

# Subject index

- 1 translocation**, Transmission of an unbalanced (Y;1) translocation in Brittany, France, **e52**
- 10p13-14 deletion**, Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic conotruncal heart defects, **e16**
- 11q23**, Prevalence of *SDHB*, *SDHC*, and *SDHD* germline mutations in clinic patients with head and neck paragangliomas, **178**
- 15q11-q13**, Prader-Willi syndrome and a deletion/duplication within the 15q11-q13 region, **202**
- 16p trisomy**, "Molecular rulers" for calibrating phenotypic effects of telomere imbalance, **734**
- 17q**, Investigation of the *GRB2*, *GRB7*, and *CSH1* genes as candidates for the Silver-Russell syndrome (SRS) on chromosome 17q, **e13**
- 20p13**, Corneal dystrophy and perceptive deafness (Harboyan syndrome): *CDPD1* maps to 20p13, **110**
- 22q11.2 deletion**, A novel atypical 22q11.2 distal deletion in father and son, **e62**
- 22q11 deletion syndrome**, A novel atypical 22q11.2 distal deletion in father and son, **e62**
- 22q11 deletion**, Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic conotruncal heart defects, **e16**
- 22q11 deletions**, Absence of 22q11 deletions in 211 patients with developmental delay analysed using PCR, **e18**
- 47,XXX**, Phenotypic effects of mosaicism for a 47,XXX cell line in Turner syndrome, **217**
- 6q21**, Sorting nexin 3 (*SNX3*) is disrupted in a patient with a translocation t(6;13)(q21;q12) and microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP) phenotype, **893**
- 7510T>C mutation**, Maternally inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial tRNA<sup>Ser(UCN)</sup> gene, **e82**
- 7-dehydrocholesterol**, Smith-Lemli-Opitz syndrome: carrier frequency and spectrum of *DHCR7* mutations in Canada, **e31**
- 8994 polymorphism**, The frequency of mtDNA 8994 polymorphism and detection of the NARP 8993 mutation, **204**
- 8p23 1**, 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation?, **769**
- 8q21.2**, A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2;8 translocation, **391**
- α-transducin**, Mapping of a novel locus for achromatopsia (*ACHM4*) to 1p and identification of a germline mutation in the α subunit of cone transducin (*GNAT2*), **656**
- aberrant splicing**, A novel aberrant splice site mutation in the *APC* gene, **754**
- absent radius**, Thrombocytopenia-absent radius syndrome: a clinical genetic study, **876**
- ACE**, An investigation of ACE as a risk factor for dementia and cognitive decline in the general population, **403**
- achromatopsia**, Mapping of a novel locus for achromatopsia (*ACHM4*) to 1p and identification of a germline mutation in the α subunit of cone transducin (*GNAT2*), **656**
- acrocallosal syndrome**, De novo *GLI3* mutation in acrocallosal syndrome: broadening the phenotypic spectrum of *GLI3* defects and overlap with murine models, **804**
- acrofacial dysostosis**, Acrofacial dysostosis in a patient with the *TSC2-PKD1* contiguous gene syndrome, **136**
- acro-renal-ocular syndrome**, Okihiro syndrome and acro-renal-ocular syndrome: clinical overlap, expansion of the phenotype, and absence of *PAX2* mutations in two new families, **68**
- ACVRL1 gene**, Identification of 13 new mutations in the *ACVRL1* gene in a group of 52 unselected Italian patients affected by hereditary haemorrhagic telangiectasia, **e39**
- adenylosuccinate lyase deficiency**, Towards a suggestive facial dysmorphism in adenylosuccinate lyase deficiency?, **440**
- age**, Cytogenetic analysis of spermatozoa from males aged between 47 and 71 years, **e63**
- aggrecan**, Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene, **634**
- aldolase B**, Molecular analysis of the aldolase B gene in patients with hereditary fructose intolerance from Spain, **e56**
- alkaptonuria**, Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the *HGO* gene, **e40**
- Alport syndrome**, Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR), **359**
- Alzheimer's disease**, An investigation of ACE as a risk factor for dementia and cognitive decline in the general population, **403**
- Association of an interleukin 1B gene polymorphism (-511) with Parkinson's disease in Finnish patients, **400**
- aminoglycoside**, In vitro analysis of aminoglycoside therapy for the Arg120stop nonsense mutation in RP2 patients, **62**
- aneuploidy**, Cytogenetic analysis of spermatozoa from males aged between 47 and 71 years, **e63**
- Angelman syndrome**, A patient with a supernumerary marker chromosome (15), Angelman syndrome, and uniparental disomy resulting from paternal meiosis II non-disjunction, **e9**
- Exceptionally mild Angelman syndrome phenotype associated with an incomplete imprinting defect, **e51**
- Severe phenotype in Angelman syndrome resulting from paternal isochromosome 15, **e4**
- anophthalmia**, National study of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland: investigation of genetic aetiology, **16**
- anticonvulsant medication**, Antiepileptic drug therapy during pregnancy: the neurologist's perspective, **248**
- Assessing epidemiological evidence for the teratogenic effects of anticonvulsant medications, **243**
- The teratogenicity of anticonvulsant drugs: a progress report, **245**
- anticonvulsants**, Long term health and neurodevelopment in children exposed to antiepileptic drugs before birth, **251**
- APC gene**, A novel aberrant splice site mutation in the *APC* gene, **754**
- Genetic characterisation of patients with multiple colonic polyps, **297**
- apolipoprotein E**, Apolipoprotein E4 is only a weak predictor of dementia and cognitive decline in the general population, **639**
- arrhythmia**, SNP S1103Y in the cardiac sodium channel gene *SCN5A* is associated with cardiac arrhythmias and sudden death in a white family, **913**
- aspartoacylase gene mutations**, Two novel aspartoacylase gene (*ASP*) missense mutations specific to Norwegian and Swedish patients with Canavan disease, **e55**
- assisted reproduction**, Preimplantation genetic diagnosis in clinical practice, **6**
- association study**, Apolipoprotein E4 is only a weak predictor of dementia and cognitive decline in the general population, **639**
- ataxia**, Calculating predictive values for the large repeat alleles at the SCA8 locus in patients with ataxia, **935**
- Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjögren syndrome, **838**
- ataxia-telangiectasia**, *ATM* mutations in Finnish breast cancer patients, **192**
- ataxia-telangiectasia variant**, A late onset variant of ataxia-telangiectasia with a compound heterozygous genotype, A8030G/7481insA, **57**
- atherosclerosis**, Association of polymorphisms and allelic combinations in the tumour necrosis factor-α-complement MHC region with coronary artery disease, **46**
- ATM**, A late onset variant of ataxia-telangiectasia with a compound heterozygous genotype, A8030G/7481insA, **57**
- ATM mutations**, *ATM* mutations in Finnish breast cancer patients, **192**
- Constitutional alterations of the *ATM* gene in early onset sporadic breast cancer, **751**
- audiometry**, Distinctive audiometric features between USH2A and USH2B subtypes of Usher syndrome, **281**
- audiotapes**, The use of audiotapes in consultations with women from high risk breast cancer families: a randomised trial, **697**
- autism**, A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2;8 translocation, **391**
- An aetiological study of 25 mentally retarded adults with autism, **205**
- No association between *HOXA1* and *HOXB1* genes and autism spectrum disorders (ASD), **e70**
- autoimmune**, Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome, **537**
- autosomal dominant**, Autosomal dominant (AD) pure spastic paraplegia (HSP) linked to locus SPG4 affects almost exclusively males in a large pedigree, **e77**
- autosomal dominant cerebellar ataxia**, Predictive testing in the context of pregnancy: experience in Huntington's disease and autosomal dominant cerebellar ataxia, **522**
- autosomal dominant inheritance**, Spondyloepimetaphyseal dysplasia with multiple dislocations (Hall type): three further cases and evidence of autosomal dominant inheritance, **666**

- autosomal dominant polycystic kidney disease**, Mutation screening of the *PKD1* transcript by RT-PCR, **422**
- Omphalocele** in three generations with autosomal dominant transmission, **184**
- autosomal recessive inheritance**, Infantile spinal muscular atrophy variant with congenital fractures in a female neonate: evidence for autosomal recessive inheritance, **74**
- autosomal recessive primary microcephaly (MCPH)**, Autosomal recessive primary microcephaly: an analysis of locus heterogeneity and phenotypic variation, **718**
- autosomal recessive retinitis pigmentosa**, Novel homozygous mutation in the alpha subunit of the rod cGMP gated channel (*CNGA1*) in two Spanish sibs affected with autosomal recessive retinitis pigmentosa, **e66**
- autosome translocations**, Transmission of an unbalanced (Y;1) translocation in Brittany, France, **e52**
- autozygosity mapping**, Mapping of a novel locus for achromatopsia (*ACHM4*) to 1p and identification of a germline mutation in the  $\alpha$  subunit of cone transducin (*GNAT2*), **656**
- AZFc locus**, A new case of Yq microdeletion transmitted from a normal father to two infertile sons, **e27**
- Bannayan-Riley-Ruvalcaba syndrome**, Germline mutation of the tumour suppressor *PTEN* in Proteus syndrome, **937**
- behavioural complaints**, Behavioural complaints in participants who underwent predictive testing for Huntington's disease, **857**
- behavioural phenotype**, Behavioural phenotype of Bardet-Biedl syndrome, **e76**
- BHD hot spot mutation**, Clinical and genetic studies of Birt-Hogg-Dubé syndrome, **906**
- Birt-Hogg-Dubé syndrome**, Exclusion of *PTEN*, *CTNNB1*, and *PTCH* as candidate genes for Birt-Hogg-Dubé syndrome, **e10**
- Birt-Hogg-Dubé syndrome**, Clinical and genetic studies of Birt-Hogg-Dubé syndrome, **906**
- blepharophimosis-ptosis-epicanthus inversus syndrome (BPES)**, *FOXL2* mutation screening in a large panel of POF patients and XX males, **e43**
- bone**, Identification of a 52 kb deletion downstream of the *SOST* gene in patients with van Buchem disease, **91**
- bone dysplasia**, Dysosteosclerosis: a report of three new cases and evolution of the radiological findings, **603**
- Book reviews**, Bahou WF. Genetics for haematologists: the molecular genetic basis of haematological disorders , **303**
- Bale SJ. Genetics for dermatologists: the molecular genetic basis of dermatological disorders, **303**
- Canepa G, Maroteaux V, Pietrogrande V. Dysmorphic syndromes and constitutional diseases of the skeleton, **78**
- Hagerman RJ, Hagerman PJ, *editors*. Fragile X syndrome: diagnosis, treatment and research, **783**
