

BIBLIOGRAPHY

- Angeli CB, Capelli LP, Auricchio MT, Leal-Mesquita ER, Ribeiro-dos-Santos AK, Ferrari I, Oliveira SF, Klautau-Guimaraes Mde N, Vianna-Morgante AM, Mingroni-Netto RC. 2005. AGG interspersion patterns in the CGG repeat of the FMR1 gene and linked DDX548/FRA-XAC1 haplotypes in Brazilian populations. *Am J Med Genet* 132: 210–4.
- Arinami T, Asano M, Kobayashi K, Yanagi H, Hamaguchi H. 1993. Data on the CGG repeat at the fragile X site in the non-retarded Japanese population and family suggest the presence of a subgroup of normal alleles predisposing to mutate. *Hum Genet* 92: 431–6.
- Arrieta I, Penagarikano O, Telez M, Ortega B, Flores P, Criado B, Veiga I, Peixoto AL, Lostao CM. 2003. The FMR1 CGG repeat and linked microsatellite markers in two Basque valleys. *Heredity* 90: 206–11.
- Ashley CT, Sutcliffe JS, Kunst CB, Leiner HA, Eichler EE, Nelson DL, Warren ST. 1993. Human and murine FMR-1: alternative splicing and translational initiation downstream of the CGG-repeat. *Nat Genet* 4: 244–51.
- Bell MV, Hirst MC, Nakahori Y, MacKinnon RN, Roche A, Flint TJ, Jacobs PA, Tommerup N, Tranebjærg L, Froster-Iskenius U, et al. 1991. Physical mapping across the fragile X: hypermethylation and clinical expression of the fragile X syndrome. *Cell* 64: 861–6.
- Berry-Kravis E, Grossman AW, Crnic LS, Greenough WT. 2002. Understanding fragile X syndrome. *Current Pediatrics* 12: 316–324.
- Bonaventure G, Torrado M, Barreiro C, Chertkoff L. 1998. Fragile X founder effects in Argentina. *Am J Med Genet* 79: 200–4.
- Bontekoe CJ, Bakker CE, Nieuwenhuizen IM, van der Linde H, Lans H, de Lange D, Hirst MC, Oostra BA. 2001. Instability of a (CGG)₉₈ repeat in the Fmr1 promoter. *Hum Mol Genet* 10: 1693–9.
- Boycott KM, Halley GR, Schlessinger D, Bech-Hansen NT. 1996. A 2-megabase physical contig incorporating 43 DNA markers on the human X chromosome at p11.23-p11.22 from ZNF21 to DDX255. *Genomics* 33: 488–97.

- Brightwell G, Wycherley R, Waghorn A. 2002a. SNP genotyping using a simple and rapid single-tube modification of ARMS illustrated by analysis of 6 SNPs in a population of males with FRAXA repeat expansions. *Mol Cell Probes* 16: 297–305.
- Brightwell G, Wycherley R, Potts G, Waghorn A. 2002b. A high-density SNP map for the FRAX region of the X chromosome. Single-nucleotide polymorphisms. *J Hum Genet* 47: 567–75.
- Brown WT, Houck GE Jr, Jezirowska A, Levinson FN, Ding X, Dobkin C, Zhong N, Henderson J, Brooks SS, Jenkins EC. 1993. Rapid fragile X carrier screening and prenatal diagnosis using a nonradioactive PCR test. *JAMA* 270: 1569–75.
- Brown WT. 1996a. The molecular biology of the fragile X mutation. In: Hagerman RJ, Cronister A, editor. *Fragile X syndrome: diagnosis, treatment, and research*, 2nd edition. Baltimore: Johns Hopkins University Press. p88–113.
- Brown WT, Zhong N, Dobkin C. 1996b. Positive fragile X microsatellite associations point to a common mechanism of dynamic mutation evolution. *Am J Hum Genet* 58: 641–3.
- Buyle S, Reyniers E, Vits L, De Boulle K, Handig I, Wuyts FL, Deelen W, Halley DJ, Oostra BA, Willems PJ. 1993. Founder effect in a Belgian–Dutch fragile X population. *Hum Genet* 92: 269–72.
- Cardon LR, Abecasis GR. 2003. Using haplotype blocks to map human complex trait loci. *Trends Genet* 19: 135–40.
