

Chapter VIII - References

- Abbasi, A.A., Paparidis, Z., Malik, S., Goode, D.K., Callaway, H., Elgar, G. and Grzeschik, K.H. (2007) Human GLI3 Infragenic Conserved Non-Coding Sequences Are Tissue-Specific Enhancers. *PLoS ONE*, **2**, e366.
- Adams, D.J., Dermitzakis, E.T., Cox, T., Smith, J., Davies, R., Banerjee, R., Bonfield, J., Mullikin, J.C., Chung, Y.J., Rogers, J., et al (2005) Complex haplotypes, copy number polymorphisms and coding variation in two recently divergent mouse strains. *Nat. Genet.*, **37**, 532-536.
- Ager, E., Suzuki, S., Pask, A., Shaw, G., Ishino, F. and Renfree, M.B. (2007) Insulin is imprinted in the placenta of the marsupial, *Macropus eugenii*. *Dev. Biol.*, **309**, 317-328.
- Allen, E., Horvath, S., Tong, F., Kraft, P., Spiteri, E., Riggs, A.D. and Marahrens, Y. (2003) High concentrations of long interspersed nuclear element sequence distinguish monoallelically expressed genes. *Proc. Natl. Acad. Sci. U. S. A.*, **100**, 9940-9945.
- Aloni, R. and Lancet, D. (2005) Conservation anchors in the vertebrate genome. *Genome Biol.*, **6**, 115.
- Altschul, S.F., Gish, W., Miller, W., Myers, E.W. and Lipman, D.J. (1990) Basic local alignment search tool. *J. Mol. Biol.*, **215**, 403-410.
- Ambatipudi, K., Joss, J., Raftery, M. and Deane, E. (2007) A proteomic approach to analysis of antimicrobial activity in marsupial pouch secretions. *Dev. Comp. Immunol.*,
- Anderson, R.J. and Spencer, H.G. (1999) Population models of genomic imprinting. I. Differential viability in the sexes and the analogy with genetic dominance. *Genetics*, **153**, 1949-1958.
- Arney, K.L. (2003) H19 and Igf2--enhancing the confusion? *Trends Genet.*, **19**, 17-23.
- Ashurst, J.L., Chen, C.K., Gilbert, J.G., Jekosch, K., Keenan, S., Meidl, P., Searle, S.M., Stalker, J., Storey, R., Trevanion, S., et al (2005) The Vertebrate Genome Annotation (Vega) database. *Nucleic Acids Res.*, **33**, D459-65.

- Bailey, J.A., Baertsch, R., Kent, W.J., Haussler, D. and Eichler, E.E. (2004) Hotspots of mammalian chromosomal evolution. *Genome Biol.*, **5**, R23.
- Banerji, J., Olson, L. and Schaffner, W. (1983) A lymphocyte-specific cellular enhancer is located downstream of the joining region in immunoglobulin heavy chain genes. *Cell*, **33**, 729-740.
- Banerji, J., Rusconi, S. and Schaffner, W. (1981) Expression of a beta-globin gene is enhanced by remote SV40 DNA sequences. *Cell*, **27**, 299-308.
- Barlow, D.P. (1995) Gametic imprinting in mammals. *Science*, **270**, 1610-1613.
- Barlow, D.P. (1993) Methylation and imprinting: from host defense to gene regulation? *Science*, **260**, 309-310.
- Barlow, D.P., Stoger, R., Herrmann, B.G., Saito, K. and Schweifer, N. (1991) The mouse insulin-like growth factor type-2 receptor is imprinted and closely linked to the Tme locus. *Nature*, **349**, 84-87.
- Bartolomei, M.S., Zemel, S. and Tilghman, S.M. (1991) Parental imprinting of the mouse H19 gene. *Nature*, **351**, 153-155.
- Barton, S.C., Surani, M.A. and Norris, M.L. (1984) Role of paternal and maternal genomes in mouse development. *Nature*, **311**, 374-376.
- Bateman, A., Coin, L., Durbin, R., Finn, R.D., Hollich, V., Griffiths-Jones, S., Khanna, A., Marshall, M., Moxon, S., Sonnhammer, E.L., et al (2004) The Pfam protein families database. *Nucleic Acids Res.*, **32**, D138-41.
- Batzoglou, S. (2005) The many faces of sequence alignment. *Brief Bioinform*, **6**, 6-22.
- Beaudet, A.L. and Jiang, Y.H. (2002) A rheostat model for a rapid and reversible form of imprinting-dependent evolution. *Am. J. Hum. Genet.*, **70**, 1389-1397.
- Beck, S., Olek, A. and Walter, J. (1999) From genomics to epigenomics: a loftier view of life. *Nat. Biotechnol.*, **17**, 1144.
- Bejerano, G., Lowe, C.B., Ahituv, N., King, B., Siepel, A., Salama, S.R., Rubin, E.M., Kent, W.J. and Haussler, D. (2006) A distal enhancer and an ultraconserved exon are derived from a novel retroposon. *Nature*, **441**, 87-90.
- Bejerano, G., Pheasant, M., Makunin, I., Stephen, S., Kent, W.J., Mattick, J.S. and Haussler, D. (2004) Ultraconserved elements in the human genome. *Science*, **304**, 1321-1325.

- Bell, A.C. and Felsenfeld, G. (2000) Methylation of a CTCF-dependent boundary controls imprinted expression of the Igf2 gene. *Nature*, **405**, 482-485.
- Bell, A.C., West, A.G. and Felsenfeld, G. (2001) Insulators and boundaries: versatile regulatory elements in the eukaryotic genome. *Science*, **291**, 447-450.
- Bell, A.C., West, A.G. and Felsenfeld, G. (1999) The protein CTCF is required for the enhancer blocking activity of vertebrate insulators. *Cell*, **98**, 387-396.
- Benson, G. (1999) Tandem repeats finder: a program to analyze DNA sequences. *Nucleic Acids Res.*, **27**, 573-580.
- Bentley, D.R. (2006) Whole-genome re-sequencing. *Curr. Opin. Genet. Dev.*, **16**, 545-552.
- Bentley, D.R. (1996) Genomic sequence information should be released immediately and freely in the public domain. *Science*, **274**, 533-534.
- Bentley, D.R., Deloukas, P., Dunham, A., French, L., Gregory, S.G., Humphray, S.J., Mungall, A.J., Ross, M.T., Carter, N.P., Dunham, I., et al (2001) The physical maps for sequencing human chromosomes 1, 6, 9, 10, 13, 20 and X. *Nature*, **409**, 942-943.
- Berg, P. (2006) Origins of the human genome project: why sequence the human genome when 96% of it is junk? *Am. J. Hum. Genet.*, **79**, 603-605.
- Bernardi, G. (2000) Isochores and the evolutionary genomics of vertebrates. *Gene*, **241**, 3-17.
- Bernardi, G. (1995) The human genome: organization and evolutionary history. *Annu. Rev. Genet.*, **29**, 445-476.
- Bernardi, G., Mouchiroud, D., Gautier, C. and Bernardi, G. (1988) Compositional patterns in vertebrate genomes: conservation and change in evolution. *J. Mol. Evol.*, **28**, 7-18.
- Bernstein, B.E., Meissner, A. and Lander, E.S. (2007) The mammalian epigenome. *Cell*, **128**, 669-681.
- Bertone, P., Stolc, V., Royce, T.E., Rozowsky, J.S., Urban, A.E., Zhu, X., Rinn, J.L., Tongprasit, W., Samanta, M., Weissman, S., et al (2004) Global identification of human transcribed sequences with genome tiling arrays. *Science*, **306**, 2242-2246.

- Bininda-Emonds, O.R., Cardillo, M., Jones, K.E., MacPhee, R.D., Beck, R.M., Grenyer, R., Price, S.A., Vos, R.A., Gittleman, J.L. and Purvis, A. (2007) The delayed rise of present-day mammals. *Nature*, **446**, 507-512.
- Bird, A.P. (1986) CpG-rich islands and the function of DNA methylation. *Nature*, **321**, 209-213.
- Birney, E., Clamp, M. and Durbin, R. (2004) GeneWise and Genomewise. *Genome Res.*, **14**, 988-995.
- Blakesley, R.W., Hansen, N.F., Mullikin, J.C., Thomas, P.J., McDowell, J.C., Maskeri, B., Young, A.C., Benjamin, B., Brooks, S.Y., Coleman, B.I., et al (2004) An intermediate grade of finished genomic sequence suitable for comparative analyses. *Genome Res.*, **14**, 2235-2244.
- Blanchette, M., Kent, W.J., Riemer, C., Elnitski, L., Smit, A.F., Roskin, K.M., Baertsch, R., Rosenbloom, K., Clawson, H., Green, E.D., et al (2004) Aligning multiple genomic sequences with the threaded blockset aligner. *Genome Res.*, **14**, 708-715.
- Boffelli, D., McAuliffe, J., Ovcharenko, D., Lewis, K.D., Ovcharenko, I., Pachter, L. and Rubin, E.M. (2003) Phylogenetic shadowing of primate sequences to find functional regions of the human genome. *Science*, **299**, 1391-1394.
- Bonfield, J.K., Smith, K. and Staden, R. (1995) A new DNA sequence assembly program. *Nucleic Acids Res.*, **23**, 4992-4999.
- Bouma, M.E., Rogier, E., Verthier, N., Labarre, C. and Feldmann, G. (1989) Further cellular investigation of the human hepatoblastoma-derived cell line HepG2: morphology and immunocytochemical studies of hepatic-secreted proteins. *In Vitro Cell. Dev. Biol.*, **25**, 267-275.
- Bourque, G., Pevzner, P.A. and Tesler, G. (2004) Reconstructing the genomic architecture of ancestral mammals: lessons from human, mouse, and rat genomes. *Genome Res.*, **14**, 507-516.
- Brannan, C.I., Dees, E.C., Ingram, R.S. and Tilghman, S.M. (1990) The product of the H19 gene may function as an RNA. *Mol. Cell. Biol.*, **10**, 28-36.
- Bray, N. and Pachter, L. (2004) MAVID: constrained ancestral alignment of multiple sequences. *Genome Res.*, **14**, 693-699.
- Brenner, S., Johnson, M., Bridgham, J., Golda, G., Lloyd, D.H., Johnson, D., Luo, S., McCurdy, S., Foy, M., Ewan, M., et al (2000) Gene expression analysis by

massively parallel signature sequencing (MPSS) on microbead arrays. *Nat. Biotechnol.*, **18**, 630-634.

Bridgham, J.T. and Johnson, A.L. (2004) Alternatively spliced variants of Gallus gallus TNFRSF23 are expressed in the ovary and differentially regulated by cell signaling pathways. *Biol. Reprod.*, **70**, 972-979.

Brudno, M., Do, C.B., Cooper, G.M., Kim, M.F., Davydov, E., NISC Comparative Sequencing Program, Green, E.D., Sidow, A. and Batzoglou, S. (2003) LAGAN and Multi-LAGAN: efficient tools for large-scale multiple alignment of genomic DNA. *Genome Res.*, **13**, 721-731.

Brudno, M., Poliakov, A., Minovitsky, S., Ratnere, I. and Dubchak, I. (2007) Multiple whole genome alignments and novel biomedical applications at the VISTA portal. *Nucleic Acids Res.*, **35**, W669-74.

