1056 Letters, Book review

- Boughman JA, Berg KA, Astemborski JA, et al. Familial risks of congenital heart defect assessed in population-based epidemiologic study. Am J Med Genet 1987;26:839-49.
   Weigel TJ, Driscoll DH, Michels VV. Occur-
- 2 Weigel TJ, Driscoll DH, Michels VV. Occurrence of congenital heart defects in siblings of patients with univentricular heart and tricuspid atresia. Am J Cardiol 1989;64:768-71.
- 3 Grant JW. Congenital malformations of the tricuspid valve. *Pediatr Cardiol* 1996;17:327-9.
- 4 Nora JJ, Nora AH. The evolution of specific genetic and environmental counseling in congenital heart diseases. *Circulation* 1978;57:205-13.
- 5 Ferencz C, Loffredo CA, Correa-Villasenor A, Wilson PD. Genetic and environmental risk factors of major cardiovascular malformations. Perspect Pediatr Cardiol 1997;5:343.

## **BOOK REVIEW**

If you wish to order or require further information regarding the titles reviewed here, please write to or telephone the BMJ Bookshop, PO Box 295, London WC1H 9JR. Tel 0171 383 6244. Fax 0171 383 6662. Books are supplied post free in the UK and for BFPO addresses. Overseas customers should add 15% for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name. (The price and availability are occasionally subject to revision by the Publishers.)

The Science behind Jeans for Genes Day: Teaching Packs for Primary (Key Stage 2) and Secondary (Key Stage 4) Schools. M Pembrey. The Progress Educational Trust.

It is often suggested that if genetics teaching within the national curriculum could be focused on practical issues that could affect students themselves (such as genetic diagnosis and screening), this change in emphasis could help them to learn by involving their natural interest, and also prepare them for the impact of current genetic advances on their adult lives. However, teachers need help from expert geneticists if this is to come about.

Professor Marcus Pembrey has an unusual ability to communicate genetic concepts in imaginative ways, and has put his ability to work in creating these two teachers' packs, adapted to specific stages of the national curriculum. The packs are intended to support teachers in teaching genetics in a realistic way that will be useful for students in later life as well as supporting educational objectives. Each pack consists of (1) a resource book for teachers, the "Progress Guide to Genetics", and (2) a file of four case studies, which can be photocopied for the children: these are the same in both the primary and the secondary packs. There are also (3) teachers' notes, and (4) four worksheets, also to be photocopied: these differ between packs.

The materials are imaginative and attractive. For example, the worksheet for primary schools shows a picture of Professor Pembrey's own family, with clues for drawing up a family tree, the common symbols used, and hints to help young children learn the principles of pedigree drawing. One worksheet

explains the structure of DNA, the way it is packed into chromosomes, and the location of the chromosomes in the nucleus of the cell. Another shows some organs of the human body, challenges the students to place them in a body outline, and explains which of the organs can be affected by the genetic disorders outlined in the case histories. The teachers' notes include suggestions for games that can be used to illustrate, for example, the pairing principle that permits faithful replication of DNA. The secondary school pack uses the same basic resources to develop the basic concepts of genetics further. Subjects tackled include Mendelian inheritance and inheritance of sex, replication of DNA and the function of genes, inheritance of genetic disease, and genetic testing in the family and its uses. The teachers' pack includes information to assist teachers in discussing complex issues, such as prenatal diagnosis and other difficult choices.

The progress guide to genetics also illustrates Professor Pembrey's flair for simple, benign, and graphic illustration, and includes the images used on the worksheets. However, the text is not as clear and simple as the diagrams. It uses a conversational tone, which may be helpful for people who have no previous understanding of genetics, but does not quite match the simplicity and clarity of the worksheets.

This is an excellent resource, not only for teachers, but for nurses, genetic counsellors, and others who sometimes go into schools to help teach pupils about genetic issues.

BERNADETTE MODELL

## **VOLUME 35 • AUTHOR INDEX**

Aaltonen L see Olschwang S et al Abeliovich D see Robinson WP et al Abernathy CR see Rasmussen SA et al Adam MF see Stoilova D et al Adamski E see Griffin DK et al Ades L see Kirk E and Ades L Adès LC see Mowat DR et al Aderts JMFG see van Royen-Kerkhof A et al Affara NA see Slaney SF et al Agid Y see Dumanchin C et al Akarsu AN see Percin EF et al Akasaka Y see Kikuchi H et al Ala-Mello S et al. Molecular studies in Finnish patients with familial juvenile nephronophthisis exclude a founder effect and support a common mutation causing mechanism, 279

Al-Awadi SA see Tayel SM et al see Sabry MA et al
Albin RL. Fuch's corneal dystrophy in a patient with mitochondrial DNA mutations, 258
Albrechts JCM see Engelen JJM et al
see Plomp AS et al
Alderman JK see Hughes HE et al
Al-Gazali LI see Bonthron DT et al Ali JBM et al. Mutations in the TSC1 gene account for a minority of patients with tuberous sclerosis, 969
Ali M et al. Hereditary fructose intolerance: review, 353 Allanson JE et al. Classical lissencephaly syndromes: does the face reflect the brain?, 920 Allinson PS see Parsons DW et al Al-Maghtheh M see Inglehearn CF et al see Vithana E et al Al-Torki N see van Steensel M et al Alvarez AI et al. Molecular study of the rhodopsin gene in retinitis pigmentosa patients in the Basque Country, 387 Amati P see Longy M et al Amman G see Berger A et al Andrews RM see Chinnery PF et al Andria G see de Franchis R et al see Melis D et al Andrikovics H see Tordai A et al Anvret M see Xiang F et al Arason A et al. A population study of mutations and LOH at breast cancer gene loci in tumours from sister pairs: two recurrent mutations seem to account for all BRCA1/BRCA2 linked breast cancer in Iceland, 446 Arinami T see Kawashima T et al Arn PH see Rasmussen SA et al Arngrimsson R see Mossey PA et al Arostegui E see Alvarez AI et al Asher Jr JH see Carey ML et al Ashley S see Watson M et al Aubry M-C see Muller F et al Austin-Ward E et al. Neonatal lupus syndrome: a case with chondrodysplasia punctata and other unusual manifestations, Avizienyte E see Olschwang S et al Aylsworth AS see Du Y-Z et al Ayuso C see Espinós C et al see Kennan AM et al Azar ST see Mustapha M et al Baala L see Kabbaj K et al Badia I see Lapunzina P et al Baiget M see Bayés M et al see Espinos C et al

Baiget M. Frequency of the HFE C282Y and H63D mutations in distinct ethnic groups living in Spain: letter, 701

Baker JC see Precht KS et al Baker PN see Morgan L et al Balassopoulou A see Papaioannou M et al Balcells S see Bayés M et al Ballabio A see Jonsson JJ et al Ballesta F see Margarit E et al Banchs I see Pujana MA et al Bankier A see Hunter AGW et al see Peters HL and Bankier A see Savarirayan R and Bankier A

Barber JCK. "Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public". Advisory Committee on Genetic Testing, 443

Barber JCK et al. Duplication of 8p23.1: a cytogenetic anomaly with no established clinical significance, 491

Barbierato L see Tupler R et al Barhoumi C see Meggouh F et al Barkardottir RB see Arason A et al Barnes C see Skirton H et al

Barois A see Guicheney P et al Barr DGD see Bonthron DT et al Barsoum-Homsy M see Stoilova D et al Bartlett K see Ryan AK et al Bartoloni L see Speer MC et al Bartolon L see Special Ce et al.
Barwell KJ see Broom MF et al.
Bateman MS see Barber JCK et al.
Baumann P et al. Myotonia congenita in northern Finland: an epidemiological and genetic study, 293 Bayés M et al. A new autosomal recessive retinitis pigmentosa locus Bayes M et al. A new autosomal recessive retinitis pigmentos maps on chromosome 2q31-q33, 141

Becher H see Chang-Claude J et al

Beemer FA see Sjarif DR et al

see van Royen-Kerkhof A et al

Beerens CEMT see Verheijen FW et al

Beesley CE et al. Identification of 12 novel mutations in the I-acetylglucosaminidase gene in 14 patients with Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), 910

Bellingham J see Papaioannou M et al

Bellingham J, Gregory-Evans K. Microsatellite markers for the cone-rod retinal dystrophy gene, CRX, on 19q13.3: letter, 527

Belmont JW see Timms KM et al Benatar A see Seneca S et al Beneyto M see Espinos C et al Benito A see Combarros O et al Benomar A see Meggouh F et al Benton EC see Porteous MEM et al Bercau G see Portnoï M-F et al Berciano J see Combarros O et al Berger A et al. Neonatal cholestasis and focal medullary dysplasia of the kidneys in a case of microcephalic osteodysplastic primordial dwarfism, 61 Bergthorsson JT see Arason A et al Bermejo E see Martínez-Frías ML et al Bernasconi F see Robinson WP et al Bessant D see Papaioannou M et al Bessant DAR et al. Further refinement of the Usher 2A locus at Bessis R see Muller F et al Bhattacharya S see Papaioannou M et al Bhattacharya SS see Bessant DAR et al see Inglehearn CF et al see Inglehearn CF et al see Vithana E et al
see Votruba M et al
Bienvenu T see Muller F et al
Billam LJ see Long FL et al
Bird AC see Bessant DAR et al see Inglehearn CF et al see Inglehearn CF et al Birouk N see Meggouh F et al Black G see Wu CL et al Black GCM et al. Locus heterogeneity in autosomal dominant congenital external ophthalmoplegia (CFEOM), 985 Blanchet P see Verloes A et al Bloomfield J see Austin-Ward E et al Bobadilla L see Rivera H et al Bodmer W see Olschwang S et al Boner AL see Trabetti E et al Bonneau D see Longy M et al Bonthron DT et al. Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome, 288 Book reviews Bartel PL, Fields S, editors. The Yeast Two-Hybrid System, 704 Bodmer W. McKie R. The Book of Man: The Human Genome Project and the Quest to Discover our Genetic Heritage, 703 Bridge PJ. The Calculation of Genetic Risks: Worked Examples in DNA Diagnostics, 704 Brown TA. Genetics. A Molecular Approach, 704 Chiocca EA, Breakfield X, editors. Gene Therapy for Neurological Disorders and Brain Tumours, 615 Ciba Foundation Symposium. Oligonncleotides as Therapeutic Agents, 703 Clarke A, Parsons E, editors. Culture, Kinship and Genes., 439 Emery AEH, editor. Neuromuscular Disorders, 792
Friedberg EC. Correcting The Blueprint of Life. An Historical Account of the Discovery of DNA Repair Mechanisms, 264
Hogben A, Hogben A, editors. Lancelot Hogben Scientific Humanist. An unauthorised autobiography, 966 Hurtley SM, editor. Protein targeting
Kaufman S. Tetrahydrobiopterin - Basic Biochemistry and Role in Human Disease., 439 King RC, Stansfield WD, editors. A Dictionary of Genetics. 5th edition, 176
March KL, editor. Gene Transfer in the Cardiovascular System, 791

Chen C-P see Devriendt K and Chen C-P

Mueller RF, Young ID, editors. Emery's Elements of Medical Chen C-P et al. De novo unbalanced translocation resulting in monosomy for proximal 14q and distal 4p in a fetus with intrauterine growth retardation, Wolf-Hirschhorn syndrome Geneics, 792 Nicolini C, editor. Genome Structure and Function: From Chromohypertrophic cardiomyopathy, and partial hemihypoplasia, 1050 Kyphomelic dysplasia in two sib fetuses, 65 Chen M-H see Chen C-P et al Chen W-L see Chen C-P et al some Characterization to Gene Technology., 439 North K, Gutman D, Korf BR. Neurofibromatosis type 1 in childhood Pembrey M. The Science Behind Jeans for Genes Day: teaching packs for primary (key stage 2) and secondary (key stage 4) Chern S-R see Chen C-P et al see Chen C-P et al
Cherniske EM see Jonsson JJ et al
Chéry M see Sloan-Béna F et al
Chevrette L see Stoilova D et al schools, 1056 Platt LD, Koch R, de la Cruz F, editors. Genetic Disorders and Pregnancy Outcome, 702 Sessa A, Conte F, Meroni M, Battini G, editors. Hereditary Kidney Chhoul H see Kabbaj K et al Diseases, 440 Chia NL see Mowat DR et al Chierakul C see Georgiades P et al Child A see Stoilova D et al Chinnery PF et al. Mitochondrial DNA mutations and Wolman SR, Sell S, editors. Human Cytogenetic Cancer Markers, Boone RA see Horsley SW et al pathogenicity: letter, 701 Chitty LS see Slaney SF et al Borrone C see di Rocco M et al Bortotto L see Pirola B et al Bottani A see Longy M et al Bowden DW see Price JA et al Chivelet P see Bayés M et al Chkili T see Meggouh F et al
Choi K-L et al. Absence of PAX2 gene mutations in patients with
primary familial vesicoureteric reflux, 338 Bowman CM see Liang M-H et al Boyadjiev SA see Flanagan N et al Boyd E see Ghaffari SR et al Chotai KA, Payne SJ. A rapid, PCR based test for differential molecular diagnosis of Prader-Willi and Angelman syndromes, see Ghaffari SR et al Brady AF et al. Outcome of chromosomally normal livebirths with Christian SL see Jacobsen J et al increased fetal nuchal translucency at 10-14 weeks' gestation, see Precht KS et al
Christiano AM see Frank J et al
Chuang C-Y see Chen C-P et al
Cinti R see Concolino D et al 222 Brewer CM et al. Beckwith-Wiedemann syndrome in a child with chromosome 18q deletion, 162 Brice A see Dumanchin C et al Clark T see Kennan AM et al see Meggouh F et al Clarke A see Ferguson BM et al Brice G see Stoilova D et al Brink P see Moolman-Smook JC et al Brock DJH see Gilfillan A et al see Webb T et al see Xiang F et al
Claustres M see de Meeus A et al
Clayton P see Ryan AK et al
Clayton-Smith J see Black GCM et al see Hayward C et al Brookfield JFY see Morgan L et al Broom JE see Broom MF et al Broom MF et al. Ovine neuronal ceroid lipofuscinosis: a large animal Clerget-Darpoux F see Dumanchin C et al Cnossen MH see van Asperen CJ et al
Cnossen MH et al. Minor disease features in neurofibromatosis type
1 (NF1) and their possible value in diagnosis of NF1 in children

< 6 years and clinically suspected of having NF1, 624 model syntenic with the human neuronal ceroid lipofuscinosis variant CLN6, 717 Brunet J-S see Pal T et al Brunner HG see Cunliffe HE et al Cochrane S see Lalloo F et al Bruttini M see Jonsson JJ et al Bulman B see Lalloo F et al Cofré-Beca J see Austin-Ward E et al Cohen Jr MM et al. New overgrowth syndrome and FGFR3 dosage Bundey S see Hutchesson ACJ et al effect: letter, 348 Collins S see Evans DGR et al Buocompagni A see di Rocco M et al Buoninconti A see de Franchis R et al Collinson MN see Barber JCK et al Burghes AHM see Parsons DW et al Burn J see Chase DS et al Colman SD see Rasmussen SA et al Colomb-Brockmann C see Teebi AS et al
Colombel J-F see Mirza MM et al
Combarros O et al. Diagnosis of the CMT1A duplication by PCR
based detection of a novel junction fragment: letter, 962 see Ryan AK et al Burrows NP see Richards AJ et al Burton PR see Laing IA et al Bush A see Davies J et al Bussaglia E see Bayés M et al Concolino D et al. Partial trisomy 1(q42-)qter): a new case with a mild phenotype, 75 Connor JM see Ghaffari SR et al see Ghaffari SR et al Cairns NJ see Kehoe PG et al see Knight SW et al Campion D see Dumanchin C et al Capizzi G see Silengo M et al Capon H see Lench NJ et al see Mossey PA et al Conway G see Murray A et al Cook Jr EH see Jacobsen J et al Coppin B see Dalton P et al Capra V see de Franchis R et al Carey ML et al. Septo-optic dysplasia and WS1 in the proband of a WS1 family segregating for a novel mutation in PAX3 exon 7, Corfield VA see Moolman-Smook JC et al Corral J see Pujana MA et al 248 Costa D see Margarit E et al Cotter PD see Glass IA et al Cottrell S see Olschwang S et al Coulon V see Longy M et al Carles S see de Meeus A et al Caron S see Marsh DJ et al Carrier L see Yu B et al Carrió A see Margarit E et al Couzin D see de Silva D et al Casartelli A see Trabetti E et al Cox TM see Ali M et al Cass DT see Mowat DR et al Crawshaw S see Morgan L et al Croaker GDH see Mowat DR et al Croce AI see Gasparini P et al Castellan C see Merienne K et al Castelló F see Chabás A et al Castello F see Chabas A et al
Castillo S see Austin-Ward E et al
Ceelen TL see Verhoef S et al
Cenani A see Silahtaroglu AN et al
Cerrillo-Hinojosa M see López-Gutiérrez AU et al
Cervera C see Pujana MA et al Crolla JA see Long FL et al see Rivera H et al Croquette MF see Stavropoulou C et al Crow Y see Reid E et al Crow YJ see Ghaffari SR et al Chabás A et al. Recurrence of the D409H mutation in Spanish see Munroe PB et al Gaucher disease patients: description of a new homozygous Crow YJ, Tolmie JL. Recurrence risks in mental retardation: review, patient and haplotype analysis, 775
Chaitow J see Mowat DR et al
Chalmers IJ see Slaney SF et al
Chang K-M see Chen C-P et al Crow YJ et al. "Cataplexy" and muscle ultrasound abnormalities in Coffin-Lowry syndrome, 94 Cruaud C see Guicheney P et al Chang-Claude J et al. Using gene carrier probability to select high risk families for identifying germline mutations in breast cancer susceptibility genes, 116

Chase DS et al. The North Cumbria Community Genetics Project, Cuchacovich M see Austin-Ward E et al Cunliffe HE et al. The prevalence of PAX2 mutations in patients with isolated colobomas or colobomas associated with urogenital anomalies, 806 Curran JL see Robinson R et al Curry C see Verloes A et al Chase NA see Pivnick EK et al

Dobyns WB see Allanson JE et al

Docherty Z see Ogilvie CM et al Curtis D see McDermott MF et al Curtis G see Skirton H et al Dokal I see Knight SW et al Cusin V see Trabetti E et al Domínguez MC see Chabás A et al Dominiczak AF see Lee WK et al Dominiczak MH see Lee WK et al Czarnecki PM et al. A mother with VCFS and unilateral dysplastic kidney and her fetus with multicystic dysplastic kidneys: Dommergues M see Muller F et al Dong J see Chang-Claude J et al additional evidence to support the association of renal malformations and VCFS: letter, 348 Donnai D see Kerr B et al Dahia PLM see Marsh DJ et al see Ryan AK et al Dahl H-HM see Sheffield LJ et al
Dalton P et al. Three patients with a 45,X/46,X,psu dic(Xp)
karyotype, 519
Daly P see McAllister MF et al Dorrance AM see Lee WK et al Douglas F see Reid E et al Drummond J see de Silva D et al Du Y-Z see Michaelis RC et al Dandamudi R see Kerr B et al Du Y-Z et al. A silent mutation, C924T (G308G), in the L1CAM David A see Longy M et al

Davidson J see Watson M et al

Davidson R see Gilfillan A et al

Davies AF et al. Further evidence for the involvement of human gene results in X linked hydrocephalus (HSAS), 456 Duboué B see Longy M et al

Duckett DP see Long FL et al

Dudokdewit AC et al. Predicting adaptation to presymptomatic chromosome 6p24 in the aetiology of orofacial clefting, 857 DNA testing for late onset disorders: who will experience Davies J et al. Distal 10q trisomy syndrome with unusual cardiac and pulmonary abnormalities, 72 Duivenvoorden HJ see Dudokdewit AC et al Dumanchin C et al. De novo presenilin 1 mutations are rare in Davies KE see Trump D et al

Davies S see Olschwang S et al

Davies SJ see Thompson PW and Davies SJ

Day INM see Lee WK et al clinically sporadic, early onset Alzheimer's disease cases, 672 **Dunlop N** see Bonthron DT et al Duran M see Alvarez AI et al Dean JCS see de Silva D et al see Gilfillan A et al see Sjarif DR et al D'Urso M see Knight SW et al Dean JCS et al. Craniosynostosis and chromosome 22q11 deletion: Dutly F see Robinson WP et al Duvivier V see Watson M et al letter, 346 **de Braekeleer M.** Is meconium ileus genetically determined or Dyson HM see Barber JCK et al associated with a more severe evolution of cystic fibrosis?: letter, Earley M see Flanagan N et al Easton DF see Arason A et al see Mirza MM et al de Die-Smulders CEM see Plomp AS et al de Fazio A see Gasparini P et al de Franchis R et al. The C677T mutation of the Eaton S see Ryan AK et al Eber E see Laing IA et al Eccles MR see Choi K-L et al see Cunliffe HE et al 5,10-methylenetetrahydrofolate reductase gene is a moderate risk factor for spina bifida in Italy, 1009 de Goede-Bolder A see Cnossen MH et al de Grandis D see Tupler R et al de Jonghe P see Nelis E et al del Gado R see de Franchis R et al Economides DL see MacDermot KD et al Eden OB see Kerr B et al Edmonds LD see Huether CA et al de la Chapelle A see Ala-Mello S et al Edström L see Xiang F et al Edwards FJ see Timms KM et al Eeles R see Watson M et al see Olschwang S et al Delatycki MB et al. Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the FRDA gene, 713

Delobel B see Stavropoulou C et al

Delozier-Blanchet CD see Xiang F et al Eeles RA see Marsh DJ et al Eggermann K see Eggermann T et al Eggermann T et al. Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome, 784
Eggers S, Zatz M. How the magnitude of clinical severity and Demaille J see de Meeus A et al de Matteo E see Lapunzina P et al

