaster, C. R. Cloning and expression of a human choline/ethanolaminephosphotransferase: synthesis of phosphatidylcholine and phosphatidylethanolamine. (1999) *Biochem J*, **339 (Pt 2)**, 291-8.
- Higgins, D. G. CLUSTAL V: multiple alignment of DNA and protein sequences. (1994) *Methods Mol Biol*, **25**, 307-18.
- Hijikata, M., Ohta, Y. and Mishiro, S. Identification of a single nucleotide polymorphism in the MxA gene promoter (G/T at nt -88) correlated with the response of hepatitis C patients to interferon. (2000) *Intervirology*, **43**, 124-7.
- Hillier, L. D., Lennon, G., Becker, M., Bonaldo, M. F., Chiapelli, B., Chissoe, S., et al. Generation and analysis of 280,000 human expressed sequence tags. (1996) *Genome Res*, **6**, 807-28.
- Hoffman, H. M., Mueller, J. L., Broide, D. H., Wanderer, A. A. and Kolodner, R. D. Mutation of a new gene encoding a putative pyrin-like protein causes familial cold autoinflammatory syndrome and Muckle-Wells syndrome. (2001) *Nat Genet*, **29**, 301-5.
- Horton, R., Niblett, D., Milne, S., Palmer, S., Tubby, B., Trowsdale, J., et al. Large-scale sequence comparisons reveal unusually high levels of variation in the HLA-DQB1 locus in the class II region of the human MHC. (1998) *Journal Of Molecular Biology*, **282**, 71-97.
- Hoskins, R. A., Nelson, C. R., Berman, B. P., Lavery, T. R., George, R. A., Ciesiolka, L., et al. A BAC-based physical map of the major autosomes of *Drosophila melanogaster*. (2000) *Science*, **287**, 2271-4.
- Hsia, D. Y.-Y., Naylor, J., Bigler, J. A. Gaucher's disease: report of two cases in father and son and review of the literature. (1959) *New Eng. J. Med.*, **261**, 164-169.

- Hu, B., Trinh, K., Figueira, W. F. and Price, P. A. Isolation and sequence of a novel human chondrocyte protein related to mammalian members of the chitinase protein family. (1996) *J Biol Chem*, **271**, 19415-20.
- Hubert, R. S., Mitchell, S., Chen, X. N., Ekmekji, K., Gadomski, C., Sun, Z., et al. BAC and PAC contigs covering 3.5 Mb of the Down syndrome congenital heart disease region between D21S55 and MX1 on chromosome 21. (1997) *Genomics*, **41**, 218-26.
- Hudson, T. J., Engelstein, M., Lee, M. K., Ho, E. C., Rubenfield, M. J., Adams, C. P., et al. Isolation and chromosomal assignment of 100 highly informative human simple sequence repeat polymorphisms. (1992) *Genomics*, **13**, 622-9.
- Hudson, T. J., Stein, L. D., Gerety, S. S., Ma, J., Castle, A. B., Silva, J., et al. An STS-based map of the human genome. (1995) *Science*, **270**, 1945-54.
- Huntington's Disease Collaborative Research Group, T. A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. (1993) *Cell*, **72**, 971-83.
- Hurst, L. D. and Eyre-Walker, A. Evolutionary genomics: reading the bands. (2000) *Bioessays*, **22**, 105-7.
- International Human Genome Sequencing Consortium, T. Initial sequencing and analysis of the human genome. (2001) *Nature*, **409**, 860-921.
- International SNP Map Working Group, T. A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms. (2001) *Nature*, **409**, 928-33.
- Ioannou, P. A., Amemiya, C. T., Garnes, J., Kroisel, P. M., Shizuya, H., Chen, C., et al. A new bacteriophage P1-derived vector for the propagation of large human DNA fragments. (1994) *Nat Genet*, **6**, 84-9.
- Irizarry, K., Kustanovich, V., Li, C., Brown, N., Nelson, S., Wong, W., et al. Genome-wide analysis of single-nucleotide polymorphisms in human expressed sequences. (2000) *Nat Genet*, **26**, 233-6.
- Isbrandt, D., Leicher, T., Waldschutz, R., Zhu, X., Luhmann, U., Michel, U., et al. Gene structures and expression profiles of three human KCND (Kv4) potassium channels mediating A-type currents I(TO) and I(SA). (2000) *Genomics*, **64**, 144-54.

- Jang, W., Hua, A., Spilson, S. V., Miller, W., Roe, B. A. and Meisler, M. H. Comparative sequence of human and mouse BAC clones from the mnd2 region of chromosome 2p13. (1999) *Genome Res*, **9**, 53-61.
- Jeffers, M., Paciucci, R. and Pellicer, A. Characterization of unr; a gene closely linked to N-ras. (1990) *Nucleic Acids Res*, **18**, 4891-9.
- Jeffreys, A. J., Wilson, V. and Thein, S. L. Hypervariable 'minisatellite' regions in human DNA. (1985) *Nature*, **314**, 67-73.
- Jones, J. M., Morrell, J. C. and Gould, S. J. Identification and characterization of HAOX1, HAOX2, and HAOX3, three human peroxisomal 2-hydroxy acid oxidases. (2000) *J Biol Chem*, **275**, 12590-7.
- Jurka, J. Sequence patterns indicate an enzymatic involvement in integration of mammalian retroposons. (1997) *Proc Natl Acad Sci U S A*, **94**, 1872-7.
- Kan, Y. W. and Dozy, A. M. Polymorphism of DNA sequence adjacent to human beta-globin structural gene: relationship to sickle mutation. (1978) *Proc Natl Acad Sci U S A*, **75**, 5631-5.
- Karvonen, M. K., Pesonen, U., Heinonen, P., Laakso, M., Rissanen, A., Naukkarinen, H., et al. Identification of new sequence variants in the leptin gene. (1998) *J Clin Endocrinol Metab*, **83**, 3239-42.
- Kent, W. J. BLAT--the BLAST-like alignment tool. (2002) *Genome Res*, **12**, 656-64.
- Kessler, M. M., Beckendorf, R. C., Westhafer, M. A. and Nordstrom, J. L. Requirement of A-A-U-A-A-A and adjacent downstream sequences for SV40 early polyadenylation. (1986) *Nucleic Acids Res*, **14**, 4939-52.
- Khan, A. S., Wilcox, A. S., Hopkins, J. A. and Sikela, J. M. Efficient double stranded sequencing of cDNA clones containing long poly(A) tails using anchored poly(dT) primers. (1991) *Nucleic Acids Res*, **19**, 1715.
- Khan, S. G., Muniz-Medina, V., Shahlavi, T., Baker, C. C., Inui, H., Ueda, T., et al. The human XPC DNA repair gene: arrangement, splice site information content and influence of a single