Bruford, E.A., Lush, M.J., Wright, M.W., Sneddon, T.P., Povey, S. and Birney, E. (2007) The HGNC Database in 2008: a resource for the human genome. *Nucleic Acids Res.*,

Burge, C. and Karlin, S. (1997) Prediction of complete gene structures in human genomic DNA. *J. Mol. Biol.*, **268**, 78-94.

Burgio, G., Szatanik, M., Guenet, J.L., Arnau, M.R., Panthier, J.J. and Montagutelli, X. (2007) Interspecific recombinant congenic strains between C57BL/6 and mice of the Mus spreitus species : a powerful tool to dissect genetic control of complex traits. *Genetics*,

Burt, A. and Trivers, R. (1998) Genetic conflicts in genomic imprinting. *Proc. Biol. Sci.*, **265**, 2393-2397.

Cai, X. and Cullen, B.R. (2007) The imprinted H19 noncoding RNA is a primary microRNA precursor. *RNA*, **13**, 313-316.

Carninci, P. (2006) Tagging mammalian transcription complexity. *Trends Genet.*, **22**, 501-510.

Cartharius, K., Frech, K., Grote, K., Klocke, B., Haltmeier, M., Klingenhoff, A., Frisch, M., Bayerlein, M. and Werner, T. (2005) MatInspector and beyond: promoter analysis based on transcription factor binding sites. *Bioinformatics*, **21**, 2933-2942.

Cattanach, B.M. and Kirk, M. (1985) Differential activity of maternally and paternally derived chromosome regions in mice. *Nature*, **315**, 496-498.

- Cavaille, J., Seitz, H., Paulsen, M., Ferguson-Smith, A.C. and Bachellerie, J.P. (2002) Identification of tandemly-repeated C/D snoRNA genes at the imprinted human 14q32 domain reminiscent of those at the Prader-Willi/Angelman syndrome region. *Hum. Mol. Genet.*, **11**, 1527-1538.
- Celniker, S.E. and Drewell, R.A. (2007) Chromatin looping mediates boundary element promoter interactions. *Bioessays*, **29**, 7-10.
- Chapman, M.A., Charchar, F.J., Kinston, S., Bird, C.P., Grafham, D., Rogers, J., Grutzner, F., Graves, J.A., Green, A.R. and Gottgens, B. (2003) Comparative and functional analyses of LYL1 loci establish marsupial sequences as a model for phylogenetic footprinting. *Genomics*, **81**, 249-259.
- Cheng, J., Kapranov, P., Drenkow, J., Dike, S., Brubaker, S., Patel, S., Long, J., Stern, D., Tammana, H., Helt, G., et al (2005) Transcriptional maps of 10 human chromosomes at 5-nucleotide resolution. *Science*, **308**, 1149-1154.
- Chimpanzee Sequencing and Analysis Consortium (2005) Initial sequence of the chimpanzee genome and comparison with the human genome. *Nature*, **437**, 69-87.
- Choi, S.H., Kim, I.C., Kim, D.S., Kim, D.W., Chae, S.H., Choi, H.H., Choi, I., Yeo, J.S., Song, M.N. and Park, H.S. (2006) Comparative genomic organization of the human and bovine PRNP locus. *Genomics*, **87**, 598-607.
- Chow, J.C., Yen, Z., Ziesche, S.M. and Brown, C.J. (2005) Silencing of the mammalian X chromosome. *Annu. Rev. Genomics Hum. Genet.*, **6**, 69-92.
- Chung, J.H., Bell, A.C. and Felsenfeld, G. (1997) Characterization of the chicken beta-globin insulator. *Proc. Natl. Acad. Sci. U. S. A.*, **94**, 575-580.
- Clamp, M., Cuff, J., Searle, S.M. and Barton, G.J. (2004) The Jalview Java alignment editor. *Bioinformatics*, **20**, 426-427.
- Clark, L., Wei, M., Cattoretti, G., Mendelsohn, C. and Tycko, B. (2002) The Tnfrh1 (Tnfrsf23) gene is weakly imprinted in several organs and expressed at the trophoblast-decidua interface. *BMC Genet.*, **3**, 11.
- Collins, J.E., Wright, C.L., Edwards, C.A., Davis, M.P., Grinham, J.A., Cole, C.G., Goward, M.E., Aguado, B., Mallya, M., Mokrab, Y., et al (2004) A genome annotation-driven approach to cloning the human ORFeome. *Genome Biol.*, **5**, R84.
- Constancia, M., Kelsey, G. and Reik, W. (2004) Resourceful imprinting. *Nature*, **432**, 53-57.

- Cooper, D.W., VandeBerg, J.L., Sharman, G.B. and Poole, W.E. (1971) Phosphoglycerate kinase polymorphism in kangaroos provides further evidence for paternal X inactivation. *Nat. New Biol.*, **230**, 155-157.
- Cooper, G.M. and Sidow, A. (2003) Genomic regulatory regions: insights from comparative sequence analysis. *Curr. Opin. Genet. Dev.*, **13**, 604-610.
- Cooper, G.M., Stone, E.A., Asimenos, G., NISC Comparative Sequencing Program, Green, E.D., Batzoglou, S. and Sidow, A. (2005) Distribution and intensity of constraint in mammalian genomic sequence. *Genome Res.*, **15**, 901-913.
- Coulondre, C., Miller, J.H., Farabaugh, P.J. and Gilbert, W. (1978) Molecular basis of base substitution hotspots in Escherichia coli. *Nature*, **274**, 775-780.
- Crawford, G.E., Davis, S., Scacheri, P.C., Renaud, G., Halawi, M.J., Erdos, M.R., Green, R., Meltzer, P.S., Wolfsberg, T.G. and Collins, F.S. (2006) DNase-chip: a high-resolution method to identify DNase I hypersensitive sites using tiled microarrays. *Nat. Methods*, **3**, 503-509.
- Cullen, B.R. (2004) Transcription and processing of human microRNA precursors. *Mol. Cell*, **16**, 861-865.
- Dallosso, A.R., Hancock, A.L., Brown, K.W., Williams, A.C., Jackson, S. and Malik, K. (2004) Genomic imprinting at the WT1 gene involves a novel coding transcript (AWT1) that shows deregulation in Wilms' tumours. *Hum. Mol. Genet.*, **13**, 405-415.
- Dannenberg, L.O. and Edenberg, H.J. (2006) Epigenetics of gene expression in human hepatoma cells: expression profiling the response to inhibition of DNA methylation and histone deacetylation. *BMC Genomics*, **7**, 181.
- Darling, A.C., Mau, B., Blattner, F.R. and Perna, N.T. (2004) Mauve: multiple alignment of conserved genomic sequence with rearrangements. *Genome Res.*, **14**, 1394-1403.
- Day, T. and Bonduriansky, R. (2004) Intralocus sexual conflict can drive the evolution of genomic imprinting. *Genetics*, **167**, 1537-1546.
- Deakin, J.E., Siddle, H.V., Cross, J.G., Belov, K. and Graves, J.A. (2007) Class I genes have split from the MHC in the tammar wallaby. *Cytogenet. Genome Res.*, **116**, 205-211.
- Dean, W., Bowden, L., Aitchison, A., Klose, J., Moore, T., Meneses, J.J., Reik, W. and Feil, R. (1998) Altered imprinted gene methylation and expression in completely

ES cell-derived mouse fetuses: association with aberrant phenotypes. *Development*, **125**, 2273-2282.

DeChiara, T.M., Robertson, E.J. and Efstratiadis, A. (1991) Parental imprinting of the mouse insulin-like growth factor II gene. *Cell*, **64**, 849-859.

Deininger, P.L. and Batzer, M.A. (1999) Alu repeats and human disease. *Mol. Genet. Metab.*, **67**, 183-193.

Dekker, J., Rippe, K., Dekker, M. and Kleckner, N. (2002) Capturing chromosome conformation. *Science*, **295**, 1306-1311.

Delbridge, M.L., Lingenfelter, P.A., Disteche, C.M. and Graves, J.A. (1999) The candidate spermatogenesis gene RBMY has a homologue on the human X chromosome. *Nat. Genet.*, **22**, 223-224.

Dewey, C.N. and Pachter, L. (2006) Evolution at the nucleotide level: the problem of multiple whole-genome alignment. *Hum. Mol. Genet.*, **15 Spec No 1**, R51-6.

Dhami, P., Coffey, A.J., Abbs, S., Vermeesch, J.R., Dumanski, J.P., Woodward, K.J., Andrews, R.M., Langford, C. and Vetrici, D. (2005) Exon array CGH: detection of copy-number changes at the resolution of individual exons in the human genome. *Am. J. Hum. Genet.*, **76**, 750-762.

Down, T.A. and Hubbard, T.J. (2005) NestedMICA: sensitive inference of over-represented motifs in nucleic acid sequence. *Nucleic Acids Res.*, **33**, 1445-1453.

Down, T.A. and Hubbard, T.J. (2002) Computational detection and location of transcription start sites in mammalian genomic DNA. *Genome Res.*, **12**, 458-461.

Drewell, R.A., Arney, K.L., Arima, T., Barton, S.C., Brenton, J.D. and Surani, M.A. (2002) Novel conserved elements upstream of the H19 gene are transcribed and act as mesodermal enhancers. *Development*, **129**, 1205-1213.

Drewell, R.A., Brenton, J.D., Ainscough, J.F., Barton, S.C., Hilton, K.J., Arney, K.L., Dandolo, L. and Surani, M.A. (2000) Deletion of a silencer element disrupts H19 imprinting independently of a DNA methylation epigenetic switch. *Development*, **127**, 3419-3428.

Duncan, B.K. and Miller, J.H. (1980) Mutagenic deamination of cytosine residues in DNA. *Nature*, **287**, 560-561.

- Dunham, I., Shimizu, N., Roe, B.A., Chissoe, S., Hunt, A.R., Collins, J.E., Bruskiewich, R., Beare, D.M., Clamp, M., Smink, L.J., et al (1999) The DNA sequence of human chromosome 22. *Nature*, **402**, 489-495.
- Dunzinger, U., Nanda, I., Schmid, M., Haaf, T. and Zechner, U. (2005) Chicken orthologues of mammalian imprinted genes are clustered on macrochromosomes and replicate asynchronously. *Trends Genet.*, **21**, 488-492.
- Duret, L., Chureau, C., Samain, S., Weissenbach, J. and Avner, P. (2006) The Xist RNA gene evolved in eutherians by pseudogenization of a protein-coding gene. *Science*, **312**, 1653-1655.
- Easton, D.F., Pooley, K.A., Dunning, A.M., Pharoah, P.D., Thompson, D., Ballinger, D.G., Struewing, J.P., Morrison, J., Field, H., Luben, R., et al (2007) Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **447**, 1087-1093.
- Eddy, S.R. (2005) A model of the statistical power of comparative genome sequence analysis. *PLoS Biol.*, **3**, e10.
- Edwards, C.A. and Ferguson-Smith, A.C. (2007) Mechanisms regulating imprinted genes in clusters. *Curr. Opin. Cell Biol.*, **19**, 281-289.
- Edwards, C.A., Rens, W., Clark, O., Mungall, A.J., Hore, T., Marshall Graves, J.A., Dunham, I., Ferguson-Smith, A.C. and Ferguson-Smith, M.A. (2007) The evolution of imprinting: chromosomal mapping of orthologues of mammalian imprinted domains in monotreme and marsupial mammals. *BMC Evol. Biol.*, **7**, 157.
- Elgar, G. (2006) Different words, same meaning: understanding the languages of the genome. *Trends Genet.*, **22**, 639-641.
- ENCODE Project Consortium. (2004) The ENCODE (ENCyclopedia Of DNA Elements) Project. *Science*, **306**, 636-640.
- ENCODE Project Consortium, Birney, E., Stamatoyannopoulos, J.A., Dutta, A., Guigo, R., Gingeras, T.R., Margulies, E.H., Weng, Z., Snyder, M., Dermitzakis, E.T., et al (2007) Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature*, **447**, 799-816.
- Engemann, S., Strodicke, M., Paulsen, M., Franck, O., Reinhardt, R., Lane, N., Reik, W. and Walter, J. (2000) Sequence and functional comparison in the Beckwith-Wiedemann region: implications for a novel imprinting centre and extended imprinting. *Hum. Mol. Genet.*, **9**, 2691-2706.