Demczuk S see Portnoï M-F et al

de Meeus A et al. Linkage disequilibrium between the M470V
variant and the IVS8 polyT alleles of the CFTR gene in recurrence risk affects reproductive decisions in adult males with different forms of progressive muscular dystrophy, 189 Egilmez H see Percin EF et al
Egilsson V see Arason A et al
Elçioglu N, Hall CM. A lethal skeletal dysplasia with features of CBAVD, 594 de Meirleir L see Seneca S et al chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a **Denamur E** see Muller F et al see Verlingue C et al rare genetic disorder, 505 Dennis NR see Barber JCK et al Maternal systemic lupus erythematosus and chrondrodysplasia de Paepe A see van Tongerloo A and de Paepe A punctata in two sibs: phenocopy or coincidence?, 690
Elcioglu NH see Hall CM et al
Elion J see Verlingue C et al
Elles R see Lalloo F et al Depetris D see Stavropoulou C et al Desai DC et al. A survey of phenotypic features in juvenile polyposis, Ellis FR see Robinson R et al Ellis I see Olschwang S et al Ellis PM see Sumner AT et al Emery A see Rahman S et al Desai T see Stoilova D et al Desgeorges M see de Meeus A et al de Silva DC see Dean JCS et al de Silva D et al. Mosaicism for a tandem duplication dup(1)(q12q22) in an 18 year old female, 600 Devlieger H see Devriendt K et al see Moerman P et al Emmerson A see Kerr B et al Eng C see Longy M et al see Marsh DJ et al
Engelen JJM see Plomp AS et al
Engelen JJM et al. A simple and efficient method for microdissection
and microFISH, 265 de Vriendt E see Nelis E et al Devriendt K see Cunliffe HE et al see Fryns J-P et al
see Knight SW et al

Devriendt K, Chen C-P. Holoprosencephaly in deletions of Engelhardt A see Kress W et al Engels H see Wieczorek D et al proximal chromosome 14q: letter, 612 Espinós C et al. Linkage analysis in Usher syndrome type I (USH1) Devriendt K et al. Oto-onycho-peroneal syndrome: confirmation of a families from Spain, 391
Espinoza A see Austin-Ward E et al
Estivill X see Pujana MA et al
Estournet B see Guicheney P et al syndrome, 508 Proteinuria in a patient with the diaphragmatic hernia-hypertelorism-myopia-deafness syndrome: further evidence that the facio-oculo-acoustico-renal syndrome represents the same European consortium see Ryan A et al Evans DGR see Lalloo F et al see McAllister MF et al entity, 70 The annual incidence of DiGeorge/velocardiofacial syndrome: letter, see Wu CL et al de Vries BBA et al. The fragile X syndrome: syndrome of the month, Evans DGR et al. Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease D'Hooge R see Fransen E et al associated with truncating mutations, 450 Dickerson C see Du Y-Z et al Dien J see Heinzlef O et al Eydoux P see Teebi AS et al di Rocco M et al. Spectrum of clinical features associated with Facher EA, Law JC. PTEN and prostate cancer: letter, 790 Fantes JA see Matsumoto N et al Farag TI see Tayel SM et al interstitial chromosome 22q11 deletions: letter, 346 Dixon PH see Trump D et al

Farah S see Tayel SM et al

Genís D see Pujana MA et al
Georgiades P et al. Parental origin effects in human trisomy for
chromosome 14q: implications for genomic imprinting, 821
Geraedts JPM see Engelen JJM et al
Geraghty MT see Flanagan N et al
Gerbitz K-D see Jaksch M et al
Gerrard M see Kerr B et al
Ghaffari SR et al. A new strategy for cryptic telomeric translocation
screening in patients with idiopathic mental retardation, 225
Mosaic supernumerary ring chromosome 19 identified by compara see van Steensel M et al Fardeau M see Guicheney P et al Farrar GJ see Kennan AM et al see Mansergh F et al Faulkner CL, Kingston HM. Knowledge, views, and experience of 25 women with myotonic dystrophy, 1020 Felbor U see Gehrig A et al Feldman GL see Czarnecki PM et al Fenske CD et al. Localisation of the gene for glycogen storage disease type 1c by homozygosity mapping to 11q, 269

Férec C see Verlingue C et al

see Muller F et al

Ferguson BM et al. Scarcity of mutations detected in families with X

linked hypohidrotic ectodermal dysplasia: diagnostic Mosaic supernumerary ring chromosome 19 identified by comparative genomic hybridisation, 836 Gibbs RA see Timms KM et al Gibson J see Reid E et al Gibson NA see Laing IA et al Giglio S see Pirola B et al implications, 112 Ferguson-Smith AC see Georgiades P et al Ferguson-Smith MA see Griffin DK et al Gilchrist JM see Speer MC et al Gilfillan A et al. P67L: a cystic fibrosis allele with mild effects found Ferlini A see Tupler R et al Fermont L see Portnoï M-F et al Fernández A see Martínez F et al at high frequency in the Scottish population, 122 Gimelli G see di Rocco M et al Gläser D see Wöhrle D et al Glaser D see Wohrle D et al
Glass IA et al. Trisomy 2q11.2 \rightarrow q21.1 resulting from an unbalanced insertion in two generations, 319
Goebel H-H see Kress W et al
Goldaracena B see Bayés M et al
Goldblatt J see Laing IA et al
Golden WL see Kelly TE et al
Goldman A et al. Molecular evidence that fragile X syndrome occurs Fernández-Luna JL see Combarros O et al Ferrari I see Pratesi R et al Ferraro L see Concolino D et al Findlay I see Toth T et al Fingert JH see Kennan AM et al Fisher AM see Law CJ et al Fitchett M see Horsley SW et al Fitzpatrick DR see Brewer CM et al in the South African black population: letter, 878 Gondos B see Müller J et al Gonzales M see Portnoï M-F et al Gonzalez-Duarte R see Bayés M et al González S see Austin-Ward E et al Flanagan N et al. Familial craniosynostosis, anal anomalies, and porokeratosis: CAP syndrome, 763
Flanders T see Pal T et al
Flint J see Horsley SW et al Goodship J. Kenny-Caffey syndrome is part of the CATCH 22 haploinsufficiency cluster: letter, 1054 Forrest SM see Delatycki MB et al see Sheffield LJ et al Forshaw K see Delatycki MB et al Foulkes WD see Pal T et al Frank J et al. Molecular basis of variegate porphyria: a missense Goodship JA see Ryan A et al Goodwin BS see Huether CA et al Gort L see Chabás A et al mutation in the protoporphyrinogen oxidase gene, 244
Franko JB see Kelly TE et al Grace E see Brewer CM et al Gratacós M see Pujana MA et al Franks J Bet al. Genotype-phenotype correlation in L1 associated diseases, 399

Frayling IM see Marsh DJ et al
Frebourg T see Dumanchin C et al
French Alzheimer's Disease Study Group see Dumanchin C et al Gray RGF, Hall SK. Autoclaving Guthrie cards does not prevent their use in PCR reactions!: letter, 702
Green A see Hutchesson ACJ et al Green AJ see Ali JBM et al see Barber JCK et al Green SL see Halkas AC et al French Alzheimer's Disease Str French JA see Yu B et al see Yu B et al French MC see Choi K-L et al French P see Yu B et al Frey V see Guicheney P et al Friedman TB see Carey ML et al Friedman TB see Carey ML et al Froster U see Meiner A et al Fryns J-P see Devriendt K et al see Devriendt K et al Green SDE see Munroe PB et al
Greening A see Gilfillan A et al
Greenough A see Brady AF et al
Gregory-Evans C see Papaioannou M et al Gregory-Evans K see Bellingham J and Gregory-Evans K Grichener J see Lapunzina P et al
Griffin DK et al. Chromosome specific comparative genome
hybridisation for determining the origin of intrachromosomal
duplications, 37 see Devriendt K et al Grimley S see Murray A et al see Moerman P et al Grinberg D see Bayés M et al Fryns J-P. Another holoprosencephaly locus at 7q21.2?: letter, 614 Fryns J-P, Smeets E. "Cataplexy" in Coffin-Lowry syndrome: letter, see Chabás A et al Grobbee DE see Cnossen MH et al Guerrini R see Pirola B et al Guicheney P et al. PCR based mutation screening of the laminin α2 Fryns J-P et al. Two adult females with a distinct familial mental chain gene (LAMA2): application to prenatal diagnosis and retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, search for founder effects in congenital muscular dystrophy, 211 Guilbert P see Skirton H et al Guilford PJ see Choi K-L et al Guiraud-Chaumeil C see Vincent M-C et al hypergonadotrophic hypogonadism, and retinal dystrophy, 333 Fujieda K see Hoshi N et al Fujimoto S see Hoshi N et al Fujino T see Hoshi N et al Guittard C see de Meeus A et al Fujita M see Hoshi N et al Guven GS see Silahtaroglu AN et al Fukuzawa R see Kikuchi H et al Hacihanefioglu S see Silahtaroglu AN et al Furman WL see Pivnick EK et al Hacking E see Glass IA et al Haddad L see Lee WK et al Furusho K see Yorifuji T et al Fusilli P see Pallotta R and Fusilli P Haenggeli C-A see Guicheney P et al Gaffney D see Lee WK et al Gahukamble DB. Familial occurrence of congenital incomplete Hahn S-H see Park M-S et al
Hainque B see Guicheney P et al
Halkas AC et al. Variants of α,-proteinase inhibitor in black and white South African patients with focal glomerulosclerosis and minimal change nephrotic syndrome, 6
Hall CM see Elçioglu N and Hall CM see Elçioglu N and Hall CM
Hall CM et al. A distinct form of spondyloepimetaphyseal dysplasia with multiple dislocations, 566
Hall SK see Gray RGF and Hall SK see Hutchesson ACJ et al
Hall WJ see Robinson R et al
Halley DJJ see de Vries BBA et al see Verhoef S et al
Halsall PJ see Robinson R et al
Halsall PJ see Robinson R et al Hahn S-H see Park M-S et al Gahukamble DB. Familial occurrence of prepyloric mucosal diaphragm, 1040 Gaillard G see Portnoï M-F et al Gaillard MC see Halkas AC et al Gallagher PG see Jonsson JJ et al Galoppo C see Lapunzina P et al Garcia A see Martinez-Frias ML et al Garchon H-J see Stoilova D et al Gardiner RM see Munroe PB et al Gardiner RM see Delatycki MB et al Gardiner RM see Munroe PB et al
Gardner RJM see Delatycki MB et al
Garssen MPJ see Cnossen MH et al
Gassell PC see Speer MC et al
Gasparini P et al. Usher syndrome type III (USH3) linked to
chromosome 3q in an Italian family, 666
Gehrig A et al. Assessment of the interphotoreceptor matrix
proteoglycan-1 (IMPG1) gene localised to 6q13-q15 in
autosomal dominant Stargardt-like disease (ADSTGD),
progressive bifocal chorioretinal atrophy (PBCRA), and North
Carolina macular dystrophy (MCDR1), 641 Halsall PJ see Robinson R et al Hamaguchi H see Kawashima T et al Hambly B see Yu B et al Hamel BCJ see Yntema HG et al Hamers AJH see Plomp AS et al

Hamers GJH see Engelen JJM et al

see Mansergh F et al

Humphries SE see Lee WK et al Hanauer A see Merienne K et al Handa Y see Hoshi N et al Handoko HY et al. Meiotic breakpoint mapping of a proposed X Hunt DM see Gehrig A et al Hunter A. Clinical features of chromosome 22q11 deletion: letter, linked visual loss susceptibility locus in Leber's hereditary optic 346 neuropathy, 668

Hanefeld F see Webb T et al

Hanihara T see Onishi H et al

Hannequin D see Dumanchin C et al Hunter AGW et al. Medical complications of achondroplasia: a multicentre patient review, 705 Huson S see Horsley SW et al

Hutcheon RG et al. Clinical features and mental development of a child with a prenatally identified 45,XX,der(5)t(5;18) Hansmann I see Xiang F et al Hansmann Tsee Alaig F et al Hanyu N see Yamamoto K et al Hareyama H see Hoshi N et al Hargreave T see Gilfillan A et al Harper J see Flanagan N et al Harper PS see Tischkowitz M et al (p15;q11.2),-18 karyotype, **865 Hutchesson ACJ** et al. A comparison of disease and gene frequencies of inborn errors of metabolism among different ethnic groups in the West Midlands, UK, 366 Hutchison WM see Sheffield LJ et al Huttenlocher PR see Precht KS et al Harper PS. Medical genetics in the UK and the National Health Service, 441

Harpey J-P see Guicheney P et al

Harriso JB see Richards AJ et al

Harrison RH see Ogilvie CM et al

Lost TC see Price IA et al Ikeda S-I see Yamamoto K et al Imachi H see Sato M et al Imaizumi K see Davies AF et al Inglehearn CF et al. A linkage survey of 20 dominant retinitis Hart TC see Price JA et al Haschke N see Berger A et al Hassan SJA see Sabry MA et al Hassold TJ see Zaragoza MV et al Hata J-I see Kikuchi H et al pigmentosa families: frequencies of the nine known loci and evidence for further heterogeneity, 1 A new dominant retinitis pigmentosa family mapping to the RP18 locus on chromosome 1q11-21: letter, 788
Innis JW see Carey ML et al Hatchwell E see Black GCM et al Hauser G. Laws regarding insurance companies: letter, 526
Havelaar AC see Verheijen FW et al
Hayden CM see Laing IA et al
Hayward C see Brewer CM et al Iragui-Madoz MI see Bayés M et al Iseki E see Onishi H et al Ishizaka A see Nagai K et al Ismail EAR see van Steensel M et al Itagaki T see Nagai K et al Ivanovich J see Huether CA et al Iwama T see Olschwang S et al Hayward C et al. Homozygosity for Asn86Ser mutation in the CuZn-superoxide dismutase gene produces a severe clinical cuzn-superoxide dismutase gene produces a severe clinical phenotype in a juvenile onset case of familial amyotrophic lateral sclerosis: letter, 174

Hazan J see Heinzlef O et al

He Y see Guicheney P et al

Heinzlef O et al. Mapping of a complicated familial spastic paraplegia to locus SPG4 on chromosome 2p, 89

Heiss NS see Knight SW et al

Helbling-Leclerc A see Guicheney P et al

Hemmirk A see Olschwapp S et al Jabs EW see Flanagan N et al Jackson SNJ et al. The diagnosis of Liddle syndrome by identification of a mutation in the  $\beta$  subunit of the epithelial sodium channel, 510 Jacobs P see Dalton P et al see Murray A et al

Jacobsen J et al. Molecular screening for proximal 15q abnormalities in a mentally retarded population, 534 Hemminki A see Olschwang S et al Henke B see Wieczorek D et al Hennekam RCM see van Asperen CJ et al Herd RM see Porteous MEM et al Herens C see Verloes A et al Hertzberg VS see Huether CA et al Hill DF see Broom MF et al Jacquot S see Merienne K et al **Jaffé A** see Davies J et al Jaksch M et al. A systematic mutation screen of 10 nuclear and 25 mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA<sup>scr(UCN)</sup> mutations in a subgroup with syndromal encephalopathy, 895

Jamar M see Verloes A et al

James R see Dalton P et al Hilton-Jones D see Horsley SW et al Hitman GA see McDermott MF et al Ho L-P see Gilfillan A et al
Ho VT see Rasmussen SA et al
Hodgson S see Olschwang S et al
Hodgson SV see Desai DC et al Janes A see Pirola B et al Jansen LAJ see Verhoef S et al Jarman PR, Warner TT. The dystonias: syndrome of the month, 314 Jedele KB see Jaksch M et al see Marsh DJ et al Jeffery S see Fenske CD et al see Mirza MM et al see MacDermot KD et al Hofmann S see Jaksch M et al Hollman A see Crow YJ et al Jenkins JJ see Pivnick EK et al Jenkins T see Goldman A et al see Reid E et al Jeremy RW see Yu B et al Holmans P see Kehoe PG et al see Yu B et al Honda M see Kikuchi H et al Johnson JP et al. A family with mental retardation, variable Hong C-H see Park M-S et al

Hopkins PM see Robinson R et al

Hopwood JJ see van Royen-Kerkhof A et al

Horrigan SK see Speer MC et al

Horsley SW et al. Del(18p) shown to be a cryptic translocation using macrocephaly and macro-orchidism, and linkage to Xq12-q21, 1026 Johnston KM see Precht KS et al Jolly RD see Broom MF et al Jonas P see Chase DS et al a multiprobe FISH assay for subtelomeric chromosome **Jonasdottir A** see Arason A et al rearrangements, 722 Jones AM see Ghaffari SR et al Jones KJ et al. Abnormalities of dystrophin, the sarcoglycans, and laminin α2 in the muscular dystrophies, 379 Hoshi N et al. Seminoma in a postmenopausal woman with a Y;15 translocation in peripheral blood lymphocytes and a t(Y;15)/45,X Turner mosaic pattern in skin fibroblasts, 852 Jones LC see McDermott MF et al Houlston R see Marsh DJ et al Jonsson JJ et al. Alport syndrome, mental retardation, midface see Olschwang S et al hypoplasia, and elliptocytosis: a new X linked contiguous gene Houlston RS see Fenske CD et al deletion syndrome?, 273

Jonveaux P see Sloan-Béna F et al

Joseluiz P see Xiang F et al

Jouanolle AM see Pinson S et al Houtman P see Jackson SNJ et al Howell A see Lalloo F et al Howell N see Chinnery PF et al Hsu E see Liang M-H et al Huang F-Y see Chen C-P et al Joyce CA see Barber JCK et al Joyé N see Portnoï M-F et al Jukkola A et al. New lethal disease involving type I and III collagen Huether CA et al. Maternal age specific risk rate estimates for Down syndrome among live births in whites and other races from Ohio and Metropolitan Atlanta, 1970-1989, 482

Hughes AE see Magee AC and Hughes AE defect resembling geroderma osteodysplastica, De Barsy syndrome, and Ehlers-Danlos syndrome IV, 513 Kabbaj K et al. Autosomal recessive anhidrotic ectodermal dysplasia in a large Moroccan family, 1043
Kader NA see Tayel SM et al
Kääriäinen H see Ala-Mello S et al
Kaiser P see Eggermann T et al
Kaji M see Yorifuji T et al
Kala U see Halkas AC et al
Kalmar I. see Tordai A et al Hughes AE et al. Fine localisation of the gene for central areolar choroidal dystrophy on chromosome 17p, 770 **Hughes HE** et al. Contracting for clinical genetic services: the Welsh model, 309
Hughes KS see Marsh DJ et al Hughes-Benzie R see Lapunzina P et al Hugot J-P see Mirza MM et al Humphries P see Kennan AM et al Kalmár L see Tordai A et al Kalsheker N see Morgan L et al

Kalter H. Congenital malformations: an inquiry into classification Law CJ et al. Distal 6p deletion syndrome: a report of a case with and nomenclature, 661
Kamiyama Y see Kikuchi H et al anterior chamber eye anomaly and review of published reports, 685 Law JC see Facher EA and Law JC
Leandro P see Rivera I et al
Lechner MC see Rivera I et al
Ledbetter DH see Allanson JE et al
see Jacobsen J et al
see Matsumoto N et al Kant JA see Hoshi N et al Kashtan CE see Jonsson JJ et al
Kaste SC, Pivnick EK. Bony orbital morphology in
neurofibromatosis type 1 (NF1), 628
Kauppila S see Jukkola A et al
Kawai M see Yorifuji T et al Kawanishi C see Onishi H et al see Precht KS et al Lee C-C see Chen C-P et al Lee J see Mirza MM et al Lee PJ see Fenske CD et al Kawashima T et al. Linkage and association of an interleukin 4 gene polymorphism with atopic dermatitis in Japanese families, 502 **Kearney L** see Horsley SW et al **Keen TJ** see Inglehearn CF et al Lee WK et al. Identification of a common low density lipoprotein Kehoe PG et al. The butyrylcholinesterase K variant and receptor mutation (C163Y) in the west of Scotland, 573 Le Gac G see Verlingue C et al Legius E see Fryns J-P et al Leguern E see Meggouh F et al Leheup B see Sloan-Béna F et al susceptibility to Alzheimer's disease, 1034 Kelly TE et al. Discordant phenotypes and 45,X/46,X,idic(Y), 862
Kelsell DP see Lench NJ et al
Kelsell RE see Gehrig A et al
Kenna PF see Kennan AM et al Leisti J see Baumann P et al Kennan AM et al. A novel Asp380Ala mutation in the see Jukkola A et al GLC1A/myocilin gene in a family with juvenile onset primary open angle glaucoma, 957

Kerr BA see Mowat DR et al Lench NJ et al. A Moroccan family with autosomal recessive Lench NJ et al. A Moroccan family with autosomal recessive sensorineural hearing loss caused by a mutation in the gap junction protein gene connexin 26 (GJB2), 151

Lennard-Jones JE see Mirza MM et al

Leonard JV see Fenske CD et al

Lerone M see Silengo M et al

Lese CM see Precht KS et al

Le Souëf PN see Laing IA et al

Leung K-Y see Munroe PB et al

Leventhal BL see Jacobsen J et al

Leventhal BL see Sloan-Béna F et al

Levs A see Devriendt K et al Kerr B et al. Costello syndrome: two cases with embryonal rhabdomyosarcoma, 1036 Kershaw A see Skirton H et al Kerzin-Storrar L see Skirton H et al
Keymolen K see Devriendt K et al
Kharrat K see Mégarbané A et al
Kikuchi H et al. Do intronic mutations affecting splicing of WT1 exon 9 cause Frasier syndrome?, 45 Kim H-S see Park M-S et al Leys A see Devriendt K et al Kim J-S see Park M-S et al Kim SS see Jones KJ et al King BH see Jacobsen J et al Liang M-H et al. Cystic fibrosis in a Puerto Rican female homozygous for the R1066C mutation: letter, 84
Lichter-Konecki U see Rivera I et al Kingston HM see Faulkner CL and Kingston HM
Kirk E, Ades L. Hypoplastic left heart in cerebrocostomandibular Liebaers I see Seneca S et al Liechti-Gallati S see Jaksch M et al Lin AE, Rosti L. Tricuspid atresia in sibs: letter, 1055 Lindhout D see Verhoef S et al Lissens W see Seneca S et al syndrome: letter, 879 Kirk J. Kenny-Caffey syndrome is part of the CATCH 22 haploinsufficiency cluster: letter, 1054
Kirk JM see Gilfillan A et al
Kittikamron K see Matsumoto N et al
see Precht KS et al Loff S see Olschwang S et al Logie LJ see Porteous MEM et al Loiselet J see Nustapha M et al

Long FL et al. Triplication of 15q11-q13 with inv dup(15) in a
female with developmental delay, 425

Longy M et al. Mutations of PTEN in patients with see Verheijen FW et al see van Royen-Kerkhof A et al see Verheijen FW et al Klein D see Liang M-H et al Klein I see Tordai A et al Bannayan-Riley-Ruvalcaba phenotype, 886
López-Gutiérrez AU et al. Uniparental disomy for chromosome 6 Kleinle S see Jaksch M et al results in steroid 21-hydroxylase deficiency: evidence of different Kneppers A see Passos-Bueno MR et al
Knight SJL see Horsley SW et al
Knight SW et al. 1.4 Mb candidate gene region for X linked
dyskeratosis congenita defined by combined haplotype and X genetic mechanisms involved in the production of the disease, 1014
Lotery AJ see Hughes AE et al
Loutradis-Anagnostou A see Papaioannou M et al
Ludewick H see Halkas AC et al chromosome inactivation analysis, 993 Kobayashi O see Nagai K et al Lye R see Wu CL et al Kohlhauser C see Berger A et al Komitowski D see Chang-Claude J et al Kosaka K see Onishi H et al Koskimies O see Ala-Mello S et al McAllister MF et al. Men in breast cancer families: a preliminary qualitative study of awareness and experience, 739 McAndrew PE see Parsons DW et al McColl J see Mossey PA et al Kosugi S see Müller J et al MacDermot KD et al. Prenatal diagnosis of autosomal dominant polycystic kidney disease (PKD1) presenting in utero and prognosis for very early onset disease, 13

McDermott EM see McDermott MF et al.