- nucleotide polymorphism in a splice acceptor site on alternative splicing and function. (2002) *Nucleic Acids Res*, **30**, 3624-31.
- Kiechle, M., Hinrichs, M., Jacobsen, A., Luttgies, J., Pfisterer, J., Kommoss, F., et al. Genetic imbalances in precursor lesions of endometrial cancer detected by comparative genomic hybridization. (2000) *Am J Pathol*, **156**, 1827-33.
- Kim, U. J., Shizuya, H., de Jong, P. J., Birren, B. and Simon, M. I. Stable propagation of cosmid sized human DNA inserts in an F factor based vector. (1992) *Nucleic Acids Res*, **20**, 1083-5.
- Kimberling, W. J., Weston, M. D., Moller, C., Davenport, S. L., Shugart, Y. Y., Priluck, I. A., et al. Localization of Usher syndrome type II to chromosome 1q. (1990) *Genomics*, **7**, 245-9.
- Kitayama, H., Sugimoto, Y., Matsuzaki, T., Ikawa, Y. and Noda, M. A ras-related gene with transformation suppressor activity. (1989) *Cell*, **56**, 77-84.
- Knight, J. C., Udalova, I., Hill, A. V., Greenwood, B. M., Peshu, N., Marsh, K., et al. A polymorphism that affects OCT-1 binding to the TNF promoter region is associated with severe malaria. (1999) *Nat Genet*, **22**, 145-50.
- Kohara, Y., Akiyama, K. and Isono, K. The physical map of the whole E. coli chromosome: application of a new strategy for rapid analysis and sorting of a large genomic library. (1987) *Cell*, **50**, 495-508.
- Kohl, S., Baumann, B., Rosenberg, T., Kellner, U., Lorenz, B., Vadala, M., et al. Mutations in the cone photoreceptor G-protein alpha-subunit gene GNAT2 in patients with achromatopsia. (2002) *Am J Hum Genet*, **71**, 422-5.
- Kondo, S., Schutte, B. C., Richardson, R. J., Bjork, B. C., Knight, A. S., Watanabe, Y., et al. Mutations in IRF6 cause Van der Woude and popliteal pterygium syndromes. (2002) *Nat Genet*, **32**, 285-9.
- Kondoh, N., Nishina, Y., Tsuchida, J., Koga, M., Tanaka, H., Uchida, K., et al. Assignment of synaptonemal complex protein 1 (SCP1) to human chromosome 1p13 by fluorescence in situ hybridization and its expression in the testis. (1997) *Cytogenet Cell Genet*, **78**, 103-4.

- Kondrashov, A. S. and Shabalina, S. A. Classification of common conserved sequences in mammalian intergenic regions. (2002) *Hum Mol Genet*, **11**, 669-74.
- Kong, A., Gudbjartsson, D. F., Sainz, J., Jonsdottir, G. M., Gudjonsson, S. A., Richardsson, B., et al. A high-resolution recombination map of the human genome. (2002) *Nat Genet*, **31**, 241-7.
- Koop, B. F. and Hood, L. Striking sequence similarity over almost 100 kilobases of human and mouse T-cell receptor DNA. (1994) *Nat Genet*, **7**, 48-53.
- Kozak, M. An analysis of 5'-noncoding sequences from 699 vertebrate messenger RNAs. (1987) *Nucleic Acids Res*, **15**, 8125-48.
- Kritzler, R. A., Terner, J. Y., Lindenbaum, J., Magidson, J., Williams, R., Preisig, R., Phillips, G. B. Chediak-Higashi syndrome: cytologic and serum lipid observations in a case and family. (1964) *Am. J. Med*, **36**, 583-594.
- Kruglyak, L. and Nickerson, D. A. Variation is the spice of life. (2001) *Nat Genet*, **27**, 234-6.
- Kudoh, J., Nagamine, K., Asakawa, S., Abe, I., Kawasaki, K., Maeda, H., et al. Localization of 16 exons to a 450-kb region involved in the autoimmune polyglandular disease type I (APECED) on human chromosome 21q22.3. (1997) *DNA Res*, **4**, 45-52.
- Kupfer, K., Smith, M. W., Quackenbush, J. and Evans, G. A. Physical mapping of complex genomes by sampled sequencing: a theoretical analysis. (1995) *Genomics*, **27**, 90-100.
- Labay, V., Raz, T., Baron, D., Mandel, H., Williams, H., Barrett, T., et al. Mutations in SLC19A2 cause thiamine-responsive megaloblastic anaemia associated with diabetes mellitus and deafness. (1999) *Nat Genet*, **22**, 300-4.
- Lachance, Y., Luu-The, V., Verreault, H., Dumont, M., Rheaume, E., Leblanc, G., et al. Structure of the human type II 3 beta-hydroxysteroid dehydrogenase/delta 5-delta 4 isomerase (3 beta-HSD) gene: adrenal and gonadal specificity. (1991) *DNA Cell Biol*, **10**, 701-11.
- Lahat, H., Pras, E., Olender, T., Avidan, N., Ben-Asher, E., Man, O., et al. A missense mutation in a highly conserved region of CASQ2 is associated with autosomal recessive catecholamine-induced polymorphic ventricular tachycardia in Bedouin families from Israel. (2001) *Am J Hum Genet*, **69**, 1378-84.