- Estivill, X., Cheung, J., Pujana, M.A., Nakabayashi, K., Scherer, S.W. and Tsui, L.C. (2002) Chromosomal regions containing high-density and ambiguously mapped putative single nucleotide polymorphisms (SNPs) correlate with segmental duplications in the human genome. *Hum. Mol. Genet.*, **11**, 1987-1995.
- Feinberg, A.P. and Vogelstein, B. (1983) A technique for radiolabeling DNA restriction endonuclease fragments to high specific activity. *Anal. Biochem.*, **132**, 6-13.
- Fichant, G.A. and Burks, C. (1991) Identifying potential tRNA genes in genomic DNA sequences. *J. Mol. Biol.*, **220**, 659-671.
- Finn, R.D., Mistry, J., Schuster-Bockler, B., Griffiths-Jones, S., Hollich, V., Lassmann, T., Moxon, S., Marshall, M., Khanna, A., Durbin, R., et al (2006) Pfam: clans, web tools and services. *Nucleic Acids Res.*, **34**, D247-51.
- Fisher, S., Grice, E.A., Vinton, R.M., Bessling, S.L. and McCallion, A.S. (2006) Conservation of RET regulatory function from human to zebrafish without sequence similarity. *Science*, **312**, 276-279.
- Fitzpatrick, G.V., Pugacheva, E.M., Shin, J.Y., Abdullaev, Z., Yang, Y., Khatod, K., Lobanenkov, V.V. and Higgins, M.J. (2007) Allele-specific binding of CTCF to the multipartite imprinting control region KvDMR1. *Mol. Cell. Biol.*, **27**, 2636-2647.
- Fitzpatrick, G.V., Soloway, P.D. and Higgins, M.J. (2002) Regional loss of imprinting and growth deficiency in mice with a targeted deletion of KvDMR1. *Nat. Genet.*, **32**, 426-431.
- Frazer, K.A., Elnitski, L., Church, D.M., Dubchak, I. and Hardison, R.C. (2003) Cross-species sequence comparisons: a review of methods and available resources. *Genome Res.*, **13**, 1-12.
- Frazer, K.A., Pachter, L., Poliakov, A., Rubin, E.M. and Dubchak, I. (2004) VISTA: computational tools for comparative genomics. *Nucleic Acids Res.*, **32**, W273-9.
- Fredlake, C.P., Hert, D.G., Mardis, E.R. and Barron, A.E. (2006) What is the future of electrophoresis in large-scale genomic sequencing? *Electrophoresis*, **27**, 3689-3702.
- Gardiner-Garden, M. and Frommer, M. (1987) CpG islands in vertebrate genomes. *J. Mol. Biol.*, **196**, 261-282.
- Gaszner, M. and Felsenfeld, G. (2006) Insulators: Exploiting transcriptional and epigenetic mechanisms. *Nat. Rev. Gen.*, **7**, 703-713.

- Gerstein, M.B., Bruce, C., Rozowsky, J.S., Zheng, D., Du, J., Korbel, J.O., Emanuelsson, O., Zhang, Z.D., Weissman, S. and Snyder, M. (2007) What is a gene, post-ENCODE? History and updated definition. *Genome Res.*, **17**, 669-681.
- Gibbs, R.A., Weinstock, G.M., Metzker, M.L., Muzny, D.M., Sodergren, E.J., Scherer, S., Scott, G., Steffen, D., Worley, K.C., Burch, P.E., et al (2004) Genome sequence of the Brown Norway rat yields insights into mammalian evolution. *Nature*, **428**, 493-521.
- Gilbert, N. and Labuda, D. (2000) Evolutionary inventions and continuity of CORE-SINEs in mammals. *J. Mol. Biol.*, **298**, 365-377.
- Gilbert, N. and Labuda, D. (1999) CORE-SINEs: eukaryotic short interspersed retropositing elements with common sequence motifs. *Proc. Natl. Acad. Sci. U. S. A.*, **96**, 2869-2874.
- Giresi, P.G., Kim, J., McDaniell, R.M., Iyer, V.R. and Lieb, J.D. (2007) FAIRE (Formaldehyde-Assisted Isolation of Regulatory Elements) isolates active regulatory elements from human chromatin. *Genome Res.*, **17**, 877-885.
- Glass, J.L., Thompson, R.F., Khulan, B., Figueroa, M.E., Olivier, E.N., Oakley, E.J., Van Zant, G., Bouhassira, E.E., Melnick, A., Golden, A., et al (2007) CG dinucleotide clustering is a species-specific property of the genome. *Nucleic Acids Res.*,
- Godbout, R., Ingram, R.S. and Tilghman, S.M. (1988) Fine-structure mapping of the three mouse alpha-fetoprotein gene enhancers. *Mol. Cell. Biol.*, **8**, 1169-1178.
- Gottgens, B., Barton, L.M., Gilbert, J.G., Bench, A.J., Sanchez, M.J., Bahn, S., Mistry, S., Grafham, D., McMurray, A., Vaudin, M., et al (2000) Analysis of vertebrate SCL loci identifies conserved enhancers. *Nat. Biotechnol.*, **18**, 181-186.
- Gould, S.J. and Vrba, E.S. (1982) Exaptation; a missing term in the science of form. *Paleobiology*, **8**, 4-15.
- Graves, J.A. and Westerman, M. (2002) Marsupial genetics and genomics. *Trends Genet.*, **18**, 517-521.
- Greally, J.M. (2002) Short interspersed transposable elements (SINEs) are excluded from imprinted regions in the human genome. *Proc. Natl. Acad. Sci. U. S. A.*, **99**, 327-332.
- Green, P. (2007) 2x Genomes--does Depth Matter? *Genome Res.*, **17**, 1547-1549.

- Gregory, S.G., Sekhon, M., Schein, J., Zhao, S., Osoegawa, K., Scott, C.E., Evans, R.S., Burridge, P.W., Cox, T.V., Fox, C.A., et al (2002) A physical map of the mouse genome. *Nature*, **418**, 743-750.
- Griffiths-Jones, S., Grocock, R.J., van Dongen, S., Bateman, A. and Enright, A.J. (2006) miRBase: microRNA sequences, targets and gene nomenclature. *Nucleic Acids Res.*, **34**, D140-4.
- Grimson, A., Farh, K.K., Johnston, W.K., Garrett-Engele, P., Lim, L.P. and Bartel, D.P. (2007) MicroRNA targeting specificity in mammals: determinants beyond seed pairing. *Mol. Cell*, **27**, 91-105.
- Grutzner, F., Rens, W., Tsendl-Ayush, E., El-Mogharbel, N., O'Brien, P.C., Jones, R.C., Ferguson-Smith, M.A. and Marshall Graves, J.A. (2004) In the platypus a meiotic chain of ten sex chromosomes shares genes with the bird Z and mammal X chromosomes. *Nature*, **432**, 913-917.
- Guenet, J.L. and Bonhomme, F. (2003) Wild mice: an ever-increasing contribution to a popular mammalian model. *Trends Genet.*, **19**, 24-31.
- Guigo, R., Flieck, P., Abril, J.F., Reymond, A., Lagarde, J., Denoeud, F., Antonarakis, S., Ashburner, M., Bajic, V.B., Birney, E., et al (2006) EGASP: the human ENCODE Genome Annotation Assessment Project. *Genome Biol.*, **7 Suppl 1**, S2.1-31.
- Haig, D. and Westoby, M. (1989) Parent-Specific Gene Expression and the Triploid Endosperm. *Am. Nat.*, **134**, 147-155.
- Haig, D. (2004) Genomic imprinting and kinship: how good is the evidence? *Annu. Rev. Genet.*, **38**, 553-585.
- Haig, D. (1994) Refusing the ovarian time bomb. *Trends Genet.*, **10**, 346-7; author reply 348-9.
- Haig, D. and Westoby, M. (2006) An earlier formulation of the genetic conflict hypothesis of genomic imprinting. *Nat. Genet.*, **38**, 271.
- Hammond, S.M., Bernstein, E., Beach, D. and Hannon, G.J. (2000) An RNA-directed nuclease mediates post-transcriptional gene silencing in Drosophila cells. *Nature*, **404**, 293-296.
- Han, J., Lee, Y., Yeom, K.H., Kim, Y.K., Jin, H. and Kim, V.N. (2004) The Drosha-DGCR8 complex in primary microRNA processing. *Genes Dev.*, **18**, 3016-3027.

- Hardison, R.C., Oeltjen, J. and Miller, W. (1997) Long human-mouse sequence alignments reveal novel regulatory elements: a reason to sequence the mouse genome. *Genome Res.*, **7**, 959-966.
- Hardison, R.C., Roskin, K.M., Yang, S., Diekhans, M., Kent, W.J., Weber, R., Elnitski, L., Li, J., O'Connor, M., Kolbe, D., et al (2003) Covariation in frequencies of substitution, deletion, transposition, and recombination during eutherian evolution. *Genome Res.*, **13**, 13-26.
- Hark, A.T., Schoenherr, C.J., Katz, D.J., Ingram, R.S., Levorse, J.M. and Tilghman, S.M. (2000) CTCF mediates methylation-sensitive enhancer-blocking activity at the H19/Igf2 locus. *Nature*, **405**, 486-489.
- Harrow, J., Denoeud, F., Frankish, A., Reymond, A., Chen, C.K., Chrast, J., Lagarde, J., Gilbert, J.G., Storey, R., Swarbreck, D., et al (2006) GENCODE: producing a reference annotation for ENCODE. *Genome Biol.*, **7 Suppl 1**, S4.1-9.
- Hayashizaki, Y., Shibata, H., Hirotsune, S., Sugino, H., Okazaki, Y., Sasaki, N., Hirose, K., Imoto, H., Okuzumi, H. and Muramatsu, M. (1994) Identification of an imprinted U2af binding protein related sequence on mouse chromosome 11 using the RLGS method. *Nat. Genet.*, **6**, 33-40.
- Heard, E. (2005) Delving into the diversity of facultative heterochromatin: the epigenetics of the inactive X chromosome. *Curr. Opin. Genet. Dev.*, **15**, 482-489.
- Heintzman, N.D., Stuart, R.K., Hon, G., Fu, Y., Ching, C.W., Hawkins, R.D., Barrera, L.O., Van Calcar, S., Qu, C., Ching, K.A., et al (2007) Distinct and predictive chromatin signatures of transcriptional promoters and enhancers in the human genome. *Nat. Genet.*, **39**, 311-318.
- Hillier, L.W., Miller, W., Birney, E., Warren, W., Hardison, R.C., Ponting, C.P., Bork, P., Burt, D.W., Groenen, M.A., Delany, M.E., et al (2004) Sequence and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution. *Nature*, **432**, 695-716.
- Hodgson, C.P. and Fisk, R.Z. (1987) Hybridization probe size control: optimized 'oligolabelling'. *Nucleic Acids Res.*, **15**, 6295.
- Horak, C.E., Mahajan, M.C., Luscombe, N.M., Gerstein, M., Weissman, S.M. and Snyder, M. (2002) GATA-1 binding sites mapped in the beta-globin locus by using mammalian chIP-chip analysis. *Proc. Natl. Acad. Sci. U. S. A.*, **99**, 2924-2929.