McDermott MF et al. Exclusion of the familial Mediterranean fever Kraimps J-L see Longy M et al Krause A see Goldman A et al Krawczak M see Hughes HE et al Kreichati G see Mégarbané A et al Kremer H see Yntema HG et al Kress W see Merienne K et al
Kress W et al. No evidence for heterogeneity in oculopharyngeal
muscular dystrophy: letter, 613
Krivchenia EL see Huether CA et al
Kula K see Price JA et al locus as a susceptibility region for autosomal dominant familial Hibernian fever, 432

McGiffert C see Griffin DK et al

McIntyre D see Gilfillan A et al

McKeown C see Olschwang S et al Macleod MJ see Lee WK et al Kum JB see Marsh DJ et al MacMillan A see Pal T et al McNoe LA see Choi K-L et al see Cunliffe HE et al Kuner R see Eggermann T et al Kunz J see Rivera H et al Kuroki Y see Davies AF et al see Cunliffe HE et al

McTaggart DR see Yu B et al
see Yu B et al
McWilliam R see Crow YJ et al
Madigand M see Heinzlef O et al
Magee AC, Hughes AE. Segregation distortion in myotonic
dystrophy, 1045
Maher ER see Brewer CM et al
Mchiany E see Heinzlef O et al Kurosawa Y see Kikuchi H et al Kyne L see Flanagan N et al Lacombe D see Longy M et al Ladusans E see Kerr B et al

Laing IA et al. A polymorphism of the CC16 gene is associated with
an increased risk of asthma, 463 Mahieux F see Heinzlef O et al Malerba G see Trabetti E et al Malfeyt GAM see Verhoef S et al Lalloo F et al. An evaluation of common breast cancer gene Lamor F et al. An evaluation of common breast cancer gene mutations in a population of Ashkenazi Jews, 10 Lam WWK see Brewer CM et al Laporte J see Vincent M-C et al Lapunzina P et al. A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma, 153 Laprègue M see L ongy M et al. Mallik A see Hutcheon RG et al Malone M see Vassal HB et al Mancini GMS see Verheijen FW et al Mandato C see de Franchis R et al Mandel J-L see Vincent M-C et al Larrègue M see Longy M et al Larsen MB see Reddy KS and Larsen MB Manouvrier-Hanu S see Vincent M-C et al Laurent-Puig P see Mirza MM et al Mansergh FC see Kennan AM et al

Mitchison HM see Munroe PB et al

Miyauchi A see Sato M et al

Moerman P et al. Severe primary pulmonary hypoplasia ("acinar Mansergh F et al. Lack of evidence for genetic heterogeneity in Best dysplasia") in sibs: a genetically determined mesodermal defect?: vitelliform macular dystrophy: letter, 85 letter, 964 Mao X. Chinese geneticists approach ethics: letter, 83 Moglabey YB see Mustapha M et al Mole SE see Munroe PB et al Molina M see Alvarez AI et al Molloy MG see McDermott MF et al Momoi T see Yorifuji T et al Monaco AP see Sloan-Béna F et al Maraschio P see Tupler R et al Marchese C see Olschwang S et al Margarit E et al. Molecular, cytogenetic, and clinical characterisation of six XX males including one prenatal diagnosis, 727 Marie SK see Moreira ES et al Moncla A see Stavropoulou C et al Markham AF see Lench NJ et al Montfort M see Chabás A et al
Moolman-Smook JC et al. Identification of a new missense
mutation in MyBP-C associated with hypertrophic see Robinson R et al Markie D see Olschwang S et al Marks J see Verloes A et al Marques I see Saraiva JM et al cardiomyopathy, 253 Moons KGM see Cnossen MH et al Marsh DJ et al. Germline PTEN mutations in Cowden Moore AT see Inglehearn CF et al see Votruba M et al Mora M see Guicheney P et al Moreau V see Dumanchin C et al syndrome-like families, 881 Marshall T see Gilfillan A et al Martinez F et al. Genetic localisation of mental retardation with spastic diplegia to the pericentromeric region of the X chromosome: X inactivation in female carriers, 284 Moreira ES et al. A first missense mutation in the  $\delta$  sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies, 951

Morgan D see Ferguson BM et al Martínez-Frías ML et al. Cyclopia and sirenomelia in a liveborn Martinez-Frias ML et al. Cyclopie infant: letter, 263 Martínez-Mir A see Bayés M et al Martin C see Dumanchin C et al Martin J-J see Nelis E et al Martin R see Alvarez AI et al Martin S see Richards AJ et al Morgan L et al. Distortion of maternal-fetal angiotensin II type 1 receptor allele transmission in pre-eclampsia, 632 Mori T see Müller J et al
Moricca MT see Concolino D et al
Morison IM see Okamoto K et al
Morlier G see Portnoï M-F et al
Mortier G see Devriendt K et al Martinati LC see Trabetti E et al Martinez M see Dumanchin C et al Marton T see Toth T et al Maruyama Y see Onishi H et al Mossey PA et al. Prediction of liability to orofacial clefting using Marzuki S see Handoko HY et al Mason PJ see Knight SW et al Massie D see de Silva D et al Mastroiacovo P see de Franchis R et al genetic and craniofacial data from parents, 371

Mowat DR et al. Hirschsprung disease, microcephaly, mental retardation, and characteristic facial features: delineation of a new syndrome and identification of a locus at chromosome new syndrome and identification of a locus at chromosome 2q22-q23, 617

Mucke J see Wöhrle D et al

Mueller RF see Lench NJ et al

Muller F et al. Cystic fibrosis screening: a fetus with hyperechogenic bowel may be the index case, 657

Muller J et al. Severe testotoxicosis phenotype associated with Mathew CG see Mirza MM et al Matoso E see Saraiva JM et al Matsubara S see Sato M et al Matsumoto N et al. Isolation of BAC clones spanning the Xq22.3 translocation breakpoint in a lissencephaly patient with a de novo X;2 translocation, \$29 Matsuno M see Davies AF et al Asp→Tyr mutation of the lutrophin/choriogonadotrophin Asp → 1yr mutation of the introphilivenoriogonauditophilir receptor gene, 340

Muller-Höcker J see Jaksch M et al

Muller-Mysok B see Kress W et al

Mumm S see Trump D et al

Munoz F see Ferguson BM et al

Munroe PB et al. Sharing of PPT mutations between distinct clinical Matsuzono Y see Nagai K et al Mattei MG see Stavropoulou C et al Matthijs G see Knight SW et al Maumenee IH see Gehrig A et al May DS see Huether CA et al Mayosi B see Moolman-Smook JC et al Mégarbané A et al. Four sibs with dislocated elbows, bowed tibiae, forms of neuronal ceroid lipofuscinoses in patients from Scotland: letter, 790 Murao K see Sato M et al Murday V see Desai DC et al scoliosis, deafness, cataract, microcephaly, and mental retardation: a new MCA/MR syndrome, 755 Meggouh F et al. The first de novo mutation of the connexin 32 gene associated with X linked Charcot-Marie-Tooth disease, 251
Meiner A et al. Instability in the normal CTG repeat range at the see Watson M et al Murday VA see Marsh DJ et al myotonic dystrophy locus: letter, 791
Meisel-Stosiek M see Wöhrle D et al Muroi J see Yorifuji T et al see Yorifuji T et al Murray A et al. Studies of FRAXA and FRAXE in women with Meitinger T see Jaksch M et al see Mansergh F et al

Melis D et al. Mild phenotype associated with an interstitial deletion
of the long arm of chromosome 1, 1047 premature ovarian failure, 637 Mustapha M et al. Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425, 202 Memmi M see Tupler R et al Mercier B see Verlingue C et al Myllylä VV see Baumann P et al Nagai K et al. Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation, 342 Mergenthaler S see Eggermann T et al
Merienne K et al. Rapid immunoblot and kinase assay tests for a
syndromal form of X linked mental retardation: Coffin-Lowry Nagao M see Nagai K et al syndrome, 890 Merlini L see Guicheney P et al see Nagai K et al Nagashima K see Hoshi N et al Nagy B see Tóth T et al Najera C see Espinós C et al Nakagawa H see Kawashima T et al Nakahori Y see Hoshi N et al Michaelis RC see Robinson WP et al Michaelis RC et al. The site of a missense mutation in the extracellular Ig or FN domains of L1CAM influences infant mortality and the severity of X linked hydrocephalus, 901

Middleton W see Gilfillan A et al

Mignon C see Stavropoulou C et al

Mikuni M see Hoshi N et al Narod SA see Pal T et al
Nassif N see Delatycki MB et al
Nassogne MC see Seneca S et al
Neal J see Kehoe PG et al
Neale K see Olschwang S et al
Neale KF see Desai DC et al
Neary W see Wu CL et al
Nelis E et al. Mutation analysis of the nerve specific promoter of the
peripheral myelin protein 22 gene in CMT1 disease and HNPP,
590
Nelson R see Johnson IP et al Narod SA see Pal T et al Milla P see Desai DC et al Miller S see Teebi AS et al Millán JM see Espinós C et al see Martinez F et al Millie E see Zaragoza MV et al Mills L see Ryan AK et al Minagawa K see Nagai K et al Minns RA see Hayward C et al Mirza G see Davies AF et al Nelson R see Johnson JP et al Nelson K see Johnson JP et al
Neri G see Cohen Jr MM et al
Newbury-Ecob R. Atelosteogenesis type 2: syndrome of the month, 49
Nicholls AC see Richards AJ et al
Nicholls RD see Robinson WP et al
Nicholson GA see Delatycki MB et al
Nicholson JC see Barber JCK et al
Nicholson MR see Yu B et al
see Yu B et al Mirza MM et al. Evidence of linkage of the inflammatory bowel disease susceptibility locus on chromosome 16 (IBD1) to ulcerative colitis, 218

Mitchell AR see Sumner AT et al

Mitchell-Lehman M see Pal T et al

see Yu B et al

Nicolaides KH see Brady AF et al Pénzes M see Tordai A et al Pepe A see de Franchis R et al Niermeijer MF see Cnossen MH et al see de Vries BBA et al Percin EF et al. Mesoaxial complete syndactyly and synostosis with see Dudokdewit AC et al Nigro V see Moreira ES et al Nixon J see Horsley SW et al Noguchi E see Kawashima T et al hypoplastic thumbs: an unusual combination or homozygous expression of syndactyly type I?, 868 Percin S see Percin EF et al Pereira J-L see Webb T et al Pérez-Garrigues H see Espinós C et al Pericak-Vance MA see Speer MC et al North KN see Jones KJ et al O'Brien P see Griffin DK et al Perone L see Melis D et al Oei PTSP see Glass IA et al Perveen R see Black GCM et al Peters HL, Bankier A. Lipomatous myelomeningocele, athyrotic Ogilvie CM et al. A new approach to the elucidation of complex hypothyroidism, and sensorineural deafness: a new form of syndromal deafness?, 948 chromosome rearrangements illustrated by a case of Rieger syndrome, 234
Ogunkolade BW see McDermott MF et al Petros AJ see Vassal HB et al
Pfeiffer R see Devriendt K et al
Phelan M see McDermott MF et al
Philip N, Sigaudy S. Costello syndrome: syndrome of the month, 238
Philippe C see Sloan-Béna F et al Ohye H see Sato M et al. Okamoto K et al. Is p57<sup>KIP2</sup> mutation a common mechanism for Beckwith-Wiedemann syndrome or somatic overgrowth?: letter, Okuyama K see Hoshi N et al O'Lague P see Verloes A et al Phillips R see Olschwang S et al Phillips RKS see Desai DC et al Oliva R see Margarit E et al Piccini M see Jonsson JJ et al Picco P see di Rocco M et al Pignatti PF see Trabetti E et al Olschwang S et al. Peutz-Jeghers disease: most, but not all, families are compatible with linkage to 19p13.3, 42 Onaindia ML see Alvarez AI et al
Onishi H et al. Pancreatic exocrine dysfunction associated with
mitochondrial tRNA<sup>Leu(UUR)</sup> mutation, 255 Pilz D see Tischkowitz M et al Pilz DT see Matsumoto N et al Pinson S et al. Non-C282Y familial iron overload: evidence for locus Oostra BA see de Vries BBA et al heterogeneity in haemochromatosis, 954 Ordoñez-Sánchez ML see López-Gutiérrez AU et al Piovan E see Pirola B et al Pipkin FB see Morgan L et al

Pirola B et al. Agenesis of the corpus callosum with Probst bundles Ormiston W see McAllister MF et al Osborn AH see Sheffield LJ et al Ostrer H see Teebi AS et al owing to haploinsufficiency for a gene in an 8 cM region of 6q25, Oterino A see Combarros O et al Otsuka F see Kawashima T et al 1031 Pitts Crick R see Stoilova D et al Pivnick EK see Kaste SC and Pivnick EK see Park VM and Pivnick EK Oudjhane K see Teebi AS et al Oury J-F see Muller F et al
Overweg-Plandsoen WCG see van Asperen CJ et al
Owen MJ see Kehoe PG et al
Ozbas F see Percin EF et al Pivnick EK et al. Simultaneous adrenocortical carcinoma and ganglioneuroblastoma in a child with Turner syndrome and germline p53 mutation, 328 Ozdemir N see Stoilova D et al Plant C see Bessant DAR et al see Inglehearn CF et al Paavola P see Salonen R and Paavola P Plauchu H see Pinson S et al Packard CJD see Lee WK et al Plomp AS et al. Two cases of partial trisomy 8p and partial Pajunen L see Jukkola A et al monosomy 21q in a family with a reciprocal translocation (8;21)(p21.1;q22.3), 604

Ploos van Amstel JK see Sjarif DR et al

Pober BR see Jonsson JJ et al

Poh-Fitzpatrick M see Frank J et al

Pohl K see Crow YJ et al

Poll-The BT see Sjarif DR et al Pal T et al. Genetic implications of double primary cancers of the colorectum and endometrium, 978
Palau F see Martínez F et al Pallotta R, Fusilli P. Unknown syndrome: peculiar face, severe hypodontia of permanent teeth, and precocious choroid calcifications, 435 Palmer LJ see Laing IA et al Pandya PP see Brady AF et al see van Royen-Kerkhof A et al Pongratz DE see Jaksch M et al Pope FM see Richards AJ et al Porschke H see Kress W et al Pannetier S see Merienne K et al
Papaikonomou M see Watson M et al
Papaioannou M et al. A new family of Greek origin maps to the Porteous MEM et al. Keratosis follicularis spinulosa decalvans: CRD locus for autosomal dominant cone-rod dystrophy on 19q, confirmation of linkage to Xp22.13-p22.2, 336 429 Portnoï M-F et al. Prenatal diagnosis by FISH of a 22q11 deletion Papp C see Tóth T et al Papp Z see Tóth T et al Parenti G see Melis D et al in two families, 165
Pouget J see Heinzlef O et al
Poulton J see Rahman S et al Poulton J see Rahman S et al

Poustka A see Knight SW et al

Powell RJ see McDermott MF et al

Pratesi R et al. Costello syndrome in two Brazilian children, 54

Precht KS et al. Two 22q telomere deletions serendipitously detected by FISH, 939

Prece MA see Hutchesson ACJ et al

Price JA et al. A common DLX3 gene mutation is responsible for tricho-dento-osseous syndrome in Virginia and North Carolin Paris D see Delatycki MB et al Park M-S et al. Extensive form of aplasia cutis congenita: a new syndrome?, 609 Park VM, Pivnick EK. Neurofibromatosis type 1 (NF1): a protein truncation assay yielding identification of mutations in 73% of patients, 813 Parker CO see Chase DS et al Parker CO see Chase DS et al
Parker Jr WD see Parsons DW et al
Parker L see Chase DS et al
Parsons DW et al. Diagnosis of spinal muscular atrophy in an SMN
non-deletion patient using a quantitative PCR screen and
mutation analysis, 674
Partington M see Cohen Jr MM et al
Partington MW. Another dystonia: letter, 879
Pasmans NMT see Cnossen MH et al
Passarge E see Wicczorek D et al
Passchier J see Dudokdewit AC et al
Passons Bueno MR see Moreira ES et al tricho-dento-osseous syndrome in Virginia and North Carolina families, 825
Priest JH see Huether CA et al Prieto F see Martínez F et al Prior TW see Parsons DW et al Prud'homme J-F see Heinzlef O et al Pujana MA et al. Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method, 99 Passos-Bueno MR see Moreira ES et al Quane KA see McDermott MF et al Passos-Bueno MR et al. Presence of the Apert canonical S252W Quarrell O see Kerr B et al FGFR2 mutation in a patient without severe syndactyly, 677

Patch C see Skirton H et al

Patel PI see Nelis E et al Quirke P see Tóth T et al Ragoussis J see Davies AF et al Paternotte C see Heinzlef O et al Rahman S et al. UK centres are not following the Royal College of Pathologists' recommendations for storage of Guthrie cards: a Patton MA see Brady AF et al Payne A see Papaioannou M et al
Payne AM see Bessant DAR et al
Payne SJ see Chotai KA and Payne SJ
Peacock RE see Inglehearn CF et al
Peet J et al. 49,XXXXY: a distinct phenotype. Three new cases and national policy is needed: letter, 263
Rajczy K see Tordai A et al Ramadan DG see Sabry MA et al Ramárez-Jiménez S see López-Gutiérrez AU et al Ramos-Arroyo MA see Bayés M et al Ramsden RT see Wu CL et al Ranke MB see Eggermann T et al review, 420

Peleg D, Yankowitz J. Choroid plexus cysts and aneuploidy, 554

Peñaherrera M see Robinson WP et al

Rash FC see Sunada F et al

Saul RA see Rasmussen SA et al Rasmussen SA et al. Constitutional and mosaic large NF1 gene deletions in neurofibromatosis type 1, 468 Savarirayan R, Bankier A. Acampomelic campomelic dysplasia with de novo 5q;17q reciprocal translocation and severe phenotype, 597 Rasool MAA see Sabry MA et al Raus P see Fryns J-P et al
Raymond FL see Ogilvie CM et al
Read AP see Wu CL et al
Reardon W see Dean JCS et al
Reck A see Black GCM et al Savarirayan R et al. Baller-Gerold syndrome associated with congenital portal venous malformation, 767
Schatteman I see Lench NJ et al
Schindler D see Wöhrle D et al Reddy KS, Larsen MB. A molecular, cytogenetic, and clinical Schinzel A see Merienne K et al Schinzel AA see Robinson WP et al
Schlessinger D see Trump D et al
Schmidt S see Chang-Claude J et al
Schrander-Stumpel CTRM see Verhoef S et al
Schwanitz G see Wieczorek D et al evaluation of mosaic tandem duplication 17p and Charcot-Marie-Tooth type 1A neuropathy, 169 Redline RW see Zaragoza MV et al Reeve AE see Okamoto K et al Reid C see Harris A and Reid C Reid E et al. Autosomal dominant juvenile recurrent parotitis, 417 Schwartz C see Rasmussen SA et al Rellos P see Ali M et al
Renieri A see Jonsson JJ et al
Riba L see López-Gutiérrez AU et al
Ribeiro RC see Pivnick EK et al Schwartz CE see Du Y-Z et al see Johnson JP et al see Michaelis RC et al Schwartz K see Guicheney P et al Richard P see Guicheney P et al see Yu B et al Richards AJ et al. A single base mutation in COL5A2 causes Scott C see Merienne K et al Scott Jr CI see Hunter AGW et al Ehlers-Danlos syndrome type II, 846 Richieri-Costa A see Passos-Bueno MR et al Richmond DR see Yu B et al Scott WK see Speer MC et al Scriven PN see Ogilvie CM et al Seal S see Olschwang S et al Sefiani A see Kabbaj K et al see Yu B et al Risteli J see Jukkola A et al Risteli L see Jukkola A et al **Sekihara H** see Onishi H et al Rivera H et al. Intrachromosomal triplication of distal 7p, 78