- Lamerdin, J. E. and Carrano, A. V. Automated fluorescence-based restriction fragment analysis. (1993) *Biotechniques*, **15**, 294-303.
- Larsen, F., Gundersen, G., Lopez, R. and Prydz, H. CpG islands as gene markers in the human genome. (1992) *Genomics*, **13**, 1095-107.
- Lee, K. A., Kim, S. H., Woo, H. Y., Hong, Y. J. and Cho, H. C. Increased frequencies of glutathione S-transferase (GSTM1 and GSTT1) gene deletions in Korean patients with acquired aplastic anemia. (2001) *Blood*, **98**, 3483-5.
- Levine, A. and Durbin, R. A computational scan for U12-dependent introns in the human genome sequence. (2001) *Nucleic Acids Res*, **29**, 4006-13.
- Levy-Lahad, E., Wijsman, E. M., Nemens, E., Anderson, L., Goddard, K. A., Weber, J. L., et al. A familial Alzheimer's disease locus on chromosome 1. (1995) *Science*, **269**, 970-3.
- Li, W. H. and Sadler, L. A. Low nucleotide diversity in man. (1991) *Genetics*, **129**, 513-23.
- Liang, F., Holt, I., Pertea, G., Karamycheva, S., Salzberg, S. L. and Quackenbush, J. Gene index analysis of the human genome estimates approximately 120,000 genes. (2000) *Nat Genet*, **25**, 239-40.
- Litt, M. and Luty, J. A. A hypervariable microsatellite revealed by in vitro amplification of a dinucleotide repeat within the cardiac muscle actin gene. (1989) *Am J Hum Genet*, **44**, 397-401.
- Little, P., Curtis, P., Coutelle, C., Van den Berg, J., Dagleish, R., Malcolm, S., et al. Isolation and partial sequence of recombinant plasmids containing human alpha-, beta- and gamma-globin cDNA fragments. (1978) *Nature*, **273**, 640-3.
- Liu, H., Nakagawa, T., Kanematsu, T., Uchida, T. and Tsuji, S. Isolation of 10 differentially expressed cDNAs in differentiated Neuro2a cells induced through controlled expression of the GD3 synthase gene. (1999) *J Neurochem*, **72**, 1781-90.
- Loetscher, P., Alvarez-Gonzalez, R. and Althaus, F. R. Poly(ADP-ribose) may signal changing metabolic conditions to the chromatin of mammalian cells. (1987) *Proc Natl Acad Sci U S A*, **84**, 1286-9.

- Makalowski, W., Zhang, J. and Boguski, M. S. Comparative analysis of 1196 orthologous mouse and human full-length mRNA and protein sequences. (1996) *Genome Res*, **6**, 846-57.
- Mallon, A. M., Platzer, M., Bate, R., Gloeckner, G., Botcherby, M. R., Nordsiek, G., et al. Comparative genome sequence analysis of the Bpa/Str region in mouse and Man. (2000) *Genome Res*, **10**, 758-75.
- Mannervik, B. and Danielson, U. H. Glutathione transferases--structure and catalytic activity. (1988) *CRC Crit Rev Biochem*, **23**, 283-337.
- Marcelino, J., Carpten, J. D., Suwairi, W. M., Gutierrez, O. M., Schwartz, S., Robbins, C., et al. CACP, encoding a secreted proteoglycan, is mutated in camptodactyly-arthropathy-coxa vara-pericarditis syndrome. (1999) *Nat Genet*, **23**, 319-22.
- Marra, M., Kucaba, T., Sekhon, M., Hillier, L., Martienssen, R., Chinwalla, A., et al. A map for sequence analysis of the *Arabidopsis thaliana* genome. (1999) *Nat Genet*, **22**, 265-70.
- Marra, M. A., Kucaba, T. A., Dietrich, N. L., Green, E. D., Brownstein, B., Wilson, R. K., et al. High throughput fingerprint analysis of large-insert clones. (1997) *Genome Res*, **7**, 1072-84.
- Marshall, E. Drug firms to create public database of genetic mutations. (1999) *Science*, **284**, 406-7.
- Marth, G., Yeh, R., Minton, M., Donaldson, R., Li, Q., Duan, S., et al. Single-nucleotide polymorphisms in the public domain: how useful are they? (2001) *Nat Genet*, **27**, 371-2.
- Mattick, J. S. Non-coding RNAs: the architects of eukaryotic complexity. (2001) *EMBO Rep*, **2**, 986-91.
- Maxam, A. M. and Gilbert, W. A new method for sequencing DNA. (1977) *Proc Natl Acad Sci U S A*, **74**, 560-4.
- McPherson, J. D., Marra, M., Hillier, L., Waterston, R. H., Chinwalla, A., Wallis, J., et al. A physical map of the human genome. (2001) *Nature*, **409**, 934-41.
- Meek, T. D., Dayton, B. D., Metcalf, B. W., Dreyer, G. B., Strickler, J. E., Gorniak, J. G., et al. Human immunodeficiency virus 1 protease expressed in *Escherichia coli* behaves as a dimeric aspartic protease. (1989) *Proc Natl Acad Sci U S A*, **86**, 1841-5.

- Mercher, T., Coniat, M. B., Monni, R., Mauchauffe, M., Khac, F. N., Gressin, L., et al. Involvement of a human gene related to the *Drosophila* spen gene in the recurrent t(1;22) translocation of acute megakaryocytic leukemia. (2001) *Proc Natl Acad Sci U S A*, **98**, 5776-9.
- Milpetz, F., Argos, P. and Persson, B. TMAP: a new email and WWW service for membrane-protein structural predictions. (1995) *Trends Biochem Sci*, **20**, 204-5.
- Mirzoeva, S., Weigand, S., Lukas, T. J., Shuvalova, L., Anderson, W. F. and Watterson, D. M. Analysis of the functional coupling between calmodulin's calcium binding and peptide recognition properties. (1999) *Biochemistry*, **38**, 3936-47.
- Misra, S., 1, 2, Madeline A Crosby³, Christopher J Mungall^{2, 4}, Beverley B Matthews³, Kathryn S Campbell³, Pavel Hradecky³, Yanmei Huang³, Joshua S Kaminker^{1, 2}, Gillian H Millburn⁵, Simon E Prochnik^{1, 2}, Christopher D Smith^{1, 2}, Jonathan L Tupy^{1, 2}, Eleanor J Whitfield⁶, Leyla Bayraktaroglu³, Benjamin P Berman¹, Brian R Bettencourt³, Susan E Celniker⁷, Aubrey DNJ de Grey⁵, Rachel A Drysdale⁵, Nomi L Harris^{2, 7}, John Richter⁴, Susan Russo³, Andrew J Schroeder³, Sheng Qiang Shu^{1, 2}, Mark Stapleton⁷, Chihiro Yamada⁵, Michael Ashburner⁵, William M Gelbart³, Gerald M Rubin^{1, 2, 4, 7} and Suzanna E Lewis^{1, 2}
Annotation of the *Drosophila melanogaster* euchromatic genome: a systematic review. (2002) *Genome Biology*, **3**.
- Mochizuki, N., Cho, G., Wen, B. and Insel, P. A. Identification and cDNA cloning of a novel human mosaic protein, LGN, based on interaction with G alpha i2. (1996) *Gene*, **181**, 39-43.
- Modrek, B. and Lee, C. A genomic view of alternative splicing. (2002) *Nat Genet*, **30**, 13-9.
- Moore, M. J. and Sharp, P. A. Evidence for two active sites in the spliceosome provided by stereochemistry of pre-mRNA splicing. (1993) *Nature*, **365**, 364-8.
- Morton, N. E. Parameters of the human genome. (1991) *Proc Natl Acad Sci U S A*, **88**, 7474-6.
- Mouse Genome Sequencing Consortium, T. Initial sequencing and comparative analysis of the mouse genome. (2002) *Nature*, **420**, 520-62.