- Harper PS. Myotonic dystrophy, **782**
- Hughes IA, Clark AJL, *editors*. Adrenal disease in childhood. Clinical and molecular aspects, **78**
- Marian AJ. Genetics for cardiologists: the molecular genetic basis for cardiovascular disorders, **454**
- Milunsky A. Your genetic destiny: know your genes, secure your health, save your life, **863**
- Passarge E. Colour atlas of genetics, **78**
- Peel RA, Timson J, *editors*. The century of Mendelism, **782**
- Rimoin DL, Connor JM, Pyeritz RE, Korf BR, *editors*. Emery and Rimoin's principles and practice of medical genetics, **454**
- Schinzl A. Catalogue of unbalanced chromosome aberrations in man, **375**
- Schneider JK. Counseling about cancer: strategies for genetic counseling, **615**
- Shepard TH. Catalog of teratogenic agents, **454**
- Winter RM, Baraitser M. Oxford medical databases. London dysmorphology database version 3.0. London neurogenetics database version 3.0. Dysmorphology photo library on CD-ROM version 3.0, **782**
- brachydactyly type A1**, A novel locus for brachydactyly type A1 on chromosome 5p13.3-p13.2, **186**
- brachydactyly**, Vitreoretinopathy with phalangeal epiphyseal dysplasia, a type II collagenopathy resulting from a novel mutation in the C-propeptide region of the molecule, **661**
- brain anomalies**, Corpus callosum hypoplasia and associated brain anomalies in Nijmegen breakage syndrome, **e25**
- branchio-oculo-facial syndrome**, Branchio-oculo-facial syndrome and branchio-otic/branchio-oto-renal syndromes are distinct entities, **71**
- BRCA1**, A breast cancer family from Spain with germline mutations in both the *BRCA1* and *BRCA2* genes, **e44**
- Bar code screening on combed DNA for large rearrangements of the *BRCA1* and *BRCA2* genes in French breast cancer families, **817**
- Change in the penetrance of founder *BRCA1/2* mutations? A retrospective cohort study, **407**
- Detection of large rearrangements of exons 13 and 22 in the *BRCA1* gene in German families, **e36**
- BRCA1/2**, A significant response to neoadjuvant chemotherapy in *BRCA1/2* related breast cancer, **608**
- Incidence of non-founder *BRCA1* and *BRCA2* mutations in high risk Ashkenazi breast and ovarian cancer families, **611**
- BRCA2**, A breast cancer family from Spain with germline mutations in both the *BRCA1* and *BRCA2* genes, **e44**
- Bar code screening on combed DNA for large rearrangements of the *BRCA1* and *BRCA2* genes in French breast cancer families, **817**
- Change in the penetrance of founder *BRCA1/2* mutations? A retrospective cohort study, **407**
- Contribution of *BRCA2* germline mutations to hereditary breast/ovarian cancer in Germany, **e12**
- Hereditary ovarian cancer resulting from a non-ovarian cancer cluster region (OCCR) *BRCA2* mutation: is the OCCR useful clinically?, **e68**
- Novel *BRCA2* mutation in a Polish family with hamartoma and two male breast cancers, **e35**
- Unique de novo mutation of *BRCA2* in a woman with early onset breast cancer, **126**
- breakpoint mapping**, A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2;8 translocation, **391**
- breast cancer**, A significant response to neoadjuvant chemotherapy in *BRCA1/2* related breast cancer, **608**
- ATM* mutations in Finnish breast cancer patients, **192**
- Change in the penetrance of founder *BRCA1/2* mutations? A retrospective cohort study, **407**
- Concerns of women presenting to a comprehensive cancer centre for genetic cancer risk assessment, **526**
- Family history of breast cancer as a determinant of the risk of developing endometrial cancer: a nationwide cohort study, **826**
- Genes other than *BRCA1* and *BRCA2* involved in breast cancer susceptibility, **225**
- Genetic counselling for familial breast and ovarian cancer in Ontario, **695**
- High frequency of skewed X inactivation in young breast cancer patients, **30**
- Incidence of non-founder *BRCA1* and *BRCA2* mutations in high risk Ashkenazi breast and ovarian cancer families, **611**
- Low rate of *TP53* germline mutations in breast cancer/sarcoma families not fulfilling classical criteria for Li-Fraumeni syndrome, **941**
- Management of women with a family history of breast cancer in the North West Region of England: training for implementing a vision of the future, **531**
- Molecular changes in skin predict predisposition to breast cancer, **e1**
- Risk assessment and management of high risk familial breast cancer, **865**
- The use of audiotapes in consultations with women from high risk breast cancer families: a randomised trial, **697**
- breast/ovarian cancer**, Detection of large rearrangements of exons 13 and 22 in the *BRCA1* gene in German families, **e36**
- campomelic dysplasia**, The phenotype of survivors of campomelic dysplasia, **597**
- Canavan disease**, Two novel aspartoacylase gene (*ASPA*) missense mutations specific to Norwegian and Swedish patients with Canavan disease, **e55**
- cancer**, Current status of human chromosome 14, **81**
- The null oncogene hypothesis and protection from cancer, **12**
- candidate gene analysis**, Mutation analysis in the candidate Möbius syndrome genes *PGT* and *GATA2* on chromosome 3 and *EGR2* on chromosome 10, **e30**
- carbohydrate deficient glycoprotein syndrome**, Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib, **849**
- carboxyl-terminal propeptide (C-propeptide)**, A single amino acid substitution (D1441Y) in the carboxyl-terminal propeptide of the  $\alpha 1(I)$  chain of type I collagen results in a lethal variant of osteogenesis imperfecta with features of dense bone diseases, **23**
- cardiac anomalies**, A locus for isolated cataract on human Xp, **105**
- cardiac defects**, 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation?, **769**
- cardiac sodium channel gene *SCN5A***, SNP S1103Y in the cardiac sodium channel gene *SCN5A* is associated with cardiac arrhythmias and sudden death in a white family, **913**
- cardiovascular manifestations**, Cardiovascular manifestations in 75 patients with Williams syndrome, **554**
- carotid body tumours**, Prevalence of *SDHB*, *SDHC*, and *SDHD* germline mutations in clinic patients with head and neck paragangliomas, **178**
- carrier frequency**, Smith-Lemli-Opitz syndrome: carrier frequency and spectrum of *DHCR7* mutations in Canada, **e31**
- carrier screening**, Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test results, **e26**
- cataract**, A locus for isolated cataract on human Xp, **105**



- Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjögren syndrome, **838**
- Novel mutations in the  $\gamma$ -crystallin genes cause autosomal dominant congenital cataracts, **352**
- cataracts**, Craniofacial anomalies, cataracts, congenital heart disease, sacral neural tube defects, and growth and developmental retardation in two sisters: a new autosomal recessive MCA/MR syndrome?, **145**
- cationic trypsinogen gene**, Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis, **347**
- CBP gene**, Molecular analysis of the *CBP* gene in 60 patients with Rubinstein-Taybi syndrome, **415**
- CCFDN syndrome**, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjögren syndrome, **838**
- CD14**, Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis, **844**
- CD36 deficiency**, Identification of cryptic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, **286**
- CDG Ib**, Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib, **849**
- cell cycle**, Expression of wild type and mutant *TSC2*, but not *TSC1*, causes an increase in the G1 fraction of the cell cycle in HEK293 cells, **676**
- centromere specific multicolour FISH**, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,+der(12)(p11→q11)/46,XX karyotype, **519**
- cerebral cortex**, Autosomal recessive primary microcephaly: an analysis of locus heterogeneity and phenotypic variation, **718**
- CFTR gene** Relationship between genotype and phenotype for the *CFTR* gene W846X mutation, **e32**
- CFTR mutations**, Splice mutation 1811+1.6kbA>G causes severe cystic fibrosis with pancreatic insufficiency: report of 11 compound heterozygous and two homozygous patients, **e73**
- CGG repeat**, A single base alteration in the CGG repeat region of *FMR1*: possible effects on gene expression and phenotype, **196**
- child abuse**, Testing for osteogenesis imperfecta in cases of suspected non-accidental injury, **382**
- chondrodysplasia**, Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene, **634**
- Vitreoretinopathy with phalangeal epiphyseal dysplasia, a type II collagenopathy resulting from a novel mutation in the C-propeptide region of the molecule, **661**
- chromosomal abnormalities**, Comparative genomic hybridisation shows a partial de novo deletion 16p11.2 in a neonate with multiple congenital malformations, **e24**
- chromosomal translocation**, Breakpoint analysis of a familial balanced translocation t(2;8)(q31;p21) associated with mesomelic dysplasia, **e34**
- chromosome**, 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation?, **769**
- chromosome 2**, Chromosome 2 aberrations in clinical cases characterised by high resolution multicolour banding and region specific FISH probes, **434**
- chromosome 5**, Familial pericentric inversion of chromosome 5 in a family with benign neonatal convulsions, **214**
- Partial trisomy 9p12p21.3 with a normal phenotype, **141**
- chromosome 5p13.3-p13.2**, A novel locus for brachydactyly type A1 on chromosome 5p13.3-p13.2, **186**
- chromosome 6**, Linkage of otosclerosis to a third locus (*OTSC3*) on human chromosome 6p21.3-22.3, **473**
- Major difference in aetiology and phenotypic abnormalities between transient and permanent neonatal diabetes, **370**
- Transient neonatal diabetes, a disorder of imprinting, **872**
- chromosome 6q deletion**, Deletion of the *SIM1* gene (6q16.2) in a patient with a Prader-Willi-like phenotype, **594**
- chromosome 9**, Partial trisomy 9p12p21.3 with a normal phenotype, **141**
- chromosome 12**, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,+der(12)(p11→q11)/46,XX karyotype, **519**
- Mosaicism for duplication 12q (12q13→12q21.2) accompanied by a pericentric inversion in a dysmorphic female infant, **e72**
- chromosome 14**, Current status of human chromosome 14, **81**
- chromosome 15**, Organisation of the pericentromeric region of chromosome 15: at least four partial gene copies are amplified in patients with a proximal duplication of 15q, **170**
- chromosome 15q26.1**, Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene, **634**
- chromosome 16p**, Mutations of the *UMOD* gene are responsible for medullary cystic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, **882**