Rivera I et al. Population genetics of hyperphenylalaninaemia
resulting from phenylalanine hydroxylase deficiency in Portugal, Semsarian C see Yu B et al see Yu B et al Seneca S et al. Pitfalls in the diagnosis of mtDNA mutations: letter, 963 Robinson R et al. Genetic heterogeneity and HOMOG analysis in Sepp T see Ali JBM et al Seri M see Silengo M et al British malignant hyperthermia families, 196
Robinson WP et al. The mechanisms involved in formation of deletions and duplications of 15q11-q13, 130
Rodolph G see Mansergh F et al
Rodrigo O see Espinos C et al Serre J-L see Muller F et al Sertié AL see Passos-Bueno MR et al Sewry CA see Tupler R et al Sezgin I see Percin EF et al Shaham M see Hutcheon RG et al Shaltout A see Sabry MA et al Shannon N see Kerr B et al Rogers C see Hughes HE et al Rogers JG see Hunter AGW et al Rogol A see Kelly TE et al Rolon A see Rivera H et al Shannon N see Kerr B et al
Shapiro B see Liang M-H et al
Shaw DG see Hall CM et al
Shayeghi M see Chang-Claude J et al
Sheffield LJ et al. Segregation of mutations in arylsulphatase E and
correlation with the clinical presentation of chondrodysplasia Romano A see Melis D et al Romeo G see Silengo M et a Ropers H-H see Yntema HG et al Rosaia L see Silengo M et al Rosell J see Margarit E et al Rosenbloom L see Webb T et al punctata, 1004 Shenker A see Müller J et al
Shih S-L see Chen C-P et al
Sigaudy S see Philip N and Sigaudy S
Silahtaroglu AN et al. Not para-, not peri-, but centric inversion of Rossi A see Merienne K et al Rosti L see Lin AE and Rosti L Rougas C see Papaioannou M et al Rouger H see Meggouh F et al Roullet E see Heinzlef O et al chromosome 12, 682 Silberman P see Lapunzina P et al Royer-Pokora B see Chang-Claude J et al Silengo M et al. Ectodermal dysplasia, primary hypothyroidism, and Ryan A et al. Reply to letters regarding clinical features of chromosome 22q11 deletion: letter, 347 agenesis of the corpus callosum: variable expression of a single syndrome?, 157 Ryan AK et al. Smith-Lemli-Opitz syndrome: a variable clinical and Sillence D see Hunter AGW et al Sillence DO see Sheffield LJ et al biochemical phenotype, 558 Rye PJ see Laing IA et al Silvestri G see Hughes AE et al Silvestro L see Silengo M et al Simon-Bouy B see Muller F et al Sabbatino MS see Melis D et al Sabry MA see Tayel SM et al Sinke RJ see Sjarif DR et al Sitepu M see Handoko HY et al see van Steensel M et al
Sabry MA et al. Genotypic/phenotypic heterogeneity of
Kenny-Caffey syndrome: letter, 1054 Sjarif DR et al. Clinical heterogeneity and novel mutations in the glycerol kinase gene in three families with isolated glycerol kinase deficiency, 650 Kenny-Čaffey syndrome CATCH of the Kenny-Caffey syndrome is part of the CATCH 22 haploinsufficiency cluster, 31

Sacks N see Watson M et al

Sagawa T see Hoshi N et al

Saggar-Malik AK see MacDermot KD et al

Sakkubai N see Xiang F et al

Salat U see Wöhrle D et al

Saleh QA see Sabry MA et al

Salonen R, Paavola P. Meckel syndrome: syndrome of the month, 497

Salvaggio F. see de Franchis R et al part Skirton H et al. Recommendations for Education and Training of Genetic Nurses and Counsellors in the United Kingdom, 410 Skuse D see Dalton P et al Slaney SF et al. An autosomal or X linked mutation results in true hermaphrodites and 46,XX males in the same family, 17 Slim R see Mustapha M et al Sloan-Béna F et al. Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH, 146 Salvaggio E see de Franchis R et al Sampson J see Olschwang S et al Sánchez A see Pujana MA et al Smeets DFCM see Yntema HG et al Sankila E-M see Ala-Mello S et al Smeets E see Fryns J-P and Smeets E Sanoudou D see Griffin DK et al Sanousi AAE see Bonthron DT et al Santos M see Pratesi R et al Smith RJH see Lench NJ et al Smits APT see Yntema HG et al Solans T see Bayés M et al Soler A see Margarit E et al Solivelles X see Austin-Ward E et al Saouda M see Mustapha M et al Saraiva JM et al. Absence of a del(22q11) in a patient with the 3C (craniocerebellocardiac) syndrome: letter, 347 Speer MC et al. Evidence for anticipation in autosomal dominant Sarda P see Verloes A et al Sarfarazi M see Stoilova D et al limb-girdle muscular dystrophy, 305 Sperandeo MP see de Franchis R et al Sarkadi B see Tordai A et al Sarojini B see Xiang F et al Sasaki H see Yorifuji T et al see Melis D et al Spigelman A see Olschwang S et al Spiro RP see Precht KS et al Sato M et al. Identification of five novel germline mutations of the MEN1 gene in Japanese multiple endocrine neoplasia type 1 (MEN1) families, 915 Stack B see Gilfillan A et al Stajich JM see Speer MC et al Standaert L see Devriendt K et al Stanziale P see Gasparini P et al Satoh S see Onishi H et al

Stavropoulou C et al. Severe phenotype resulting from an active ring Trembath RC see Jackson SNJ et al X chromosome in a female with a complex karyotype: Trent RJ see Yu B et al characterisation and replication study, 932 Steinbach P see Wöhrle D et al Stephens RS see Davies AF et al see Yu B et al Trivier E see Merienne K et al
Trueman L see Evans DGR et al
Trump D et al. Localisation of X linked recessive idiopathic
hypoparathyroidism to a 1.5 Mb region on Xq26-q27, 905 Stephens RS see Davies AF et al
Stephenson JBP see Munroe PB et al
Stephenson JBP see Crow YJ et al
Stewart AD see Robinson R et al
Stoffelen D see Devriendt K et al
Stoilova D et al. Novel TIGR/MYOC mutations in families with
juvenile onset primary open angle glaucoma, 989
Stormer P see Glass IA et al
Strain T see Evans DGR et al
Strain I. see Porteous MEM et al Tryggvason K see Guicheney P et al Tuddenham EGD see Waccy AI and Tuddenham EGD
Tumer Z see Silahtaroglu AN et al
Tupler R et al. Identical de novo mutation at the D4F104S1 locus in
monozygotic male twins affected by facioscapulohumeral muscular dystrophy (FSHD) with different clinical expression, Strain L see Porteous MEM et al Stratton M see Olschwang S et al Stratton MR see Chang-Claude J et al Turlin B see Pinson S et al Turnbull DM see Chinnery PF et al Turner G see Cohen Jr MM et al Tusié-Luna MT see López-Gutiérrez AU et al Strenge S see Meiner A et al Strisciuglio P see Concolino D et al Sudoyo HA see Handoko HY et al Tuzzi MR see Melis D et al Sugiyama N see Onishi H et al Uematsu A see Yorifuji T et al see Yorifuji T et al Sumner AT et al. A FISH study of chromosome fusion in the ICF syndrome: involvement of paracentric heterochromatin but not of the centromeres themselves, 833 Unterberger U see Berger A et al Sunada F et al. MRI findings in a patient with partial monosomy Vainzof M see Moreira ES et al 10p, 159 van Asperen CJ et al. Familial neurofibromatosis type 1 associated Swingler RJ see Hayward C et al with an overgrowth syndrome resembling Weaver syndrome, 323 van Bokhoven H see Yntema HG et al van Broeckhoven C see Nelis E et al Taillemite J-L see Portnoï M-F et al Takahara J see Sato M et al Takahashi Y see Nagai K et al van Camp G see Fransen E et al see Lench NJ et al Takata A see Kikuchi H et al Takata S see Yamamoto K et al Takekoshi Y see Nagai K et al Talbot I see Olschwang S et al Tam DA see Sunada F et al Vance GH see Peet J et al van de Heyning PJ see Lench NJ et al van den Bergh P see Seneca S et al van den Helm B see Yntema HG et al Tardieu S see Meggouh F et al van den Ouweland AMW see Verhoef S et al van den Ouweland AMW see Verhoef S et al
van Diggelen OP see van Royen-Kerkhof A et al
van Dyke DL see Czarnecki PM et al
Vanhole C see Moerman P et al
van Hole C see Devriendt K et al
van Lingen C see Fryns J-P et al
van Roosmalen T see Yntema HG et al
van Royen-Kerkhof A et al. Coexistence of Gaucher disease type 1
and Joubert syndrome: letter, 965
van Steensel M et al. Robinow syndrome: letter, 349 Tarttelin EE see Inglehearn CF et al see Inglehearn CF et al
Tassin J see Meggouh F et al
Tavares de Almeida I see Rivera I et al
Tawn EJ see Chase DS et al Tayel SM et al. Triophthalmia and facial clefting: a case report, 875 Taylor R see Inglehearn CF et al Teare D see Mirza MM et al van Steensel M et al. Robinow syndrome: letter, 349 van Thienen M-N see Devriendt K et al van Tijn DA see van Asperen CJ et al van Tongerloo A, de Paepe A. Psychosocial adaptation in Teare MD see Arason A et al

Teebi AS et al. Spastic paraplegia, optic atrophy, microcephaly with
normal intelligence, and XY sex reversal: a new autosomal
recessive syndrome?, 759 Tejada MI see Alvarez AI et al adolescents and young adults with Marfan syndrome: an Tello A see Lapunzina P et al Tempelaars A see Verhoef S et al Temple IK see Law CJ et al Thakker N see Wu CL et al exploratory study, 405

Váradi A see Tordai A et al

Varley J see Lalloo F et al

Vassal HB et al. Familial persistent pulmonary hypertension of the Thakker RV see Trump D et al newborn resulting from misalignment of the pulmonary vessels (congenital alveolar capillary dysplasia), 58
Vats S see Czarnecki PM et al
Velagaleti GVN see Pivnick EK et al
Vellodi A see Beesley CE et al
Vendrell T see Margarit E et al Thamm B see Meiner A et al The neurofibromatosis team of Sophia Children's Hospital see Cnossen MH et al Thomas G see Mirza MM et al see Olschwang S et al Thomas NST see Ferguson BM et al Verheijen FW et al. Fibroblast silver loading for the diagnosis of Menkes disease, 849
Verhoef S et al. Familial cylindromatosis mimicking tuberous sclerosis complex and confirmation of the cylindromatosis locus, Thompson E see Savarirayan R et al Thompson PW, Davies SJ. Frequency of inherited deletions of 22q11: letter, 789 Thomson PD see Halkas AC et al CYLD1, in a large family, 841

Verlingue C et al. Absence of mutations in the interspecies conserved regions of the CFTR promoter region in cystic fibrosis (CF) and CF related patients, 137

Verloes A et al. Juvenile rheumatoid arthritis and del(22q11) Tibben A see Dudokdewit AC et al Tiepolo L see Tupler R et al Timms KM et al. Reassessment of biochemically determined Hunter syndrome carrier status by DNA testing, 646

Tischkowitz M et al. Familial gonadal tumours: letter, 84

Tolmie JL see Crow YJ and Tolmie JL

see Crow YJ et al

see Ghaffari SR et al syndrome: a non-random association, 943 Viersbach R see Wieczorek D et al Vignier N see Guicheney P et al **Vignola S** see di Rocco M et al see Ghaffari SR et al Vilageliu L see Bayés M et al Tomás M see Martinez F et al Tomé FMS see Guicheney P et al Tomlinson I see Olschwang S et al see Chabás A et al Vilela C see Espinos C et al
Vincent M-C et al. Extensive germinal mosaicism in a family with X linked myotubular myopathy simulates genetic heterogeneity, 241
Vintiner GM see Mossey PA et al
Vitelli F see Jonsson JJ et al
Vithana E see Inglehearn CF et al
Vithana E et al. RP11 is the second most common locus for Tomlinson IPM see Marsh DJ et al Tomlinson P see Savarirayan R et al Tommerup N see Okamoto K et al see Silahtaroglu AN et al Topaloglu H see Guicheney P et al dominant retinitis pigmentosa: letter, 174
Vits L see Fransen E et al
Vogel W see Wöhrle D et al
Volpini V see Pujana MA et al Tordai A et al. High frequency of the haemochromatosis C282Y mutation in Hungary could argue against a Celtic origin of the mutation: letter, 878

Toriello HV. Chondrodysplasia punctata and maternal systemic lupus erythematosus: commentary, 698

Toth T et al. Prenatal detection of trisomy 21 and 18 from amniotic Votruba M et al. Clinical features, molecular genetics, and votruba M et al. Clinical features, molecular geneucs, and pathophysiology of dominant optic atrophy: review, 793
Vuillard E see Muller F et al
Vuillaumier S see Verlingue C et al
Vulliamy TJ see Knight SW et al
Vuopala K see Jukkola A et al
Vuzevski VD see Verhoef S et al fluid by quantitative fluorescent polymerase chain reaction, 126 Toth-Pai E see Toth T et al
Trabetti E et al. Association of the FcεRIβ gene with bronchial hyper-responsiveness in an Italian population, 680 Trainer AH see Ghaffari SR et al

Wacev AI, Tuddenham EGD. Mutation databases on the web: edito-Wade Walsh M see Watson M et al Waldron-Lynch F see McDermott MF et al Walford-Moore J see Skirton H et al Wallace A see Evans DGR et al Wallace MR see Rasmussen SA et al Ward S see Ali JBM et al Ward TA see Cunliffe HE et al
Warner JP see Gilfillan A et al
Warner TT see Jarman PR and Warner TT
Watson M et al. Family history of breast cancer: what do women understand and recall about their genetic risk?, 731 Watson R see Flanagan N et al Watters G see Teebi AS et al Weaver DD see Peet J et al Webb J see Murray A et al Webb T et al. Linkage analysis in Rett syndrome families suggests that there may be a critical region at Xq28, 997 Weber BHF see Gehrig A et al Weber JL see Fenske CD et al Weiss L see Rasmussen SA et al Weissenbach J see Heinzlef O et al Weninger M see Berger A et al West SP see Robinson R et al Westbrook CA see Speer MC et al White SJ see Sheffield LJ et al Whyte MP see Trump D et al Wieczorek D et al. Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs, 545 Wiedemann HR see Okamoto K et al Wienberg J see Griffin DK et al Wilcock G see Kehoe PG et al Willatt LR see Barber JCK et al Willems PJ see Fransen E et al see Lench NJ et al
Williams B see Jackson SNJ et al
Williams DK see Long FL et al
Williams H see Kehoe PG et al Williamson R see Delatycki MB et al Wilson DI see Ryan A et al
Wilson DJ see Lee WK et al
Wilson MJ see Mowat DR et al
Winchester BG see Beesley CE et al Winter RM see Ryan AK et al winter KW see Kyan AK et al
see Vassal HB et al
Wirapati PJ see Handoko HY et al
Wirth J see Silahtaroglu AN et al
Wöhrle D et al. Unusual mutations in high functioning fragile X males: apparent instability of expanded unmethylated CGG repeats, 103
Wollmann HA see Eggermann T et al Wong L-JC see Liang M-H et al see Liang M-H et al Wooding C see Trump D et al Woods CG see Webb T et al

Wright JT see Price JA et al

Wu CL et al. Differential diagnosis of type 2 neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas, 973 Wuilque F see Sloan-Béna F et al Wyke J see Olschwang S et al Xiang F et al. Chromosome mapping of Rett syndrome: a likely candidate region on the telomere of Xq, 297 Yagishita S see Onishi H et al Yahyaoui M see Meggouh F et al Yamada H see Hoshi N et al Yamada Y see Onishi H et al Yamakawa-Kobayashi K see Kawashima T et al Yamamoto K et al. A pedigree analysis with minimised ascertainment bias shows anticipation in Met30-transthyretin related familial amyloid polyneuropathy, 23 Yamanaka C see Yorifuji T et al Yamaoka LH see Speer MC et al Yanagisawa N see Yamamoto K et al Yanai S see Nagai K et al Yankowitz J see Peleg D and Yankowitz J Yaouanq J see Pinson S et al Yates JRW see Ali JBM et al see Barber JCK et al see Timms KM et al Yntema HG et al. Localisation of a gene for non-specific X linked mental retardation (MRX46) to Xq25-q26, 801 Yoneyama H see Kikuchi H et al Yorifuji T et al. Kenny-Caffey syndrome without the CATCH 22 deletion: letter, 1054 Uniparental and functional X disomy in Turner syndrome patients with unexplained mental retardation and X derived marker chromosomes, 539

Young EP see Beesley CE et al

Yu B et al. Counselling issues in familial hypertrophic cardiomyopathy, 183 Molecular pathology of familial hypertrophic cardiomyopathy caused by mutations in the cardiac myosin binding protein C gene, 205 Yuksel B see Brady AF et al Zaider E see Frank J et al Zaki M see Sabry MA et al see Sabry MA et al

Zaragoza MV et al. Studies of non-disjunction in trisomies 2, 7, 15, and 22: does the parental origin of trisomy influence placental morphology?, 924

Zatz M see Eggers S and Zatz M see Moreira ES et al Zauber P see Olschwang S et al Zeitoun G see Mustapha M et al Zelante L see Gasparini P et al

Zhang X see Guicheney P et al Zhang Z see Xiang F et al

Zhou C see Broom MF et al Zonana J see Ferguson BM et al Zuberi SM see Crow YJ et al

Zuffardi O see Pirola B et al

## VOLUME 35 • SUBJECT INDEX

- 18p-, Clinical features and mental development of a child with a prenatally identified 45,XX,der(5)t(5;18)(p15;q11.2),-18 karyotype, 865
- 19p13.3, Peutz-Jeghers disease: most, but not all, families are compatible with linkage to 19p13.3, 42
- 22q11 deletion, Clinical features of chromosome 22q11 deletion: let-
  - Craniosynostosis and chromosome 22q11 deletion: letter, 346
- Reply to letters regarding clinical features of chromosome 22q11 deletion: letter, 347
- 22q11.2 deletion, Absence of a del(22q11) in a patient with the 3C (craniocerebellocardiac) syndrome: letter, 347
- 22q11.2 haploinsufficiency cluster, Kenny-Caffey syndrome is part of the CATCH 22 haploinsufficiency cluster, 31
- 2q13 deletion, Molecular studies in Finnish patients with familial juvenile nephronophthisis exclude a founder effect and support a common mutation causing mechanism, 279
- 3C syndrome, Absence of a del(22q11) in a patient with the 3C (craniocerebellocardiac) syndrome: letter, 34'
- 46,XX male, An autosomal or X linked mutation results in true hermaphrodites and 46,XX males in the same family, 17
- -, Clinical features and mental development of a child with a prenatally identified 45,XX,der(5)t(5;18)(p15;q11.2),-18 karyotype,
- 6q deletion, Agenesis of the corpus callosum with Probst bundles owing to haploinsufficiency for a gene in an 8 cM region of 6q25,
- 7q21.2, Another holoprosencephaly locus at 7q21.2?: letter, 614
- absent speech, Two 22q telomere deletions serendipitously detected by FÎSH, 939
- acampomelic campomelic dysplasia, Acampomelic campomelic dysplasia with de novo 5q;17q reciprocal translocation and severe phenotype, 597
- achondroplasia, Medical complications of achondroplasia: a multicentre patient review, 705
- acinar dysplasia, Severe primary pulmonary hypoplasia ("acinar dysplasia") in sibs: a genetically determined mesodermal defect?: letter, 964
- active r(X) chromosome. Severe phenotype resulting from an active ring X chromosome in a female with a complex karyotype: characterisation and replication study, 932
- adolescents, Psychosocial adaptation in adolescents and young adults with Marfan syndrome: an exploratory study, 405
- adrenocortical carcinoma, Simultaneous adrenocortical carcinoma and ganglioneuroblastoma in a child with Turner syndrome and germline p53 mutation, 328
- ADRP, A linkage survey of 20 dominant retinitis pigmentosa families: frequencies of the nine known loci and evidence for further
- heterogeneity, 1

  Advisory Committee on Genetic Testing, "Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public". Advisory Committee on Genetic Testing, 443
- affected women, Knowledge, views, and experience of 25 women
- with myotonic dystrophy, 1020
  agenesis of the corpus callosum, Agenesis of the corpus callosum
  with Probst bundles owing to haploinsufficiency for a gene in an 8
- cM region of 6q25, 1031 aldolase B, Hereditary fructose intolerance: review, 353
- alpha-1-proteinase inhibitor, Variants of  $\alpha_1$ -proteinase inhibitor in black and white South African patients with focal glomeruloscle-
- rosis and minimal change nephrotic syndrome, 6
  Alport syndrome, Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene
- deletion syndrome?, 273
  ALS, Homozygosity for Asn86Ser mutation in the CuZn-superoxide dismutase gene produces a severe clinical phenotype in a juvenile onset case of familial amyotrophic lateral sclerosis: letter, 174
- Alzheimer's disease, De novo presenilin 1 mutations are rare in clinically sporadic, early onset Alzheimer's disease cases, 672
- K variant and susceptibility butyrylcholinesterase
- Alzheimer's disease, 1034

  anal anomalies, Familial craniosynostosis, anal anomalies, and porokeratosis: CAP syndrome, 763

  aneuploidy, 49,XXXXY: a distinct phenotype. Three new cases and review, 420

  Cheroid playes guest and aneuploidy, 554
  - Choroid plexus cysts and aneuploidy, 554
- Angelman syndrome, Molecular screening for proximal 15q abnormalities in a mentally retarded population, 534
  - The mechanisms involved in formation of deletions and duplications of 15q11-q13, 130