- Mulligan, L. M., Kwok, J. B., Healey, C. S., Elsdon, M. J., Eng, C., Gardner, E., et al. Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. (1993) *Nature*, **363**, 458-60.
- Mullikin, J. C., Hunt, S. E., Cole, C. G., Mortimore, B. J., Rice, C. M., Burton, J., et al. An SNP map of human chromosome 22. (2000) *Nature*, **407**, 516-20.
- Mungall, A. J., Edwards, C. A., Ranby, S. A., Humphray, S. J., Heathcott, R. W., Clee, C. M., et al. Physical mapping of chromosome 6: a strategy for the rapid generation of sequence-ready contigs. (1996) *DNA Seq*, **7**, 47-9.
- Murray, J. C., Buetow, K. H., Weber, J. L., Ludwigsen, S., Scherpbier-Heddema, T., Manion, F., et al. A comprehensive human linkage map with centimorgan density. Cooperative Human Linkage Center (CHLC). (1994) *Science*, **265**, 2049-54.
- Murzin, A. G., Brenner, S. E., Hubbard, T. and Chothia, C. SCOP: a structural classification of proteins database for the investigation of sequences and structures. (1995) *J Mol Biol*, **247**, 536-40.
- Myers, E. W., Sutton, G. G., Smith, H. O., Adams, M. D. and Venter, J. C. On the sequencing and assembly of the human genome. (2002) *Proc Natl Acad Sci U S A*, **99**, 4145-6.
- Nagasaki, K., Maass, N., Manabe, T., Hanzawa, H., Tsukada, T., Kikuchi, K., et al. Identification of a novel gene, DAM1, amplified at chromosome 1p13.3-21 region in human breast cancer cell lines. (1999) *Cancer Lett*, **140**, 219-26.
- Nagase, T., Ishikawa, K., Suyama, M., Kikuno, R., Hirose, M., and Miyajima, N., Tanaka, A., Kotani, H., Nomura, N. and Ohara, O. Prediction of the coding sequences of unidentified human genes.
- XII. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. (1998) *Dna Research*, **5**, 355-364.
- Nagata, T., Mugishima, H., Shichino, H., Suzuki, T., Chin, M., Koshinaga, S., et al. Karyotypic analyses of hepatoblastoma. Report of two cases and review of the literature suggesting

- chromosomal loci responsible for the pathogenesis of this disease. (1999) *Cancer Genet Cytogenet*, **114**, 42-50.
- Nakachi, K., Imai, K., Hayashi, S., Watanabe, J. and Kawajiri, K. Genetic susceptibility to squamous cell carcinoma of the lung in relation to cigarette smoking dose. (1991) *Cancer Res*, **51**, 5177-80.
- Nakamura, Y., Leppert, M., O'Connell, P., Wolff, R., Holm, T., Culver, M., et al. Variable number of tandem repeat (VNTR) markers for human gene mapping. (1987) *Science*, **235**, 1616-22.
- Navia, M. A., Fitzgerald, P. M., McKeever, B. M., Leu, C. T., Heimbach, J. C., Herber, W. K., et al. Three-dimensional structure of aspartyl protease from human immunodeficiency virus HIV-1. (1989) *Nature*, **337**, 615-20.
- Nicholas, K. B., Nicholas H.B. , and Deerfield, D.W. GeneDoc: Analysis and Visualization of Genetic Variation. (1997) *EMBNEW.NEWS*, **4**.
- Ning, Z., Cox, A. J. and Mullikin, J. C. SSAHA: a fast search method for large DNA databases. (2001) *Genome Res*, **11**, 1725-9.
- Nitta, N., Ochiai, M., Nagao, M. and Sugimura, T. Amino-acid substitution at codon 13 of the N-ras oncogene in rectal cancer in a Japanese patient. (1987) *Jpn J Cancer Res*, **78**, 21-6.
- Okubo, K., Hori, N., Matoba, R., Niiyama, T., Fukushima, A., Kojima, Y., et al. Large scale cDNA sequencing for analysis of quantitative and qualitative aspects of gene expression. (1992) *Nat Genet*, **2**, 173-9.
- Oliver, J. L. and Marín, A. A relationship between GC content and coding-sequence length. (1996) *Journal Of Molecular Evolution*, **43**, 216-23.
- Olson, M., Hood, L., Cantor, C. and Botstein, D. A common language for physical mapping of the human genome. (1989) *Science*, **245**, 1434-5.
- Olson, M. V., Dutchik, J. E., Graham, M. Y., Brodeur, G. M., Helms, C., Frank, M., et al. Random-clone strategy for genomic restriction mapping in yeast. (1986) *Proc Natl Acad Sci U S A*, **83**, 7826-30.