- chromosome 17**, *PRKAR1A*, one of the Carney complex genes, and its locus (17q22-24) are rarely altered in pituitary tumours outside the Carney complex, **e78**
- chromosome 17q23**, Association between markers in chromosomal region 17q23 and young onset hypertension: a TDT study, **42**
- chromosome 18q21.1**, Homozygosity mapping of a Dyggve-Melchior-Clausen syndrome gene to chromosome 18q21.1, **714**
- chromosome 22**, Constitutional de novo interstitial deletion of 8 Mb on chromosome 22q12.1-12.3 encompassing the neurofibromatosis type 2 (NF2) locus in a dysmorphic girl with severe malformations, **e6**
- chromosome abnormality**, A school based study of children with learning disability indicates poor levels of genetic investigation, **e19**
- Phenotypic effects of mosaicism for a 47,XXX cell line in Turner syndrome, **217**
- chronic mucocutaneous candidiasis**, Familial chronic nail candidiasis with ICAM-1 deficiency: a new form of chronic mucocutaneous candidiasis, **671**
- cleft**, Analysis of the *p63* gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts, **559**
- cleft lip/palate**, Maternal *MTHFR* genotype contributes to the risk of non-syndromic cleft lip and palate, **368**
- clinical criteria**, Relationship between clinical and genetic diagnosis of Prader-Willi syndrome, **926**
- CLN2 gene**, Identification of novel *CLN2* mutations shows Canadian specific NCL2 alleles, **822**
- CNGA1**, Novel homozygous mutation in the alpha subunit of the rod cGMP gated channel (*CNGA1*) in two Spanish sibs affected with autosomal recessive retinitis pigmentosa, **e66**
- CNS dysfunction**, Comorbid *VHL* and *SCA2* mutations in a large kindred: confounding diagnosis of neurological dysfunction caused by CNS *VHL* vascular tumours versus *SCA2* atrophic neurodegeneration, **e37**
- coeliac disease**, Genome screening of coeliac disease, **328**
- CTLA-4/CD28* gene region is associated with genetic susceptibility to coeliac disease in UK families, **51**
- Coffin-Lowry syndrome**, Coffin-Lowry phenotype in a patient with a complex chromosome rearrangement, **e41**
- Coffin-Lowry syndrome: clinical and molecular features, **705**
- cognitive decline**, Apolipoprotein E4 is only a weak predictor of dementia and cognitive decline in the general population, **639**
- cohort study**, Family history of breast cancer as a determinant of the risk of developing endometrial cancer: a nationwide cohort study, **826**
- The effect of a single *BRCA2* mutation on cancer in Iceland, **457**
- COL1A2 mutation**, A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel *COL1A2* mutation, **128**
- COL2A1**, Vitreoretinopathy with phalangeal epiphyseal dysplasia, a type II collagenopathy resulting from a novel mutation in the C-propeptide region of the molecule, **661**
- coloboma**, National study of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland: investigation of genetic aetiology, **16**
- colorectal cancer**, Hereditary non-polyposis colorectal cancer: current risks of colorectal cancer largely overestimated, **335**
- Mismatch repair gene analysis in Catalan families with colorectal cancer, **e29**
- Naturally occurring mutations and functional polymorphisms in multidrug resistance 1 gene: correlation with microsatellite instability and lymphoid infiltration in colorectal cancers, **340**
- colorectal carcinoma**, Screening for microsatellite instability target genes in colorectal cancers, **785**
- colorectal neoplasia**, Clinical and genetic studies of Birt-Hogg-Dubé syndrome, **906**
- common ancestral haplotype**, A common ancestral haplotype in carrier chromosomes from different ethnic backgrounds in vacuolating megalencephalic leucoencephalopathy with subcortical cysts, **54**
- communication**, What do women really want to know? Motives for attending familial breast cancer clinics, **410**
- comparative genomic hybridisation (CGH)**, Comparative genomic hybridisation shows a partial de novo deletion 16p11.2 in a neonate with multiple congenital malformations, **e24**
- comparative maps**, Current status of human chromosome 14, **81**
- complex chromosome rearrangement**, Coffin-Lowry phenotype in a patient with a complex chromosome rearrangement, **e41**
- concordant deletion 22q11**, Monozygotic twins with chromosome 22q11 deletion and discordant phenotypes: updates with an epigenetic hypothesis, **e71**
- conformation sensitive gel electrophoresis**, Sensitivity of conformation sensitive gel electrophoresis in detecting mutations in Marfan syndrome and related conditions, **34**
- congenital fractures**, Infantile spinal muscular atrophy variant with congenital fractures in a female neonate: evidence for autosomal recessive inheritance, **74**

- congenital heart disease**, Craniofacial anomalies, cataracts, congenital heart disease, sacral neural tube defects, and growth and developmental retardation in two sisters: a new autosomal recessive MCA/MR syndrome?, **145**
- congenital hypomyelination**, Frequency of mutations in the early growth response 2 gene associated with peripheral demyelinating neuropathies, **e81**
- congenital malformations**, Comparative genomic hybridisation shows a partial de novo deletion 16p11.2 in a neonate with multiple congenital malformations, **e24**
- congenitally bowed femora**, A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel *COL1A2* mutation, **128**
- conotruncal heart defects**, Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic conotruncal heart defects, **e16**
- contiguous gene syndrome**, Monozygotic twins with chromosome 22q11 deletion and discordant phenotypes: updates with an epigenetic hypothesis, **e71**
- continuous medical education**, Continuous medical education approaches for clinical genetics: a postal survey of general practitioners, **e69**
- convulsions**, Familial pericentric inversion of chromosome 5 in a family with benign neonatal convulsions, **214**
- coping style**, Coping style, psychological distress, risk perception, and satisfaction in subjects attending genetic counselling for hereditary cancer, **689**
- copy number**, High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH), **790**
- corneal dystrophy**, Corneal dystrophy and perceptive deafness (Harboyan syndrome): *CDPD1* maps to 20p13, **110**
- coronary disease**, Association of polymorphisms and allelic combinations in the tumour necrosis factor- $\alpha$ -complement MHC region with coronary artery disease, **46**
- corpus callosum agenesis**, Report of a new case of "genitopatellar" syndrome which challenges the importance of absent patellae as a defining feature, **933**
- correlation**, Germline mutations in the von Hippel-Lindau (VHL) gene in patients from Poland: disease presentation in patients with deletions of the entire *VHL* gene, **e38**
- Costello syndrome**, A case of Costello syndrome and glycogen storage disease type III, **e8**
- Cowden syndrome**, Germline mutation of the tumour suppressor *PTEN* in Proteus syndrome, **937**
- COXIII stop mutation**, Childhood onset mitochondrial myopathy and lactic acidosis caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene, **812**
- cranial nerves**, Frequency of mutations in the early growth response 2 gene associated with peripheral demyelinating neuropathies, **e81**
- craniofacial anomalies**, Craniofacial anomalies, cataracts, congenital heart disease, sacral neural tube defects, and growth and developmental retardation in two sisters: a new autosomal recessive MCA/MR syndrome?, **145**
- cranosynostosis**, Heterozygous P250L mutation of fibroblast growth factor receptor 3 in a case of isolated cranosynostosis, **764**
- CREB**, Coffin-Lowry syndrome: clinical and molecular features, **705**
- CREBBP**, Molecular studies in 10 cases of Rubinstein-Taybi syndrome, including a mild variant showing a missense mutation in codon 1175 of *CREBBP*, **496**
- cri du chat syndrome**, Partial trisomy 9p12p21.3 with a normal phenotype, **141**
- Crohn's disease**, Association of Crohn's disease and ulcerative colitis with haplotypes of the *MLH1* gene in Italian inflammatory bowel disease patients, **332**
- cryptic splice acceptor site**, A novel aberrant splice site mutation in the *APC* gene, **754**
- cryptophthalmos**, Fraser syndrome and cryptophthalmos: review of the diagnostic criteria and evidence for phenotypic modules in complex malformation syndromes, **623**
- CSH1**, Investigation of the *GRB2*, *GRB7*, and *CSH1* genes as candidates for the Silver-Russell syndrome (SRS) on chromosome 17q, **e13**
- CTLA-4**, *CTLA-4/CD28* gene region is associated with genetic susceptibility to coeliac disease in UK families, **51**
- CtIP**, Screening for microsatellite instability target genes in colorectal cancers, **785**
- cystic fibrosis**, Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test results, **e26**
- Prenatal detection of cystic fibrosis by ultrasonography: a retrospective study of more than 346 000 pregnancies, **443**
- Relationship between genotype and phenotype for the *CFTR* gene W846X mutation, **e32**
- Splice mutation 1811+1.6kbA>G causes severe cystic fibrosis with pancreatic insufficiency: report of 11 compound heterozygous and two homozygous patients, **e73**
- Cx32 polymorphism**, Allelic variants in the 5' non-coding region of the connexin32 gene: possible pitfalls in the diagnosis of X linked Charcot-Marie-Tooth neuropathy (CMTX), **e58**
- Cx32 promoter P2**, Allelic variants in the 5' non-coding region of the connexin32 gene: possible pitfalls in the diagnosis of X linked Charcot-Marie-Tooth neuropathy (CMTX), **e58**
- de novo dup(19q)**, A rare case of a de novo dup(19q) associated with a mild phenotype, **e61**
- decision making process**, Parental attitude towards genetic testing for familial hypercholesterolaemia in children, **e49**
- deletion**, A novel 2 bp deletion in the *TM4SF2* gene is associated with MRX58, **430**
- Identification of a 52 kb deletion downstream of the *SOST* gene in patients with van Buchem disease, **91**
- deletion 9p**, Deletion of 9p associated with gonadal dysfunction in 46,XY but not in 46,XX human fetuses, **514**
- deletion/duplication**, Prader-Willi syndrome and a deletion/duplication within the 15q11-q13 region, **202**
- dementia**, An investigation of ACE as a risk factor for dementia and cognitive decline in the general population, **403**
- Apolipoprotein E4 is only a weak predictor of dementia and cognitive decline in the general population, **639**
- Spastic paraparesis and atypical dementia caused by *PSEN1* mutation (P264L), responsible for Alzheimer's disease, **e2**
- Denys-Drash syndrome**, A necropsy case of Denys-Drash syndrome with a *WT1* mutation in exon 7, **e48**