- Two 22q telomere deletions serendipitously detected by FISH, 939 angiotensin II type 1 receptor, Distortion of maternal-fetal angiotensin II type 1 receptor allele transmission in pre-eclampsia,
- anhidrotic ectodermal dysplasia, Autosomal recessive anhidrotic ectodermal dysplasia in a large Moroccan family, 1043
- animal models, Ovine neuronal ceroid lipofuscinosis: a large animal model syntenic with the human neuronal ceroid lipofuscinosis variant CLN6, 717
- velocardiofacial syndrome: letter, 789 annual incidence,
- anterior chamber eye defects, Distal 6p deletion syndrome: a report of a case with anterior chamber eye anomaly and review of published reports, 685
- anticipation, A pedigree analysis with minimised ascertainment bias shows anticipation in Met30-transthyretin related familial amyloid polyneuropathy, 23
  - Evidence for anticipation in autosomal dominant limb-girdle muscular dystrophy, 305
- antiphospholipid antibodies, Neonatal lupus syndrome: a case with chondrodysplasia punctata and other unusual manifestations, 695
- anti-Ro/SSA antibodies, Neonatal lupus syndrome: a case with chondrodysplasia punctata and other unusual manifestations, 695
- antithrombin III, Mild phenotype associated with an interstitial deletion of the long arm of chromosome 1, 1047
- Apert syndrome, Presence of the Apert canonical S252W FGFR2 mutation in a patient without severe syndactyly, 677
- aplasia cutis congenita, Extensive form of aplasia cutis congenita: a new syndrome?, 609
- apnoeic spells, Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation, 342
- arylsulphatase, Segregation of mutations in arylsulphatase E and correlation with the clinical presentation of chondrodysplasia punctata, 1004
- ascertainment bias, A pedigree analysis with minimised ascertainment bias shows anticipation in Met30-transthyretin related familial amyloid polyneuropathy, 23
- Ashkenazi Jews, An evaluation of common breast cancer gene mutations in a population of Ashkenazi Jews, 10
- asthma, A polymorphism of the CC16 gene is associated with an
- increased risk of asthma, 463
  Association of the FcεRIβ gene with bronchial hyper-responsiveness in an Italian population, 680
- Astley-Kendall dysplasia, A lethal skeletal dysplasia with features of chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a rare genetic disorder, 505
- ataxia, Two adult females with a distinct familial mental retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, hypergonadotrophic hypogonadism, and retinal dystrophy, 333
- atelosteogenesis type 2, Atelosteogenesis type 2: syndrome of the month, 49
- atopic dermatitis, Linkage and association of an interleukin 4 gene
- polymorphism with atopic dermatitis in Japanese families, 502 audiotape, Family history of breast cancer: what do women understand and recall about their genetic risk?, 731
- autoclaving, Autoclaving Guthrie cards does not prevent their use in PCR reactions!: letter, 702
- autosomal dominant, Autosomal dominant juvenile recurrent parotitis, 417
  Evidence for anticipation in autosomal dominant limb-girdle
- muscular dystrophy, 305
- Exclusion of the familial Mediterranean fever locus as a susceptibility region for autosomal dominant familial Hibernian fever, 432
- autosomal dominant polycystic kidney disease, Prenatal diagnosis of autosomal dominant polycystic kidney disease (PKD1) presenting in utero and prognosis for very early onset disease, 1
- autosomal dominant retinitis pigmentosa, RP11 is the second most common locus for dominant retinitis pigmentosa: letter, 174
- autosomal recessive, Familial persistent pulmonary hypertension of the newborn resulting from misalignment of the pulmonary vessels (congenital alveolar capillary dysplasia), 58
- Four sibs with dislocated elbows, bowed tibiae, scoliosis, deafness cataract, microcephaly, and mental retardation: a new MCA/MR syndrome, 755
- Oto-onycho-peroneal syndrome: confirmation of a syndrome, 508 in a patient with the diaphragmatic hernia-Proteinuria hypertelorism-myopia-deafness syndrome: further evidence that the facio-oculo-acoustico-renal syndrome represents the same
- entity, 70 autosomal recessive retinitis pigmentosa. A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33,

autosomal recessive transmission, Autosomal recessive anhidrotic

ectodermal dysplasia in a large Moroccan family, 1043 awareness, Men in breast cancer families: a preliminary qualitative study of awareness and experience, 739

Baller-Gerold syndrome, Baller-Gerold syndrome associated with congenital portal venous malformation, 767

Bannayan-Riley-Ruvalcaba syndrome, A survey of phenotypic features in juvenile polyposis, 476

Mutations of PTEN in patients with Bannayan-Riley-Ruvalcaba phenotype, 886

Batten disease, Ovine neuronal ceroid lipofuscinosis: a large animal model syntenic with the human neuronal ceroid lipofuscinosis variant CLN6, 717

Beckwith-Wiedemann syndrome, Beckwith-Wiedemann syndrome in a child with chromosome 18q deletion, 162
Is p57<sup>KIP2</sup> mutation a common mechanism for Beckwith-Wiedemann

syndrome or somatic overgrowth?: letter, 86
bentiromide test, Pancreatic exocrine dysfunction associated with mitochondrial tRNA<sup>Leu(UUR)</sup> mutation, 255

Best vitelliform macular dystrophy, Lack of evidence for genetic heterogeneity in Best vitelliform macular dystrophy: letter, 85

BHR, Association of the FcεRIβ gene with hyper-responsiveness in an Italian population, 680 with bronchial

bone fracture, Neonatal cholestasis and focal medullary dysplasia of the kidneys in a case of microcephalic osteodysplastic primordial dwarfism, 61

bowed tibiae, Four sibs with dislocated elbows, bowed tibiae, scoliosis, deafness, cataract, microcephaly, and mental retardation: a new MCA/MR syndrome, 755

brachytelephalangic type, Maternal systemic lupus erythematosus and chrondrodysplasia punctata in two sibs: phenocopy or coincidence?, 690

BRCA1, A population study of mutations and LOH at breast cancer gene loci in tumours from sister pairs: two recurrent mutations seem to account for all BRCA1/BRCA2 linked breast cancer in

BRCA1/2 mutations, An evaluation of common breast cancer gene mutations in a population of Ashkenazi Jews, 10

BRCA2, A population study of mutations and LOH at breast cancer gene loci in tumours from sister pairs: two recurrent mutations seem to account for all BRCA1/BRCA2 linked breast cancer in Iceland, 446

breast, Germline PTEN mutations in Cowden syndrome-like families, 881

breast cancer, Family history of breast cancer: what do women understand and recall about their genetic risk?, 731

butyrylcholinesterase K variant, The butyrylcholinesterase K variant and susceptibility to Alzheimer's disease, 1034

C282Y mutation, Frequency of the HFE C282Y and H63D mutations in distinct ethnic groups living in Spain: letter, 701 High frequency of the haemochromatosis C282Y mutation in Hun-

gary could argue against a Celtic origin of the mutation: letter, 878 CAG repeats, Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method, 99

cancer risks, Using gene carrier probability to select high risk families for identifying germline mutations in breast cancer susceptibility genes, 116

cardiac abnormalities, Distal 10q trisomy syndrome with unusual cardiac and pulmonary abnormalities, 72

carrier analysis, Reassessment of biochemically determined Hunter

syndrome carrier status by DNA testing, 646
cataplexy, "Cataplexy" and muscle ultrasound abnormalities in Coffin-Lowry syndrome, 94
"Cataplexy" in Coffin-Lowry syndrome: letter, 702

cataract, Four sibs with dislocated elbows, bowed tibiae, scoliosis, deafness, cataract, microcephaly, and mental retardation: a new MCA/MR syndrome, 755

CATCH 22, Genotypic/phenotypic heterogeneity of Kenny-Caffey syndrome: letter, 1054

Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster, 31

Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster: letter, 1054

Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster: letter, 1054

Kenny-Caffey syndrome without the CATCH 22 deletion: letter,

CBAVD, Linkage disequilibrium between the M470V variant and the IVS8 polyT alleles of the CFTR gene in CBAVD, 594
CC16, A polymorphism of the CC16 gene is associated with an increased risk of asthma, 463

cell adhesion molecule, Genotype-phenotype correlation in L1 associated diseases, 399

The site of a missense mutation in the extracellular Ig or FN domains of L1CAM influences infant mortality and the severity of X linked hydrocephalus, 901

Central areolar choroidal dystrophy, Fine localisation of the gene for central areolar choroidal dystrophy on chromosome 17p, 770 centric inversion. Not para-, not peri-, but centric inversion of chromosome 12, 682

centromere fission, Not para-, not peri-, but centric inversion of chromosome 12, 682

centromeres, A FISH study of chromosome fusion in the ICF syndrome: involvement of paracentric heterochromatin but not of the centromeres themselves, 833

cephalometrics, Prediction of liability to orofacial clefting using genetic and craniofacial data from parents, 371

cerebrocostomandibular syndrome, Hypoplastic left heart in cerebrocostomandibular syndrome: letter, 879

CFEOM, Locus heterogeneity in autosomal dominant congenital external ophthalmoplegia (CFEOM), 985

CFTR, Cystic fibrosis and mucins: letter, 82

CFTR gene, Linkage disequilibrium between the M470V variant and the IVS8 polyT alleles of the CFTR gene in CBAVD, 594

CFTR mutations, Absence of mutations in the interspecies conserved regions of the CFTR promoter region in cystic fibrosis (CF) and CF related patients, 137

CFTR promoter, Absence of mutations in the interspecies conserved regions of the CFTR promoter region in cystic fibrosis (CF) and CF related patients, 137

CGH, Chromosome specific comparative genome hybridisation for determining the origin of intrachromosomal duplications, 3

Charcot-Marie-Tooth disease, Diagnosis of the duplication by PCR based detection of a novel junction fragment: letter, 962

The first de novo mutation of the connexin 32 gene associated with X linked Charcot-Marie-Tooth disease, 251

China, Chinese geneticists approach ethics: letter, 83

choanal atresia, Extensive form of aplasia cutis congenita: a new syndrome?, 609

cholesterol, Smith-Lemli-Opitz syndrome: a variable clinical and biochemical phenotype, 558

chondrodysplasia, Chondrodysplasia punctata and maternal systemic lupus erythematosus: commentary, 698

chondrodysplasia punctata, Maternal systemic lupus erythematosus and chrondrodysplasia punctata in two sibs: phenocopy or coincidence?, 690

Neonatal lupus syndrome: a case with chondrodysplasia punctata and other unusual manifestations, 695

egregation of mutations in arylsulphatase E and correlation with the clinical presentation of chondrodysplasia punctata, 1004

chorionic somatomammotrophin hormone 1 (CSH1), Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome, 784

choroid plexus calcifications, Unknown syndrome: peculiar face, severe hypodontia of permanent teeth, and precocious choroid calcifications, 435

choroid plexus cyst, Choroid plexus cysts and aneuploidy, 554

**chromosomal abnormalities,** Outcome of chromosomally normal livebirths with increased fetal nuchal translucency at 10-14 weeks' gestation, 222

chromosome 11, Lack of evidence for genetic heterogeneity in Best vitelliform macular dystrophy: letter, 85

chromosome 12, Locus heterogeneity in autosomal dominant congenital external ophthalmoplegia (CFEOM), 985

chromosome 14, De novo unbalanced translocation resulting in monosomy for proximal 14q and distal 4p in a fetus with intrauterine growth retardation, Wolf-Hirschhorn syndrome, hypertrophic cardiomyopathy, and partial hemihypoplasia, 1050 chromosome 16, Evidence of linkage of the inflammatory bowel dis-

ease susceptibility locus on chromosome 16 (IBD1) to ulcerative colitis, 218

Familial cylindromatosis mimicking tuberous sclerosis complex and confirmation of the cylindromatosis locus, CYLD1, in a large family, 841

chromosome 17p, Fine localisation of the gene for central areolar choroidal dystrophy on chromosome 17p, 770

chromosome 18q22.1, Beckwith-Wiedemann syndrome in a child with chromosome 18q deletion, 162

chromosome 19, Mosaic supernumerary ring chromosome 19 identified by comparative genomic hybridisation, 836 chromosome 19q13, A new family of Greek origin maps to the CRD

locus for autosomal dominant cone-rod dystrophy on 19q, 429 chromosome 1q, Mild phenotype associated with an interstitial dele-

tion of the long arm of chromosome 1, 1047

chromosome 2, Hirschsprung disease, microcephaly, mental retarda-tion, and characteristic facial features: delineation of a new syndrome and identification of a locus at chromosome 2q22-q23, 617

Trisomy 2q11.2-q21.1 resulting from an unbalanced insertion in two generations, 319

chromosome 22q11, Prenatal diagnosis by FISH of a 22q11 deletion in two families, 165

chromosome 2q, A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33, 141

chromosome 2q31, Mesoaxial complete syndactyly and synostosis with hypoplastic thumbs: an unusual combination or homozygous expression of syndactyly type I?, 868

chromosome 4, De novo unbalanced translocation resulting in monosomy for proximal 14q and distal 4p in a fetus with intrauterine growth retardation, Wolf-Hirschhorn syndrome, hypertrophic cardiomyopathy, and partial hemihypoplasia, 1050

- chromosome 6, Distal 6p deletion syndrome: a report of a case with anterior chamber eye anomaly and review of published reports,
- chromosome 6p, Further evidence for the involvement of human chromosome 6p24 in the aetiology of orofacial clefting, 857
- chromosome 7, Intrachromosomal triplication of distal 7p, 78
  chromosome 8, Trisomy 2q11.2→q21.1 resulting from an
  unbalanced insertion in two generations, 319
  chromosome 8p23.1, Duplication of 8p23.1: a cytogenetic anomaly
- with no established clinical significance, 491
- chromosome 9p, Further evidence for the involvement of human chromosome 6p24 in the aetiology of orofacial clefting, 857
- chromosome aberration, Two cases of partial trisomy 8p and partial monosomy 21q in a family with a reciprocal translocation (8;21)(p21.1;q22.3), 604

  chromosome deletion, Beckwith-Wiedemann syndrome in a child
- with chromosome 18q deletion, 162
- classification, Congenital malformations: an inquiry classification and nomenclature, 661
- clinical features, Clinical features, molecular genetics, pathophysiology of dominant optic atrophy: review, 793
- clinical genetics, Contracting for clinical genetic services: the Welsh model, 309
- CMT1, Mutation analysis of the nerve specific promoter of the peripheral myelin protein 22 gene in CMT1 disease and HNPP, 590
- CMT1A, A molecular, cytogenetic, and clinical evaluation of mosaic tandem duplication 17p and Charcot-Marie-Tooth type 1A
- neuropathy, 169
  Diagnosis of the CMT1A duplication by PCR based detection of a
- novel junction fragment: *letter*, 962

  CNS malformation, Ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum: variable expression of a single syndrome?, 157
- Coffin-Lowry syndrome, "Cataplexy" and muscle ultrasound abnormalities in Coffin-Lowry syndrome, 94
  - "Cataplexy" in Coffin-Lowry syndrome: letter, 702
- Rapid immunoblot and kinase assay tests for a syndromal form of X linked mental retardation: Coffin-Lowry syndrome, 890 COL5A2, A single base mutation in COL5A2 causes Ehlers-Danlos
- syndrome type II, 846
- coloboma, The prevalence of PAX2 mutations in patients with isolated colobomas or colobomas associated with urogenital anomalies, 806
- colorectal cancer, Genetic implications of double primary cancers of the colorectum and endometrium, 978 comparative genomic hybridisation, A new strategy for cryptic
- telomeric translocation screening in patients with idiopathic mental retardation, 225
  - Mosaic supernumerary ring chromosome 19 identified by comparative genomic hybridisation, 836
- complex chromosome rearrangement (CCR), A new approach to the elucidation of complex chromosome rearrangements illustrated by a case of Rieger syndrome, 234
- computer generated colour ideograms, A new approach to the elucidation of complex chromosome rearrangements illustrated by a case of Rieger syndrome, 234
- cone-rod dystrophy, A new family of Greek origin maps to the CRD locus for autosomal dominant cone-rod dystrophy on 19q, 429
- cone-rod retinal dystrophy gene, Microsatellite markers for the cone-rod retinal dystrophy gene, CRX, on 19q13.3: letter, 527 congenital adrenal hyperplasia, Uniparental disomy for chromosome 6 results in steroid 21-hydroxylase deficiency: evidence of different genetic mechanisms involved in the production of the disease, 1014
- congenital alveolar capillary dysplasia, Familial persistent pulmonary hypertension of the newborn resulting from misalignment of the pulmonary vessels (congenital alveolar capillary dysplasia), 58 congenital heart defect, Tricuspid atresia in sibs: letter, 1055
- congenital malformations, Congenital malformations: an inquiry into classification and nomenclature, 661
- congenital muscular dystrophy, PCR based mutation screening of the laminin α2 chain gene (LAMA2): application to prenatal diagnosis and search for founder effects in congenital muscular
- dystrophy, 211
  connexin 26 (GJB2), A Moroccan family with autosomal recessive sensorineural hearing loss caused by a mutation in the gap junction protein gene connexin 26 (GJB2), 151
- connexin 32, The first de novo mutation of the connexin 32 gene associated with X linked Charcot-Marie-Tooth disease, 251
- consanguinity, Autosomal recessive anhidrotic ectodermal dysplasia in a large Moroccan family, 1043
  contracting, Contracting for clinical genetic services: the Welsh model, 309
- copper, Fibroblast silver loading for the diagnosis of Menkes disease,
- Costello syndrome, Costello syndrome in two Brazilian children, 54

- Costello syndrome: two cases with embryonal rhabdomyosarcoma,
- Costello syndrome: syndrome of the month, 238
- counselling, Counselling issues familial hypertrophic
- cardiomyopathy, 183
  counsellors, Recommendations for Education and Training of
  Genetic Nurses and Counsellors in the United Kingdom, 410
- Cowden disease, Mutations of PTEN Bannayan-Riley-Ruvalcaba phenotype, 886 in patients
- Cowden syndrome, A survey of phenotypic features in juvenile polyposis, 476
- Germline PTEN mutations in Cowden syndrome-like families, 881 Craniosynostosis and chromosome craniosynostosis, deletion: letter, 346
  - Familial craniosynostosis, anal anomalies, and porokeratosis: CAP
- syndrome, 763 cri du chat, Clinical features and mental development of a child with a prenatally identified 45,XX,der(5)t(5;18)(p15;q11.2),-18 karyotype, 865
- Crohn's disease, Evidence of linkage of the inflammatory bowel disease susceptibility locus on chromosome 16 (IBD1) to ulcerative colitis, 218
- CRX, Microsatellite markers for the cone-rod retinal dystrophy gene, CRX, on 19q13.3: letter, 527
- cryptic splice site, A silent mutation, C924T (G308G), in the
- L1CAM gene results in X linked hydrocephalus (HSAS), 456 cryptic translocations, Del(18p) shown to be a cryptic translocation using a multiprobe FISH assay for subtelomeric chromosome
- rearrangements, 722 CTG repeat instability, Instability in the normal CTG repeat range at the myotonic dystrophy locus: letter, 791
- lopia, Cyclopia and sirenomelia in a liveborn infant: letter, 263
- CYLD1 gene, Familial cylindromatosis mimicking tuberous sclerosis complex and confirmation of the cylindromatosis locus, CYLD1, in a large family, 841
- cylindromatosis, Familial cylindromatosis mimicking tuberous sclerosis complex and confirmation of the cylindromatosis locus, CYLD1, in a large family, 841
- cystic fibrosis, Cystic fibrosis and mucins: letter, 82
  - Cystic fibrosis in a Puerto Rican female homozygous for the R1066C mutation: letter, 84

    Cystic fibrosis screening: a fetus with hyperechogenic bowel may be
  - the index case, 657
  - Is meconium ileus genetically determined or associated with a more severe evolution of cystic fibrosis?: letter, 262
- P67L: a cystic fibrosis allele with mild effects found at high frequency in the Scottish population, 122 cytochrome c oxidase (COX) deficiency, A systematic mutation screen of 10 nuclear and 25 mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA ser(UCX) mutations in a subgroup with syndromal in a subgroup mutations syndromal encephalopathy, 895
- Dandy-Walker malformation, Oral-facial-digital syndrome type IX
- in a patient with Dandy-Walker malformation, 342

  DC1, 1.4 Mb candidate gene region for X linked dyskeratosis congenita defined by combined haplotype and X chromosome inactivation analysis, 993
- De Barsy syndrome, New lethal disease involving type I and III collagen defect resembling geroderma osteodysplastica, De Barsy syndrome, and Ehlers-Danlos syndrome IV, 513
- de novo mutation, The first de novo mutation of the connexin 32 gene associated with X linked Charcot-Marie-Tooth disease, 251 deafness, A systematic mutation screen of 10 nuclear and 25
- mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA<sup>sertUC(N)</sup> mutations in a
- subgroup with syndromal encephalopathy, 895
  Four sibs with dislocated elbows, bowed tibiae, scoliosis, deafness, cataract, microcephaly, and mental retardation: a new MCA/MR syndrome, 755
- Proteinuria in a patient with the diaphragmatic herniahypertelorism-myopia-deafness syndrome: further evidence that the facio-oculo-acoustico-renal syndrome represents the same entity, 70
- Usher syndrome type III (USH3) linked to chromosome 3q in an
- 7-dehydrocholesterol, Smith-Lemli-Opitz syndrome: a variable clinical and biochemical phenotype, 558

  del(18p), Del(18p) shown to be a cryptic translocation using a multiprobe FISH assay for subtelomeric chromosome rearrangements, 722
- del(22q11) syndrome, Juvenile rheumatoid arthritis and del(22q11) syndrome: a non-random association, 943
- del(22)(q13.3), Two 22q telomere deletions serendipitously detected by FISH, 939
- deletion, Distal 6p deletion syndrome: a report of a case with anterior chamber eye anomaly and review of published reports, 685
- The mechanisms involved in formation of deletions and duplications
- of 15q11-q13, 130 deletions, Constitutional and mosaic large NF1 gene deletions in neurofibromatosis type 1, 468

- dementia, Mapping of a complicated familial spastic paraplegia to locus SPG4 on chromosome 2p, 89
- DFNB1/DFNA3, A Moroccan family with autosomal recessive sensorineural hearing loss caused by a mutation in the gap junction protein gene connexin 26 (GJB2), 151
  diagnosis, A rapid, PCR based test for differential molecular diagnosis of Prader-Willi and Angelman syndromes, 472
- - Rapid immunoblot and kinase assay tests for a syndromal form of X linked mental retardation: Coffin-Lowry syndrome, 890
- Scarcity of mutations detected in families with X linked hypohidrotic ectodermal dysplasia: diagnostic implications, 112
- diagnostic pitfalls, Pitfalls in the diagnosis of mtDNA mutations: let-
- diaphragmatic hernia, Proteinuria in a patient with the diaphragmatic hernia-hypertelorism-myopia-deafness syndrome: further evidence that the facio-oculo-acoustico-renal syndrome represents the same entity, 70
- diastrophic dysplasia, Atelosteogenesis type 2: syndrome of the month, 49
- chromosome, Discordant phenotypes dicentric 45,X/46,X,idic(Y), 862
- DiGeorge syndrome, Prenatal diagnosis by FISH of a 22q11
- deletion in two families, 165
  The annual incidence of DiGeorge/velocardiofacial syndrome: letter,
- diplegia, Genetic localisation of mental retardation with spastic diplegia to the pericentromeric region of the X chromosome: X inactivation in female carriers, 284
- disease frequencies, A comparison of disease and gene frequencies of inborn errors of metabolism among different ethnic groups in the West Midlands, UK, 366
  distal 10q trisomy, Distal 10q trisomy syndrome with unusual cardiac and pulmonary abnormalities, 72
- distal trisomy 14q, Parental origin effects in human trisomy for chro-
- mosome 14q: implications for genomic imprinting, 821 distal-less gene (DLX), A common DLX3 gene mutation is responsible for tricho-dento-osseous syndrome in Virginia and North Carolina families, 825
- DNA methylation, Unusual mutations in high functioning fragile X males: apparent instability of expanded unmethylated CGG repeats, 103
- DNA resource, The North Cumbria Community Genetics Project,
- dominant ataxia, Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method, 99
- dominant effect, P67L: a cystic fibrosis allele with mild effects found at high frequency in the Scottish population, 122
- dominant optic atrophy, Clinical features, molecular genetics, and pathophysiology of dominant optic atrophy: review, 793