- Olson, M. V. and Green, P. Criterion for the completeness of large-scale physical maps of DNA. (1993) *Cold Spring Harb Symp Quant Biol*, **58**, 349-55.
- Orias, M., Bray-Ward, P., Curran, M. E., Keating, M. T. and Desir, G. V. Genomic localization of the human gene for KCNA10, a cGMP-activated K channel. (1997) *Genomics*, **42**, 33-7.
- Pan, S. S., Han, Y., Farabaugh, P. and Xia, H. Implication of alternative splicing for expression of a variant NAD(P)H:quinone oxidoreductase-1 with a single nucleotide polymorphism at 465C>T. (2002) *Pharmacogenetics*, **12**, 479-88.
- Parvari, R., HersHKovitz, E., Grossman, N., Gorodischer, R., Loeys, B., Zecic, A., et al. Mutation of TBCE causes hypoparathyroidism-retardation-dysmorphism and autosomal recessive Kenny-Caffey syndrome. (2002) *Nat Genet*, **32**, 448-52.
- Pearl, L. H. and Taylor, W. R. Sequence specificity of retroviral proteases. (1987a) *Nature*, **328**, 482.
- Pearl, L. H. and Taylor, W. R. A structural model for the retroviral proteases. (1987b) *Nature*, **329**, 351-4.
- Peitsch, M. C. and Jongeneel, C. V. A 3-D model for the CD40 ligand predicts that it is a compact trimer similar to the tumor necrosis factors. (1993) *Int Immunol*, **5**, 233-8.
- Pemble, S., Schroeder, K. R., Spencer, S. R., Meyer, D. J., Hallier, E., Bolt, H. M., et al. Human glutathione S-transferase theta (GSTT1): cDNA cloning and the characterization of a genetic polymorphism. (1994) *Biochem J*, **300 (Pt 1)**, 271-6.
- Pennacchio, L. A., Olivier, M., Hubacek, J. A., Cohen, J. C., Cox, D. R., Fruchart, J. C., et al. An apolipoprotein influencing triglycerides in humans and mice revealed by comparative sequencing. (2001) *Science*, **294**, 169-73.
- Pennacchio, L. A. and Rubin, E. M. Genomic strategies to identify mammalian regulatory sequences. (2001) *Nat Rev Genet*, **2**, 100-9.
- Pericak-Vance, M. A., Bebout, J. L., Gaskell, P. C., Jr., Yamaoka, L. H., Hung, W. Y., Alberts, M. J., et al. Linkage studies in familial Alzheimer disease: evidence for chromosome 19 linkage. (1991) *Am J Hum Genet*, **48**, 1034-50.

- Persson, B. and Argos, P. Prediction of transmembrane segments in proteins utilising multiple sequence alignments. (1994) *J Mol Biol*, **237**, 182-92.
- Petersen, C. M., Nielsen, M. S., Nykjaer, A., Jacobsen, L., Tommerup, N., Rasmussen, H. H., et al. Molecular identification of a novel candidate sorting receptor purified from human brain by receptor-associated protein affinity chromatography. (1997) *J Biol Chem*, **272**, 3599-605.
- Pfost, D. R., Boyce-Jacino, M. T. and Grant, D. M. A SNPshot: pharmacogenetics and the future of drug therapy. (2000) *Trends Biotechnol*, **18**, 334-8.
- Picoult-Newberg, L., Ideker, T. E., Pohl, M. G., Taylor, S. L., Donaldson, M. A., Nickerson, D. A., et al. Mining SNPs from EST databases. (1999) *Genome Res*, **9**, 167-74.
- Pinkel, D., Landegent, J., Collins, C., Fuscoe, J., Segraves, R., Lucas, J., et al. Fluorescence in situ hybridization with human chromosome-specific libraries: detection of trisomy 21 and translocations of chromosome 4. (1988) *Proc Natl Acad Sci U S A*, **85**, 9138-42.
- Pinkel, D., Straume, T. and Gray, J. W. Cytogenetic analysis using quantitative, high-sensitivity, fluorescence hybridization. (1986) *Proc Natl Acad Sci U S A*, **83**, 2934-8.
- Platzer, M., Rotman, G., Bauer, D., Uziel, T., Savitsky, K., Bar-Shira, A., et al. Ataxia-telangiectasia locus: sequence analysis of 184 kb of human genomic DNA containing the entire ATM gene. (1997) *Genome Res*, **7**, 592-605.
- Rabbitts, T. H. Bacterial cloning of plasmids carrying copies of rabbit globin messenger RNA. (1976) *Nature*, **260**, 221-5.
- Ragoussis, J., and Olavesen, M.G. (1997) *Chromosome walking*. O.U.P.
- Raich, N., Mattei, M. G., Romeo, P. H. and Beaupain, D. PHTF, a novel atypical homeobox gene on chromosome 1p13, is evolutionarily conserved. (1999) *Genomics*, **59**, 108-9.
- Ramashwami, M., Gautam, M., Kamb, A.A., Rudy, B., Tanouye, M.A. and Mathew, M.K. Human potassium channel genes: molecular cloning and functional expression. (1990) *Mol Cell Biol Neurosci*, **1**, 214 - 223.

- Rao, P. H., Houldsworth, J., Dyomina, K., Parsa, N. Z., Cigudosa, J. C., Louie, D. C., et al. Chromosomal and gene amplification in diffuse large B-cell lymphoma. (1998) *Blood*, **92**, 234-40.
- Reymond, A., Meroni, G., Fantozzi, A., Merla, G., Cairo, S., Luzi, L., et al. The tripartite motif family identifies cell compartments. (2001) *Embo J*, **20**, 2140-51.
- Riles, L., Dutchik, J. E., Baktha, A., McCauley, B. K., Thayer, E. C., Leckie, M. P., et al. Physical maps of the six smallest chromosomes of *Saccharomyces cerevisiae* at a resolution of 2.6 kilobase pairs. (1993) *Genetics*, **134**, 81-150.
- Roach, J. C., Boysen, C., Wang, K. and Hood, L. Pairwise end sequencing: a unified approach to genomic mapping and sequencing. (1995) *Genomics*, **26**, 345-53.
- Roest Crolius, H., Jaillon, O., Bernot, A., Dasilva, C., Bouneau, L., Fischer, C., et al. Estimate of human gene number provided by genome-wide analysis using *Tetraodon nigroviridis* DNA sequence. (2000) *Nat Genet*, **25**, 235-8.
- Roses, A. D. Pharmacogenetics and the practice of medicine. (2000) *Nature*, **405**, 857-65.
- Roses, A. D. Genome-based pharmacogenetics and the pharmaceutical industry. (2002) *Nat Rev Drug Discov*, **1**, 541-9.
- Royle, N. J., Clarkson, R. E., Wong, Z. and Jeffreys, A. J. Clustering of hypervariable minisatellites in the proterminal regions of human autosomes. (1988) *Genomics*, **3**, 352-60.
- Sabina, R. L., Fishbein, W. N., Pezeshkpour, G., Clarke, P. R. and Holmes, E. W. Molecular analysis of the myoadenylate deaminase deficiencies. (1992) *Neurology*, **42**, 170-9.
- Saiki, R. K., Gelfand, D. H., Stoffel, S., Scharf, S. J., Higuchi, R., Horn, G. T., et al. Primer-directed enzymatic amplification of DNA with a thermostable DNA polymerase. (1988) *Science*, **239**, 487-91.
- Salvatore, C. A., Jacobson, M. A., Taylor, H. E., Linden, J. and Johnson, R. G. Molecular cloning and characterization of the human A3 adenosine receptor. (1993) *Proc Natl Acad Sci U S A*, **90**, 10365-9.