- Cargill M, Altshuler D, Ireland J, Sklar P, Ardlie K, Patil N, Shaw N, Lane CR, Lim EP, Kalyanaraman N, Nemesh J, Ziaugra L, Friedland L, Rolfe A, Warrington J, Lipshutz R, Daley GQ, Lander ES. 1999. Characterization of single-nucleotide polymorphisms in coding regions of human genes. *Nat Genet* 22: 231–8.
- Chen SH, Schoof JM, Buroker NE, Scott CR. 1997. The identification of a (CGG)6AGG insertion within the CGG repeat of the FMR1 gene in Asians. *Hum Genet* 99: 793–5.
- Chiurazzi P, Macpherson J, Sherman S, Neri G. 1996a. Significance of linkage disequilibrium between the fragile X locus and its flanking markers. *Am J Med Genet* 64: 203–8.

- Chiurazzi P, Genuardi M, Kozak L, Giovannucci-Uzielli ML, Bussani C, Dagna-Bricarelli F, Grasso M, Perroni L, Sebastio G, Sperandeo MP, Oostra BA, Neri G. 1996b. Fragile X founder chromosomes in Italy: a few initial events and possible explanation for their heterogeneity. *Am J Med Genet* 64:209–15.
- Chiurazzi P, Destro-Bisol G, Genuardi M, Oostra BA, Spedini G, Neri G. 1996c. Extended gene diversity at the FMR1 locus and neighbouring CA repeats in a sub-Saharan population. *Am J Med Genet* 64: 216–9.
- Chiurazzi P, Pomponi MG, Sharrock A, Macpherson J, Lormeau S, Morel ML, Rousseau F. 1999. DNA panel for interlaboratory standardization of haplotype studies on the fragile X syndrome and proposal for a new allele nomenclature. *Am J Med Genet* 83: 347–9.
- Cleary JD, Nichol K, Wang YH, Pearson CE. 2002. Evidence of cis-acting factors in replication-mediated trinucleotide repeat instability in primate cells. *Nat Genet* 31: 37–46.
- Crawford DC, Schwartz CE, Meadows KL, Newman JL, Taft LF, Gunter C, Brown WT, Carpenter NJ, Howard-Peebles PN, Monaghan KG, Nolin SL, Reiss AL, Feldman GL, Rohlfs EM, Warren ST, Sherman SL. 2000a. Survey of the fragile X syndrome CGG repeat and the short-tandem-repeat and single-nucleotide-polymorphism haplotypes in an African American population. *Am J Hum Genet* 66: 480–93.
- Crawford DC, Zhang F, Wilson B, Warren ST, Sherman SL. 2000b. Fragile X CGG repeat structures among African-Americans: identification of a novel factor responsible for repeat instability. *Hum Mol Genet* 9: 1759–69.
- Cummings CJ, Zoghbi HY. 2000. Trinucleotide repeats: mechanisms and pathophysiology. *Annu Rev Genomics Hum Genet* 1: 281–328.
- Curlis Y, Zhang C, Holden JJ, Loesch PK, Mitchell RJ. 2005. Haplotype study of intermediate-length alleles at the fragile X (FMR1) gene: ATL1, FMRb, and microsatellite haplotypes differ from those found in common-size FMR1 alleles. *Hum Biol* 77: 137–51.
- de Diego Y, Hmadcha A, Moron F, Lucas M, Carrasco M, Pintado E. 2002. Fragile X founder effect and distribution of CGG repeats among the mentally retarded population of Andalusia, South Spain. *Genet Mol Bio* 25: 1–6.

- Dombrowski C, Levesque S, Morel ML, Rouillard P, Morgan K, Rousseau F. 2002. Premutation and intermediate-size FMR1 alleles in 10572 males from the general population: loss of an AGG interruption is a late event in the generation of fragile X syndrome alleles. *Hum Mol Genet* 11: 371–8.
- Eichler EE, Richards S, Gibbs RA, Nelson DL. 1993. Fine structure of the human FMR1 gene. *Hum Mol Genet* 2: 1147–53.
- Eichler EE, Holden JJ, Popovich BW, Reiss AL, Snow K, Thibodeau SN, Richards CS, Ward PA, Nelson DL. 1994. Length of uninterrupted CGG repeats determines instability in the FMR1 gene. *Nat Genet* 8: 88–94.