  DOP-PCR, A simple and efficient method for microdissection and
- microFISH, 265
  - Chromosome specific comparative genome hybridisation
- determining the origin of intrachromosomal duplications, 37 **Down syndrome**, Maternal age specific risk rate estimates for Down syndrome among live births in whites and other races from Ohio and Metropolitan Atlanta, 1970-1989, 482
- Prenatal detection of trisomy 21 and 18 from amniotic fluid by quantitative fluorescent polymerase chain reaction, 126
- DTDST gene, Atelosteogenesis type 2: syndrome of the month, 49 dup(17)(p11.2p13.3), A molecular, cytogenetic, and clinical evaluation of mosaic tandem duplication 17p and Charcot-Marie-Tooth type 1A neuropathy, 169
- dup(1)(q12q22), Mosaicism for a tand dup(1)(q12q22) in an 18 year old female, 600 tandem
- duplication, Chromosome specific comparative genome hybridisa-tion for determining the origin of intrachromosomal duplications,
- Duplication of 8p23.1: a cytogenetic anomaly with no established clinical significance, 491
- duplication 15q11-q13, The mechanisms involved in formation of deletions and duplications of 15q11-q13, 130
- duplication 7p, Intrachromosomal triplication of distal 7p, 78 dyskeratosis congenita, 1.4 Mb candidate gene region for X linked dyskeratosis congenita defined by combined haplotype and X chromosome inactivation analysis, 993
  dysplastic kidney, A mother with VCFS and unilateral dysplastic
- kidney and her fetus with multicystic dysplastic kidneys: additional evidence to support the association of renal malformations and VCFS: letter, 348
- dystonia, Another dystonia: letter, 879
- The dystonias: syndrome of the month, 314 dystrophin, Abnormalities of dystrophin, the sarcoglycans, and laminin  $\alpha 2$  in the muscular dystrophies, 379
- ectodermal dysplasia, Ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum: variable expression of a single syndrome?, 157
- Ehlers-Danlos IV, New lethal disease involving type I and III collagen defect resembling geroderma osteodysplastica, De Barsy syndrome, and Ehlers-Danlos syndrome IV, 513

- Ehlers-Danlos syndrome, A single base mutation in COL5A2 causes Ehlers-Danlos syndrome type II, 846
- elliptocytosis, Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene deletion syndrome?, 273
- endometrial cancer, Genetic implications of double primary cancers of the colorectum and endometrium, 978
- entire gene deletion, Familial neurofibromatosis type 1 associated
- with an overgrowth syndrome resembling Weaver syndrome, 323 enzyme replacement therapy, Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis, 775
- epidemiology, Myotonia congenita in northern Finland: an epidemiological and genetic study, 293
  epilepsy, Mapping of a complicated familial spastic paraplegia to locus SPG4 on chromosome 2p, 89
  ethics, Chinese geneticists approach ethics: letter, 83
  ethnic groups. A comparison of disease and cons fractions of
- ethnic groups, A comparison of disease and gene frequencies of inborn errors of metabolism among different ethnic groups in the West Midlands, UK, 366
- eugenics, Chinese geneticists approach ethics: letter, 83
- exon skipping, Neurofibromatosis type 1 (NF1): a protein truncation assay yielding identification of mutations in 73% of patients, 813 external ear, Oto-onycho-peroneal syndrome: confirmation of a
- syndrome, 508
- facial clefting, Triophthalmia and facial clefting: a case report, 875 familial 22q11 deletions, Frequency of inherited deletions of 22q11: letter, 789
- familial amyloid polyneuropathy, A pedigree analysis with minimised ascertainment bias shows anticipation in Met30-transthyretin related familial amyloid polyneuropathy, 23
- familial breast cancer, Men in breast cancer families: a preliminary qualitative study of awareness and experience, 739
- familial gonadal tumours, Familial gonadal tumours: letter, 84
- familial hypercholesterolaemia (FH), Identification of a common low density lipoprotein receptor mutation (C163Y) in the west of Scotland, 573
- familial hypertrophic cardiomyopathy, Counselling issues in familial hypertrophic cardiomyopathy, 183

  Molecular pathology of familial hypertrophic cardiomyopathy caused by mutations in the cardiac myosin binding protein C ene, **205**
- familial inheritance, Kyphomelic dysplasia in two sib fetuses, 65
- familial juvenile nephronophthisis, Molecular studies in Finnish patients with familial juvenile nephronophthisis exclude a founder effect and support a common mutation causing mechanism, 279
- familial Mediterranean fever, Exclusion of the familial Mediterranean fever locus as a susceptibility region for autosomal dominant familial Hibernian fever, 432
- familial Hibernian fever, 432

  familial spastic paraplegia (FSP), Mapping of a complicated familial spastic paraplegia to locus SPG4 on chromosome 2p, 89

  family communication, Men in breast cancer families: a preliminary qualitative study of awareness and experience, 739
- family history, Family history of breast cancer: what do women understand and recall about their genetic risk?, 731
- FcεRIβ, Association of the FcεRIβ gene with hyper-responsiveness in an Italian population, 680 with
- fetal nuchal translucency, Outcome of chromosomally normal live-births with increased fetal nuchal translucency at 10-14 weeks' gestation, 222
- renal cysts, Prenatal diagnosis of autosomal dominant polycystic kidney disease (PKD1) presenting in utero and prognosis for very early onset disease, 13

  FGFR2 mutations, Presence of the Apert canonical S252W FGFR2 mutation in a patient without severe syndactyly, 677
- FGFR3, New overgrowth syndrome and FGFR3 dosage effect: letter, 348
- fibroblast growth factor receptor, Familial craniosynostosis, anal anomalies, and porokeratosis: CAP syndrome, 763
- Finland, Myotonia congenita in northern Finland: an epidemiological
- and genetic study, 293
  FISH, A FISH study of chromosome fusion in the ICF syndrome: involvement of paracentric heterochromatin but not of the centromeres themselves, 833
  - Prenatal diagnosis by FISH of a 22q11 deletion in two families, 165 Severe phenotype resulting from an active ring X chromosome in a female with a complex karyotype: characterisation and replication study, 932
- fluorescence in situ hybridisation (FISH), Del(18p) shown to be a cryptic translocation using a multiprobe FISH assay for subtelomeric chromosome rearrangements, 722 fluorescent polymerase chain reaction, Prenatal detection of
- trisomy 21 and 18 from amniotic fluid by quantitative fluorescent polymerase chain reaction, 126
- fluorescence in situ hybridisation, A simple and efficient method for microdissection and microFISH, 265
- FMR1 gene, The fragile X syndrome: syndrome of the month, 579 focal glomerulosclerosis, Variants of  $\alpha_i$ -proteinase inhibitor in black and white South African patients with focal glomerulosclerosis and minimal change nephrotic syndrome, 6

- focal medullary dysplasia of kidney, Neonatal cholestasis and focal medullary dysplasia of the kidneys in a case of microcephalic osteodysplastic primordial dwarfism, 61 cacid, The C677T mutation of the 5,10-
- acid, The C677T mutation of the 5,10-methylenetetrahydrofolate reductase gene is a moderate risk factor for spina bifida in Italy, 1009
- fragile X, Studies of FRAXA and FRAXE in women with premature ovarian failure, 637
- fragile X syndrome, Molecular evidence that fragile X syndrome occurs in the South African black population: letter, 878 The fragile X syndrome: syndrome of the month, 579
- Unusual mutations in high functioning fragile X males: apparent instability of expanded unmethylated CGG repeats, 103
- Frasier syndrome, Do intronic mutations affecting splicing of WT1
- exon 9 cause Frasier syndrome?, 45
  frequency, Frequency of inherited deletions of 22q11: letter, 789
  Friedreich ataxia, Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the FRDA care 713 in the FRDA gene, 713
- fructose intolerance, Hereditary fructose intolerance: review, 353 FSHD, Identical de novo mutation at the D4F104S1 locus in monozygotic male twins affected by facioscapulohumeral muscular dystrophy (FSHD) with different clinical expression, 778
- ganglioneuroblastoma, Simultaneous adrenocortical carcinoma and ganglioneuroblastoma in a child with Turner syndrome and germline p53 mutation, 328
- Gardner's syndrome, Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene, 244 gastric outlet obstruction, Familial occurrence of congenital incom-
- plete prepyloric mucosal diaphragm, 1040
- Gaucher disease, Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis, 775

  Gaucher disease type 1, Coexistence of Gaucher disease type 1 and
- Joubert syndrome: letter, 965
- genetics, Further refinement of the Usher 2A locus at 1q41, 773
- gene frequencies, A comparison of disease and gene frequencies of inborn errors of metabolism among different ethnic groups in the
- West Midlands, UK, 366
  gene mapping, Chromosome mapping of Rett syndrome: a likely candidate region on the telomere of Xq, 297
  gene mutation, Molecular study of the rhodopsin gene in retinitis
- pigmentosa patients in the Basque Country, 387
- gene structure, Clinical heterogeneity and novel mutations in the glycerol kinase gene in three families with isolated glycerol kinase deficiency, 650
- gene/environment interaction, The North Cumbria Community Genetics Project, 413 genetic and epidemiological research, The North Cumbria Com-
- munity Genetics Project, 413
- genetic aspects, Myotonia congenita in northern Finland: an epidemiological and genetic study, 293
- genetic counselling, How the magnitude of clinical severity and recurrence risk affects reproductive decisions in adult males with different forms of progressive muscular dystrophy, 189
- genetic heterogeneity, Lack of evidence for genetic heterogeneity in Best vitelliform macular dystrophy: letter, 85 Linkage analysis in Usher syndrome type I (USH1) families from
- Spain, **391** genetic heterogeneity, Genetic heterogeneity and HOMOG analysis
- in British malignant hyperthermia families, 196
- genetic linkage, Fine localisation of the gene for central areolar
- choroidal dystrophy on chromosome 17p, 770
  genetic nurses, Recommendations for Education and Training of
  Genetic Nurses and Counsellors in the United Kingdom, 410
- genetic risk, Family history of breast cancer: what do women understand and recall about their genetic risk?, 731
- genetics, The butyrylcholinesterase K variant and susceptibility to Alzheimer's disease, 1034
- The dystonias: syndrome of the month, 314
  genomic imprinting, Parental origin effects in human trisomy for chromosome 14q: implications for genomic imprinting, 821
  Studies of non-disjunction in trisomies 2, 7, 15, and 22: does the
  - parental origin of trisomy influence placental morphology?, 924
- genotype-phenotype, The site of a missense mutation in the extracellular Ig or FN domains of L1CAM influences infant mortality and the severity of X linked hydrocephalus, 901
- genotype-phenotype correlation, Genotype-phenotype correlation in L1 associated diseases, 399
- Severe testotoxicosis phenotype associated with Asp—Tyr mutation of the lutrophin/choriogonadotrophin receptor gene, 340
- germinal mosaicism, Extensive germinal mosaicism in a family with X linked myotubular myopathy simulates genetic heterogeneity, 241
- germline mutations, Uniparental disomy for chromosome 6 results in steroid 21-hydroxylase deficiency: evidence of different genetic mechanisms involved in the production of the disease, 1014 geroderma osteodysplastica, New lethal disease involving type I and III collagen defect resembling geroderma osteodysplastica,
- De Barsy syndrome, and Ehlers-Danlos syndrome IV, 513

- glaucoma, A novel Asp380Ala mutation in the GLC1A/myocilin gene in a family with juvenile onset primary open angle glaucoma, 957
- glycogen storage disease, Localisation of the gene for glycogen storage disease type 1c by homozygosity mapping to 11q, 269
- Gorlin syndrome, A survey of phenotypic features in juvenile polyposis, 476
- GPC3 gene, A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma, 153
- growth and developmental retardation, Costello syndrome in two Brazilian children, 54
- growth hormone gene cluster, Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome, 784
- Guthrie cards, Autoclaving Guthrie cards does not prevent their use in PCR reactions!: letter, 702

  UK centres are not following the Royal College of Pathologists' rec
  - ommendations for storage of Guthrie cards: a national policy is needed: letter, 263
- H63D mutation, Frequency of the HFE C282Y and H63D mutations in distinct ethnic groups living in Spain: letter, 701
- haemochromatosis, High frequency of the haemochromatosis C282Y mutation in Hungary could argue against a Celtic origin of the mutation: letter, 878
- haplotype analysis, Molecular studies in Finnish patients with familial juvenile nephronophthisis exclude a founder effect and support a common mutation causing mechanism, 279
  - Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis, 775
- haplotypes, Population genetics of hyperphenylalaninaemia resulting from phenylalanine hydroxylase deficiency in Portugal, 301
- hepatocellular carcinoma, A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma, 153
- hereditary breast cancer, Using gene carrier probability to select high risk families for identifying germline mutations in breast cancer susceptibility genes, 116
- hereditary haemochromatosis, Frequency of the HFE C282Y and H63D mutations in distinct ethnic groups living in Spain: letter,
- hereditary late onset disorders, Predicting adaptation to presymptomatic DNA testing for late onset disorders: who will experience distress?, 745
- hereditary non-polyposis colorectal cancer, Genetic implications of double primary cancers of the colorectum and endometrium, 978
- heterochromatin, A FISH study of chromosome fusion in the ICF syndrome: involvement of paracentric heterochromatin but not of the centromeres themselves, 833
- heterogeneity, Locus heterogeneity in autosomal dominant congenital external ophthalmoplegia (CFEOM), 985
  - No evidence for heterogeneity in oculopharyngeal muscular dystrophy: letter, 613
- HFE gene, Frequency of the HFE C282Y and H63D mutations in distinct ethnic groups living in Spain: letter, 701 **HFE mutations**, Non-C282Y familial iron overload: evidence for
- locus heterogeneity in haemochromatosis, 954
- hβENaC, The diagnosis of Liddle syndrome by identification of a mutation in the  $\beta$  subunit of the epithelial sodium channel, 510
- Hirschsprung disease, Hirschsprung disease, microcephaly, mental retardation, and characteristic facial features: delineation of a new syndrome and identification of a locus at chromosome 2q22-q23,
- HLA, Non-C282Y familial iron overload: evidence for locus heterogeneity in haemochromatosis, 954
- HNPP, Mutation analysis of the nerve specific promoter of the peripheral myelin protein 22 gene in CMT1 disease and HNPP, 590
- holoprosencephaly, Another holoprosencephaly locus at 7q21.2?: letter, **614**
- Holoprosencephaly in deletions of proximal chromosome 14q: letter, 612
- homeobox, A common DLX3 gene mutation is responsible for tricho-dento-osseous syndrome in Virginia and North Carolina families, 825
- HOMOG, Genetic heterogeneity and HOMOG analysis in British malignant hyperthermia families, 196
- homozygosity analysis, Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425, 202
- homozygosity mapping, Localisation of the gene for glycogen storage disease type 1c by homozygosity mapping to 11q, 269
- homozygous phenotype, Mesoaxial complete syndactyly and synostosis with hypoplastic thumbs: an unusual combination or homozygous expression of syndactyly type I?, 868
- HOXD13, Mesoaxial complete syndactyly and synostosis with hypoplastic thumbs: an unusual combination or homozygous
- expression of syndactyly type I?, 868
  human genetic testing services, "Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public". Advisory Committee on Genetic Testing, 443

- Hungary, High frequency of the haemochromatosis C282Y mutation in Hungary could argue against a Celtic origin of the mutation: letter, 878
- Hunter syndrome, Reassessment of biochemically determined Hunter syndrome carrier status by DNA testing, 646
- hydatidiform mole, Studies of non-disjunction in trisomies 2, 7, 15, and 22: does the parental origin of trisomy influence placental morphology?, 924
- hydrocephalus, Costello syndrome in two Brazilian children, 54
- hyperechogenic fetal bowel, Cystic fibrosis screening: a fetus with hyperechogenic bowel may be the index case, 657
- hypergonadotrophic hypogonadism, Two adult females with a distinct familial mental retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, hypergonadotrophic hypogonadism, and retinal dystrophy, 333
- hyperketotic hypoglycaemia, Clinical heterogeneity and novel mutations in the glycerol kinase gene in three families with isolated glycerol kinase deficiency, 650
- hyperphenylalaninaemia, Population genetics of hyperphenylalaninaemia resulting from phenylalanine hydroxylase deficiency in Portugal, 301
- hypertelorism, Neurofibromatosis type 1 (NF1): a protein truncation assay yielding identification of mutations in 73% of patients, 813
- hypertrophic cardiomyopathy, De novo unbalanced translocation resulting in monosomy for proximal 14q and distal 4p in a fetus with intrauterine growth retardation, Wolf-Hirschhorn syndrome, hypertrophic cardiomyopathy, and partial hemihypoplasia, 1050
- Identification of a new missense mutation in MyBP-C associated with hypertrophic cardiomyopathy, 253

  hypocalcaemia, Localisation of X linked recessive idiopathic hypoparathyroidism to a 1.5 Mb region on Xq26-q27, 905
- hypohidrotic ectodermal dysplasia, Scarcity of mutations detected in families with X linked hypohidrotic ectodermal dysplasia: diagnostic implications, 112
- hypoplastic left heart, Hypoplastic left heart in cerebrocostoman-dibular syndrome: letter, 879
- hypothyroidism, Ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum: variable expression of a single syndrome?, 157
  - Lipomatous myelomeningocele, athyrotic hypothyroidism, and sensorineural deafness: a new form of syndromal deafness?, 948
- ICF syndrome, A FISH study of chromosome fusion in the ICF syndrome: involvement of paracentric heterochromatin but not of the centromeres themselves, 833
- iduronate-2-sulphatase, Reassessment of biochemically determined Hunter syndrome carrier status by DNA testing, 646
- imprinting, Beckwith-Wiedemann syndrome in a child with chromosome 18q deletion, 162
- inborn error of metabolism, Hereditary fructose intolerance: review, 353
- inborn errors of metabolism, A comparison of disease and gene frequencies of inborn errors of metabolism among different ethnic groups in the West Midlands, UK, 366
- insertion, Trisomy 2q11.2→q21.1 resulting from an unbalanced insertion in two generations, 319
- insterstitial 22q11 deletions, Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: letter, 346
- insurance companies, Laws regarding insurance companies: letter, 526
- interleukin-4 gene, Linkage and association of an interleukin 4 gene polymorphism with atopic dermatitis in Japanese families, 502
- interphotoreceptor matrix proteoglycan-1, Assessment of the interphotoreceptor matrix proteoglycan-1, Assessment of the interphotoreceptor matrix proteoglycan-1 (IMPG1) gene localised to 6q13-q15 in autosomal dominant Stargardt-like disease (ADSTGD), progressive bifocal chorioretinal atrophy (PBCRA), and North Carolina macular dystrophy (MCDR1),
- interstitial deletion, Mild phenotype associated with an interstitial deletion of the long arm of chromosome 1, 1047
- intrachromosomal triplication, Intrachromosomal triplication of distal 7p, 78
- intronic mutation, Do intronic mutations affecting splicing of WT1 exon 9 cause Frasier syndrome?, 45
- **dup(15) chromosome,** Triplication of 15q11-q13 with inv dup(15) in a female with developmental delay, 425
- inv(12), Not para-, not peri-, but centric inversion of chromosome 12, 682
- isodisomy, Uniparental disomy for chromosome 6 results in steroid 21-hydroxylase deficiency: evidence of different g mechanisms involved in the production of the disease, 1014
- isolated glycerol kinase deficiency, Clinical heterogeneity and novel mutations in the glycerol kinase gene in three families with isolated glycerol kinase deficiency, 650

Japanese, Identification of five novel germline mutations of the MEN1 gene in Japanese multiple endocrine neoplasia type 1 (MEN1) families, 915

joint dislocations, A distinct form of spondyloepimetaphyseal dysplasia with multiple dislocations, 566

- Joubert syndrome, Coexistence of Gaucher disease type 1 and Joubert syndrome: letter, 965
- junction fragment, Diagnosis of the CMT1A duplication by PCR
- based detection of a novel junction fragment: letter, 962 juvenile haemochromatosis, Non-C282Y familial iron overload:
- evidence for locus heterogeneity in haemochromatosis, 954 juvenile onset glaucoma, Novel TIGR/MYOC mutations in families with juvenile onset primary open angle glaucoma, 989
- juvenile polyposis, A survey of phenotypic features in juvenile polyposis, 476
- juvenile recurrent parotitis, Autosomal dominant juvenile recurrent parotitis, 417
- juvenile rheumatoid arthritis, Juvenile rheumatoid arthritis and del(22q11) syndrome: a non-random association, 943
  Spectrum of clinical features associated with interstitial
  - chromosome 22q11 deletions: letter, 346
- Kenny-Caffey syndrome, Genotypic/phenotypic heterogeneity of Kenny-Caffey syndrome: letter, 1054
  - Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster, 31
  - Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster: letter, 1054
  - Kenny-Caffey syndrome is part of the CATCH 22 haplo-insufficiency cluster: letter, 1054
- Kenny-Caffey syndrome without the CATCH 22 deletion: letter,
- 1054
- KFSD, Keratosis follicularis spinulosa decalvans: confirmation of linkage to Xp22.13-p22.2, 336
   Klinefelter syndrome, 49,XXXXY: a distinct phenotype. Three new
- cases and review, 420
  Not para-, not peri-, but centric inversion of chromosome 12, 682
  kyphomelic dysplasia, Kyphomelic dysplasia in two sib fetuses, 65
- L1, Genotype-phenotype correlation in L1 associated diseases, 399 L1CAM, A silent mutation, C924T (G308G), in the L1CAM gene results in X linked hydrocephalus (HSAS), 456

  The site of a missense mutation in the extracellular Ig or FN
  - domains of L1CAM influences infant mortality and the severity of X linked hydrocephalus, 901
- laminin  $\alpha 2$ , Abnormalities of dystrophin, the sarcoglycans, and laminin  $\alpha 2$  in the muscular dystrophies, 379
- laminin-2, PCR based mutation screening of the laminin α2 chain gene (LAMA2): application to prenatal diagnosis and search for founder effects in congenital muscular dystrophy, 211
- law, Laws regarding insurance companies: letter, 526
- Leber's hereditary optic neuropathy, Fuch's corneal dystrophy in a patient with mitochondrial DNA mutations, 258