- Sanger, F., Air, G. M., Barrell, B. G., Brown, N. L., Coulson, A. R., Fiddes, C. A., et al. Nucleotide sequence of bacteriophage phi X174 DNA. (1977) *Nature*, **265**, 687-95.
- Sanger, F., Coulson, A. R., Friedmann, T., Air, G. M., Barrell, B. G., Brown, N. L., et al. The nucleotide sequence of bacteriophage phiX174. (1978) *J Mol Biol*, **125**, 225-46.
- Sanger, F., Coulson, A. R., Hong, G. F., Hill, D. F. and Petersen, G. B. Nucleotide sequence of bacteriophage lambda DNA. (1982) *J Mol Biol*, **162**, 729-73.
- Saupe, S., Roizes, G., Peter, M., Boyle, S., Gardiner, K. and De Sario, A. Molecular cloning of a human cDNA IGSF3 encoding an immunoglobulin-like membrane protein: expression and mapping to chromosome band 1p13. (1998) *Genomics*, **52**, 305-11.
- Scherf, M., Klingenhoff, A. and Werner, T. Highly specific localization of promoter regions in large genomic sequences by PromoterInspector: a novel context analysis approach. (2000) *J Mol Biol*, **297**, 599-606.
- Schuler, G. D. Sequence mapping by electronic PCR. (1997) *Genome Res*, **7**, 541-50.
- Schuler, G. D., Boguski, M. S., Stewart, E. A., Stein, L. D., Gyapay, G., Rice, K., et al. A gene map of the human genome. (1996) *Science*, **274**, 540-6.
- Schutte, B. C., Carpten, J. D., Forus, A., Gregory, S. G., Horii, A. and White, P. S. Report and abstracts of the sixth international workshop on human chromosome 1 mapping 2000. Iowa City, Iowa, USA. 30 September-3 October 2000. (2001) *Cytogenet Cell Genet*, **92**, 23-41.
- Sedlacek, Z., Korn, B., Konecki, D. S., Siebenhaar, R., Coy, J. F., Kioschis, P., et al. Construction of a transcription map of a 300 kb region around the human G6PD locus by direct cDNA selection. (1993) *Hum Mol Genet*, **2**, 1865-9.
- Seed, B., Aruffo A. Molecular cloning of the CD2 antigen, the T-cell erythrocyte receptor, by a rapid immunoselection procedure. (1987) *Proc Natl Acad Sci U S A*, **84**.
- Seidegard, J., Vorachek, W. R., Pero, R. W. and Pearson, W. R. Hereditary differences in the expression of the human glutathione transferase active on trans-stilbene oxide are due to a gene deletion. (1988) *Proc Natl Acad Sci U S A*, **85**, 7293-7.

- Shackleton, S., Lloyd, D. J., Jackson, S. N., Evans, R., Niermeijer, M. F., Singh, B. M., et al. LMNA, encoding lamin A/C, is mutated in partial lipodystrophy. (2000) *Nat Genet*, **24**, 153-6.
- Shen, L. X., Basilion, J. P. and Stanton, V. P., Jr. Single-nucleotide polymorphisms can cause different structural folds of mRNA. (1999) *Proc Natl Acad Sci U S A*, **96**, 7871-6.
- Sherry, S. T., Ward, M. H., Kholodov, M., Baker, J., Phan, L., Smigielski, E. M., et al. dbSNP: the NCBI database of genetic variation. (2001) *Nucleic Acids Res*, **29**, 308-11.
- Shimada, T., Fujii, H. and Lin, H. A 165-base pair sequence between the dihydrofolate reductase gene and the divergently transcribed upstream gene is sufficient for bidirectional transcriptional activity. (1989) *J Biol Chem*, **264**, 20171-4.
- Shizuya, H., Birren, B., Kim, U. J., Mancino, V., Slepak, T., Tachiiri, Y., et al. Cloning and stable maintenance of 300-kilobase-pair fragments of human DNA in *Escherichia coli* using an F-factor-based vector. (1992) *Proc Natl Acad Sci U S A*, **89**, 8794-7.
- Skolnick, J. and Fetrow, J. S. From genes to protein structure and function: novel applications of computational approaches in the genomic era. (2000) *Trends Biotechnol*, **18**, 34-9.
- Slim, R., Le Paslier, D., Compain, S., Levilliers, J., Ougen, P., Billault, A., et al. Construction of a yeast artificial chromosome contig spanning the pseudoautosomal region and isolation of 25 new sequence-tagged sites. (1993) *Genomics*, **16**, 691-7.
- Smit, A. F. Interspersed repeats and other mementos of transposable elements in mammalian genomes. (1999) *Curr Opin Genet Dev*, **9**, 657-63.
- Soderlund, C., Longden, I. and Mott, R. FPC: a system for building contigs from restriction fingerprinted clones. (1997) *Comput Appl Biosci*, **13**, 523-35.
- Solovyev, V. V., Salamov, A. A. and Lawrence, C. B. Predicting internal exons by oligonucleotide composition and discriminant analysis of spliceable open reading frames. (1994) *Nucleic Acids Res*, **22**, 5156-63.
- Solovyev, V. V., Salamov, A. A. and Lawrence, C. B. Identification of human gene structure using linear discriminant functions and dynamic programming. (1995) *Proc Int Conf Intell Syst Mol Biol*, **3**, 367-75.