- Eichler EE, Hammond HA, Macpherson JN, Ward PA, Nelson DL. 1995a. Population survey of the human FMR1 CGG repeat substructure suggests biased polarity for the loss of AGG interruptions. *Hum Mol Genet* 4: 2199–208.
- Eichler EE, Kunst CB, Lugenbeel KA, Ryder OA, Davison D, Warren ST, Nelson DL. 1995b. Evolution of the cryptic FMR1 CGG repeat. *Nat Genet* 11:301–8.
- Eichler EE, Macpherson JN, Murray A, Jacobs PA, Chakravarti A, Nelson DL. 1996a. Haplotype and interspersion analysis of the FMR1 CGG repeat identifies two different mutational pathways for the origin of the fragile X syndrome. *Hum Mol Genet* 5: 319–30.
- Eichler EE, Nelson DL. 1996b. Genetic variation and evolutionary stability of the FMR1 CGG repeat in six closed human populations. *Am J Med Genet* 64:220–5.
- Ennis S, Murray A, Morton NE. 2001. Haplotypic determinants of instability in the FRAX region: Concatenated mutation or founder effect? *Hum Mutat* 18: 61–9.
- Falik-Zaccai TC, Shachak E, Yalon M, Lis Z, Borochowitz Z, Macpherson JN, Nelson DL, Eichler EE. 1997. Predisposition to the fragile X syndrome in Jews of Tunisian descent is due to the absence of AGG interruptions on a rare Mediterranean haplotype. *Am J Hum Genet* 60: 103–12.
- Faradz SM, Pattiha MZ, Leigh DA, Jenkins M, Leggo J, Buckley MF, Holden JJ. 2000. Genetic diversity at the FMR1 locus in the Indonesian population. *Ann Hum Genet* 64: 329–39.
- Faradz SM, Leggo J, Murray A, Lam-Po-Tang PR, Buckley MF, Holden JJ. 2001. Distribution of FMR1 and FMR2 alleles in Javanese individuals with developmental disability and confirmation of a specific AGG-interruption pattern in Asian populations. *Ann Hum Genet* 65: 127–35.

- Fu YH, Kuhl DP, Pizzuti A, Pieretti M, Sutcliffe JS, Richards S, Verkerk AJ, Holden JJ, Fenwick RG Jr, Warren ST, et al. 1991. Variation of the CGG repeat at the fragile X site results in genetic instability: resolution of the Sherman paradox. *Cell* 67: 1047–58.
- Garcia Arocena D, Breece KE, Hagerman PJ. 2003. Distribution of CGG repeat sizes within the fragile X mental retardation 1 (FMR1) homologue in a non-human primate population. *Hum Genet* 113: 371–6.
- Gunter C, Warren ST. 1998a. Polymorphism in the FMR1 gene. *Hum Genet* 103:365–6.
- Gunter C, Paradee W, Crawford DC, Meadows KA, Newman J, Kunst CB, Nelson DL, Schwartz C, Murray A, Macpherson JN, Sherman SL, Warren ST. 1998b. Re-examination of factors associated with expansion of CGG repeats using a single nucleotide polymorphism in FMR1. *Hum Mol Genet* 7: 1935–46.
- Haataja R, Vaisanen ML, Li M, Ryyynanen M, Leisti J. 1994. The fragile X syndrome in Finland: demonstration of a founder effect by analysis of microsatellite haplotypes. *Hum Genet* 94: 479–83.
- Halushka MK, Fan JB, Bentley K, Hsie L, Shen N, Weder A, Cooper R, Lipshutz R, Chakravarti A. 1999. Patterns of single-nucleotide polymorphisms in candidate genes for blood-pressure homeostasis. *Nat Genet* 22: 239–47.
- Hamajima N. 2001. PCR with confronting two-pair primers: a new method for rapid genotyping. *Qiagen News* Issue No 4 Aug.
- Hammond LS, Macias MM, Tarleton JC, Shashidhar Pai G. 1997. Fragile X syndrome and deletions in FMR1: new case and review of the literature. *Am J Med Genet* 72: 430–4.
- Hastbacka J, de la Chapelle A, Kaitila I, Sistonen P, Weaver A, Lander E. 1992. Linkage disequilibrium mapping in isolated founder populations: diastrophic dysplasia in Finland. *Nat Genet* 2: 204–11.