- Humphray, S.J., Scott, C.E., Clark, R., Marron, B., Bender, C., Camm, N., Davis, J., Jenks, A., Noon, A., Patel, M., et al (2007) A high utility integrated map of the pig genome. *Genome Biol.*, **8**, R139.
- Hurst, L.D. (1998) Peromysci, promiscuity and imprinting. *Nat. Genet.*, **20**, 315-316.
- Hurst, L.D. and McVean, G.T. (1998) Do we understand the evolution of genomic imprinting? *Curr. Opin. Genet. Dev.*, **8**, 701-708.
- Hurst, L.D. and McVean, G.T. (1997) Growth effects of uniparental disomes and the conflict theory of genomic imprinting. *Trends Genet.*, **13**, 436-443.
- Hutvagner, G., McLachlan, J., Pasquinelli, A.E., Balint, E., Tuschl, T. and Zamore, P.D. (2001) A cellular function for the RNA-interference enzyme Dicer in the maturation of the let-7 small temporal RNA. *Science*, **293**, 834-838.
- Huynh, K.D. and Lee, J.T. (2005) X-chromosome inactivation: a hypothesis linking ontogeny and phylogeny. *Nat. Rev. Genet.*, **6**, 410-418.
- International Human Genome Sequencing Consortium. (2004) Finishing the euchromatic sequence of the human genome. *Nature*, **431**, 931-945.
- International Human Genome Sequencing Consortium. (2001) Initial sequencing and analysis of the human genome. *Nature*, **409**, 860-921.
- Ishihara, K., Hatano, N., Furuumi, H., Kato, R., Iwaki, T., Miura, K., Jinno, Y. and Sasaki, H. (2000) Comparative genomic sequencing identifies novel tissue-specific enhancers and sequence elements for methylation-sensitive factors implicated in Igf2/H19 imprinting. *Genome Res.*, **10**, 664-671.
- Ishihara, K., Oshimura, M. and Nakao, M. (2006) CTCF-dependent chromatin insulator is linked to epigenetic remodeling. *Mol. Cell*, **23**, 733-742.
- Jabbari, K. and Bernardi, G. (2004) Cytosine methylation and CpG, TpG (CpA) and TpA frequencies. *Gene*, **333**, 143-149.
- Jabbari, K., Caccio, S., Pais de Barros, J.P., Desgres, J. and Bernardi, G. (1997) Evolutionary changes in CpG and methylation levels in the genome of vertebrates. *Gene*, **205**, 109-118.
- Jinno, Y., Yun, K., Nishiwaki, K., Kubota, T., Ogawa, O., Reeve, A.E. and Niikawa, N. (1994) Mosaic and polymorphic imprinting of the WT1 gene in humans. *Nat. Genet.*, **6**, 305-309.

- Jones, B.K., Levorse, J.M. and Tilghman, S.M. (1998) Igf2 imprinting does not require its own DNA methylation or H19 RNA. *Genes Dev.*, **12**, 2200-2207.
- Ke, X., Thomas, N.S., Robinson, D.O. and Collins, A. (2002) The distinguishing sequence characteristics of mouse imprinted genes. *Mamm. Genome*, **13**, 639-645.
- Kellis, M., Patterson, N., Endrizzi, M., Birren, B. and Lander, E.S. (2003) Sequencing and comparison of yeast species to identify genes and regulatory elements. *Nature*, **423**, 241-254.
- Kent, W.J. (2002) BLAT--the BLAST-like alignment tool. *Genome Res.*, **12**, 656-664.
- Ketting, R.F., Fischer, S.E., Bernstein, E., Sijen, T., Hannon, G.J. and Plasterk, R.H. (2001) Dicer functions in RNA interference and in synthesis of small RNA involved in developmental timing in *C. elegans*. *Genes Dev.*, **15**, 2654-2659.
- Killian, J.K., Byrd, J.C., Jirtle, J.V., Munday, B.L., Stoskopf, M.K., MacDonald, R.G. and Jirtle, R.L. (2000) M6P/IGF2R imprinting evolution in mammals. *Mol. Cell*, **5**, 707-716.
- Killian, J.K., Nolan, C.M., Stewart, N., Munday, B.L., Andersen, N.A., Nicol, S. and Jirtle, R.L. (2001) Monotreme IGF2 expression and ancestral origin of genomic imprinting. *J. Exp. Zool.*, **291**, 205-212.
- Kim, T.H., Abdullaev, Z.K., Smith, A.D., Ching, K.A., Loukinov, D.I., Green, R.D., Zhang, M.Q., Lobanenkov, V.V. and Ren, B. (2007) Analysis of the vertebrate insulator protein CTCF-binding sites in the human genome. *Cell*, **128**, 1231-1245.
- Kimura, M. (1983) *The Neutral Theory of Molecular Evolution*. Cambridge University Press, Cambridge.
- Koch, C.M., Andrews, R.M., Flieck, P., Dillon, S.C., Karaoz, U., Clelland, G.K., Wilcox, S., Beare, D.M., Fowler, J.C., Couttet, P., et al (2007) The landscape of histone modifications across 1% of the human genome in five human cell lines. *Genome Res.*, **17**, 691-707.
- Kono, T. (2006) Genomic imprinting is a barrier to parthenogenesis in mammals. *Cytogenet. Genome Res.*, **113**, 31-35.
- Kono, T., Obata, Y., Wu, Q., Niwa, K., Ono, Y., Yamamoto, Y., Park, E.S., Seo, J.S. and Ogawa, H. (2004) Birth of parthenogenetic mice that can develop to adulthood. *Nature*, **428**, 860-864.

- Kumar, S. and Hedges, S.B. (1998) A molecular timescale for vertebrate evolution. *Nature*, **392**, 917-920.
- Kurtz, S., Phillippy, A., Delcher, A.L., Smoot, M., Shumway, M., Antonescu, C. and Salzberg, S.L. (2004) Versatile and open software for comparing large genomes. *Genome Biol.*, **5**, R12.
- Kurukuti, S., Tiwari, V.K., Tavoosidana, G., Pugacheva, E., Murrell, A., Zhao, Z., Lobanenkov, V., Reik, W. and Ohlsson, R. (2006) CTCF binding at the H19 imprinting control region mediates maternally inherited higher-order chromatin conformation to restrict enhancer access to Igf2. *Proc. Natl. Acad. Sci. U. S. A.*, **103**, 10684-10689.
- Lachner, M., O'Sullivan, R.J. and Jenuwein, T. (2003) An epigenetic road map for histone lysine methylation. *J. Cell. Sci.*, **116**, 2117-2124.
- Laemmli, U.K., Kas, E., Poljak, L. and Adachi, Y. (1992) Scaffold-associated regions: cis-acting determinants of chromatin structural loops and functional domains. *Curr. Opin. Genet. Dev.*, **2**, 275-285.
- Lalande, M. (1996) Parental imprinting and human disease. *Annu. Rev. Genet.*, **30**, 173-195.
- Lamesch, P., Li, N., Milstein, S., Fan, C., Hao, T., Szabo, G., Hu, Z., Venkatesan, K., Bethel, G., Martin, P., et al (2007) hORFeome v3.1: a resource of human open reading frames representing over 10,000 human genes. *Genomics*, **89**, 307-315.
- Landy, A. (1989) Dynamic, structural, and regulatory aspects of lambda site-specific recombination. *Annu. Rev. Biochem.*, **58**, 913-949.
- Larkin, M.A., Blackshields, G., Brown, N.P., Chenna, R., McGgettigan, P.A., McWilliam, H., Valentin, F., Wallace, I.M., Wilm, A., Lopez, R., et al (2007) Clustal W and Clustal X version 2.0. *Bioinformatics*, **23**, 2947-2948.
- Lawton, B.R., Obergfell, C., O'Neill, R.J. and O'Neill, M.J. (2007) Physical mapping of the IGF2 locus in the South American opossum *Monodelphis domestica*. *Cytogenet. Genome Res.*, **116**, 130-131.
- Lee, J.T. (2003) Molecular links between X-inactivation and autosomal imprinting: X-inactivation as a driving force for the evolution of imprinting? *Curr. Biol.*, **13**, R242-54.

- Lefevre, C.M., Digby, M.R., Whitley, J.C., Strahm, Y. and Nicholas, K.R. (2007) Lactation transcriptomics in the Australian marsupial, *Macropus eugenii*: transcript sequencing and quantification. *BMC Genomics*, **8**, 417.
- Leighton, P.A., Saam, J.R., Ingram, R.S., Stewart, C.L. and Tilghman, S.M. (1995) An enhancer deletion affects both H19 and Igf2 expression. *Genes Dev.*, **9**, 2079-2089.
- Lettice, L.A., Heaney, S.J., Purdie, L.A., Li, L., de Beer, P., Oostra, B.A., Goode, D., Elgar, G., Hill, R.E. and de Graaff, E. (2003) A long-range Shh enhancer regulates expression in the developing limb and fin and is associated with preaxial polydactyly. *Hum. Mol. Genet.*, **12**, 1725-1735.
- Lewis, A. and Reik, W. (2006) How imprinting centres work. *Cytogenet. Genome Res.*, **113**, 81-89.
- Li, E., Beard, C., Forster, A.C., Bestor, T.H. and Jaenisch, R. (1993a) DNA methylation, genomic imprinting, and mammalian development. *Cold Spring Harb. Symp. Quant. Biol.*, **58**, 297-305.
- Li, E., Beard, C. and Jaenisch, R. (1993b) Role for DNA methylation in genomic imprinting. *Nature*, **366**, 362-365.
- Lindblad-Toh, K., Winchester, E., Daly, M.J., Wang, D.G., Hirschhorn, J.N., Laviolette, J.P., Ardlie, K., Reich, D.E., Robinson, E., Sklar, P., et al (2000) Large-scale discovery and genotyping of single-nucleotide polymorphisms in the mouse. *Nat. Genet.*, **24**, 381-386.
- Lingenfelter, P.A., Delbridge, M.L., Thomas, S., Hoekstra, H.E., Mitchell, M.J., Graves, J.A. and Disteche, C.M. (2001) Expression and conservation of processed copies of the RBMX gene. *Mamm. Genome*, **12**, 538-545.
- Long, L. and Spear, B.T. (2004) FoxA proteins regulate H19 endoderm enhancer E1 and exhibit developmental changes in enhancer binding in vivo. *Mol. Cell. Biol.*, **24**, 9601-9609.
- Loots, G. and Ovcharenko, I. (2007) ECRbase: database of evolutionary conserved regions, promoters, and transcription factor binding sites in vertebrate genomes. *Bioinformatics*, **23**, 122-124.
- Loots, G.G., Locksley, R.M., Blanksespoor, C.M., Wang, Z.E., Miller, W., Rubin, E.M. and Frazer, K.A. (2000) Identification of a coordinate regulator of interleukins 4, 13, and 5 by cross-species sequence comparisons. *Science*, **288**, 136-140.