- derivative chromosomes**, Chromosome 2 aberrations in clinical cases characterised by high resolution multicolour banding and region specific FISH probes, **434**
- developmental abnormalities**, Major difference in aetiology and phenotypic abnormalities between transient and permanent neonatal diabetes, **370**
- developmental delay**, 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation?, **769**
- Absence of 22q11 deletions in 211 patients with developmental delay analysed using PCR, **e18**
- developmental dyslexia**, Supportive evidence for the *DYX3* dyslexia susceptibility gene in Canadian families, **125**
- DFNA41**, A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter, **567**
- DHCR7 mutations**, Smith-Lemli-Opitz syndrome: carrier frequency and spectrum of *DHCR7* mutations in Canada, **e31**
- DHPLC**, Twelve novel *FBN1* mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of *FBN1* mutation testing in clinical practice, **589**
- dilated cardiomyopathy**, Dilated cardiomyopathy, sudden cardiac death, hypoplastic discs, and retinal detachment: a new autosomal dominant syndrome, **221**
- disc herniation**, Genetic mapping of a susceptibility locus for disc herniation and spastic paraplegia on 6q23.3-q24.1, **387**
- disclosure**, Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test results, **e26**
- discordant phenotype**, Monozygotic twins with chromosome 22q11 deletion and discordant phenotypes: updates with an epigenetic hypothesis, **e71**
- distichiasis**, Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with *FOXC2* mutations or linkage to 16q24, **478**
- DMRT1**, Deletion of 9p associated with gonadal dysfunction in 46,XY but not in 46,XX human fetuses, **514**
- DNA repair**, Genetic variants of NHEJ DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, **900**
- DTDST mutations**, *DTDST* mutations are not a frequent cause of idiopathic talipes equinovarus (club foot), **e20**
- duplication**, Mosaicism for duplication 12q (12q13→12q21.2) accompanied by a pericentric inversion in a dysmorphic female infant, **e72**
- Organisation of the pericentromeric region of chromosome 15: at least four partial gene copies are amplified in patients with a proximal duplication of 15q, **170**
- duplication 15q11-13**, An aetiological study of 25 mentally retarded adults with autism, **205**
- duplication of proximal chromosome 1q**, Hereditary duplication of proximal chromosome 1q (q11q22) in a patient with T lymphoblastic lymphoma/leukaemia: a family study using G banding and comparative genomic hybridisation, **e79**
- Dyggve-Melchior-Clausen disease**, Homozygosity mapping of a Dyggve-Melchior-Clausen syndrome gene to chromosome 18q21.1, **714**
- dysgenetic testes**, A necropsy case of Denys-Drash syndrome with a *WT1* mutation in exon 7, **e48**
- dysmorphism**, Dysmorphism, variable overgrowth, normal bone age, and severe developmental delay: a "Sotos-like" syndrome?, **148**
- dysosteosclerosis**, Dysosteosclerosis: a report of three new cases and evolution of the radiological findings, **603**
- DYX3**, Supportive evidence for the *DYX3* dyslexia susceptibility gene in Canadian families, **125**
- early onset breast cancer**, Constitutional alterations of the *ATM* gene in early onset sporadic breast cancer, **751**



- echogenic bowel**, Prenatal detection of cystic fibrosis by ultrasonography: a retrospective study of more than 346 000 pregnancies, **443**
- ectopia lentis**, Sensitivity of conformation sensitive gel electrophoresis in detecting mutations in Marfan syndrome and related conditions, **34**
- ectrodactyly**, Sorting nexin 3 (*SNX3*) is disrupted in a patient with a translocation t(6;13)(q21;q12) and microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP) phenotype, **893**
- education**, Education improves general practitioner (GP) management of familial breast/ovarian cancer: findings from a cluster randomised controlled trial, **779**
- EEC syndrome**, Analysis of the *p63* gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts, **559**  
The *p63* gene in EEC and other syndromes, **377**
- EGR2 mutation**, Frequency of mutations in the early growth response 2 gene associated with peripheral demyelinating neuropathies, **e81**
- elastin vasculopathy**, Cardiovascular manifestations in 75 patients with Williams syndrome, **554**
- endometrial cancer**, Family history of breast cancer as a determinant of the risk of developing endometrial cancer: a nationwide cohort study, **826**
- enteropathy**, Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome, **537**
- epidemiology**, Assessing epidemiological evidence for the teratogenic effects of anticonvulsant medications, **243**  
The effect of a single *BRCA2* mutation on cancer in Iceland, **457**
- epilepsy**, Antiepileptic drug therapy during pregnancy: the neurologist's perspective, **248**  
Assessing epidemiological evidence for the teratogenic effects of anticonvulsant medications, **243**  
Long term health and neurodevelopment in children exposed to antiepileptic drugs before birth, **251**  
The teratogenicity of anticonvulsant drugs: a progress report, **245**
- ethics**, Parental attitude towards genetic testing for familial hypercholesterolaemia in children, **e49**
- evolution**, Evolution and expression of *FOXL2*, **916**
- externalising behaviour**, Behavioural phenotype of Bardet-Biedl syndrome, **e76**
- facial dysmorphism**, Towards a suggestive facial dysmorphism in adenylosuccinate lyase deficiency?, **440**
- facial nerve paralysis**, Mutation analysis in the candidate Möbius syndrome genes *PGT* and *GATA2* on chromosome 3 and *EGR2* on chromosome 10, **e30**
- familial**, Risk assessment and management of high risk familial breast cancer, **865**
- familial adenomatous polyposis**, A novel aberrant splice site mutation in the *APC* gene, **754**
- familial breast cancer**, What do women really want to know? Motives for attending familial breast cancer clinics, **410**
- familial breast/ovarian cancer**, Education improves general practitioner (GP) management of familial breast/ovarian cancer: findings from a cluster randomised controlled trial, **779**
- familial chronic nail candidiasis**, Familial chronic nail candidiasis with ICAM-1 deficiency: a new form of chronic mucocutaneous candidiasis, **671**
- familial hypercholesterolaemia**, CYS127S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, **e74**
- familial juvenile hyperuricaemic nephropathy**, Mutations of the *UMOD* gene are responsible for medullary cystic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, **882**
- familiarity**, The effect of a single *BRCA2* mutation on cancer in Iceland, **457**
- family history**, Family history of breast cancer as a determinant of the risk of developing endometrial cancer: a nationwide cohort study, **826**  
Management of women with a family history of breast cancer in the North West Region of England: training for implementing a vision of the future, **531**
- FBN1***, Sensitivity of conformation sensitive gel electrophoresis in detecting mutations in Marfan syndrome and related conditions, **34**
- FBN1* mutation detection**, Twelve novel *FBN1* mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of *FBN1* mutation testing in clinical practice, **589**
- FEO**, Familial expansile osteolysis in a large Spanish kindred resulting from an insertion mutation in the *TNFRSF11A* gene, **e67**
- fetal facies**, Robinow syndrome, **305**
- FGFR3* P250L mutation**, Heterozygous P250L mutation of fibroblast growth factor receptor 3 in a case of isolated craniosynostosis, **764**
- fibre diffraction**, Molecular changes in skin predict predisposition to breast cancer, **e1**
- fibrillin-1**, Twelve novel *FBN1* mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of *FBN1* mutation testing in clinical practice, **589**
- fibroblast growth factor type 3**, Heterozygous P250L mutation of fibroblast growth factor receptor 3 in a case of isolated craniosynostosis, **764**
- fibrocalculus pancreatic diabetes**, Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis, **347**
- fibulin-1**, The fibulin-1 gene (*FBLN1*) is disrupted in a t(12;22) associated with a complex type of synpolydactyly, **98**
- fingers**, Multifocal glomus tumours of the fingers in two patients with neurofibromatosis type 1, **e45**
- Finnish**, Four novel mutations in the *OFD1* (*Cxorf5*) gene in Finnish patients with oral-facial-digital syndrome 1, **292**
- FISH**, Chromosome 2 aberrations in clinical cases characterised by high resolution multicolour banding and region specific FISH probes, **434**  
Pure terminal duplication of the short arm of chromosome 19 in a boy with mild microcephaly, **e60**  
Subtelomeric rearrangements detected by FISH in three of 33 families with idiopathic mental retardation and minor physical anomalies, **e53**
- fluorescent genotyping**, Automated fluorescent genotyping detects 10% of cryptic subtelomeric rearrangements in idiopathic syndromic mental retardation, **266**
- FMR1***, A single base alteration in the CGG repeat region of *FMR1*: possible effects on gene expression and phenotype, **196**  
Mosaicism for *FMR1* and *FMR2* deletion: a new case, **200**
- FMR2***, Mosaicism for *FMR1* and *FMR2* deletion: a new case, **200**
- folate**, Inositol and folate resistant neural tube defects, **e5**  
Maternal *MTHFR* genotype contributes to the risk of non-syndromic cleft lip and palate, **368**
- follic acid supplementation**, Changes in frequencies of heterozygous thermolabile 5,10-methylenetetrahydrofolate reductase gene in fetuses with neural tube defects, **366**
- founder effect**, Identification of novel *CLN2* mutations shows Canadian specific NCL2 alleles, **822**
- founder mutation**, Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the *HGO* gene, **e40**
- FOXC2***, Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with *FOXC2* mutations or linkage to 16q24, **478**
- FOXL2***, Evolution and expression of *FOXL2*, **916**  
*FOXL2* mutation screening in a large panel of POF patients and XX males, **e43**
- fragile X syndrome**, A school based study of children with learning disability indicates poor levels of genetic investigation, **e19**  
A single base alteration in the CGG repeat region of *FMR1*: possible effects on gene expression and phenotype, **196**  
Mosaicism for *FMR1* and *FMR2* deletion: a new case, **200**
- frameshift mutation**, Two novel frameshift mutations in *NKX2.5* result in novel features including visceral inversus and sinus venosus type ASD, **807**
- Fraser syndrome**, Fraser syndrome and cryptophthalmos: review of the diagnostic criteria and evidence for phenotypic modules in complex malformation syndromes, **623**