- Heitz D, Rousseau F, Devys D, Saccone S, Abderrahim H, Le Paslier D, Cohen D, Vincent 1A, Toniolo D, Della Valle G, et al. 1991. Isolation of sequences that span the fragile X and identification of a fragile X-related CpG island. *Science* 251: 1236–9.
- Heitz D, Devys D, Imbert G, Kretz C, Mandel JL. 1992. Inheritance of the fragile X syndrome: size of the fragile X premutation is a major determinant of the transition to full mutation. *J Med Genet* 29: 794–801.

- Hirst MC, Knight SJ, Christodoulou Z, Grewal PK, Fryns JP, Davies KE. 1993. Origins of the fragile X syndrome mutation. *J Med Genet* 30: 647–50.
- Hirst MC, Grewal PK, Davies KE. 1994. Precursor arrays for triplet repeat expansion at the fragile X locus. *Hum Mol Genet* 3: 1553–60.
- Hirst MC, Arinami T, Laird CD. 1997. Sequence analysis of long FMR1 arrays in the Japanese population: insights into the generation of long (CGG)_n tracts. *Hum Genet* 101: 214–8.
- Huggins RM, Loesch DZ, Qian GQ, Bui QM, Mitchell RJ, Dobson M, Taylor AK. 2004. Hierarchical Bayes model for random haplotype and family effects in the transmission of fragile-X. *Genet Epidemiol* 26 : 294–304.
- Jacobs PA, Bullman H, Macpherson J, Youings S, Rooney V, Watson A, Dennis NR. 1993. Population studies of the fragile X: a molecular approach. *J Med Genet* 30: 454–9.
- Jara L, Aspíllaga M, Avendano I, Obreque V, Blanco R, Valenzuela CY. 1998. Distribution of (CGG)_n and FMR-1 associated microsatellite alleles in a normal Chilean population. *Am J Med Genet* 75: 277–82.
- Jiang Z, Shi J, Yang C, Jiang H, Chen Z, Jin L, Lu D, Huang W. 2001. A simple and rapid method for SNP typing by single tube bi-directional allele specific amplification. *Zhonghua Yi Xue Za Zhi* 18: 306–9.
- Jin P, Warren ST. 2000. Understanding the molecular basis of fragile X syndrome. *Hum Mol Genet* 9: 901–8.
- Kremer EJ, Pritchard M, Lynch M, Yu S, Holman K, Baker E, Warren ST, Schlessinger D, Sutherland GR, Richards RI. 1991. Mapping of DNA instability at the fragile X to a trinucleotide repeat sequence p(CCG)_n. *Science* 252: 1711–4.
- Kunst CB, Warren ST. 1994. Cryptic and polar variation of the fragile X repeat could result in predisposing normal alleles. *Cell* 77: 853–61.
- Kunst CB, Zerynick C, Karickhoff L, Eichler E, Bullard J, Chalifoux M, Holden JJ, Torroni A, Nelson DL, Warren ST. 1996. FMR1 in global populations. *Am J Hum Genet* 58: 513–22.

- Larsen LA, Armstrong JS, Gronskov K, Hjalgrim H, Brondum-Nielsen K, Hasholt L, Norgaard-Pedersen B, Vuust J. 1999. Analysis of FMR1 (CGG)_n alleles and FRAXA microsatellite haplotypes in the population of Greenland: implications for the population of the New World from Asia. *Eur J Hum Genet* 7: 771–7.
- Larsen LA, Armstrong JS, Gronskov K, Hjalgrim H, Macpherson JN, Brondum-Nielsen K, Hasholt L, Norgaard-Pedersen B, Vuust J. 2000. Haplotype and AGG-interspersion analysis of FMR1 (CGG)(n) alleles in the Danish population: implications for multiple mutational pathways towards fragile X alleles. *Am J Med Genet* 93: 99–106.
- Limprasert P, Nouri N, Heyman RA, Nopparatana C, Kamonsilp M, Deininger PL, Keats BJ. 1996. Analysis of CAG repeat of the Machado-Joseph gene in human, chimpanzee and monkey populations: a variant nucleotide is associated with the number of CAG repeats. *Hum Mol Genet* 5: 207–13.
- Limprasert P, Nouri N, Nopparatana C, Deininger PL, Keats BJ. 1997. Comparative studies of the CAG repeats in the spinocerebellar ataxia type 1 (SCA1) gene. *Am J Med Genet* 74: 488–93.