  Leber's hereditary optic neuropathy (LHON), Meiotic breakpoint mapping of a proposed X linked visual loss susceptibility locus in Leber's hereditary optic neuropathy, 668
- lethal, A lethal skeletal dysplasia with features of chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a rare genetic disorder, 505
- LHON, Fuch's corneal dystrophy in a patient with mitochondrial DNA mutations, 258
- **Liddle syndrome,** The diagnosis of Liddle syndrome by identification of a mutation in the  $\beta$  subunit of the epithelial sodium channel, 510
- limb-girdle muscular dystrophies, A first missense mutation in the δ sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies, 951
- limb-girdle muscular dystrophy, Evidence for anticipation in autosomal dominant limb-girdle muscular dystrophy, 305
- linkage, Evidence of linkage of the inflammatory bowel disease susceptibility locus on chromosome 16 (IBD1) to ulcerative
  - Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425, 202
- Linkage and association of an interleukin 4 gene polymorphism with atopic dermatitis in Japanese families, 502

  Localisation of a gene for non-specific X linked mental retardation
- (MRX46) to Xq25-q26, **801**
- Meiotic breakpoint mapping of a proposed X linked visual loss susceptibility locus in Leber's hereditary optic neuropathy, 668
  Peutz-Jeghers disease: most, but not all, families are compatible with
- linkage to 19p13.3, 42
- linkage analysis, A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33, 141

  Linkage analysis in Rett syndrome families suggests that there may
  - be a critical region at Xq28, 997
  - Linkage analysis in Usher syndrome type I (USH1) families from Spain, 391

- linkage disequilibrium, A first missense mutation in the  $\delta$  sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies, 951
- lipomatous myelomeningocele, Lipomatous myelomeningocele, athyrotic hypothyroidism, and sensorineural deafness: a new form of syndromal deafness?, 948
- lissencephaly, Classical lissencephaly syndromes: does the face reflect the brain?, 920
- locus heterogeneity, Non-C282Y familial iron overload: evidence for
  - locus heterogeneity in haemochromatosis, 954
    Novel TIGR/MYOC mutations in families with juvenile onset primary open angle glaucoma, 989
- LOH, A population study of mutations and LOH at breast cancer gene loci in tumours from sister pairs: two recurrent mutations seem to account for all BRCA1/BRCA2 linked breast cancer in Iceland,
- loss of heterozygosity, Constitutional and mosaic large NF1 gene deletions in neurofibromatosis type 1, 468
- low density lipoprotein receptor (LDLR), Identification of a common low density lipoprotein receptor mutation (C163Y) in the west of Scotland, 573
- lutrophin/choriogonadotrophin receptor, Severe testotoxicosis phenotype associated with Asp→Tyr mutation of the lutrophin/choriogonadotrophin receptor gene, 340
- M470V, Linkage disequilibrium between the M470V variant and the IVS8 polyT alleles of the CFTR gene in CBAVD, 594
- macrocephaly, Minor disease features in neurofibromatosis type 1 (NF1) and their possible value in diagnosis of NF1 in children  $\leq 6$  years and clinically suspected of having NF1, 624
- macro-orchidism, A family with mental retardation, variable macrocephaly and macro-orchidism, and linkage to Xq12-q21, 1026
- male sexual development, An autosomal or X linked mutation results in true hermaphrodites and 46,XX males in the same family, 17
- malignant hyperthermia, Genetic heterogeneity and HOMOG analysis in British malignant hyperthermia families, 196

  Marfan syndrome, Psychosocial adaptation in adolescents and
- young adults with Marfan syndrome: an exploratory study, 405
- maternal systemic lupus erythematosus, Chondrodysplasia punctata and maternal systemic lupus erythematosus: commentary, 698
- MCDR1, Assessment of the interphotoreceptor matrix proteoglycan-1 (IMPG1) gene localised to 6q13-q15 in autosomal dominant Stargardt-like disease (ADSTGD), progressive bifocal chorioretinal atrophy (PBCRA), and North Carolina macular dystrophy (MCDR1), 641
- Meckel syndrome: syndrome of the month, 497 meconium ileus, Is meconium ileus genetically determined or
- associated with a more severe evolution of cystic fibrosis?: letter,
- medical genetics, Medical genetics in the UK and the National Health Service, 441
- MELAS, UK centres are not following the Royal College of Pathologists' recommendations for storage of Guthrie cards: a national policy is needed: *letter*, 263
- men, Men in breast cancer families: a preliminary qualitative study of awareness and experience, 739
- MEN1, Identification of five novel germline mutations of the MEN1 gene in Japanese multiple endocrine neoplasia type 1 (MEN1) families, 915
- Menkes disease, Fibroblast silver loading for the diagnosis of Menkes disease, 849
- mental retardation, A family with mental retardation, variable macrocephaly and macro-orchidism, and linkage to Xq12-q21, 1026 A new strategy for cryptic telomeric translocation screening in
  - patients with idiopathic mental retardation, 225 Alport syndrome, mental retardation, midface hypoplasia, and ellip-
  - tocytosis: a new X linked contiguous gene deletion syndrome?,
  - Ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum: variable expression of a single syndrome?, 15
  - Genetic localisation of mental retardation with spastic diplegia to the pericentromeric region of the X chromosome: X inactivation in female carriers, 284
  - Genotype-phenotype correlation in L1 associated diseases, 399
  - Hirschsprung disease, microcephaly, mental retardation, and characteristic facial features: delineation of a new syndrome and identification of a locus at chromosome 2q22-q23, 617
  - Molecular screening for proximal 15q abnormalities in a mentally retarded population, 534
  - Recurrence risks in mental retardation: review, 177
  - Smith-Lemli-Opitz syndrome: a variable clinical and biochemical phenotype, 558
    The fragile X syndrome: syndrome of the month, 579

  - Two adult females with a distinct familial mental retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, hypergonadotrophic hypogonadism, and retinal dystrophy, 333

merosin, PCR based mutation screening of the laminin a2 chain gene (LAMA2): application to prenatal diagnosis and search for

founder effects in congenital muscular dystrophy, 211

meta-analysis, The C677T mutation of the 5,10methylenetetrahydrofolate reductase gene is a moderate risk
factor for spina bifida in Italy, 1009

metacarpal type, Maternal systemic lupus erythematosus and chron-

- drodysplasia punctata in two sibs: phenocopy or coincidence?, 690 methylation specific PCR, Molecular screening for proximal 15q
- abnormalities in a mentally retarded population, 534 microcephalic osteodysplastic dwarfism, Neonatal cholestasis and focal medullary dysplasia of the kidneys in a case of microcephalic osteodysplastic primordial dwarfism, 61
- microcephaly, Hirschsprung disease, microcephaly, retardation, and characteristic facial features: delineation of a new syndrome and identification of a locus at chromosome 2q22-q23,
  - Spastic paraplegia, optic atrophy, microcephaly with normal intelligence, and XY sex reversal: a new autosomal recessive syndrome?, 759
- microdeletion, Prenatal diagnosis by FISH of a 22q11 deletion in two families, 165
- microdissection, A simple and efficient method for microdissection and microFISH, 265
- microplate array diagonal gel electrophoresis (MADGE), Identification of a common low density lipoprotein receptor mutation (C163Y) in the west of Scotland, 573
- microsatellite markers, Microsatellite markers for the cone-rod retinal dystrophy gene, CRX, on 19q13.3: letter, 527
- midface hypoplasia, Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene deletion syndrome?, 273
- Miller-Dieker syndrome, Classical lissencephaly syndromes: does the face reflect the brain?, 920
- minimal change nephrotic syndrome, Variants of  $\alpha_1$ -proteinase inhibitor in black and white South African patients with focal glomerulosclerosis and minimal change nephrotic syndrome, 6
- minor disease features, Minor disease features in neurofibromatosis type 1 (NF1) and their possible value in diagnosis of NF1 in children ≤6 years and clinically suspected of having NF1, 624
- misalignment of the pulmonary vessels, Familial persistent pulmonary hypertension of the newborn resulting from misalignment of the pulmonary vessels (congenital alveolar capillary dysplasia), 58
- miscarriages, Duplication of 8p23.1: a cytogenetic anomaly with no established clinical significance, 491
- missense mutation, Identification of a new missense mutation in MyBP-C associated with hypertrophic cardiomyopathy, 253
- missense mutations, The site of a missense mutation in the extracel-lular Ig or FN domains of L1CAM influences infant mortality and the severity of X linked hydrocephalus, 901
- mitochondrial DNA, Meiotic breakpoint mapping of a proposed X linked visual loss susceptibility locus in Leber's hereditary optic neuropathy, 668
- Pancreatic exocrine dysfunction associated with mitochondrial tRNA<sup>Lu(UUR)</sup> mutation, 255
- mitochondrial DNA mutations, Mitochondrial DNA mutations and pathogenicity: letter, 701
- mitochondrial tRNA mutations, A systematic mutation screen of 10 nuclear and 25 mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA<sup>ser</sup> mutations in a subgroup with syndromal encephalopathy, 895
- mixed gonadal dysgenesis, Discordant phenotypes and 45,X/46,X,idic(Y), 862 molecular, A rapid, PCR based test for differential molecular diagno-
- sis of Prader-Willi and Angelman syndromes, 472
- molecular genetics, Clinical features, molecular genetics, and patho-
- physiology of dominant optic atrophy: review, 793
  monosomy 21q, Two cases of partial trisomy 8p and partial
  monosomy 21q in a family with a reciprocal translocation
  (8;21)(p21.1;q22.3), 604
- monozygotic twins, Identical de novo mutation at the D4F104S1 locus in monozygotic male twins affected by facioscapulohumeral muscular dystrophy (FSHD) with different clinical expression,
- mosaic, A molecular, cytogenetic, and clinical evaluation of mosaic tandem duplication 17p and Charcot-Marie-Tooth type 1A neuropathy, 169
- mosaicism, Mosaic supernumerary ring chromosome 19 identified by comparative genomic hybridisation, 836
- Mosaicism for a tandem duplication dup(1)(q12q22) in an 18 year old female, 600
- MRI, Bony orbital morphology in neurofibromatosis type 1 (NF1),
- MRI findings, MRI findings in a patient with partial monosomy 10p,
- mtDNA mutations, Pitfalls in the diagnosis of mtDNA mutations: letter, 963
  The
- MTHFR, C677T mutation of the methylenetetrahydrofolate reductase gene is a moderate risk factor for spina bifida in Italy, 1009

- mucin genes, Cystic fibrosis and mucins: letter, 82
- mucopolysaccharidosis IIIB, Identification of 12 novel mutations in the  $\alpha$ -N-acetylglucosaminidase gene in 14 patients with Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), 910 multiplex PCR, Molecular screening for proximal 15q abnormalities
- in a mentally retarded population, 534
- multiprobe fluorescence in situ hybridisation (FISH), A new approach to the elucidation of complex chromos rearrangements illustrated by a case of Rieger syndrome, 234 scle ultrasound abnormalities, "Cataplexy" and mu chromosome
- muscle ultrasound abnormalities, "Cataplexy" and ultrasound abnormalities in Coffin-Lowry syndrome, 94
- muscular dystrophies, How the magnitude of clinical severity and recurrence risk affects reproductive decisions in adult males with different forms of progressive muscular dystrophy, 189
- muscular dystrophy, Abnormalities of dystrophin, the sarcoglycans, and laminin α2 in the muscular dystrophies, 379
- mutation, A single base mutation in COL5A2 causes Ehlers-Danlos syndrome type II, 846

  Differential diagnosis of type 2 neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas, 973
  - Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations, 450
  - Homozygosity for Asn86Ser mutation in the CuZn-superoxide dismutase gene produces a severe clinical phenotype in a juvenile onset case of familial amyotrophic lateral sclerosis: *letter*, 174
  - Japanese multiple endocrine neoplasia type 1 (MEN1) families, 915 Identification of five novel germline mutations of the MEN1 gene in
- of mutations detected in families with X linked Scarcity hypohidrotic ectodermal dysplasia: diagnostic implications, 112
- mutation D409H, Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis, 775
- mutation databases, Mutation databases on the web: editorial, 529 mutation detection, Mutations in the TSC1 gene account for a minority of patients with tuberous sclerosis, 969
- mutations, De novo presenilin 1 mutations are rare in clinically sporadic, early onset Alzheimer's disease cases, 672

  Identification of 12 novel mutations in the α-N-acetylglucosaminidase gene in 14 patients with Sanfilippo
- syndrome type B (mucopolysaccharidosis type IIIB), 910

  Population genetics of hyperphenylalaninaemia resulting from phenylalanine hydroxylase deficiency in Portugal, 301

  Segregation of mutations in arylsulphatase E and correlation with
- the clinical presentation of chondrodysplasia punctata, 1004
- MYBPC3 gene, Molecular pathology of familial hypertrophic cardiomyopathy caused by mutations in the cardiac myosin binding protein C gene, 205
- myocilin, A novel Asp380Ala mutation in the GLC1A/myocilin gene in a family with juvenile onset primary open angle glaucoma, 957
- myoclonus epilepsy, A systematic mutation screen of 10 nuclear and 25 mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA ser(UCIX) mutations in a subgroup with syndromal encephalopathy, 895
- ppia, Proteinuria in a patient with the diaphragmatic hernia-hypertelorism-myopia-deafness syndrome: further evithe diaphragmatic dence that the facio-oculo-acoustico-renal syndrome represents the same entity, 70
- myosin binding protein C, Identification of a new missense mutation in MyBP-C associated with hypertrophic cardiomyopathy, 253
- myotonia congenita, Myotonia congenita in northern Finland: an epidemiological and genetic study, 293
- myotonic dystrophy, Instability in the normal CTG repeat range at the myotonic dystrophy locus: letter, 791
- Knowledge, views, and experience of 25 women with myotonic dystrophy, 1020
  Segregation distortion in myotonic dystrophy, 1045
- myotubular myopathy, Extensive germinal mosaicism in a family with X linked myotubular myopathy simulates genetic heterogeneity, 241
- myotubularin, Extensive germinal mosaicism in a family with X linked myotubular myopathy simulates genetic heterogeneity, 241
- α-N-acetylglucosaminidase Identification of 12 novel mutations in the α-N-acetylglucosaminidase gene in 14 patients with Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), 910
- nail hypoplasia, Oto-onycho-peroneal syndrome: confirmation of a syndrome, 508
- National Health Service, Medical genetics in the UK and the National Health Service, 441
- neonatal cholestasis, Neonatal cholestasis and focal medullary dysplasia of the kidneys in a case of microcephalic osteodysplastic primordial dwarfism, 61
- neonatal diabetes, Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome, 288
- neonatal lupus erythematosus, Neonatal lupus syndrome: a case chondrodysplasia punctata and other manifestations, 695

- neurofibromatosis type 1, Bony orbital morphology neurofibromatosis type 1 (NF1), 628 Constitutional and mosaic large NF1 gene deletions neurofibromatosis type 1, 468
  - Familial neurofibromatosis type 1 associated with an overgrowth
  - syndrome resembling Weaver syndrome, 323
    Minor disease features in neurofibromatosis type 1 (NF1) and their possible value in diagnosis of NF1 in children ≤6 years and clinically suspected of having NF1, 624
- Neurofibromatosis type 1 (NF1): a protein truncation assay yielding identification of mutations in 73% of patients, 813 neurofibromatosis type 2, Differential diagnosis of type 2
- neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas, 973
- Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations, 450
- neurological signs, Medical complications of achondroplasia: a multicentre patient review, 705
- neuronal ceroid lipofuscinoses, Sharing of PPT mutations between distinct clinical forms of neuronal ceroid lipofuscinoses in patients from Scotland: letter, 790
- neuronal ceroid lipofuscinosis, Ovine neuronal ceroid lipofuscinosis: a large animal model syntenic with the human neuronal ceroid lipofuscinosis variant CLN6, 717
- neutropenia, Localisation of the gene for glycogen storage disease
- type 1c by homozygosity mapping to 11q, 269
  nomenclature, Congenital malformations: an inquiry classification and nomenclature, 661
- non-mendelian inheritance, Uniparental disomy for chromosome 6 results in steroid 21-hydroxylase deficiency: evidence of different genetic mechanisms involved in the production of the disease, 1014
- non-specific mental retardation, Localisation of a gene for non-specific X linked mental retardation (MRX46) to Xq25-q26,
- non-specific X linked mental retardation, Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH, 146
- normal phenotype, Duplication of 8p23.1: a cytogenetic anomaly with no established clinical significance, 491
- novel mutation, Clinical heterogeneity and novel mutations in the glycerol kinase gene in three families with isolated glycerol kinase deficiency, 650
- oculopharyngeal muscular dystrophy, No evidence for heterogeneity in oculopharyngeal muscular dystrophy: letter, 613
- OFDS type IX, Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation, 342
- oligodontia, Unknown syndrome: peculiar face, severe hypodontia of permanent teeth, and precocious choroid calcifications, 435
- OPA1, Clinical features, molecular genetics, and pathophysiology of dominant optic atrophy: review, 793

  optic atrophy, Spastic paraplegia, optic atrophy, microcephaly with normal intelligence, and XY sex reversal: a new autosomal recessive syndrome?, 759
- orbital morphology, Bony orbital morphology in neurofibromatosis type 1 (NF1), 628
- orofacial clefting, Further evidence for the involvement of human chromosome 6p24 in the aetiology of orofacial clefting, 857 orofacial clefts, Prediction of liability to orofacial clefting using
- genetic and craniofacial data from parents, 371 osteochondrodysplasia, A lethal skeletal dysplasia with features of
- chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a rare genetic disorder, 505
- otitis media, Medical complications of achondroplasia: a multicentre patient review, 705
- overgrowth, Familial neurofibromatosis type 1 associated with an
- overgrowth syndrome resembling Weaver syndrome, 323 overgrowth syndrome, A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma, 153
  New overgrowth syndrome and FGFR3 dosage effect: letter, 348
- ovine, Ovine neuronal ceroid lipofuscinosis: a large animal model syntenic with the human neuronal ceroid lipofuscinosis variant CLN6, 717
- Simultaneous adrenocortical carcinoma and mutation, ganglioneuroblastoma in a child with Turner syndrome and
- germline p53 mutation, 328
  p57<sup>KIP2</sup>, Is p57<sup>KIP2</sup> mutation a common mechanism for Beckwith-Wiedemann syndrome or somatic overgrowth?: letter, 86 P67L, P67L: a cystic fibrosis allele with mild effects found at high fre-
- quency in the Scottish population, 122

  pancreatic exocrine dysfunction, Pancreatic exocrine dysfunction
  associated with mitochondrial tRNA<sup>Leu(UUR)</sup> mutation, 255
- papillomata, Costello syndrome: syndrome of the month, 238
- parathyroid gland development, Localisation of X linked recessive idiopathic hypoparathyroidism to a 1.5 Mb region on Xq26-q27,
- parental origin, Parental origin effects in human trisomy for chromosome 14q: implications for genomic imprinting, 821

- partial monosomy 10p, MRI findings in a patient with partial mono-
- partial monosomy 6 (q26-yqter), Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs, 545
- partial trisomy 15 (q26.1-)qter), Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs, 545
- partial trisomy 3 (q29-yqter), Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs, 545
- pathogenicity, Mitochondrial DNA mutations and pathogenicity: letter, 701
- pattern profiles, Classical lissencephaly syndromes: does the face reflect the brain?, 920
- PAX2, The prevalence of PAX2 mutations in patients with isolated colobomas or colobomas associated with urogenital anomalies,
- PAX2 gene, Absence of PAX2 gene mutations in patients with primary familial vesicoureteric reflux, 338
- PAX3, Septo-optic dysplasia and WS1 in the proband of a WS1 family segregating for a novel mutation in PAX3 exon 7, 248
- PAX4, Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome, 288
- PBCRA, Assessment of the interphotoreceptor matrix proteoglycan-1 (IMPG1) gene localised to 6q13-q15 in autosomal dominant Stargardt-like disease (ADSTGD), progressive bifocal chorioretinal atrophy (PBCRA), and North Carolina macular dystrophy (MCDR1), 641
- PCR, A rapid, PCR based test for differential molecular diagnosis of Prader-Willi and Angelman syndromes, 472
- pedigree, Using gene carrier probability to select high risk families for identifying germline mutations in breast cancer susceptibility genes, 116
- Pendred syndrome, Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425, 202
- penetrance, Counselling familial issues in hypertrophic cardiomyopathy, 183
- pericentric inversion of X chromosome, Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH, 146
- periodic fever, Exclusion of the familial Mediterranean fever locus as a susceptibility region for autosomal dominant familial Hibernian fever, **432**
- persistent deciduous teeth, Unknown syndrome: peculiar face, severe hypodontia of permanent teeth, and precocious choroid calcifications, 435
- persistent pulmonary hypertension of the newborn, Familial persistent pulmonary hypertension of the newborn resulting from misalignment of the pulmonary vessels (congenital alveolar capillary dysplasia), 58
- Peutz-Jeghers disease, Peutz-Jeghers disease: most, but not all, families are compatible with linkage to 19p13.3, 42
- Pfeiffer syndrome, Presence of the Apert canonical S252W FGFR2 mutation in a patient without severe syndactyly, 677
- phosphate transporter, Localisation of the gene for glycogen storage disease type 1c by homozygosity mapping to 11q, 269
- phylogenetic footprints, Absence of mutations in the interspecies conserved regions of the CFTR promoter region in cystic fibrosis (CF) and CF related patients, 137
- physical mapping, Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH, 146
- PMP22 promoter, Mutation analysis of the nerve specific promoter of the peripheral myelin protein 22 gene in CMT1 disease and HNPP, 590
- polymorphism, A polymorphism of the CC16 gene is associated with an increased risk of asthma, 463
- population, The North Cumbria Community Genetics Project, 413 porokeratosis, Familial craniosynostosis, anal anomalies, and porokeratosis: CAP syndrome, 763
  portal venous malformation, Baller-Gerold syndrome associated
- with congenital portal venous malformation, 767
- Portuguese, Population genetics of hyperphenylalaninaemia resulting from phenylalanine hydroxylase deficiency in Portugal, 301
- positional cloning, Isolation of BAC clones spanning the Xq22.3 translocation breakpoint in a lissencephaly patient with a de novo X;2 translocation, 829
- PPT mutations, Sharing of PPT mutations between distinct clinical forms of neuronal ceroid lipofuscinoses in patients from Scotland: letter, 790
- Prader-Willi syndrome, The mechanisms involved in formation of deletions and duplications of 15q11-q13, 130
- Prader-Willi/Angelman syndrome, A rapid, PCR based test for differential molecular diagnosis of Prader-Willi and Angelman syndromes, 472

precocious puberty, Severe testotoxicosis phenotype associated with Asp-Tyr mutation of the lutrophin/choriogonadotrophin receptor gene, 340