- Sonnhammer, E. L. and Durbin, R. A dot-matrix program with dynamic threshold control suited for genomic DNA and protein sequence analysis. (1995) *Gene*, **167**, GC1-10.
- Sonnhammer, E. L., von Heijne, G. and Krogh, A. A hidden Markov model for predicting transmembrane helices in protein sequences. (1998) *Proc Int Conf Intell Syst Mol Biol*, **6**, 175-82.
- Soriano, P., Meunier-Rotival, M. and Bernardi, G. The distribution of interspersed repeats is nonuniform and conserved in the mouse and human genomes. (1983) *Proc Natl Acad Sci U S A*, **80**, 1816-20.
- Sparkes, R. S. C., V. H.; Mohandas, T.; Zollman, S.; Cire-Eversole, P.; Amatruda, T. T.; Reed, R. R.; Lochrie, M. A.; Simon, M. I Mapping of genes encoding the subunits of guanine nucleotide-binding protein (G-proteins) in humans. (1987) *Cytogenet Cell Genet*, **46**, 696.
- Staden, R. A new computer method for the storage and manipulation of DNA gel reading data. (1980) *Nucleic Acids Res*, **8**, 3673-94.
- Strittmatter, W. J., Saunders, A. M., Schmechel, D., Pericak-Vance, M., Enghild, J., Salvesen, G. S., et al. Apolipoprotein E: high-avidity binding to beta-amyloid and increased frequency of type 4 allele in late-onset familial Alzheimer disease. (1993) *Proc Natl Acad Sci U S A*, **90**, 1977-81.
- Sudo, K., Chinen, K. and Nakamura, Y. 2058 expressed sequence tags (ESTs) from a human fetal lung cDNA library. (1994) *Genomics*, **24**, 276-9.
- Sunyaev, S., Ramensky, V., Koch, I., Lathe, W., 3rd, Kondrashov, A. S. and Bork, P. Prediction of deleterious human alleles. (2001) *Hum Mol Genet*, **10**, 591-7.
- Taillon-Miller, P., Gu, Z., Li, Q., Hillier, L. and Kwok, P. Y. Overlapping genomic sequences: a treasure trove of single-nucleotide polymorphisms. (1998) *Genome Res*, **8**, 748-54.
- Takahashi, Y., Campbell, E. A., Hirata, Y., Takayama, T. and Listowsky, I. A basis for differentiating among the multiple human Mu-glutathione S-transferases and molecular cloning of brain GSTM5. (1993) *J Biol Chem*, **268**, 8893-8.

- Tang, X., Wang, Y., Li, H. O., Sakatsume, O., Sarai, A. and Yokoyama, K. DNA fingerprinting involving fluorescence-labeled termini of any enzymatically generated fragments of DNA. (1994) *Jpn J Hum Genet*, **39**, 379-91.
- Taniguchi, T., Fujii-Kuriyama, Y. and Muramatsu, M. Molecular cloning of human interferon cDNA. (1980) *Proc Natl Acad Sci U S A*, **77**, 4003-6.
- Tao, Q., Chang, Y-L., Wang, J., Huaming, C., Islam-Faridi, M.N., Scheuring, C. Wang, B., Stelly, D.M. Zhang, H-B. Bacterial Artificial Chromosome-Based Physical Map of the Rice Genome Constructed by Restriction Fingerprint Analysis. (2001) *Genetics*, **158**, 1711-1724.
- Taylor, K., Hornigold, N., Conway, D., Williams, D., Ulinowski, Z., Agochiya, M., et al. Mapping the human Y chromosome by fingerprinting cosmid clones. (1996) *Genome Res*, **6**, 235-48.
- Thierry-Mieg, D. a. A C. elegans DataBase. (1994) *unpublished*.
- Thiery, J. P., Macaya, G. and Bernardi, G. An analysis of eukaryotic genomes by density gradient centrifugation. (1976) *J Mol Biol*, **108**, 219-35.
- Thomas, J. W., Prasad, A. B., Summers, T. J., Lee-Lin, S. Q., Maduro, V. V., Idol, J. R., et al. Parallel construction of orthologous sequence-ready clone contig maps in multiple species. (2002) *Genome Res*, **12**, 1277-85.
- Thompson, J. D., Higgins, D. G. and Gibson, T. J. CLUSTAL W: improving the sensitivity of progressive multiple sequence alignment through sequence weighting, position-specific gap penalties and weight matrix choice. (1994) *Nucleic Acids Res*, **22**, 4673-80.
- Thornton, J. M., Orengo, C. A., Todd, A. E. and Pearl, F. M. Protein folds, functions and evolution. (1999) *J Mol Biol*, **293**, 333-42.
- Tilghman, S. M., Tiemeier, D. C., Seidman, J. G., Peterlin, B. M., Sullivan, M., Maizel, J. V., et al. Intervening sequence of DNA identified in the structural portion of a mouse beta-globin gene. (1978) *Proc Natl Acad Sci U S A*, **75**, 725-9.
- Tkachuk, D. C., Westbrook, C. A., Andreeff, M., Donlon, T. A., Cleary, M. L., Suryanarayan, K., et al. Detection of bcr-abl fusion in chronic myelogeneous leukemia by in situ hybridization. (1990) *Science*, **250**, 559-62.

- Tremblay, L. O., Campbell Dyke, N. and Herscovics, A. Molecular cloning, chromosomal mapping and tissue-specific expression of a novel human alpha 1,2-mannosidase gene involved in N-glycan maturation. (1998) *Glycobiology*, **8**, 585-95.
- Trenkle, T., McClelland, M., Adlkofer, K. and Welsh, J. Major transcript variants of VAV3, a new member of the VAV family of guanine nucleotide exchange factors. (2000) *Gene*, **245**, 139-49.
- Trofatter, J. A., MacCollin, M. M., Rutter, J. L., Murrell, J. R., Duyao, M. P., Parry, D. M., et al. A novel moesin-, ezrin-, radixin-like gene is a candidate for the neurofibromatosis 2 tumor suppressor. (1993) *Cell*, **75**, 826.
- Tsukino, H., Kuroda, Y., Qiu, D., Nakao, H., Imai, H. and Katoh, T. Effects of cytochrome P450 (CYP) 2A6 gene deletion and CYP2E1 genotypes on gastric adenocarcinoma. (2002) *Int J Cancer*, **100**, 425-8.
- Uberbacher, E. C., Xu, Y. and Mural, R. J. Discovering and understanding genes in human DNA sequence using GRAIL. (1996) *Methods Enzymol*, **266**, 259-81.
- Van den Bergh, F., Sabina RL. Characterization of human AMP deaminase 2 (AMPD2) gene expression reveals alternative transcripts encoding variable N-terminal extensions of isoform L. (1995) *Biochem J*, **312**, 401-410.
- Vega-Saenz de Miera, E., Moreno H, Fruhling D, Kentros C, Rudy B. Cloning of ShIII (Shaw-like) cDNAs encoding a novel high-voltage-activating, TEA-sensitive, type-A K⁺ channel. (1992) *Proc R Soc Lond B Biol Sci*, **248**, 9-18.
- Venter, J. C., Adams, M. D., Myers, E. W., Li, P. W., Mural, R. J., Sutton, G. G., et al. The sequence of the human genome. (2001) *Science*, **291**, 1304-51.
- Verma, I. M., Temple, G. F., Fan, H. and Baltimore, D. In vitro synthesis of DNA complementary to rabbit reticulocyte 10S RNA. (1972) *Nat New Biol*, **235**, 163-7.
- Vetrie, D. Isolation of the defective gene in X linked agammaglobulinaemia. (1993) *J Med Genet*, **30**, 452-3.