- Limprasert P, Ruangdaraganon N, Sura T, Vasiknanonte P, Jinorose U. 1999. Molecular screening for fragile X syndrome in Thailand. *Southeast Asian J Trop Med Public Health* 30 Suppl 2: 114–8.
- Limprasert P, Saechan V, Ruangdaraganon N, Sura T, Vasiknanote P, Jaruratasirikul S, Brown WT. 2001. Haplotype analysis at the FRAXA locus in Thai subjects. *Am J Med Genet* 98: 224–9.
- Macpherson JN, Bullman H, Youings SA, Jacobs PA. 1994. Insert size and flanking haplotype in fragile X and normal populations: possible multiple origins for the fragile X mutation. *Hum Mol Genet* 3: 399–405.
- Maddalena A, Richards CS, McGinniss MJ, Brothman A, Desnick RJ, Grier RE, Hirsch B, Jacky P, McDowell GA, Popovich B, Watson M, Wolff DJ. 2001. Technical standards and guidelines for fragile X: the first of a series of disease-specific supplements to the Standards and Guidelines for Clinical Genetics Laboratories of the American College of Medical Genetics. Quality Assurance Subcommittee of the Laboratory Practice Committee. *Genet Med* 3: 200–5.

- Mathews DJ, Kashuk C, Brightwell G, Eichler EE, Chakravarti A. 2001. Sequence variation within the fragile X locus. *Genome Res* 11: 1382–91.
- Mingroni-Netto RC, Costa SS, Angeli CB, Vianna-Morgante AM. 1999. DXS548/FRA(X)C1 haplotypes in fragile X chromosomes in the Brazilian population. *Am J Med Genet* 84: 204–7.
- Mirkin SM. 2006. DNA structures, repeat expansions and human hereditary disorders. *Curr Opin Struct Biol* 16: 351–8.
- Mogk RL, Carson NL, Chudley AE, Dawson AJ. 1998. Transmission of the FRA(X)A haplotype from three nonpenetrant brothers to their affected grandsons: an update with AGG interspersion analysis. *Am J Med Genet* 75: 28–34.
- Morton JE, Bunney S, Webb TP, MacDonald F, Rindl PM, Bullock S. 1997. Fragile X syndrome is less common than previously estimated. *J Med Genet* 34: 1–5.
- Murray A, Youings S, Dennis N, Latsky L, Linehan P, McKechnie N, Macpherson J, Pound M, Jacobs P. 1996. Population screening at the FRA(X)A and FRA(X)E loci: molecular analyses of boys with learning difficulties and their mothers. *Hum Mol Genet* 5: 727–35.
- Murray A, Macpherson JN, Pound MC, Sharrock A, Youings SA, Dennis NR, McKechnie N, Linehan P, Morton NE, Jacobs PA. 1997. The role of size, sequence and haplotype in the stability of FRA(X)A and FRA(X)E alleles during transmission. *Hum Mol Genet* 6: 173–84.
- Napierala M, Michalowski D, de Mezer M, Krzyzosiak WJ. 2005. Facile FMR1 mRNA structure regulation by interruptions in CGG repeats. *Nucleic Acids Res* 33: 451–63.
- Newton CR, Graham A, Heptinstall LE, Powell SJ, Summers C, Kalsheker N, Smith JC, Markham AF. 1989. Analysis of any point mutation in DNA. The amplification refractory mutation system. (ARMS). *Nucleic Acid Research* 17: 2503–16.
- Nolin SL, Lewis FA, Ye LL, Houck GE Jr, Glicksman AE, Limprasert P, Li SY, Zhong N, Ashley AE, Feingold E, Sherman SL, Brown WT. 1996. Familial transmission of the FMR1 CGG repeat. *Am J Hum Genet* 59: 1252–61.

- Nolin SL, Brown WT, Glicksman A, Houck GE Jr, Gargano AD, Sullivan A, Biancalana V, Brondum-Nielsen K, Hjalgrim H, Holinski-Feder E, Kooy F, Longshore J, Macpherson J, Mandel JL, Matthijs G, Rousseau F, Steinbach P, Vaisanen ML, von Koskull H, Sherman SL. 2003. Expansion of the fragile X CGG repeat in females with premutation or intermediate alleles. *Am J Hum Genet* 72: 454–64.