- predicting distress, Predicting adaptation to presymptomatic DNA testing for late onset disorders: who will experience distress?, 745
- pre-eclampsia, Distortion of maternal-fetal angiotensin II type 1 receptor allele transmission in pre-eclampsia, 632
- premature ovarian failure, Studies of FRAXA and FRAXE in
- women with premature ovarian failure, 637 premutation, Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the FRDA gene, 713
  Studies of FRAXA and FRAXE in women with premature ovarian
  - failure, 637
- prenatal diagnosis, Cystic fibrosis screening: a fetus with hyperecho
  - genic bowel may be the index case, 657
    De novo unbalanced translocation resulting in monosomy for proximal 14q and distal 4p in a fetus with intrauterine growth retardation, Wolf-Hirschhorn syndrome, hypertrophic cardiomyopathy, and partial hemihypoplasia, 1050
  - Kyphomelic dysplasia in two sib fetuses, 65
  - Molecular, cytogenetic, and clinical characterisation of six XX males including one prenatal diagnosis, 727
    Prenatal detection of trisomy 21 and 18 from amniotic fluid by
- quantitative fluorescent polymerase chain reaction, 126
- Prenatal diagnosis of autosomal dominant polycystic kidney disease (PKD1) presenting in utero and prognosis for very early onset
- disease, 13
  prepyloric membrane, Familial occurrence congenital incomplete prepyloric mucosal diaphragm, 1040
- prepyloric web, Familial occurrence of congenital incomplete prepyloric mucosal diaphragm, 1040
- presenilin 1, De novo presenilin 1 mutations are rare in clinically sporadic, early onset Alzheimer's disease cases, 672
- presymptomatic DNA testing, Predicting adaptation to presymptomatic DNA testing for late onset disorders: who will experience distress?, 745
- **primary pulmonary hypoplasia**, Severe primary pulmonary hypoplasia ("acinar dysplasia") in sibs: a genetically determined mesodermal defect?: letter, 964
- Probst bundles, Agenesis of the corpus callosum with Probst bundles owing to haploinsufficiency for a gene in an 8 cM region of 6q25, 1031
- progressive sensorineural deafness, Distal 6p deletion syndrome: a report of a case with anterior chamber eye anomaly and review of published reports, 685
- prostate cancer, PTEN and prostate cancer: letter, 790
- protein, Molecular pathology of familial hypertrophic cardiomyopathy caused by mutations in the cardiac myosin binding protein C gene, **205**
- **protein assays**, Rapid immunoblot and kinase assay tests for a syndromal form of X linked mental retardation: Coffin-Lowry syndrome, 890
- protein truncation, Neurofibromatosis type 1 (NF1): a protein truncation assay yielding identification of mutations in 73% of patients, 813
- protoporphyrinogen oxidase gene, Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene, 244
- proximal 14q deletions, Holoprosencephaly in deletions of proximal
- chromosome 14q: letter, 612

  proximal 15q triplication, Triplication of 15q11-q13 with inv dup(15) in a female with developmental delay, 425
- psu dic(Xp), Three patients with a 45,X/46,X,psu dic(Xp) karyotype,
- psychological consequences, Psychosocial adaptation in adolescents and young adults with Marfan syndrome: an exploratory study, 405
- psychosocial, How the magnitude of clinical severity and recurrence risk affects reproductive decisions in adult males with different forms of progressive muscular dystrophy, 189
- psychosocial adaptation, Psychosocial adaptation in adolescents and young adults with Marfan syndrome: an exploratory study,
- PTEN, Germline PTEN mutations in Cowden syndrome-like families, 881
- Mutations of PTEN in patients with Bannayan-Riley-Ruvalcaba phenotype, 886
  PTEN and prostate cancer: letter, 790
- pulmonary abnormalities, Distal 10q trisomy syndrome with unusual cardiac and pulmonary abnormalities, 72
- quantitative PCR, Diagnosis of spinal muscular atrophy in an SMN non-deletion patient using a quantitative PCR screen and mutation analysis, 674
- R1066C CFTR mutation, Cystic fibrosis in a Puerto Rican female homozygous for the R1066C mutation: letter, 84
- recommendations education and training, Recommendations for Education and Training of Genetic Nurses and Counsellors in the United Kingdom, 410
- recurrence risks, Recurrence risks in mental retardation: review, 177

- redundant skin, Costello syndrome in two Brazilian children, 54 reflux nephropathy, Absence of PAX2 gene mutations in patients with primary familial vesicoureteric reflux, 338
- renal-coloboma syndrome, The prevalence of PAX2 mutations in patients with isolated colobomas or colobomas associated with urogenital anomalies, 806
- repeat expansion detection, Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method, 99 repeat instability, Unusual mutations in high functioning fragile X
- males: apparent instability of expanded unmethylated CGG repeats, 103
- replication study, Severe phenotype resulting from an active ring X chromosome in a female with a complex karyotype: characterisation and replication study, 932
- reproductive decision making, Knowledge, views, and experience of 25 women with myotonic dystrophy, 1020
  restriction endonuclease fingerprinting (REF), A silent mutation, C924T (G308G), in the L1CAM gene results in X linked hydrocephalus (HSAS), 456
- retina, A linkage survey of 20 dominant retinitis pigmentosa families: frequencies of the nine known loci and evidence for further heterogeneity, 1
  - A new family of Greek origin maps to the CRD locus for autosomal dominant cone-rod dystrophy on 19q, 429
- retinal degeneration, A linkage survey of 20 dominant retinitis pigmentosa families: frequencies of the nine known loci and evidence for further heterogeneity, 1
- retinal dystrophy, Two adult females with a distinct familial mental retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, with ataxia and hypotonia, similar facial appearance hypergonadotrophic hypogonadism, and retinal dystrophy, 333
- retinitis pigmentosa, A linkage survey of 20 dominant retinitis pigmentosa families: frequencies of the nine known loci and evidence for further heterogeneity, 1
- A new dominant retinitis pigmentosa family mapping to the RP18 locus on chromosome 1q11-21: letter, 788
  Further refinement of the Usher 2A locus at 1q41, 773
- Molecular study of the rhodopsin gene in retinitis pigmentosa patients in the Basque Country, 387
  Usher syndrome type III (USH3) linked to chromosome 3q in an Italian family, 666
- retrobulbar cysts, Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation, 342
- Rett syndrome, Chromosome mapping of Rett syndrome: a likely candidate region on the telomere of Xq, 297
- Linkage analysis in Rett syndrome families suggests that there may be a critical region at Xq28, 997
- reverse chromosome painting, Chromosome specific comparative genome hybridisation for determining the origin of intrachromosomal duplications, 37
- rhabdomyosarcoma, Costello syndrome: two cases with embryonal rhabdomyosarcoma, 1036
- rhodopsin, Molecular study of the rhodopsin gene in retinitis pigmentosa patients in the Basque Country, 387
- Rieger syndrome, A new approach to the elucidation of complex chromosome rearrangements illustrated by a case of Rieger syndrome, 234
- ring chromosome, Mosaic supernumerary ring chromosome 19 identified by comparative genomic hybridisation, 836
- risk rate estimates, Maternal age specific risk rate estimates for Down syndrome among live births in whites and other races from Ohio and Metropolitan Atlanta, 1970-1989, 482
- Robinow syndrome, Robinow syndrome: letter, 349
- RP11, RP11 is the second most common locus for dominant retinitis pigmentosa: letter, 174
- RP18 locus, A new dominant retinitis pigmentosa family mapping to the RP18 locus on chromosome 1q11-21: letter, 788
- Sanfilippo syndrome type B, Identification of 12 novel mutations in the α-N-acetylglucosaminidase gene in 14 patients with Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), 910
- $\delta$  sarcoglycan, A first missense mutation in the  $\delta$  sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian muscular dystrophy type sarcoglycanopathies, 951
  - Abnormalities of dystrophin, the sarcoglycans, and laminin  $\alpha 2$  in the muscular dystrophies, 379
- sarcoglycanopathies, A first missense mutation in the  $\delta$  sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies, 951
- segregation distortion, Segregation distortion in myotonic dystrophy, 1045
- seminoma, Seminoma in a postmenopausal woman with a Y;15 translocation in peripheral blood lymphocytes and a t(Y;15)/45,X Turner mosaic pattern in skin fibroblasts, 852
- sensorineural deafness, Lipomatous myelomeningocele, athyrotic hypothyroidism, and sensorineural deafness: a new form of syndromal deafness?, 948

- sensorineural hearing loss, A Moroccan family with autosomal recessive sensorineural hearing loss caused by a mutation in the gap junction protein gene connexin 26 (GJB2), 151
- septo-optic dysplasia, Septo-optic dysplasia and WS1 in the proband of a WS1 family segregating for a novel mutation in PAX3 exon 7, 248
- severe phenotype, Severe phenotype resulting from an active ring X chromosome in a female with a complex karyotype: characterisation and replication study, 932
- sex chromosome, 49,XXXXY: a distinct phenotype. Three new cases and review, 420
- sex determining region of Y (SRY), Seminoma in a postmenopausal woman with a Y;15 translocation in peripheral blood lymphocytes and a t(Y;15)/45,X Turner mosaic pattern in skin fibroblasts, 852
- sex reversal, Spastic paraplegia, optic atrophy, microcephaly with normal intelligence, and XY sex reversal: a new autosomal recessive syndrome?, 759

  short limbed dwarfism, A lethal skeletal dysplasia with features of
- chondrodysplasia punctata and osteogenesis imperfecta: an example of Astley-Kendall dysplasia. Further delineation of a rare genetic disorder, 505
- short stature, Minor disease features in neurofibromatosis type 1 (NF1) and their possible value in diagnosis of NF1 in children ≤6 years and clinically suspected of having NF1, 624 shoulder malformation, Oto-onycho-peroneal syndrome: confirma-
- tion of a syndrome, 508
- silent mutation, A silent mutation, C924T (G308G), in the L1CAM gene results in X linked hydrocephalus (HSAS), 456
- silver, Fibroblast silver loading for the diagnosis of Menkes disease,
- Silver-Russell syndrome, Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome, 784
- Simpson-Golabi-Behmel syndrome, A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma, 153
- sirenomelia, Cyclopia and sirenomelia in a liveborn infant : letter, 263 skeletal dysplasia, Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome, 288
- Smith-Lemli-Opitz syndrome, Smith-Lemli-Opitz syndrome: a variable clinical and biochemical phenotype, 558
- SOD1, Homozygosity for Asn86Ser mutation in the CuZn-superoxide dismutase gene produces a severe clinical phenotype in a juvenile onset case of familial amyotrophic lateral sclerosis: letter, 174 somatic mosaicism, Constitutional and mosaic large NF1 gene
- deletions in neurofibromatosis type 1, 468
  - Differential diagnosis of type 2 neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas, 973
  - Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations, 450
- South African black population, Molecular evidence that fragile X syndrome occurs in the South African black population: letter, 878
- spastic paraplegia, Spastic paraplegia, optic atrophy, microcephaly with normal intelligence, and XY sex reversal: a new autosomal recessive syndrome?, 759
- sperm, Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the FRDA gene, 713
- SPG4 locus, Mapping of a complicated familial spastic paraplegia to locus SPG4 on chromosome 2p, 89
  spina bifida, The C677T mutation of the 5,10-
- methylenetetrahydrofolate reductase gene is a moderate risk factor for spina bifida in Italy, 1009
- spinal muscular atrophy, Diagnosis of spinal muscular atrophy in an SMN non-deletion patient using a quantitative PCR screen and mutation analysis, 674
- spondyloepimetaphyseal dysplasia, distinct spondyloepimetaphyseal dysplasia with multiple dislocations, 566 sporadic cases, De novo presenilin 1 mutations are rare in clinically sporadic, early onset Alzheimer's disease cases, 672
- SRY, Molecular, cytogenetic, and clinical characterisation of six XX males including one prenatal diagnosis, 727
- SRY gene, An autosomal or X linked mutation results in true hermaphrodites and 46,XX males in the same family, 17
- SSCP, Mutation analysis of the nerve specific promoter of the peripheral myelin protein 22 gene in CMT1 disease and HNPP, 590
- Stargardt-like macular dystrophy, Assessment of the interphotoreceptor matrix proteoglycan-1 (IMPG1) gene localised to 6q13-q15 in autosomal dominant Stargardt-like disease (ADSTGD), progressive bifocal chorioretinal atrophy (PBCRA), and North Carolina macular dystrophy (MCDR1), 641
- storage, UK centres are not following the Royal College of Pathologists' recommendations for storage of Guthrie cards: a national policy is needed: letter, 263
- subtelomeric chromosome rearrangements, Del(18p) shown to be a cryptic translocation using a multiprobe FISH assay for subtelomeric chromosome rearrangements, 722
- survival motor neurone gene, Diagnosis of spinal muscular atrophy in an SMN non-deletion patient using a quantitative PCR screen and mutation analysis, 674

syndactyly, Extensive form of aplasia cutis congenita: a new syndrome?, 609

- syndactyly type I, Mesoaxial complete syndactyly and synostosis with hypoplastic thumbs: an unusual combination or homozygous expression of syndactyly type I?, 868
- systemic lupus erythematosus, Maternal systemic lupus erythematosus and chrondrodysplasia punctata in two sibs: phenocopy or coincidence?, 690
- tandem duplication, Mosaicism for a tandem duplication dup(1)(q12q22) in an 18 year old female, 600
- telomere, A new strategy for cryptic telomeric translocation screening in patients with idiopathic mental retardation, 225
- is determining fa 45,X/46,X,idic(Y), 862 testis factor, Discordant phenotypes
- testotoxicosis, Severe testotoxicosis phenotype associated with Asp→Tyr mutation of the lutrophin/choriogonadotrophin receptor gene, 340
- tetralogy of Fallot, Clinical features and mental development of a child with a prenatally identified 45,XX,der(5)t(5;18) (p15;q11.2),-18 karyotype, 865
- TGFa, Prediction of liability to orofacial clefting using genetic and craniofacial data from parents, 371
- three way translocation, Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs, 545
- thyroid, Germline PTEN mutations in Cowden syndrome-like families, 881
- tibial bowing, Medical complications of achondroplasia: a multicentre patient review, 705
- TIGR, A novel Asp380Ala mutation in the GLC1A/myocilin gene in a
- family with juvenile onset primary open angle glaucoma, 957 TIGR/MYOC mutations, Novel TIGR/MYOC mutations in families with juvenile onset primary open angle glaucoma, 989
- translocation, Acampomelic campomelic dysplasia with de novo 5q;17q reciprocal translocation and severe phenotype, 597
- translocation (18)(q42.1-p23.3), Partial trisomy 1(q42-qter): a new case with a mild phenotype, 75
- translocation (6;9), Further evidence for the involvement of human chromosome 6p24 in the aetiology of orofacial clefting, 857
- translocation (8p;21q), Two cases of partial trisomy 8p and partial monosomy 21q in a family with a reciprocal translocation (8;21)(p21.1;q22.3), 604
- transmission disequilibrium test, Linkage and association of an interleukin 4 gene polymorphism with atopic dermatitis in Japanese families, 502
- transthyretin, A pedigree analysis with minimised ascertainment bias shows anticipation in Met30-transthyretin related familial amyloid polyneuropathy, 23
- tricho-dento-osseous syndrome (TDO), A common DLX3 gene mutation is responsible for tricho-dento-osseous syndrome in Virginia and North Carolina families, 825
- tricuspid atresia, Tricuspid atresia in sibs: letter, 1055
- trinucleotide repeats, Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the FRDA gene, 713
- triophthalmia, Triophthalmia and facial clefting: a case report, 875 trisomy, Studies of non-disjunction in trisomies 2, 7, 15, and 22: does the parental origin of trisomy influence placental morphology?,
  - Trisomy  $2q11.2\rightarrow q21.1$  resulting from an unbalanced insertion in two generations, 319
- trisomy 18, Choroid plexus cysts and aneuploidy, 554
- trisomy 1q42-qter, Partial trisomy 1(q42-qter): a new case with a mild phenotype, 75 trisomy 21, Choroid plexus cysts and aneuploidy, 554
- trisomy 8p, Two cases of partial trisomy 8p and partial monosomy 21q in a family with a reciprocal translocation 21q in a family (8;21)(p21.1;q22.3), 604
- trophoblast, Studies of non-disjunction in trisomies 2, 7, 15, and 22: does the parental origin of trisomy influence placental morphology?, 924
- true hermaphrodite, An autosomal or X linked mutation results in true hermaphrodites and 46,XX males in the same family, 17
- TSC1 gene, Mutations in the TSC1 gene account for a minority of patients with tuberous sclerosis, 969
- TTF-1 gene, Holoprosencephaly in deletions of proximal chromosome 14q: letter, 612
- tuberculosis, Variants of  $\alpha$ -proteinase inhibitor in black and white South African patients with focal glomerulosclerosis and minimal change nephrotic syndrome, 6
- tuberous sclerosis, Familial cylindromatosis mimicking tuberous sclerosis complex and confirmation of the cylindromatosis locus, CYLD1, in a large family, 841

  Mutations in the TSC1 gene account for a minority of patients with tuberous sclerosis, 969
- Turner mosaicism, Seminoma in a postmenopausal woman with a Y;15 translocation in peripheral blood lymphocytes and a t(Y;15)/ 45,X Turner mosaic pattern in skin fibroblasts, 852

Turner syndrome, Discordant phenotypes and 45,X/46,X,idic(Y),

- Simultaneous adrenocortical carcinoma and ganglioneuroblastoma in a child with Turner syndrome and germline p53 mutation, 328 Three patients with a 45,X/46,X,psu dic(Xp) karyotype, 519 Uniparental and functional X disomy in Turner syndrome patients
- with unexplained mental retardation and X derived marker chromosomes, 539
- types I and III collagen defect, New lethal disease involving type I and III collagen defect resembling geroderma osteodysplastica, De Barsy syndrome, and Ehlers-Danlos syndrome IV, 513
- ulcerative colitis, Evidence of linkage of the inflammatory bowel disease susceptibility locus on chromosome 16 (IBD1) to ulcerative colitis, 218
- ultrasound, Cystic fibrosis screening: a fetus with hyperechogenic bowel may be the index case, 657
- Kyphomelic dysplasia in two sib fetuses, 65
- unaffected family, Instability in the normal CTG repeat range at the myotonic dystrophy locus: letter, 791
  uniparental disomy, Parental origin effects in human trisomy for
- chromosome 14q: implications for genomic imprinting, 821
- uniparental X disomy, Uniparental and functional X disomy in Turner syndrome patients with unexplained mental retardation and X derived marker chromosomes, 539
- Usher syndrome, Further refinement of the Usher 2A locus at 1q41,
  - Linkage analysis in Usher syndrome type I (USH1) families from Spain, 391
- Usher syndrome type III (USH3) linked to chromosome 3q in an Italian family, 666
- variable expression, A common DLX3 gene mutation is responsible for tricho-dento-osseous syndrome in Virginia and North Carolina families, 825
- variegate porphyria, Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene, 244
- velocardiofacial syndrome, A mother with VCFS and unilateral dysplastic kidney and her fetus with multicystic dysplastic kidneys: additional evidence to support the association of renal malformations and VCFS: letter, 348
- The annual incidence of DiGeorge/velocardiofacial syndrome: letter,
- vesicoureteric reflux, Absence of PAX2 gene mutations in patients with primary familial vesicoureteric reflux, 338
- The prevalence of PAX2 mutations in patients with isolated colobomas or colobomas associated with urogenital anomalies, 806
- vestibular schwannoma, Differential diagnosis of type 2 neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas, 973
- Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations, 450
- Waardenburg syndrome, Septo-optic dysplasia and WS1 in the proband of a WS1 family segregating for a novel mutation in PAX3 exon 7, 248
- Wales, Contracting for clinical genetic services: the Welsh model, 309 Weaver syndrome, Familial neurofibromatosis type 1 associated with an overgrowth syndrome resembling Weaver syndrome, 323
- eb, Mutation databases on the web: editorial, 529
- Wolcott-Rallison syndrome, Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome, 288
- WT1 gene, Do intronic mutations affecting splicing of WT1 exon 9 cause Frasier syndrome?, 45
- X;autosome translocation, Isolation of BAC clones spanning the Xq22.3 translocation breakpoint in a lissencephaly patient with a de novo X;2 translocation, 829
- X chromosome, Chromosome mapping of Rett syndrome: a likely candidate region on the telomere of Xq, 297
- Localisation of a gene for non-specific X linked mental retardation (MRX46) to Xq25-q26, 801
- X chromosome inactivation pattern, 1.4 Mb candidate gene region for X linked dyskeratosis congenita defined by combined haplotype and X chromosome inactivation analysis, 993
- X inactivation, Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH, 146 Genetic localisation of mental retardation with spastic diplegia to the pericentromeric region of the X chromosome: X inactivation in
  - female carriers, 284 Uniparental and functional X disomy in Turner syndrome patients with unexplained mental retardation and X derived marker chromosomes, 539
- X linked, Keratosis follicularis spinulosa decalvans: confirmation of linkage to Xp22.13-p22.2, 336
- X linked lissencephaly, Isolation of BAC clones spanning the Xq22.3 translocation breakpoint in a lissencephaly patient with a de novo X;2 translocation, 829

- X linked mental retardation, A family with mental retardation, variable macrocephaly and macro-orchidism, and linkage to Xq12-q21, 1026
   X linked susceptibility locus, Meiotic breakpoint mapping of a proposed X linked visual loss susceptibility locus in Leber's hereditary optic neuropathy, 668
   XLMR, A family with mental retardation, variable macrocephaly and macro-orchidism, and linkage to Xq12-q21, 1026
   Xp22.13-p22.2, Keratosis follicularis spinulosa decalvans: confirmation of linkage to Xp22.13-p22.2, 336

- **Xp-Yp interchanges,** Molecular, cytogenetic, and clinical characterisation of six XX males including one prenatal diagnosis, 727
- Xq28, 1.4 Mb candidate gene region for X linked dyskeratosis congenita defined by combined haplotype and X chromosome inactivation analysis, 993
  - Linkage analysis in Rett syndrome families suggests that there may be a critical region at Xq28, 997
- XX males, Molecular, cytogenetic, and clinical characterisation of six XX males including one prenatal diagnosis, 727