- Loots, G.G. and Ovcharenko, I. (2005) Dcode.org anthology of comparative genomic tools. *Nucleic Acids Res.*, **33**, W56-64.
- Loots, G.G. and Ovcharenko, I. (2004) rVISTA 2.0: evolutionary analysis of transcription factor binding sites. *Nucleic Acids Res.*, **32**, W217-21.
- Lowe, C.B., Bejerano, G. and Haussler, D. (2007) Thousands of human mobile element fragments undergo strong purifying selection near developmental genes. *Proc. Natl. Acad. Sci. U. S. A.*, **104**, 8005-8010.
- Lowe, T.M. and Eddy, S.R. (1997) tRNAscan-SE: a program for improved detection of transfer RNA genes in genomic sequence. *Nucleic Acids Res.*, **25**, 955-964.
- Luedi, P.P., Dietrich, F.S., Weidman, J.R., Bosko, J.M., Jirtle, R.L. and Hartemink, A.J. (2007) Computational and experimental identification of novel human imprinted genes. *Genome Res.*, **17**, 1723-1730.
- Luedi, P.P., Hartemink, A.J. and Jirtle, R.L. (2005) Genome-wide prediction of imprinted murine genes. *Genome Res.*, **15**, 875-884.
- Lyko, F., Brenton, J.D., Surani, M.A. and Paro, R. (1997) An imprinting element from the mouse H19 locus functions as a silencer in Drosophila. *Nat. Genet.*, **16**, 171-173.
- Lyon, M.F. (1998) X-chromosome inactivation: a repeat hypothesis. *Cytogenet. Cell Genet.*, **80**, 133-137.
- Ma, B., Tromp, J. and Li, M. (2002) PatternHunter: faster and more sensitive homology search. *Bioinformatics*, **18**, 440-445.
- Maglott, D., Ostell, J., Pruitt, K.D. and Tatusova, T. (2007) Entrez Gene: gene-centered information at NCBI. *Nucleic Acids Res.*, **35**, D26-31.
- Maizel, J.V., Jr and Lenk, R.P. (1981) Enhanced graphic matrix analysis of nucleic acid and protein sequences. *Proc. Natl. Acad. Sci. U. S. A.*, **78**, 7665-7669.
- Mancini-Dinardo, D., Steele, S.J., Levorse, J.M., Ingram, R.S. and Tilghman, S.M. (2006) Elongation of the Kcnq1ot1 transcript is required for genomic imprinting of neighboring genes. *Genes Dev.*, **20**, 1268-1282.
- Margueron, R., Trojer, P. and Reinberg, D. (2005) The key to development: interpreting the histone code? *Curr. Opin. Genet. Dev.*, **15**, 163-176.

- Margulies, E.H., Blanchette, M., Haussler, D., Green, E.D. and NISC Comparative Sequencing Program. (2003a) Identification and characterization of multi-species conserved sequences. *Genome Res.*, **13**, 2507-2518.
- Margulies, E.H., Green, E.D. and NISC Comparative Sequencing Program. (2003b) Detecting highly conserved regions of the human genome by multispecies sequence comparisons. *Cold Spring Harb. Symp. Quant. Biol.*, **68**, 255-263.
- Margulies, E.H., Maduro, V.V., Thomas, P.J., Tomkins, J.P., Amemiya, C.T., Luo, M., Green, E.D. and NISC Comparative Sequencing Program. (2005a) Comparative sequencing provides insights about the structure and conservation of marsupial and monotreme genomes. *Proc. Natl. Acad. Sci. U. S. A.*, **102**, 3354-3359.
- Margulies, E.H., Vinson, J.P., Miller, W., Jaffe, D.B., Lindblad-Toh, K., Chang, J.L., Green, E.D., Lander, E.S., Mullikin, J.C., Clamp, M., et al (2005b) An initial strategy for the systematic identification of functional elements in the human genome by low-redundancy comparative sequencing. *Proc. Natl. Acad. Sci. U. S. A.*, **102**, 4795-4800.
- Marra, M.A., Kucaba, T.A., Dietrich, N.L., Green, E.D., Brownstein, B., Wilson, R.K., McDonald, K.M., Hillier, L.W., McPherson, J.D. and Waterston, R.H. (1997) High throughput fingerprint analysis of large-insert clones. *Genome Res.*, **7**, 1072-1084.
- Marsters, S.A., Frutkin, A.D., Simpson, N.J., Fendly, B.M. and Ashkenazi, A. (1992) Identification of cysteine-rich domains of the type 1 tumor necrosis factor receptor involved in ligand binding. *J. Biol. Chem.*, **267**, 5747-5750.
- Martinez, J., Patkaniowska, A., Urlaub, H., Luhrmann, R. and Tuschl, T. (2002) Single-stranded antisense siRNAs guide target RNA cleavage in RNAi. *Cell*, **110**, 563-574.
- Maston, G.A., Evans, S.K. and Green, M.R. (2006) Transcriptional Regulatory Elements in the Human Genome. *Annu. Rev. Genomics Hum. Genet.*, **7**, 29-59.
- Matys, V., Kel-Margoulis, O.V., Fricke, E., Liebich, I., Land, S., Barre-Dirrie, A., Reuter, I., Chekmenev, D., Krull, M., Hornischer, K., et al (2006) TRANSFAC and its module TRANSCompel: transcriptional gene regulation in eukaryotes. *Nucleic Acids Res.*, **34**, D108-10.
- Mayor, C., Brudno, M., Schwartz, J.R., Poliakov, A., Rubin, E.M., Frazer, K.A., Pachter, L.S. and Dubchak, I. (2000) VISTA : visualizing global DNA sequence alignments of arbitrary length. *Bioinformatics*, **16**, 1046-1047.

- McDonald, J.F., Matzke, M.A. and Matzke, A.J. (2005) Host defenses to transposable elements and the evolution of genomic imprinting. *Cytogenetic and Genome Research*, **110**, 242-249.
- McGowan, R.A. and Martin, C.C. (1997) DNA methylation and genome imprinting in the zebrafish, *Danio rerio*: some evolutionary ramifications. *Biochem. Cell Biol.*, **75**, 499-506.
- McGrath, J. and Solter, D. (1984) Completion of mouse embryogenesis requires both the maternal and paternal genomes. *Cell*, **37**, 179-183.
- McPherson, J.D., Marra, M., Hillier, L., Waterston, R.H., Chinwalla, A., Wallis, J., Sekhon, M., Wylie, K., Mardis, E.R., Wilson, R.K., et al (2001) A physical map of the human genome. *Nature*, **409**, 934-941.
- MENDEL, G. (1950) Gregor Mendel's letters to Carl Nageli, 1866-1873. *Genetics*, **35**, 1-29.
- Mikkelsen, T.S., Wakefield, M.J., Aken, B., Amemiya, C.T., Chang, J.L., Duke, S., Garber, M., Gentles, A.J., Goodstadt, L., Heger, A., et al (2007) Genome of the marsupial *Monodelphis domestica* reveals innovation in non-coding sequences. *Nature*, **447**, 167-177.
- Mineno, J., Okamoto, S., Ando, T., Sato, M., Chono, H., Izu, H., Takayama, M., Asada, K., Mirochnitchenko, O., Inouye, M., et al (2006) The expression profile of microRNAs in mouse embryos. *Nucleic Acids Res.*, **34**, 1765-1771.
- Mitsuya, K., Sui, H., Meguro, M., Kugoh, H., Jinno, Y., Niikawa, N. and Oshimura, M. (1997) Paternal expression of WT1 in human fibroblasts and lymphocytes. *Hum. Mol. Genet.*, **6**, 2243-2246.
- Moore, T. (1994) Refusing the ovarian time bomb. *Trends Genet.*, **10**, 347-349.
- Moore, T. and Haig, D. (1991) Genomic imprinting in mammalian development: a parental tug-of-war. *Trends Genet.*, **7**, 45-49.
- Morgan, T.H. (1915) Localization of the Hereditary Material in the Germ Cells. *Proc. Natl. Acad. Sci. U. S. A.*, **1**, 420-429.
- Morgenstern, B. (1999) DIALIGN 2: improvement of the segment-to-segment approach to multiple sequence alignment. *Bioinformatics*, **15**, 211-218.
- Morison, I.M., Ramsay, J.P. and Spencer, H.G. (2005) A census of mammalian imprinting. *Trends Genet.*, **21**, 457-465.

- Morrow, C.P., Curtin, J.P. and Townsend, D.E. (1993) Tumors of the ovary: classification; the adnexal mass. *Synopsis of gynecologic oncology*. 4th ed. New York: Churchill Livingstone, **224**,
- Mott, R. (1997) EST_GENOME: a program to align spliced DNA sequences to unspliced genomic DNA. *Comput. Appl. Biosci.*, **13**, 477-478.
- Mungall, A.J. and Humphray, S.J. (2003) Assembling physical maps and sequence clone selection. In Dunham, I. (ed) *Genome Mapping and Sequencing*. Horizon Scientific Press, Cambridge, UK, 167-200.
- Mungall, A.J., Humphray, S.J., Ranby, S.A., Edwards, C.A., Heathcott, R.W., Clee, C.M., Holloway, E., Peck, A.I., Harrison, P., Green, L.D., et al (1997) From long range mapping to sequence-ready contigs on human chromosome 6. *DNA Seq.*, **8**, 151-154.
- Murphy, S.K. and Jirtle, R.L. (2003) Imprinting evolution and the price of silence. *Bioessays*, **25**, 577-588.
- Murphy, W.J., Larkin, D.M., Everts-van der Wind, A., Bourque, G., Tesler, G., Auvil, L., Beever, J.E., Chowdhary, B.P., Galibert, F., Gatzke, L., et al (2005) Dynamics of mammalian chromosome evolution inferred from multispecies comparative maps. *Science*, **309**, 613-617.
- Murrell, A., Heeson, S. and Reik, W. (2004) Interaction between differentially methylated regions partitions the imprinted genes Igf2 and H19 into parent-specific chromatin loops. *Nat. Genet.*, **36**, 889-893.
- Nardone, J., Lee, D.U., Ansel, K.M. and Rao, A. (2004) Bioinformatics for the 'bench biologist': how to find regulatory regions in genomic DNA. *Nat. Immunol.*, **5**, 768-774.
- Needleman, S.B. and Wunsch, C.D. (1970) A general method applicable to the search for similarities in the amino acid sequence of two proteins. *J. Mol. Biol.*, **48**, 443-453.
- Nilsson, M.A., Arnason, U., Spencer, P.B. and Janke, A. (2004) Marsupial relationships and a timeline for marsupial radiation in South Gondwana. *Gene*, **340**, 189-196.
- Nobrega, M.A., Ovcharenko, I., Afzal, V. and Rubin, E.M. (2003) Scanning human gene deserts for long-range enhancers. *Science*, **302**, 413.