- Oberle I, Rousseau F, Heitz D, Kretz C, Devys D, Hanauer A, Boue J, Bertheas MF, Mandel JL. 1991. Instability of a 550-base pair DNA segment and abnormal methylation in fragile X syndrome. *Science* 252: 1097–102.
- Oostra BA, Chiurazzi P. 2001. The fragile X gene and its function. *Clin Genet* 60: 399–408.
- Orrico A, Galli L, Dotti MT, Plewnia K, Censini S, Federico A. 1998. Mosaicism for full mutation and normal-sized allele of the FMR1 gene: a new case. *Am J Med Genet* 78: 341–4.
- Oudet C, Mornet E, Serre JL, Thomas F, Lentes-Zengerling S, Kretz C, Deluchat C, Tejada I, Boue J, Boue A, et al. 1993. Linkage disequilibrium between the fragile X mutation and two closely linked CA repeats suggests that fragile X chromosomes are derived from a small number of founder chromosomes. *Am J Hum Genet* 52: 297–304.
- Pekarik V, Blazkova M, Kozak L. 1999. Haplotype analysis of the fragile X syndrome gene FMR1 in the Czech Republic. *Am J Med Genet* 84: 214–6.
- Penagarikano O, Gil A, Telez M, Ortega B, Flores P, Veiga I, Peixoto A, Criado B, Arrieta I. 2004. A new insight into fragile X syndrome among Basque population. *Am J Med Genet* 128: 250–5.
- Pesso R, Barkai G, Ravia Y, Gak E, Frydman M, Goldman B, Friedman E. 1997. No founder effect detected in Jewish Ashkenazi patients with fragile-X syndrome. *Hum Genet* 101: 186–9.
- Petronis A. 1996. Genomic imprinting in unstable DNA diseases. *Bioessays* 18: 587–90.
- Pieretti M, Zhang FP, Fu YH, Warren ST, Oostra BA, Caskey CT, Nelson DL. 1991. Absence of expression of the FMR-1 gene in fragile X syndrome. *Cell* 66: 817–22.
- Poon PM, Pang CP, Chen QL, Zhong N, Lai KY, Lau CH, Wong CK, Brown WT. 1999. FRAXAC1 and DXS548 polymorphisms in the Chinese population. *Am J Med Genet* 84: 208–13.

- Rerkamnuaychoke B, Thanakitgosate J, Purisa W. 1998. Length polymorphism of the CGG repeats in FMR1 gene in normal Thais. *Rama Med J* 21: 79–83.
- Reyniers E, Vits L, De Boulle K, Van Roy B, Van Velzen D, de Graaff E, Verkerk AJ, Jorens HZ, Darby JK, Oostra B, et al. 1993. The full mutation in the FMR-1 gene of male fragile X patients is absent in their sperm. *Nat Genet* 4: 143–6.
- Richards RI, Holman K, Kozman H, Kremer E, Lynch M, Pritchard M, Yu S, Mulley J, Sutherland GR. 1991. Fragile X syndrome: genetic localisation by linkage mapping of two microsatellite repeats FRAXAC1 and FRAXAC2 which immediately flank the fragile site. *J Med Genet* 28: 818–23.
- Richards RI, Holman K, Friend K, Kremer E, Hillen D, Staples A, Brown WT, Goonewardena P, Tarleton J, Schwartz C, et al. 1992. Evidence of founder chromosomes in fragile X syndrome. *Nat Genet* 1: 257–60.
- Richards RI, Kondo I, Holman K, Yamauchi M, Seki N, Kishi K, Staples A, Sutherland GR, Hori T. 1994a. Haplotype analysis at the FRAXA locus in the Japanese population. *Am J Med Genet* 51: 412–6.
- Richards RI, Holman K, Friend K, Staples A, Sutherland GR, Oudet C, Biancalana V, Mandel JL. 1994b. FRAXAC2 instability. *Nat Genet* 7:122
- Richards RI, Crawford J, Narahara K, Mangelsdorf M, Friend K, Staples A, Denton M, Easteal S, Hori TA, Kondo I, Jenkins T, Goldman A, Panich V, Ferakova E, Sutherland GR. 1996. Dynamic mutation loci: allele distributions in different populations. *Ann Hum Genet* 60: 391–400.