- Vorachek, W. R., Pearson, W. R. and Rule, G. S. Cloning, expression, and characterization of a class-mu glutathione transferase from human muscle, the product of the GST4 locus. (1991) *Proc Natl Acad Sci U S A*, **88**, 4443-7.
- Walker, A. P., Muscatelli, F. and Monaco, A. P. Isolation of the human Xp21 glycerol kinase gene by positional cloning. (1993) *Hum Mol Genet*, **2**, 107-14.
- Wallace, M. R., Marchuk, D. A., Andersen, L. B., Letcher, R., Odeh, H. M., Saulino, A. M., et al. Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients. (1990) *Science*, **249**, 181-6.
- Wallner, B. P., Frey, A. Z., Tizard, R., Mattaliano, R. J., Hession, C., Sanders, M. E., et al. Primary structure of lymphocyte function-associated antigen 3 (LFA-3). The ligand of the T lymphocyte CD2 glycoprotein. (1987) *J Exp Med*, **166**, 923-32.
- Wang, D. G., Fan, J. B., Siao, C. J., Berno, A., Young, P., Sapolsky, R., et al. Large-scale identification, mapping, and genotyping of single-nucleotide polymorphisms in the human genome. (1998) *Science*, **280**, 1077-82.
- Wang, M., Duell, T., Gray, J. W. and Weier, H.-U. G. High sensitivity, high resolution physical mapping by fluorescence *in situ* hybridisation on to individually straightened DNA molecules. (1996) *Bioimaging*, **4**, 73-83.
- Wang, X., Zuckerman, B., Pearson, C., Kaufman, G., Chen, C., Wang, G., et al. Maternal cigarette smoking, metabolic gene polymorphism, and infant birth weight. (2002) *Jama*, **287**, 195-202.
- Wang, Z. and Moulton, J. SNPs, protein structure, and disease. (2001) *Hum Mutat*, **17**, 263-70.
- Waterston, R. H., Lander, E. S. and Sulston, J. E. On the sequencing of the human genome. (2002) *Proc Natl Acad Sci U S A*, **99**, 3712-6.
- Watson, J. D. a. C., F. A Structure for Deoxyribose Nucleic Acid. (1953) *Nature*, **171**, 171.
- Weber, J. L. and May, P. E. Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction. (1989) *Am J Hum Genet*, **44**, 388-96.
- Weissenbach, J., Gyapay, G., Dib, C., Vignal, A., Morissette, J., Millasseau, P., et al. A second-generation linkage map of the human genome. (1992) *Nature*, **359**, 794-801.

- White, P. S., Forus, A., Matise, T. C., Schutte, B. C., Spieker, N., Stanier, P., et al. Report of the fifth international workshop on human chromosome 1 mapping 1999. (1999) *Cytogenet Cell Genet*, **87**, 143-71.
- Wickens, M. P., Buell, G. N. and Schimke, R. T. Synthesis of double-stranded DNA complementary to lysozyme, ovomucoid, and ovalbumin mRNAs. Optimization for full length second strand synthesis by *Escherichia coli* DNA polymerase I. (1978) *J Biol Chem*, **253**, 2483-95.
- Wilke, C. M., Guo, S. W., Hall, B. K., Boldog, F., Gemmill, R. M., Chandrasekharappa, S. C., et al. Multicolor FISH mapping of YAC clones in 3p14 and identification of a YAC spanning both FRA3B and the t(3;8) associated with hereditary renal cell carcinoma. (1994) *Genomics*, **22**, 319-26.
- Wyman, A. R. and White, R. A highly polymorphic locus in human DNA. (1980) *Proc Natl Acad Sci U S A*, **77**, 6754-8.
- Xiang, Z., Morse, E., Hu, X. L., Flint, J., Chi, H. C., Grady, D. L., et al. A sequence-ready map of the human chromosome 1q telomere. (2001) *Genomics*, **72**, 105-7.
- Xie, Y. G., Han, F. Y., Peyrard, M., Rutledge, M. H., Fransson, I., DeJong, P., et al. Cloning of a novel, anonymous gene from a megabase-range YAC and cosmid contig in the neurofibromatosis type 2/meningioma region on human chromosome 22q12. (1993) *Hum Mol Genet*, **2**, 1361-8.
- Xu, C. F., Chambers, J. A. and Solomon, E. Complex regulation of the BRCA1 gene. (1997) *J Biol Chem*, **272**, 20994-7.
- Xu, S., Wang, Y., Roe, B. and Pearson, W. R. Characterization of the human class Mu glutathione S-transferase gene cluster and the GSTM1 deletion. (1998) *J Biol Chem*, **273**, 3517-27.
- Zachmann, M. and Prader, A. Unusual heterozygotes of congenital adrenal hyperplasia due to 21-hydroxylase deficiency confirmed by HLA tissue typing. (1979) *Acta Endocrinol (Copenh)*, **92**, 542-6.

Zattara-Cannoni, H., Roll, P., Figarella-Branger, D., Lena, G., Dufour, H., Grisoli, F., et al.

Cytogenetic study of six cases of radiation-induced meningiomas. (2001) *Cancer Genet*

Cytogenet, **126**, 81-4.

Zoubak, S., Clay, O. and Bernardi, G. The gene distribution of the human genome. (1996) *Gene*, **174**,

95-102.