- Nolan, C.M., Killian, J.K., Petitte, J.N. and Jirtle, R.L. (2001) Imprint status of M6P/IGF2R and IGF2 in chickens. *Dev. Genes Evol.*, **211**, 179-183.
- Obernosterer, G., Leuschner, P.J., Alenius, M. and Martinez, J. (2006) Post-transcriptional regulation of microRNA expression. *RNA*, **12**, 1161-1167.
- Ohlsson, R., Renkawitz, R. and Lobanenkov, V. (2001) CTCF is a uniquely versatile transcription regulator linked to epigenetics and disease. *Trends Genet.*, **17**, 520-527.
- Ohshima, K. and Okada, N. (2005) SINEs and LINEs: symbionts of eukaryotic genomes with a common tail. *Cytogenet. Genome Res.*, **110**, 475-490.
- Olek, A. and Walter, J. (1997) The pre-implantation ontogeny of the H19 methylation imprint. *Nat. Genet.*, **17**, 275-276.
- O'Neill, M.J., Ingram, R.S., Vrana, P.B. and Tilghman, S.M. (2000) Allelic expression of IGF2 in marsupials and birds. *Dev. Genes Evol.*, **210**, 18-20.
- Ono, R., Kobayashi, S., Wagatsuma, H., Aisaka, K., Kohda, T., Kaneko-Ishino, T. and Ishino, F. (2001) A retrotransposon-derived gene, PEG10, is a novel imprinted gene located on human chromosome 7q21. *Genomics*, **73**, 232-237.
- Osoegawa, K., Tateno, M., Woon, P.Y., Frengen, E., Mammoser, A.G., Catanese, J.J., Hayashizaki, Y. and de Jong, P.J. (2000) Bacterial artificial chromosome libraries for mouse sequencing and functional analysis. *Genome Res.*, **10**, 116-128.
- Ovcharenko, I., Loots, G.G., Giardine, B.M., Hou, M., Ma, J., Hardison, R.C., Stubbs, L. and Miller, W. (2005) Mulan: multiple-sequence local alignment and visualization for studying function and evolution. *Genome Res.*, **15**, 184-194.
- Ovcharenko, I., Loots, G.G., Hardison, R.C., Miller, W. and Stubbs, L. (2004a) zPicture: dynamic alignment and visualization tool for analyzing conservation profiles. *Genome Res.*, **14**, 472-477.
- Ovcharenko, I., Stubbs, L. and Loots, G.G. (2004b) Interpreting mammalian evolution using Fugu genome comparisons. *Genomics*, **84**, 890-895.
- Owen, R. (1849) *On parthenogenesis: or the successive production of procreating individuals from a single ovum*. John Van Voorst, London, London.
- Pauler, F.M. and Barlow, D.P. (2006) Imprinting mechanisms--it only takes two. *Genes Dev.*, **20**, 1203-1206.
- Pauler, F.M., Koerner, M.V. and Barlow, D.P. (2007) Silencing by imprinted noncoding RNAs: is transcription the answer? *Trends Genet.*, **23**, 284-292.

- Paulsen, M., Khare, T., Burgard, C., Tierling, S. and Walter, J. (2005) Evolution of the Beckwith-Wiedemann syndrome region in vertebrates. *Genome Res.*, **15**, 146-153.
- Pennacchio, L.A., Ahituv, N., Moses, A.M., Prabhakar, S., Nobrega, M.A., Shoukry, M., Minovitsky, S., Dubchak, I., Holt, A., Lewis, K.D., et al (2006) In vivo enhancer analysis of human conserved non-coding sequences. *Nature*, **444**, 499-502.
- Pennisi, E. (2003) Human genome. A low number wins the GeneSweep Pool. *Science*, **300**, 1484.
- Peters, A.H. and Schubeler, D. (2005) Methylation of histones: playing memory with DNA. *Curr. Opin. Cell Biol.*, **17**, 230-238.
- Pevzner, P. and Tesler, G. (2003) Human and mouse genomic sequences reveal extensive breakpoint reuse in mammalian evolution. *Proc. Natl. Acad. Sci. U. S. A.*, **100**, 7672-7677.
- Plass, C., Yu, F., Yu, L., Strout, M.P., El-Rifai, W., Elonen, E., Knuutila, S., Marcucci, G., Young, D.C., Held, W.A., et al (1999) Restriction landmark genome scanning for aberrant methylation in primary refractory and relapsed acute myeloid leukemia; involvement of the WIT-1 gene. *Oncogene*, **18**, 3159-3165.
- Prabhakar, S., Poulin, F., Shoukry, M., Afzal, V., Rubin, E.M., Couronne, O. and Pennacchio, L.A. (2006) Close sequence comparisons are sufficient to identify human cis-regulatory elements. *Genome Res.*, **16**, 855-863.
- Pruitt, K.D., Tatusova, T. and Maglott, D.R. (2007) NCBI reference sequences (RefSeq): a curated non-redundant sequence database of genomes, transcripts and proteins. *Nucleic Acids Res.*, **35**, D61-5.
- Ptashne, M. (1992) *A Genetic Switch: Phage [lambda] and Higher Organisms*. Blackwell Publishing, Cambridge, MA.
- Puttagunta, R., Gordon, L.A., Meyer, G.E., Kapfhamer, D., Lamerdin, J.E., Kantheti, P., Portman, K.M., Chung, W.K., Jenne, D.E., Olsen, A.S., et al (2000) Comparative maps of human 19p13.3 and mouse chromosome 10 allow identification of sequences at evolutionary breakpoints. *Genome Res.*, **10**, 1369-1380.
- Rada-Iglesias, A., Wallerman, O., Koch, C., Ameur, A., Enroth, S., Clelland, G., Wester, K., Wilcox, S., Dovey, O.M., Ellis, P.D., et al (2005) Binding sites for metabolic disease related transcription factors inferred at base pair resolution by chromatin immunoprecipitation and genomic microarrays. *Hum. Mol. Genet.*, **14**, 3435-3447.

- Rapkins, R.W., Hore, T., Smithwick, M., Ager, E., Pask, A.J., Renfree, M.B., Kohn, M., Hameister, H., Nicholls, R.D., Deakin, J.E., et al (2006) Recent assembly of an imprinted domain from non-imprinted components. *PLoS Genet.*, **2**, e182.
- Reik, W., Constancia, M., Fowden, A., Anderson, N., Dean, W., Ferguson-Smith, A., Tycko, B. and Sibley, C. (2003) Regulation of supply and demand for maternal nutrients in mammals by imprinted genes. *J. Physiol.*, **547**, 35-44.
- Reik, W. and Lewis, A. (2005) Co-evolution of X-chromosome inactivation and imprinting in mammals. *Nat. Rev. Genet.*, **6**, 403-410.
- Reik, W. and Walter, J. (2001) Genomic imprinting: parental influence on the genome. *Nat. Rev. Genet.*, **2**, 21-32.
- Reik, W. and Walter, J. (1998) Imprinting mechanisms in mammals. *Curr. Opin. Genet. Dev.*, **8**, 154-164.
- Renda, M., Baglivo, I., Burgess-Beusse, B., Esposito, S., Fattorusso, R., Felsenfeld, G. and Pedone, P.V. (2007) Critical DNA binding interactions of the insulator protein CTCF: a small number of zinc fingers mediate strong binding, and a single finger-dna interaction controls binding at imprinted loci. *J. Biol. Chem.*, **282**, 33336-33345.
- Renfree, M.B. and Blanden, D.R. (2000) Progesterone and oestrogen receptors in the female genital tract throughout pregnancy in tammar wallabies. *J. Reprod. Fertil.*, **119**, 121-128.
- Renfree, M.B. and Shaw, G. (2000) Diapause. *Annu. Rev. Physiol.*, **62**, 353-375.
- Rens, W., O'Brien, P.C., Grutzner, F., Clarke, O., Graphodatskaya, D., Tsend-Ayush, E., Trifonov, V.A., Skelton, H., Wallis, M.C., Johnston, S., et al (2007) The multiple sex chromosomes of platypus and echidna are not completely identical and several share homology with the avian Z. *Genome Biol.*, **8**, R243.
- Rice, P., Longden, I. and Bleasby, A. (2000) EMBOSS: the European Molecular Biology Open Software Suite. *Trends Genet.*, **16**, 276-277.
- Rice, W.R. and Chippindale, A.K. (2001) Sexual recombination and the power of natural selection. *Science*, **294**, 555-559.
- Ross, M.T., LaBrie, S., McPherson, J. and Stanton, V.P. (1999) Screening large-insert libraries by hybridization. In Dracopoli, N. C., Haines, J. L., Korf, B. R., et al (eds) *In Current protocols in human genetics*. John Wiley and Sons, New York, 1-52.

- Roth, S.Y., Denu, J.M. and Allis, C.D. (2001) Histone acetyltransferases. *Annu. Rev. Biochem.*, **70**, 81-120.
- Rozen, S. and Skaletsky, H. (2000) Primer3 on the WWW for general users and for biologist programmers. *Methods Mol. Biol.*, **132**, 365-386.
- Rual, J.F., Hirozane-Kishikawa, T., Hao, T., Bertin, N., Li, S., Dricot, A., Li, N., Rosenberg, J., Lamesch, P., Vidalain, P.O., et al (2004) Human ORFeome version 1.1: a platform for reverse proteomics. *Genome Res.*, **14**, 2128-2135.
- Ruiz-Herrera, A., Castresana, J. and Robinson, T.J. (2006) Is mammalian chromosomal evolution driven by regions of genome fragility? *Genome Biol.*, **7**, R115.
- Ryan, D., Rahimi, M., Lund, J., Mehta, R. and Parviz, B.A. (2007) Toward nanoscale genome sequencing. *Trends Biotechnol.*, **25**, 385-389.
- Sabo, P.J., Kuehn, M.S., Thurman, R., Johnson, B.E., Johnson, E.M., Cao, H., Yu, M., Rosenzweig, E., Goldy, J., Haydock, A., et al (2006) Genome-scale mapping of DNase I sensitivity in vivo using tiling DNA microarrays. *Nat. Methods*, **3**, 511-518.
- Salamov, A.A. and Solovyev, V.V. (2000) Ab initio gene finding in *Drosophila* genomic DNA. *Genome Res.*, **10**, 516-522.
- Salcedo, T., Gerald, A. and Nachman, M.W. (2007) Nucleotide variation in wild and inbred mice. *Genetics*, **177**, 2277-2291.
- Sandelin, A., Carninci, P., Lenhard, B., Ponjavic, J., Hayashizaki, Y. and Hume, D.A. (2007) Mammalian RNA polymerase II core promoters: insights from genome-wide studies. *Nat. Rev. Genet.*, **8**, 424-436.
- Sanger, F., Nicklen, S. and Coulson, A.R. (1977) DNA sequencing with chain-terminating inhibitors. *Proc. Natl. Acad. Sci. U. S. A.*, **74**, 5463-5467.
- Santangelo, A.M., de Souza, F.S., Franchini, L.F., Bumaschny, V.F., Low, M.J. and Rubinstein, M. (2007) Ancient exaptation of a CORE-SINE retroposon into a highly conserved mammalian neuronal enhancer of the proopiomelanocortin gene. *PLoS Genet.*, **3**, 1813-1826.
- Sasaki, H., Jones, P.A., Chaillet, J.R., Ferguson-Smith, A.C., Barton, S.C., Reik, W. and Surani, M.A. (1992) Parental imprinting: potentially active chromatin of the repressed maternal allele of the mouse insulin-like growth factor II (*Igf2*) gene. *Genes Dev.*, **6**, 1843-1856.