- Richards RI, Sutherland GR. 1997. Dynamic mutation: possible mechanisms and significance in human disease. *Trends Biochem Sci* 22: 432–6.
- Riggins GJ, Sherman SL, Oostra BA, Sutcliffe JS, Feitell D, Nelson DL, van Oost BA, Smits AP, Ramos FJ, Pfendner E, et al. 1992. Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. *Am J Med Genet* 43: 237–43.
- Rubinstein DC, Leggo J, Coetzee GA, Irvine RA, Buckley M, Ferguson-Smith MA. 1995. Sequence variation and size ranges of CAG repeats in the Machado–Joseph disease, spinocerebellar atrophy type 1 and androgen receptor genes. *Hum Mol Genet* 4:1585–90.
- Saul RA, Tarleton JC. 2004. Fragile X syndrome. www.genetests.org

- Schaeffer C, Beaulande M, Ehresmann C, Ehresmann B, Moine H. 2003. The RNA binding protein FMRP: new connections and missing links. *Biol Cell* 95: 221–8.
- Schmucker B, Seidel J. 1999. Mosaicism for a full mutation and a normal size allele in two fragile X males. *Am J Med Genet* 84: 221–5.
- Sharma D, Gupta M, Thelma BK. 2003. FMR1 haplotype analyses among Indians: a weak founder effect and other findings. *Hum Genet* 112: 262–71.
- Siomi H, Siomi MC, Nussbaum RL, Dreyfuss G. 1993. The protein product of the fragile X gene, FMR1, has characteristics of an RNA-binding protein. *Cell* 74: 291–8.
- Siomi H, Choi M, Siomi MC, Nussbaum RL, Dreyfuss G. 1994. Essential role for KH domains in RNA binding: impaired RNA binding by a mutation in the KH domain of FMR1 that causes fragile X syndrome. *Cell* 77: 33–9.
- Snow K, Doud LK, Hagerman R, Pergolizzi RG, Erster SH, Thibodeau SN. 1993. Analysis of a CGG sequence at the FMR-1 locus in fragile X families and in the general population. *Am J Hum Genet* 53: 1217–28.
- Snow K, Tester DJ, Kruckeberg KE, Schaid DJ, Thibodeau SN. 1994. Sequence analysis of the fragile X trinucleotide repeat: implications for the origin of the fragile X mutation. *Hum Mol Genet* 3: 1543–51.
- Sturzeneker R, Haddad LA, Bevilacqua RA, Simpson AJ, Pena SD. 1998. Polarity of mutations in tumor-associated microsatellite instability. *Hum Genet* 102: 231–5.
- Syrrou M, Patsalis PC, Georgiou I, Hadjimarcou MI, Constantinou-Deltas CD, Pagoulatos G. 1996. Evidence for high-risk haplotypes and (CGG)_n expansion in fragile X syndrome in the Hellenic population of Greece and Cyprus. *Am J Med Genet* 64: 234–8.
- Taillon-Miller P, Kwok PY. 2000. A high-density single-nucleotide polymorphism map of Xq25–q28. *Genomics* 65: 195–202.
- Tarleton J, Kenneson A, Taylor AK, Crandall K, Fletcher R, Casey R, Hart PS, Hatton D, Fisch G, Warren ST. 2002. A single base alteration in the CGG repeat region of FMR1: possible effects on gene expression and phenotype. *J Med Genet* 39: 196–200.
- The International HapMap Consortium. 2003. The International HapMap Project. *Nature* 426: 789–96.

- Turner AM, Robinson H, Wake S, Laing SJ, Leigh D, Turner G. 1994. Counselling risk figures for fragile X carrier females of varying band sizes for use in predicting the likelihood of retardation in their offspring. *Am J Med Genet* 51: 458–62.
- Turner G, Webb T, Wake S, Robinson H. 1996. Prevalence of fragile X syndrome. *Am J Med Genet* 64: 196–7.
- Tzeng CC, Tsai LP, Hwu WL, Lin SJ, Chao MC, Jong YJ, Chu SY, Chao WC, Lu CL. 2005. Prevalence of the FMR1 mutation in Taiwan assessed by large-scale screening of newborn boys and analysis of DXS548–FRAXAC1 haplotype. *Am J Med Genet* 133: 37–43.