- frequency**, Molecular screening for Smith-Magenis syndrome among patients with mental retardation of unknown cause, **e59**
- FRN**, Correlation between a specific Wilms tumour suppressor gene (*WT1*) mutation and the histological findings in Wilms tumour (WT), **e83**
- fructose intolerance**, Molecular analysis of the aldolase B gene in patients with hereditary fructose intolerance from Spain, **e56**
- $\gamma$ -crystallin**, Novel mutations in the  $\gamma$ -crystallin genes cause autosomal dominant congenital cataracts, **352**
- GCY* gene**, Localisation of the Y chromosome stature gene to a 700 kb interval in close proximity to the centromere, **507**
- gene mapping**, Identification of candidate lung cancer susceptibility genes in mouse using oligonucleotide arrays, **644**
- gene mutation**, CYS127S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, **e74**
- general practitioner**, Education improves general practitioner (GP) management of familial breast/ovarian cancer: findings from a cluster randomised controlled trial, **779**
- general practitioners**, Continuous medical education approaches for clinical genetics: a postal survey of general practitioners, **e69**
- genetic aetiology**, National study of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland: investigation of genetic aetiology, **16**
- genetic cancer risk assessment**, Concerns of women presenting to a comprehensive cancer centre for genetic cancer risk assessment, **526**
- genetic counselling**, Coping style, psychological distress, risk perception, and satisfaction in subjects attending genetic counselling for hereditary cancer, **689**

- Genetic counselling for familial breast and ovarian cancer in Ontario, **695**
- Genetic testing and genetic counselling in hypertrophic cardiomyopathy: the French experience, **741**
- What do women really want to know? Motives for attending familial breast cancer clinics, **410**
- genetic diagnosis**, Relationship between clinical and genetic diagnosis of Prader-Willi syndrome, **926**
- genetic polymorphisms**, Genes other than *BRCA1* and *BRCA2* involved in breast cancer susceptibility, **225**
- genetic registers**, Comparison of genetic services with and without genetic registers: access and attitudes to genetic counselling services among relatives of genetic clinic patients, **e85**
- Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes about genetic counselling among probands referred to three genetic clinics, **e84**
- genetic services**, Genetic counselling for familial breast and ovarian cancer in Ontario, **695**
- Preimplantation genetic diagnosis in clinical practice, **6**
- genetic testing**, Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss, **449**
- Calculating predictive values for the large repeat alleles at the SCA8 locus in patients with ataxia, **935**
- Genetic testing in hereditary non-polyposis colorectal cancer families with a *MSH2*, *MLH1*, or *MSH6* mutation, **833**
- Parental attitude towards genetic testing for familial hypercholesterolaemia in children, **e49**
- genetics**, A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter, **567**
- CTLA-4/CD28* gene region is associated with genetic susceptibility to coeliac disease in UK families, **51**
- Molecular analysis of the aldolase B gene in patients with hereditary fructose intolerance from Spain, **e56**
- genetics counselling**, Management of women with a family history of breast cancer in the North West Region of England: training for implementing a vision of the future, **531**
- genital**, Report of a new case of "genitopatellar" syndrome which challenges the importance of absent patellae as a defining feature, **933**
- genitopatellar syndrome**, Report of a new case of "genitopatellar" syndrome which challenges the importance of absent patellae as a defining feature, **933**
- genome screening**, Genome screening of coeliac disease, **328**
- genomic imprinting**, Prevalence of *SDHB*, *SDHC*, and *SDHD* germline mutations in clinic patients with head and neck paragangliomas, **178**
- genotype-phenotype**, Germline mutations in the von Hippel-Lindau (VHL) gene in patients from Poland: disease presentation in patients with deletions of the entire *VHL* gene, **e38**
- genotype-phenotype correlation**, Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy, **722**
- Maternally inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial tRNA<sup>Ser(UCN)</sup> gene, **e82**
- Splice mutation 1811+1.6kbA>G causes severe cystic fibrosis with pancreatic insufficiency: report of 11 compound heterozygous and two homozygous patients, **e73**
- genotype/phenotype correlations**, "Molecular rulers" for calibrating phenotypic effects of telomere imbalance, **734**
- germline mutation**, Germline mutations in the von Hippel-Lindau (VHL) gene in patients from Poland: disease presentation in patients with deletions of the entire *VHL* gene, **e38**
- SDHB* mutation analysis in familial and sporadic pheochromocytoma identifies a novel mutation, **e64**
- germline mutations**, Contribution of *BRCA2* germline mutations to hereditary breast/ovarian cancer in Germany, **e12**
- GLI3*, De novo *GLI3* mutation in acrocallosal syndrome: broadening the phenotypic spectrum of *GLI3* defects and overlap with murine models, **804**
- glutathione S-transferase P1-1**, Paternal contribution to the risk for pre-eclampsia, **44**
- glycogen storage disease type III**, A case of Costello syndrome and glycogen storage disease type III, **e8**
- GNAT2*, Mapping of a novel locus for achromatopsia (*ACHM4*) to 1p and identification of a germline mutation in the  $\alpha$  subunit of cone transducin (*GNAT2*), **656**
- gonadal dysfunction**, Deletion of 9p associated with gonadal dysfunction in 46,XY but not in 46,XX human fetuses, **514**
- gout**, Mutations of the *UMOD* gene are responsible for medullary cystic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, **882**
- GPs' attitudes**, Continuous medical education approaches for clinical genetics: a postal survey of general practitioners, **e69**
- GRB2*, Investigation of the *GRB2*, *GRB7*, and *CSH1* genes as candidates for the Silver-Russell syndrome (SRS) on chromosome 17q, **e13**
- GRB7*, Investigation of the *GRB2*, *GRB7*, and *CSH1* genes as candidates for the Silver-Russell syndrome (SRS) on chromosome 17q, **e13**
- GSH2* gene, Heterozygous truncating mutation in the human homeobox gene *GSH2* has no discernable phenotypic effect, **686**
- GSTP1*, Paternal contribution to the risk for pre-eclampsia, **44**
- hamartoma**, Novel *BRCA2* mutation in a Polish family with hamartoma and two male breast cancers, **e35**
- hand-foot-genital syndrome (HFGS)**, Severe digital abnormalities in a patient heterozygous for both a novel missense mutation in *HOXD13* and a polyalanine tract expansion in *HOXA13*, **852**
- haploinsufficiency**, Deletion of the *OPA1* gene in a dominant optic atrophy family: evidence that haploinsufficiency is the cause of disease, **e47**
- Evolution and expression of *FOXL2*, **916**
- Harboyan syndrome**, Corneal dystrophy and perceptive deafness (Harboyan syndrome): *CDPD1* maps to 20p13, **110**
- hearing loss**, Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss, **449**
- Novel autosomal dominant mandibulofacial dysostosis with ptosis: clinical description and exclusion of *TCOF1*, **484**
- Novel *ATP6V1B1* and *ATP6V0A4* mutations in autosomal recessive distal renal tubular acidosis with new evidence for hearing loss, **796**
- height**, Localisation of the Y chromosome stature gene to a 700 kb interval in close proximity to the centromere, **507**
- hereditary**, Familial adult renal neoplasia, **1**
- hereditary breast cancer**, Unique de novo mutation of *BRCA2* in a woman with early onset breast cancer, **126**
- hereditary breast/ovarian cancer**, Contribution of *BRCA2* germline mutations to hereditary breast/ovarian cancer in Germany, **e12**
- hereditary cancer**, Coping style, psychological distress, risk perception, and satisfaction in subjects attending genetic counselling for hereditary cancer, **689**
- hereditary haemorrhagic telangiectasia**, Identification of 13 new mutations in the *ACVRL1* gene in a group of 52 unselected Italian patients affected by hereditary haemorrhagic telangiectasia, **e39**
- hereditary motor and sensory neuropathy**, Allelic variants in the 5' non-coding region of the connexin32 gene: possible pitfalls in the diagnosis of X linked Charcot-Marie-Tooth neuropathy (CMTX), **e58**
- hereditary ovarian cancer**, Hereditary ovarian cancer resulting from a non-ovarian cancer cluster region (OCCR) *BRCA2* mutation: is the OCCR useful clinically?, **e68**
- hereditary spastic paraplegia**, Autosomal dominant (AD) pure spastic paraplegia (HSP) linked to locus SPG4 affects almost exclusively males in a large pedigree, **e77**
- Spastin* gene mutation in Japanese with hereditary spastic paraplegia, **e46**
- heterogeneity**, Autosomal recessive primary microcephaly: an analysis of locus heterogeneity and phenotypic variation, **718**
- HGO gene**, Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the *HGO* gene, **e40**
- high myopia**, A genome wide scan for familial high myopia suggests a novel locus on chromosome 7q36, **118**
- high risk**, Risk assessment and management of high risk familial breast cancer, **865**
- Hirschsprung disease**, Hirschsprung disease and *L1CAM*: is the disturbed sex ratio caused by *L1CAM* mutations?, **e11**
- histone acetyltransferase domain**, Molecular studies in 10 cases of Rubinstein-Taybi syndrome, including a mild variant showing a missense mutation in codon 1175 of *CREBBP*, **496**
- HLA region**, Linkage of otosclerosis to a third locus (*OTSC3*) on human chromosome 6p21.3-22.3, **473**
- hMLH1 mutations**, Mismatch repair gene analysis in Catalanian families with colorectal cancer, **e29**
- HNPCC**, An *MLH1* haplotype is over-represented on chromosomes carrying an HNPCC predisposing mutation in *MLH1*, **323**
- Description and functional analysis of a novel in frame mutation linked to hereditary non-polyposis colorectal cancer, **747**
- Genetic testing in hereditary non-polyposis colorectal cancer families with a *MSH2*, *MLH1*, or *MSH6* mutation, **833**
- Germline *MSH2* and *MLH1* mutational spectrum in HNPCC families from Poland and the Baltic States, **e65**
- Hereditary non-polyposis colorectal cancer: current risks of colorectal cancer largely overestimated, **335**
- homeobox domain**, Heterozygous truncating mutation in the human homeobox gene *GSH2* has no discernable phenotypic effect, **686**
- homozygosity mapping**, Genetic mapping of a susceptibility locus for disc herniation and spastic paraplegia on 6q23.3-q24.1, **387**