Schiltz, R.L., Mizzen, C.A., Vassilev, A., Cook, R.G., Allis, C.D. and Nakatani, Y. (1999) Overlapping but distinct patterns of histone acetylation by the human coactivators p300 and PCAF within nucleosomal substrates. *J. Biol. Chem.*, **274**, 1189-1192.

Schmutz, J., Wheeler, J., Grimwood, J., Dickson, M., Yang, J., Caoile, C., Bajorek, E., Black, S., Chan, Y.M., Denys, M., et al (2004) Quality assessment of the human genome sequence. *Nature*, **429**, 365-368.

Schneider, P., Olson, D., Tardivel, A., Browning, B., Lugovskoy, A., Gong, D., Dobles, M., Hertig, S., Hofmann, K., Van Vlijmen, H., et al (2003) Identification of a new murine tumor necrosis factor receptor locus that contains two novel murine receptors for tumor necrosis factor-related apoptosis-inducing ligand (TRAIL). *J. Biol. Chem.*, **278**, 5444-5454.

Schuler, G.D. (1997) Sequence mapping by electronic PCR. *Genome Res.*, **7**, 541-550.

Schwartz, S., Elnitski, L., Li, M., Weirauch, M., Riemer, C., Smit, A., NISC Comparative Sequencing Program, Green, E.D., Hardison, R.C. and Miller, W. (2003a) MultiPipMaker and supporting tools: Alignments and analysis of multiple genomic DNA sequences. *Nucleic Acids Res.*, **31**, 3518-3524.

Schwartz, S., Kent, W.J., Smit, A., Zhang, Z., Baertsch, R., Hardison, R.C., Haussler, D. and Miller, W. (2003b) Human-mouse alignments with BLASTZ. *Genome Res.*, **13**, 103-107.

Schwartz, S., Zhang, Z., Frazer, K.A., Smit, A., Riemer, C., Bouck, J., Gibbs, R., Hardison, R. and Miller, W. (2000) PipMaker--a web server for aligning two genomic DNA sequences. *Genome Res.*, **10**, 577-586.

Schwarz, D.S., Hutvagner, G., Haley, B. and Zamore, P.D. (2002) Evidence that siRNAs function as guides, not primers, in the Drosophila and human RNAi pathways. *Mol. Cell*, **10**, 537-548.

Searle, S.M., Gilbert, J., Iyer, V. and Clamp, M. (2004) The otter annotation system. *Genome Res.*, **14**, 963-970.

Seitz, H., Royo, H., Bortolin, M.L., Lin, S.P., Ferguson-Smith, A.C. and Cavaille, J. (2004) A large imprinted microRNA gene cluster at the mouse Dlk1-Gtl2 domain. *Genome Res.*, **14**, 1741-1748.

Seitz, H., Youngson, N., Lin, S.P., Dalbert, S., Paulsen, M., Bachellerie, J.P., Ferguson-Smith, A.C. and Cavaille, J. (2003) Imprinted microRNA genes

transcribed antisense to a reciprocally imprinted retrotransposon-like gene. *Nat. Genet.*, **34**, 261-262.

Sharman, G.B. (1971) Late DNA replication in the paternally derived X chromosome of female kangaroos. *Nature*, **230**, 231-232.

She, X., Jiang, Z., Clark, R.A., Liu, G., Cheng, Z., Tuzun, E., Church, D.M., Sutton, G., Halpern, A.L. and Eichler, E.E. (2004) Shotgun sequence assembly and recent segmental duplications within the human genome. *Nature*, **431**, 927-930.

Shen, J.C., Rideout, W.M.,3rd and Jones, P.A. (1994) The rate of hydrolytic deamination of 5-methylcytosine in double-stranded DNA. *Nucleic Acids Res.*, **22**, 972-976.

Shendure, J., Mitra, R.D., Varma, C. and Church, G.M. (2004) Advanced sequencing technologies: methods and goals. *Nat. Rev. Genet.*, **5**, 335-344.

Shirohzu, H., Yokomine, T., Sato, C., Kato, R., Toyoda, A., Purbowasito, W., Suda, C., Mukai, T., Hattori, M., Okumura, K., et al (2004) A 210-kb segment of tandem repeats and retroelements located between imprinted subdomains of mouse distal chromosome 7. *DNA Res.*, **11**, 325-334.

Shizuya, H., Birren, B., Kim, U.J., Mancino, V., Slepak, T., Tachiiri, Y. and Simon, M. (1992) Cloning and stable maintenance of 300-kilobase-pair fragments of human DNA in Escherichia coli using an F-factor-based vector. *Proc. Natl. Acad. Sci. U. S. A.*, **89**, 8794-8797.

Siddle, H.V., Deakin, J.E., Baker, M.L., Miller, R.D. and Belov, K. (2006) Isolation of major histocompatibility complex Class I genes from the tammar wallaby (*Macropus eugenii*). *Immunogenetics*, **58**, 487-493.

Siepel, A., Bejerano, G., Pedersen, J.S., Hinrichs, A.S., Hou, M., Rosenbloom, K., Clawson, H., Spieth, J., Hillier, L.W., Richards, S., et al (2005) Evolutionarily conserved elements in vertebrate, insect, worm, and yeast genomes. *Genome Res.*, **15**, 1034-1050.

Sleutels, F., Zwart, R. and Barlow, D.P. (2002) The non-coding Air RNA is required for silencing autosomal imprinted genes. *Nature*, **415**, 810-813.

Smith, T.F. and Waterman, M.S. (1981) Identification of common molecular subsequences. *J. Mol. Biol.*, **147**, 195-197.

Soderlund, C., Longden, I. and Mott, R. (1997) FPC: a system for building contigs from restriction fingerprinted clones. *Comput. Appl. Biosci.*, **13**, 523-535.

- Soejima, H. and Wagstaff, J. (2005) Imprinting centers, chromatin structure, and disease. *J. Cell. Biochem.*, **95**, 226-233.
- Solovyev, V.V., Salamov, A.A. and Lawrence, C.B. (1994) Predicting internal exons by oligonucleotide composition and discriminant analysis of spliceable open reading frames. *Nucleic Acids Res.*, **22**, 5156-5163.
- Solter, D. (1994) Refusing the ovarian time bomb. *Trends Genet.*, **10**, 346; author reply 348-9.
- Solter, D. (1988) Differential imprinting and expression of maternal and paternal genomes. *Annu. Rev. Genet.*, **22**, 127-146.
- Sonnhammer, E.L. and Durbin, R. (1995) A dot-matrix program with dynamic threshold control suited for genomic DNA and protein sequence analysis. *Gene*, **167**, GC1-10.
- Spillane, C., Schmid, K.J., Laoueille-Duprat, S., Pien, S., Escobar-Restrepo, J.M., Baroux, C., Gagliardini, V., Page, D.R., Wolfe, K.H. and Grossniklaus, U. (2007) Positive darwinian selection at the imprinted MEDEA locus in plants. *Nature*, **448**, 349-352.
- Staden, R. (1979) A strategy of DNA sequencing employing computer programs. *Nucleic Acids Res.*, **6**, 2601-2610.
- Staden, R., Beal, K.F. and Bonfield, J.K. (2000) The Staden package, 1998. *Methods Mol. Biol.*, **132**, 115-130.
- Stankiewicz, P., Park, S.S., Inoue, K. and Lupski, J.R. (2001) The evolutionary chromosome translocation 4;19 in Gorilla gorilla is associated with microduplication of the chromosome fragment syntenic to sequences surrounding the human proximal CMT1A-REP. *Genome Res.*, **11**, 1205-1210.
- Strahl, B.D. and Allis, C.D. (2000) The language of covalent histone modifications. *Nature*, **403**, 41-45.
- Sueoka, N. (1988) Directional mutation pressure and neutral molecular evolution. *Proc. Natl. Acad. Sci. U. S. A.*, **85**, 2653-2657.
- Surani, M.A., Barton, S.C. and Norris, M.L. (1984) Development of reconstituted mouse eggs suggests imprinting of the genome during gametogenesis. *Nature*, **308**, 548-550.

- Suzuki, S., Ono, R., Narita, T., Pask, A.J., Shaw, G., Wang, C., Kohda, T., Alsop, A.E., Marshall Graves, J.A., Kohara, Y., et al (2007) Retrotransposon silencing by DNA methylation can drive mammalian genomic imprinting. *PLoS Genet.*, **3**, e55.
- Suzuki, S., Renfree, M.B., Pask, A.J., Shaw, G., Kobayashi, S., Kohda, T., Kaneko-Ishino, T. and Ishino, F. (2005) Genomic imprinting of IGF2, p57(KIP2) and PEG1/MEST in a marsupial, the tammar wallaby. *Mech. Dev.*, **122**, 213-222.
- Sved, J. and Bird, A. (1990) The expected equilibrium of the CpG dinucleotide in vertebrate genomes under a mutation model. *Proc. Natl. Acad. Sci. U. S. A.*, **87**, 4692-4696.
- Szabo, Z., Levi-Minzi, S.A., Christiano, A.M., Struminger, C., Stoneking, M., Batzer, M.A. and Boyd, C.D. (1999) Sequential loss of two neighboring exons of the tropoelastin gene during primate evolution. *J. Mol. Evol.*, **49**, 664-671.
- Tagle, D.A., Koop, B.F., Goodman, M., Slightom, J.L., Hess, D.L. and Jones, R.T. (1988) Embryonic epsilon and gamma globin genes of a prosimian primate (*Galago crassicaudatus*). Nucleotide and amino acid sequences, developmental regulation and phylogenetic footprints. *J. Mol. Biol.*, **203**, 439-455.
- The Arabidopsis Genome Initiative. (2000) Analysis of the genome sequence of the flowering plant *Arabidopsis thaliana*. *Nature*, **408**, 796-815.
- The C. elegans Sequencing Consortium. (1998) Genome Sequence of the Nematode *C. elegans*: A Platform for Investigating Biology. *Science*, **282**, 2012-2018.
- Thomas, J.W., Touchman, J.W., Blakesley, R.W., Bouffard, G.G., Beckstrom-Sternberg, S.M., Margulies, E.H., Blanchette, M., Siepel, A.C., Thomas, P.J., McDowell, J.C., et al (2003) Comparative analyses of multi-species sequences from targeted genomic regions. *Nature*, **424**, 788-793.
- Thomson, J.M., Newman, M., Parker, J.S., Morin-Kensicki, E.M., Wright, T. and Hammond, S.M. (2006) Extensive post-transcriptional regulation of microRNAs and its implications for cancer. *Genes Dev.*, **20**, 2202-2207.
- Thorvaldsen, J.L., Mann, M.R., Nwoko, O., Duran, K.L. and Bartolomei, M.S. (2002) Analysis of sequence upstream of the endogenous H19 gene reveals elements both essential and dispensable for imprinting. *Mol. Cell. Biol.*, **22**, 2450-2462.
- Toder, R., Wilcox, S.A., Smithwick, M. and Graves, J.A. (1996) The human/mouse imprinted genes IGF2, H19, SNRPN and ZNF127 map to two conserved autosomal clusters in a marsupial. *Chromosome Res.*, **4**, 295-300.