- Verkerk AJ, Pieretti M, Sutcliffe JS, Fu YH, Kuhl DP, Pizzuti A, Reiner O, Richards S, Victoria MF, Zhang FP, et al. 1991. Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. *Cell* 65: 905–14.
- Vincent JB, Gurling HM. 1998. Point mutation in intron 10 of FMR1 is unlikely to be a cause of fragile X syndrome. *Hum Mutat* 12: 431–2.
- Vincent JB, Thevarkunnel S, Kolozsvari D, Paterson AD, Roberts W, Scherer SW. 2004. Association and transmission analysis of the FMR1 IVS10 + 14C-T variant in autism. *Am J Med Genet B Neuropsychiatr Genet* 125: 54–6.
- Wang YC, Lin ML, Lin SJ, Li YC, Li SY. 1997. Novel point mutation within intron 10 of FMR-1 gene causing fragile X syndrome. *Hum Mutat* 10: 393–9.
- Wang DG, Fan JB, Siao CJ, Berno A, Young P, Sapolsky R, Ghandour G, Perkins N, Winchester E, Spencer J, Kruglyak L, Stein L, Hsie L, Topaloglu T, Hubbell E, Robinson E, Mittmann M, Morris MS, Shen N, Kilburn D, Rioux J, Nusbaum C, Rozen S, Hudson TJ, Lander ES, et al. 1998. Large-scale identification, mapping, and genotyping of single-nucleotide polymorphisms in the human genome. *Science* 280: 1077–82.
- Weisman-Shomer P, Cohen E, Fry M. 2000. Interruption of the fragile X syndrome expanded sequence d(CGG)(n) by interspersed d(AGG) trinucleotides diminishes the formation and stability of d(CGG)(n) tetrahelical structures. *Nucleic Acids Res* 28: 1535–41.

- Xu B, School JM, Buroker NE, Scott CR, Chen SH. 1999. High frequency of the FMR1 IVS10+14C/T polymorphism in Asians, and its association with the fragile X syndrome in Caucasians. *Am J Hum Genet* 65(Suppl): A403 (abstract).
- Ye S, Dhillon S, Ke X, Collins AR, Day IN. 2001. An efficient procedure for genotyping single nucleotide polymorphisms. *Nucleic Acid Research* 29: E88–8.
- Zhong N, Dobkin C, Brown WT. 1993. A complex mutable polymorphism located within the fragile X gene. *Nat Genet* 5: 248–53.
- Zhong N, Ye L, Dobkin C, Brown WT. 1994a. Fragile X founder chromosome effects: linkage disequilibrium or microsatellite heterogeneity? *Am J Med Genet* 51: 405–11.
- Zhong N, Liu X, Gou S, Houck GE Jr, Li S, Dobkin C, Brown WT. 1994b. Distribution of FMR-1 and associated microsatellite alleles in a normal Chinese population. *Am J Med Genet* 51: 417–22.
- Zhong N, Yang W, Dobkin C, Brown WT. 1995. Fragile X gene instability: anchoring AGGs and linked microsatellites. *Am J Hum Genet* 57: 351–61.
- Zhong N, Kajanoja E, Smits B, Pietrofesa J, Curley D, Wang D, Ju W, Nolin S, Dobkin C, Ryynanen M, Brown WT. 1996a. Fragile X founder effects and new mutations in Finland. *Am J Med Genet* 64: 226–33.
- Zhong N, Ju W, Pietrofesa J, Wang D, Dobkin C, Brown WT. 1996b. Fragile X "gray zone" alleles: AGG patterns, expansion risks, and associated haplotypes. *Am J Med Genet* 64: 261–5.
- Zhong N, Ju W, Xu W, Ye L, Shen Y, Wu G, Chen SH, Jin R, Hu XF, Yang A, Liu X, Poon P, Pang C, Zheng Y, Song L, Zhao P, Fu B, Gu H, Brown WT. 1999. Frequency of the fragile X syndrome in Chinese mentally retarded populations is similar to that in Caucasians. *Am J Med Genet* 84: 191–4.
- Zhou Y, Tang K, Law HY, Ng IS, Lee CG, Chong SS. 2006. FMR1 CGG Repeat Patterns and Flanking Haplotypes in Three Asian Populations and Their Relationship With Repeat Instability. *Ann Hum Genet* 70: 784–96.