- Homozygosity mapping of a Dyggve-Melchior-Clausen syndrome gene to chromosome 18q21.1, **714**



- HOX genes**, Severe digital abnormalities in a patient heterozygous for both a novel missense mutation in *HOXD13* and a polyalanine tract expansion in *HOXA13*, **852**
- HOXA1**, No association between *HOXA1* and *HOXB1* genes and autism spectrum disorders (ASD), **e70**
- HOXB1**, No association between *HOXA1* and *HOXB1* genes and autism spectrum disorders (ASD), **e70**
- HOXD**, Breakpoint analysis of a familial balanced translocation t(2;8)(q31;p21) associated with mesomelic dysplasia, **e34**
- H<sup>+</sup>-ATPase** Novel *ATP6V1B1* and *ATP6V0A4* mutations in autosomal recessive distal renal tubular acidosis with new evidence for hearing loss, **796**
- human gene mutations**, Human gene mutations causing infertility, **153**
- Huntington's disease**, Behavioural complaints in participants who underwent predictive testing for Huntington's disease, **857**  
Predictive testing in the context of pregnancy: experience in Huntington's disease and autosomal dominant cerebellar ataxia, **522**
- hypercholesterolaemia**, Identification of mutations in the gene encoding sterol regulatory element binding protein (SREBP)-2 in hypercholesterolaemic subjects, **271**
- hypertension**, Association between markers in chromosomal region 17q23 and young onset hypertension: a TDT study, **42**
- hypertrophic cardiomyopathy**, Genetic testing and genetic counselling in hypertrophic cardiomyopathy: the French experience, **741**
- hypoplastic discs**, Dilated cardiomyopathy, sudden cardiac death, hypoplastic discs, and retinal detachment: a new autosomal dominant syndrome, **221**
- ICAM-1 deficiency**, Familial chronic nail candidiasis with ICAM-1 deficiency: a new form of chronic mucocutaneous candidiasis, **671**
- idiopathic mental retardation**, Subtelomeric rearrangements detected by FISH in three of 33 families with idiopathic mental retardation and minor physical anomalies, **e53**
- IGF2**, Characterisation of the growth regulating gene *IMP3*, a candidate for Silver-Russell syndrome, **575**
- IMP3**, Characterisation of the growth regulating gene *IMP3*, a candidate for Silver-Russell syndrome, **575**
- imprinting**, Characterisation of the growth regulating gene *IMP3*, a candidate for Silver-Russell syndrome, **575**  
Current status of human chromosome 14, **81**  
Major difference in aetiology and phenotypic abnormalities between transient and permanent neonatal diabetes, **370**  
Transient neonatal diabetes, a disorder of imprinting, **872**
- in situ hybridisation**, Cytogenetic analysis of spermatozoa from males aged between 47 and 71 years, **e63**
- inborn errors**, Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib, **849**
- incomplete imprinting defect**, Exceptionally mild Angelman syndrome phenotype associated with an incomplete imprinting defect, **e51**
- infantile spinal muscular atrophy**, Infantile spinal muscular atrophy variant with congenital fractures in a female neonate: evidence for autosomal recessive inheritance, **74**
- infertility**, Human gene mutations causing infertility, **153**
- inheritance**, Mutation in *KCNQ1* that has both recessive and dominant characteristics, **681**
- inositol**, Inositol and folate resistant neural tube defects, **e5**
- interleukin 1B gene polymorphism**, Association of an interleukin 1B gene polymorphism (-511) with Parkinson's disease in Finnish patients, **400**
- internalising behaviour**, Behavioural phenotype of Bardet-Biedl syndrome, **e76**
- interstitial deletion**, Constitutional de novo interstitial deletion of 8 Mb on chromosome 22q12.1-12.3 encompassing the neurofibromatosis type 2 (NF2) locus in a dysmorphic girl with severe malformations, **e6**
- interstitial telomeres**, Interstitial telomeres of an inv(9)(p11.2;q34) involved in a jumping translocation found in a woman through a stable unbalanced translocation in her malformed child, **e42**
- intrahepatic arteriovenous shunts**, Identification of 13 new mutations in the *ACVRL1* gene in a group of 52 unselected Italian patients affected by hereditary haemorrhagic telangiectasia, **e39**
- intron 14**, The intron 14 2140+5G>A variant in the low density lipoprotein receptor gene has no effect on plasma cholesterol levels, **e57**
- inv(9)**, Interstitial telomeres of an inv(9)(p11.2;q34) involved in a jumping translocation found in a woman through a stable unbalanced translocation in her malformed child, **e42**
- investigations**, A school based study of children with learning disability indicates poor levels of genetic investigation, **e19**
- IPEX syndrome**, Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome, **537**
- ischaemic heart disease**, Identification of cryptic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, **286**
- isochromosome 15**, Severe phenotype in Angelman syndrome resulting from paternal isochromosome 15, **e4**
- jumping translocation**, Interstitial telomeres of an inv(9)(p11.2;q34) involved in a jumping translocation found in a woman through a stable unbalanced translocation in her malformed child, **e42**
- KCNQ1**, Mutation in *KCNQ1* that has both recessive and dominant characteristics, **681**
- kidney**, Novel *ATP6V1B1* and *ATP6V0A4* mutations in autosomal recessive distal renal tubular acidosis with new evidence for hearing loss, **796**
- KLF8**, Abnormal expression of the *KLF8* (*ZNF741*) gene in a female patient with an X;autosomal translocation t(X;21)(p11.2;q22.3) and non-syndromic mental retardation, **113**
- kidney tumours**, Familial adult renal neoplasia, **1**
- Klinefelter syndrome**, Klinefelter-like phenotype and primary infertility in a male with a paracentric Xq inversion, **e28**
- kyphomelic dysplasia**, A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel *COL1A2* mutation, **128**
- L1CAM**, Hirschsprung disease and *L1CAM*: is the disturbed sex ratio caused by *L1CAM* mutations?, **e11**
- lack of association**, No association between *HOXA1* and *HOXB1* genes and autism spectrum disorders (ASD), **e70**
- lactic acidosis**, Childhood onset mitochondrial myopathy and lactic acidosis caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene, **812**
- language delay**, Molecular characterisation of a ring chromosome 22 in a patient with severe language delay: a contribution to the refinement of the subtelomeric 22q deletion syndrome, **e17**
- late infantile onset neuronal ceroid lipofuscinosis (NCL2, LINCL)**, Identification of novel *CLN2* mutations shows Canadian specific NCL2 alleles, **822**
- Laurence-Moon-Bardet-Biedl syndrome**, Behavioural phenotype of Bardet-Biedl syndrome, **e76**
- LDL receptor**, CYS127S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, **e74**
- learning disability**, A school based study of children with learning disability indicates poor levels of genetic investigation, **e19**  
High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH), **790**
- Leber hereditary optic neuropathy**, Leber hereditary optic neuropathy, **162**
- Leber optic atrophy**, Leber hereditary optic neuropathy, **162**
- LEOPARD syndrome**, *PTPN11* mutations in LEOPARD syndrome, **571**
- Leri-Weill dyschondrosteosis**, *SHOX* point mutations and deletions in Leri-Weill dyschondrosteosis, **e33**  
Prevalence of mutations in the short stature homeobox containing gene (*SHOX*) in Madelung deformity of childhood, **758**
- leukaemia**, Hereditary duplication of proximal chromosome 1q (q11q22) in a patient with T lymphoblastic lymphoma/leukaemia: a family study using G banding and comparative genomic hybridisation, **e79**
- Li-Fraumeni syndrome**, Low rate of *TP53* germline mutations in breast cancer/sarcoma families not fulfilling classical criteria for Li-Fraumeni syndrome, **941**
- ligase IV**, Genetic variants of NHEJ DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, **900**
- limb**, Thrombocytopenia-absent radius syndrome: a clinical genetic study, **876**
- limb development**, The fibulin-1 gene (*FBLN1*) is disrupted in a t(12;22) associated with a complex type of synpolydactyly, **98**
- limb malformations**, Severe digital abnormalities in a patient heterozygous for both a novel missense mutation in *HOXD13* and a polyalanine tract expansion in *HOXA13*, **852**
- linkage**, Linkage stratification and mutation analysis at the *parkin* locus identifies mutation positive Parkinson's disease families, **489**  
Supportive evidence for the *DYX3* dyslexia susceptibility gene in Canadian families, **125**
- linkage analysis**, A genome wide scan for familial high myopia suggests a novel locus on chromosome 7q36, **118**  
A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter, **567**  
Identification of candidate lung cancer susceptibility genes in mouse using oligonucleotide arrays, **644**  
Linkage of otosclerosis to a third locus (*OTSC3*) on human chromosome 6p21.3-22.3, **473**
- linkage disequilibrium**, An *MLH1* haplotype is over-represented on chromosomes carrying an HNPCC predisposing mutation in *MLH1*, **323**

- Specific haplotypes of the *RET* proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma, **260**
- Supportive evidence for the *DYX3* dyslexia susceptibility gene in Canadian families, **125**
- lipodystrophy**, Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy, **722**
- lipopolysaccharide**, Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis, **844**
- locus heterogeneity**, A genome wide scan for familial high myopia suggests a novel locus on chromosome 7q36, **118**
- LOH**, A breast cancer family from Spain with germline mutations in both the *BRCA1* and *BRCA2* genes, **e44**
- long QT syndrome**, Mutation in *KCNQ1* that has both recessive and dominant characteristics, **681**
- SNP S1103Y in the cardiac sodium channel gene *SCN5A* is associated with cardiac arrhythmias and sudden death in a white family, **913**
- low density lipoprotein receptor gene (LDLR)**, The intron 14 2140+5G>A variant in the low density lipoprotein receptor gene has no effect on plasma cholesterol levels, **e57**
- low penetrance gene**, Genes other than *BRCA1* and *BRCA2* involved in breast cancer susceptibility, **225**
- low penetrance mutation**, A T to C mutation in the polypyrimidine tract of the exon 9 splicing site of the *RB1* gene responsible for low penetrance hereditary retinoblastoma, **e21**
- lymphangiomyomatosis**, Expression of wild type and mutant *TSC2*, but not *TSC1*, causes an increase in the G1 fraction of the cell cycle in HEK293 cells, **676**
- Lynch syndrome**, Hereditary non-polyposis colorectal cancer: current risks of colorectal cancer largely overestimated, **335**
- Madelung deformity**, Prevalence of mutations in the short stature homeobox containing gene (*SHOX*) in Madelung deformity of childhood, **758**