- Tremblay, K.D., Duran, K.L. and Bartolomei, M.S. (1997) A 5' 2-kilobase-pair region of the imprinted mouse H19 gene exhibits exclusive paternal methylation throughout development. *Mol. Cell. Biol.*, **17**, 4322-4329.
- Tuiskula-Haavisto, M. and Vilkki, J. (2007) Parent-of-origin specific QTL--a possibility towards understanding reciprocal effects in chicken and the origin of imprinting. *Cytogenet. Genome Res.*, **117**, 305-312.
- Tycko, B. and Morison, I.M. (2002) Physiological functions of imprinted genes. *J. Cell. Physiol.*, **192**, 245-258.
- Tyndale-Biscoe, H. and Renfree, M. (1987) *Reproductive Physiology of Marsupials*. Cambridge University Press,
- Vakoc, C.R., Sachdeva, M.M., Wang, H. and Blobel, G.A. (2006) Profile of histone lysine methylation across transcribed mammalian chromatin. *Mol. Cell. Biol.*, **26**, 9185-9195.
- Varmuza, S. and Mann, M. (1994) Genomic imprinting--defusing the ovarian time bomb. *Trends Genet.*, **10**, 118-123.
- Veevers, J.J. (1991) Phanerozoic Australia in the changing configuration of Proto-Pangea through Gondwanaland and Pangea to the present dispersed continents. *Aust. Syst. Bot.*, **4**, 1-11.
- Venter, J.C., Adams, M.D., Myers, E.W., Li, P.W., Mural, R.J., Sutton, G.G., Smith, H.O., Yandell, M., Evans, C.A., Holt, R.A., et al (2001) The sequence of the human genome. *Science*, **291**, 1304-1351.
- Visel, A., Bristow, J. and Pennacchio, L.A. (2007) Enhancer identification through comparative genomics. *Semin. Cell Dev. Biol.*, **18**, 140-152.
- Vollrath, D. (1999) DNA Markers for Physical Mapping. In Birren, B., Green, E. D., Hieter, P., et al (eds) *Genome Analysis: A Laboratory Manual*. Cold Spring Harbor Laboratory Press, 187-216.
- Vu, T.H., Jirtle, R.L. and Hoffman, A.R. (2006) Cross-species clues of an epigenetic imprinting regulatory code for the IGF2R gene. *Cytogenet. Genome Res.*, **113**, 202-208.
- Waddington, C. (1942) The Epigenotype. *Endeavour*, **1**, 18-20.
- Wakefield, M.J. and Graves, J.A. (2005) Marsupials and monotremes sort genome treasures from junk. *Genome Biol.*, **6**, 218.

- Walter, J., Hutter, B., Khare, T. and Paulsen, M. (2006) Repetitive elements in imprinted genes. *Cytogenet. Genome Res.*, **113**, 109-115.
- Walter, J. and Paulsen, M. (2003) The potential role of gene duplications in the evolution of imprinting mechanisms. *Hum. Mol. Genet.*, **12 Spec No 2**, R215-20.
- Waterston, R.H., Lindblad-Toh, K., Birney, E., Rogers, J., Abril, J.F., Agarwal, P., Agarwala, R., Ainscough, R., Andersson, M., An, P., et al (2002) Initial sequencing and comparative analysis of the mouse genome. *Nature*, **420**, 520-562.
- Weber, M., Hagege, H., Murrell, A., Brunel, C., Reik, W., Cathala, G. and Forne, T. (2003) Genomic imprinting controls matrix attachment regions in the Igf2 gene. *Mol. Cell. Biol.*, **23**, 8953-8959.
- Weidman, J.R., Maloney, K.A. and Jirtle, R.L. (2006) Comparative phylogenetic analysis reveals multiple non-imprinted isoforms of opossum Dlk1. *Mammalian Genome*, **17**, 157-167.
- Weidman, J.R., Murphy, S.K., Nolan, C.M., Dietrich, F.S. and Jirtle, R.L. (2004) Phylogenetic footprint analysis of IGF2 in extant mammals. *Genome Res.*, **14**, 1726-1732.
- Wentworth, B.M., Schaefer, I.M., Villa-Komaroff, L. and Chirgwin, J.M. (1986) Characterization of the two nonallelic genes encoding mouse preproinsulin. *J. Mol. Evol.*, **23**, 305-312.
- West, A.G., Gaszner, M. and Felsenfeld, G. (2002) Insulators: many functions, many mechanisms. *Genes Dev.*, **16**, 271-288.
- Wilkins, J.F. (2005) Genomic imprinting and methylation: epigenetic canalization and conflict. *Trends Genet.*, **21**, 356-365.
- Wilkins, J.F. and Haig, D. (2003) What good is genomic imprinting: the function of parent-specific gene expression. *Nat. Rev. Genet.*, **4**, 359-368.
- Willson, M.F. and Burley, N. (1983) *Mate Choice in Plants: Tactics, Mechanisms, and Consequences*. Princeton University Press,
- Wolfe, K.H., Sharp, P.M. and Li, W.H. (1989) Mutation rates differ among regions of the mammalian genome. *Nature*, **337**, 283-285.
- Wood, A.J., Roberts, R.G., Monk, D., Moore, G.E., Schulz, R. and Oakey, R.J. (2007) A screen for retrotransposed imprinted genes reveals an association between X chromosome homology and maternal germ-line methylation. *PLoS Genet.*, **3**, e20.

- Woolfe, A., Goodson, M., Goode, D.K., Snell, P., McEwen, G.K., Vavouri, T., Smith, S.F., North, P., Callaway, H., Kelly, K., et al (2005) Highly conserved non-coding sequences are associated with vertebrate development. *PLoS Biol.*, **3**, e7.
- Wu, H.K., Squire, J.A., Song, Q. and Weksberg, R. (1997) Promoter-dependent tissue-specific expressive nature of imprinting gene, insulin-like growth factor II, in human tissues. *Biochem. Biophys. Res. Commun.*, **233**, 221-226.
- Xi, H., Shulha, H.P., Lin, J.M., Vales, T.R., Fu, Y., Bodine, D.M., McKay, R.D., Chenoweth, J.G., Tesar, P.J., Furey, T.S., et al (2007) Identification and characterization of cell type-specific and ubiquitous chromatin regulatory structures in the human genome. *PLoS Genet.*, **3**, e136.
- Xie, X., Mikkelsen, T.S., Gnirke, A., Lindblad-Toh, K., Kellis, M. and Lander, E.S. (2007) Systematic discovery of regulatory motifs in conserved regions of the human genome, including thousands of CTCF insulator sites. *Proc. Natl. Acad. Sci. U. S. A.*, **104**, 7145-7150.
- Yahagi, S., Shibuya, K., Obayashi, I., Masaki, H., Kurata, Y., Kudoh, J. and Shimizu, N. (2004) Identification of two novel clusters of ultrahigh-sulfur keratin-associated protein genes on human chromosome 11. *Biochem. Biophys. Res. Commun.*, **318**, 655-664.
- Yang, P.K. and Kuroda, M.I. (2007) Noncoding RNAs and intranuclear positioning in monoallelic gene expression. *Cell*, **128**, 777-786.
- Ye, L. and Huang, X. (2005) MAP2: multiple alignment of syntenic genomic sequences. *Nucleic Acids Res.*, **33**, 162-170.
- Ylstra, B., van den Ijssel, P., Carvalho, B., Brakenhoff, R.H. and Meijer, G.A. (2006) BAC to the future! or oligonucleotides: a perspective for micro array comparative genomic hybridization (array CGH). *Nucleic Acids Res.*, **34**, 445-450.
- Yoder, J.A., Walsh, C.P. and Bestor, T.H. (1997) Cytosine methylation and the ecology of intragenomic parasites. *Trends Genet.*, **13**, 335-340.
- Yokomine, T., Hata, K., Tsudzuki, M. and Sasaki, H. (2006) Evolution of the vertebrate DNMT3 gene family: a possible link between existence of DNMT3L and genomic imprinting. *Cytogenet. Genome Res.*, **113**, 75-80.
- Yokomine, T., Kuroiwa, A., Tanaka, K., Tsudzuki, M., Matsuda, Y. and Sasaki, H. (2001) Sequence polymorphisms, allelic expression status and chromosome locations of the chicken IGF2 and MPR1 genes. *Cytogenet. Cell Genet.*, **93**, 109-113.

- Yokomine, T., Shirohzu, H., Purbowasito, W., Toyoda, A., Iwama, H., Ikeo, K., Hori, T., Mizuno, S., Tsudzuki, M., Matsuda, Y., et al (2005) Structural and functional analysis of a 0.5-Mb chicken region orthologous to the imprinted mammalian Ascl2/Mash2-Igf2-H19 region. *Genome Res.*, **15**, 154-165.
- Yoo-Warren, H., Pachnis, V., Ingram, R.S. and Tilghman, S.M. (1988) Two regulatory domains flank the mouse H19 gene. *Mol. Cell. Biol.*, **8**, 4707-4715.
- Youngson, N.A., Kocialkowski, S., Peel, N. and Ferguson-Smith, A.C. (2005) A small family of sushi-class retrotransposon-derived genes in mammals and their relation to genomic imprinting. *J. Mol. Evol.*, **61**, 481-490.
- Yusufzai, T.M. and Felsenfeld, G. (2004) The 5'-HS4 chicken beta-globin insulator is a CTCF-dependent nuclear matrix-associated element. *Proc. Natl. Acad. Sci. U. S. A.*, **101**, 8620-8624.
- Zaratiegui, M., Irvine, D.V. and Martienssen, R.A. (2007) Noncoding RNAs and gene silencing. *Cell*, **128**, 763-776.
- Zeng, Y., Yi, R. and Cullen, B.R. (2005) Recognition and cleavage of primary microRNA precursors by the nuclear processing enzyme Drosha. *EMBO J.*, **24**, 138-148.
- Zhao, Y. and Srivastava, D. (2007) A developmental view of microRNA function. *Trends Biochem. Sci.*, **32**, 189-197.
- Zubair, M., Hilton, K., Saam, J.R., Surani, M.A., Tilghman, S.M. and Sasaki, H. (1997) Structure and expression of the mouse L23mrp gene downstream of the imprinted H19 gene: biallelic expression and lack of interaction with the H19 enhancers. *Genomics*, **45**, 290-296.