- magnetic resonance imaging**, Corpus callosum hypoplasia and associated brain anomalies in Nijmegen breakage syndrome, **e25**
- male breast cancer**, Novel *BRCA2* mutation in a Polish family with hamartoma and two male breast cancers, **e35**
- male infertility**, A new case of Yq microdeletion transmitted from a normal father to two infertile sons, **e27**
- malformation**, Long term health and neurodevelopment in children exposed to antiepileptic drugs before birth, **251**
- malignant peripheral nerve sheath tumours**, Malignant peripheral nerve sheath tumours in neurofibromatosis 1, **311**
- mandibulofacial dysostosis**, Novel autosomal dominant mandibulofacial dysostosis with ptosis: clinical description and exclusion of *TCOF1*, **484**
- MAPH**, High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH), **790**
- mapping**, A locus for isolated cataract on human Xp, **105**
- Marfan syndrome**, Sensitivity of conformation sensitive gel electrophoresis in detecting mutations in Marfan syndrome and related conditions, **34**
- Twelve novel *FBN1* mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of *FBN1* mutation testing in clinical practice, **589**
- Marinesco-Sjögren syndrome**, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjögren syndrome, **838**
- maternal serum triple test**, Physical and psychomotor development of 1799 children born after second trimester amniocentesis for maternal serum positive triple test screening and normal prenatal karyotype, **e75**
- MCAMR**, Molecular screening for Smith-Magenis syndrome among patients with mental retardation of unknown cause, **e59**
- MeCP2**, Functional characterisation of *MeCP2* mutations found in male patients with X linked mental retardation, **132**
- MDR1 mutations**, Naturally occurring mutations and functional polymorphisms in multidrug resistance 1 gene: correlation with microsatellite instability and lymphoid infiltration in colorectal cancers, **340**
- MECP2**, *MECP2* gene nucleotide changes and their pathogenicity in males: proceed with caution, **586**
- medullary cystic kidney disease 2**, Mutations of the *UMOD* gene are responsible for medullary cystic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, **882**
- MELAS**, Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, **e22**
- mental retardation**, Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR), **359**
- An aetiological study of 25 mentally retarded adults with autism, **205**
- Automated fluorescent genotyping detects 10% of cryptic subtelomeric rearrangements in idiopathic syndromic mental retardation, **266**
- Autosomal recessive primary microcephaly: an analysis of locus heterogeneity and phenotypic variation, **718**
- Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy, **722**
- Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience, **546**
- MERRF**, Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, **e22**
- mesomelic dysplasia**, Breakpoint analysis of a familial balanced translocation t(2;8)(q31;p21) associated with mesomelic dysplasia, **e34**
- methylation**, Monozygotic twins with chromosome 22q11 deletion and discordant phenotypes: updates with an epigenetic hypothesis, **e71**
- MHC polymorphisms**, Association of polymorphisms and allelic combinations in the tumour necrosis factor- $\alpha$ -complement MHC region with coronary artery disease, **46**
- microarray**, Identification of candidate lung cancer susceptibility genes in mouse using oligonucleotide arrays, **644**
- microcephaly**, Pure terminal duplication of the short arm of chromosome 19 in a boy with mild microcephaly, **e60**
- microdeletion**, A new case of Yq microdeletion transmitted from a normal father to two infertile sons, **e27**
- microphthalmia**, National study of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland: investigation of genetic aetiology, **16**
- microsatellite instability**, Naturally occurring mutations and functional polymorphisms in multidrug resistance 1 gene: correlation with microsatellite instability and lymphoid infiltration in colorectal cancers, **340**
- Screening for microsatellite instability target genes in colorectal cancers, **785**
- mild phenotype**, A rare case of a de novo dup(19q) associated with a mild phenotype, **e61**
- Exceptionally mild Angelman syndrome phenotype associated with an incomplete imprinting defect, **e51**
- mismatch repair gene**, Mismatch repair gene analysis in Catalan families with colorectal cancer, **e29**
- missense mutation**, De novo *GLI3* mutation in acrocallosal syndrome: broadening the phenotypic spectrum of *GLI3* defects and overlap with murine models, **804**
- mitochondrial DNA**, Leber hereditary optic neuropathy, **162**
- Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, **e22**
- mitochondrial tRNA<sup>Ser(UCN)</sup> gene**, Maternally inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial tRNA<sup>Ser(UCN)</sup> gene, **e82**
- MLH1**, An *MLH1* haplotype is over-represented on chromosomes carrying an HNPCC predisposing mutation in *MLH1*, **323**
- Description and functional analysis of a novel in frame mutation linked to hereditary non-polyposis colorectal cancer, **747**
- MLH1 gene**, Association of Crohn's disease and ulcerative colitis with haplotypes of the *MLH1* gene in Italian inflammatory bowel disease patients, **332**
- MLH1/MSH2**, Germline *MSH2* and *MLH1* mutational spectrum in HNPCC families from Poland and the Baltic States, **e65**
- MMR**, Description and functional analysis of a novel in frame mutation linked to hereditary non-polyposis colorectal cancer, **747**
- MMR genes**, Genetic characterisation of patients with multiple colonic polyps, **297**
- Möbius syndrome**, Mutation analysis in the candidate Möbius syndrome genes *PGT* and *GATA2* on chromosome 3 and *EGR2* on chromosome 10, **e30**
- mosaicism**, Mosaicism for duplication 12q (12q13→12q21.2) accompanied by a pericentric inversion in a dysmorphic female infant, **e72**
- Mosaicism for *FMR1* and *FMR2* deletion: a new case, **200**
- Phenotypic effects of mosaicism for a 47,XXX cell line in Turner syndrome, **217**
- motives**, What do women really want to know? Motives for attending familial breast cancer clinics, **410**
- MRXS locus**, A new *MRXS* locus maps to the X chromosome pericentromeric region: a new syndrome or narrow definition of Sutherland-Haas genetic locus?, **276**
- mtDNA**, Childhood onset mitochondrial myopathy and lactic acidosis caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene, **812**
- The frequency of mtDNA 8994 polymorphism and detection of the NARP 8993 mutation, **204**
- MTHFR**, Human *T* and risk for neural tube defects, **e14**
- MTHFR gene**, Changes in frequencies of heterozygous thermolabile 5,10-methylenetetrahydrofolate reductase gene in fetuses with neural tube defects, **366**
- MTHFR genotype**, Maternal *MTHFR* genotype contributes to the risk of non-syndromic cleft lip and palate, **368**
- mucopolysaccharidosis type IIIB**, Identification and characterisation of mutations underlying Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), **e3**



- multicolour banding**, Chromosome 2 aberrations in clinical cases characterised by high resolution multicolour banding and region specific FISH probes, **434**
- multifocal glomus tumours**, Multifocal glomus tumours of the fingers in two patients with neurofibromatosis type 1, **e45**
- multiple colorectal adenomas**, Genetic characterisation of patients with multiple colonic polyps, **297**
- multiple congenital anomaly syndromes**, Physical and psychomotor development of 1799 children born after second trimester amniocentesis for maternal serum positive triple test screening and normal prenatal karyotype, **e75**
- multiple myeloma**, Genetic variants of NHEJ DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, **900**
- multipoise analysis**, A genome wide scan for familial high myopia suggests a novel locus on chromosome 7q36, **118**
- mutation**, A single amino acid substitution (D1441Y) in the carboxyl-terminal propeptide of the  $\alpha 1(I)$  chain of type I collagen results in a lethal variant of osteogenesis imperfecta with features of dense bone diseases, **23**
- Four novel mutations in the *OFD1* (*Cxorf5*) gene in Finnish patients with oral-facial-digital syndrome 1, **292**
- Identification of mutations in the gene encoding sterol regulatory element binding protein (SREBP)-2 in hypercholesterolaemic subjects, **271**
- Molecular genetic analysis of the NF2 gene in young patients with unilateral vestibular schwannomas, **315**
- Novel homozygous mutation in the alpha subunit of the rod cGMP gated channel (*CNGA1*) in two Spanish sibs affected with autosomal recessive retinitis pigmentosa, **e66**
- Novel mutations in the  $\gamma$ -crystallin genes cause autosomal dominant congenital cataracts, **352**
- The intron 14 2140+5G>A variant in the low density lipoprotein receptor gene has no effect on plasma cholesterol levels, **e57**
- mutation 1811+1 6kbA>G**, Splice mutation 1811+1.6kbA>G causes severe cystic fibrosis with pancreatic insufficiency: report of 11 compound heterozygous and two homozygous patients, **e73**
- mutation analysis**, A late onset variant of ataxia-telangiectasia with a compound heterozygous genotype, A8030G/T481insA, **57**
- Germline *MSH2* and *MLH1* mutational spectrum in HNPCC families from Poland and the Baltic States, **e65**
- mutation carriers**, Hereditary non-polyposis colorectal cancer: current risks of colorectal cancer largely overestimated, **335**
- mutation hotspots**, Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the *HGO* gene, **e40**
- mutation screening**, Mutation screening of the *PKD1* transcript by RT-PCR, **422**
- mutational analysis**, Analysis of the *p63* gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts, **559**
- mutations**, Identification of 13 new mutations in the *ACVRL1* gene in a group of 52 unselected Italian patients affected by hereditary haemorrhagic telangiectasia, **e39**
- Identification of novel *CLN2* mutations shows Canadian specific NCL2 alleles, **822**
- myotonic dystrophy**, Reproductive counselling for women with myotonic dystrophy, **e15**
- NAG**, Identification and characterisation of mutations underlying Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), **e3**
- NARP**, The frequency of mtDNA 8994 polymorphism and detection of the NARP 8993 mutation, **204**
- NBS1 gene**, Corpus callosum hypoplasia and associated brain anomalies in Nijmegen breakage syndrome, **e25**
- neoadjuvant chemotherapy**, A significant response to neoadjuvant chemotherapy in *BRCA1/2* related breast cancer, **608**
- neonatal diabetes**, Major difference in aetiology and phenotypic abnormalities between transient and permanent neonatal diabetes, **370**
- neural tube defects**, Changes in frequencies of heterozygous thermolabile 5,10-methylenetetrahydrofolate reductase gene in fetuses with neural tube defects, **366**
- Craniofacial anomalies, cataracts, congenital heart disease, sacral neural tube defects, and growth and developmental retardation in two sisters: a new autosomal recessive MCA/MR syndrome?, **145**
- Human *T* and risk for neural tube defects, **e14**
- Inositol and folate resistant neural tube defects, **e5**
- neurodevelopment**, Long term health and neurodevelopment in children exposed to antiepileptic drugs before birth, **251**
- neuroendocrine system**, Hereditary paraganglioma targets diverse paraganglia, **617**
- neurofibromatosis 1**, Malignant peripheral nerve sheath tumours in neurofibromatosis 1, **311**
- neurofibromatosis type 1**, Multifocal glomus tumours of the fingers in two patients with neurofibromatosis type 1, **e45**
- neurofibromatosis type 2**, Molecular genetic analysis of the NF2 gene in young patients with unilateral vestibular schwannomas, **315**
- new mutation**, Unique de novo mutation of *BRCA2* in a woman with early onset breast cancer, **126**
- NF2 locus**, Constitutional de novo interstitial deletion of 8 Mb on chromosome 22q12.1-12.3 encompassing the neurofibromatosis type 2 (NF2) locus in a dysmorphic girl with severe malformations, **e6**
- Nijmegen breakage syndrome**, Corpus callosum hypoplasia and associated brain anomalies in Nijmegen breakage syndrome, **e25**
- NKX2.5**, Two novel frameshift mutations in *NKX2.5* result in novel features including visceral inversions and sinus venosus type ASD, **807**
- non-founder mutations**, Incidence of non-founder *BRCA1* and *BRCA2* mutations in high risk Ashkenazi breast and ovarian cancer families, **611**
- nonsense**, In vitro analysis of aminoglycoside therapy for the Arg120stop nonsense mutation in RP2 patients, **62**
- non-syndromic hearing impairment**, Maternally inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial tRNA<sup>Ser(UCN)</sup> gene, **e82**
- non-syndromic hearing loss**, A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter, **567**
- Noonan syndrome**, *PTPN11* mutations in LEOPARD syndrome, **571**
- null oncogene**, The null oncogene hypothesis and protection from cancer, **12**
- OCCR**, Hereditary ovarian cancer resulting from a non-ovarian cancer cluster region (OCCR) *BRCA2* mutation: is the OCCR useful clinically?, **e68**
- OFD1**, Four novel mutations in the *OFD1* (*Cxorf5*) gene in Finnish patients with oral-facial-digital syndrome 1, **292**
- Okihiro syndrome**, Okihiro syndrome and acro-renal-ocular syndrome: clinical overlap, expansion of the phenotype, and absence of *PAX2* mutations in two new families, **68**
- omphalocele**, Omphalocele in three generations with autosomal dominant transmission, **184**
- OPA1 gene**, Deletion of the *OPA1* gene in a dominant optic atrophy family: evidence that haploinsufficiency is the cause of disease, **e47**
- optic atrophy**, Deletion of the *OPA1* gene in a dominant optic atrophy family: evidence that haploinsufficiency is the cause of disease, **e47**
- Leber hereditary optic neuropathy, **162**
- optic nerve**, Leber hereditary optic neuropathy, **162**
- oral-facial-digital syndrome 1**, Four novel mutations in the *OFD1* (*Cxorf5*) gene in Finnish patients with oral-facial-digital syndrome 1, **292**
- osteoarthritis**, Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene, **634**
- osteogenesis imperfecta**, Testing for osteogenesis imperfecta in cases of suspected non-accidental injury, **382**
- osteogenesis imperfecta (OI)**, A single amino acid substitution (D1441Y) in the carboxyl-terminal propeptide of the  $\alpha 1(I)$  chain of type I collagen results in a lethal variant of osteogenesis imperfecta with features of dense bone diseases, **23**
- osteogenesis imperfecta type IV**, A variant of osteogenesis imperfecta type IV with resolving kyphomelia is caused by a novel *COL1A2* mutation, **128**
- osteolysis**, Familial expansile osteolysis in a large Spanish kindred resulting from an insertion mutation in the *TNFRSF11A* gene, **e67**
- otoferlin**, Q829X, a novel mutation in the gene encoding otoferlin (*OTOF*), is frequently found in Spanish patients with prelingual non-syndromic hearing loss, **502**
- OTOF**, Q829X, a novel mutation in the gene encoding otoferlin (*OTOF*), is frequently found in Spanish patients with prelingual non-syndromic hearing loss, **502**
- otopalatodigital syndrome type I**, Refined mapping of the gene for otopalatodigital syndrome type I, **e7**
- otosclerosis**, Linkage of otosclerosis to a third locus (*OTSC3*) on human chromosome 6p21.3-22.3, **473**
- ovarian cancer**, Concerns of women presenting to a comprehensive cancer centre for genetic cancer risk assessment, **526**
- Genetic counselling for familial breast and ovarian cancer in Ontario, **695**
- Incidence of non-founder *BRCA1* and *BRCA2* mutations in high risk Ashkenazi breast and ovarian cancer families, **611**
- overgrowth**, Dysmorphism, variable overgrowth, normal bone age, and severe developmental delay: a "Sotos-like" syndrome?, **148**
- P glycoprotein function**, Naturally occurring mutations and functional polymorphisms in multidrug resistance 1 gene: correlation with microsatellite instability and lymphoid infiltration in colorectal cancers, **340**

- p63**, Analysis of the *p63* gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts, **559**
- p63 gene**, The *p63* gene in EEC and other syndromes, **377**
- pancreatic secretory trypsin inhibitor gene**, Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis, **347**
- papillary thyroid carcinoma**, Specific haplotypes of the *RET* proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma, **260**
- paraganglia**, Hereditary paraganglioma targets diverse paraganglia, **617**
- paraganglioma**, Hereditary paraganglioma targets diverse paraganglia, **617**
- SDHB* mutation analysis in familial and sporadic pheochromocytoma identifies a novel mutation, **e64**
- parental attitudes**, Parental attitude towards genetic testing for familial hypercholesterolaemia in children, **e49**
- parkin** locus Linkage stratification and mutation analysis at the *parkin* locus identifies mutation positive Parkinson's disease families, **489**
- Parkinson's disease**, Association of an interleukin 1B gene polymorphism (–511) with Parkinson's disease in Finnish patients, **400**
- Linkage stratification and mutation analysis at the *parkin* locus identifies mutation positive Parkinson's disease families, **489**
- partial deletion 4p**, Partial deletion of 4p and 4q in a fetus with ring chromosome 4: phenotype and molecular mapping of the breakpoints, **e23**
- partial deletion 4q**, Partial deletion of 4p and 4q in a fetus with ring chromosome 4: phenotype and molecular mapping of the breakpoints, **e23**
- partial monosomy**, Comparative genomic hybridisation shows a partial de novo deletion 16p11.2 in a neonate with multiple congenital malformations, **e24**
- partial trisomy 9p**, Partial trisomy 9p12p21.3 with a normal phenotype, **141**
- patient satisfaction**, Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes about genetic counselling among probands referred to three genetic clinics, **e84**
- PAX3 gene**, A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2:8 translocation, **391**
- PCR**, A breast cancer family from Spain with germline mutations in both the *BRCA1* and *BRCA2* genes, **e44**
- Absence of 22q11 deletions in 211 patients with developmental delay analysed using PCR, **e18**
- perceptive deafness**, Corneal dystrophy and perceptive deafness (Harboyan syndrome): *CDPD1* maps to 20p13, **110**
- pericentric inversion**, Familial pericentric inversion of chromosome 5 in a family with benign neonatal convulsions, **214**
- pericentromeric region**, Organisation of the pericentromeric region of chromosome 15: at least four partial gene copies are amplified in patients with a proximal duplication of 15q, **170**
- periodontitis**, Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis, **844**
- pheochromocytoma**, *SDHB* mutation analysis in familial and sporadic pheochromocytoma identifies a novel mutation, **e64**
- phenotype of survivors**, The phenotype of survivors of campomelic dysplasia, **597**
- phenotypic sex**, A comparative study between infertile males and patients with Turner syndrome to determine the influence of sex chromosome mosaicism and the breakpoints of structurally abnormal Y chromosomes on phenotypic sex, **e80**
- phosphomannose isomerase**, Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib, **849**
- physical and psychomotor development**, Physical and psychomotor development of 1799 children born after second trimester amniocentesis for maternal serum positive triple test screening and normal prenatal karyotype, **e75**
- pituitary tumours**, *PRKAR1A*, one of the Carney complex genes, and its locus (17q22-24) are rarely altered in pituitary tumours outside the Carney complex, **e78**
- PKD1**, Mutation screening of the *PKD1* transcript by RT-PCR, **422**
- Poland/Baltic States**, Germline *MSH2* and *MLH1* mutational spectrum in HNPCC families from Poland and the Baltic States, **e65**
- polled intersex syndrome (PIS)**, *FOXL2* mutation screening in a large panel of POF patients and XX males, **e43**
- polyendocrinopathy**, Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome, **537**
- polymorphism**, An *MLH1* haplotype is over-represented on chromosomes carrying an HNPCC predisposing mutation in *MLH1*, **323**
- Identification of mutations in the gene encoding sterol regulatory element binding protein (SREBP)-2 in hypercholesterolaemic subjects, **271**
- Novel mutations in the  $\gamma$ -crystallin genes cause autosomal dominant congenital cataracts, **352**
- Organisation of the pericentromeric region of chromosome 15: at least four partial gene copies are amplified in patients with a proximal duplication of 15q, **170**
- Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis, **844**
- polymorphisms**, Specific haplotypes of the *RET* proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma, **260**
- polypyrimidine tract**, A T to C mutation in the polypyrimidine tract of the exon 9 splicing site of the *RB1* gene responsible for low penetrance hereditary retinoblastoma, **e21**
- pooled analysis**, Genes other than *BRCA1* and *BRCA2* involved in breast cancer susceptibility, **225**
- PPAR $\alpha$  gene**, A Val227Ala polymorphism in the peroxisome proliferator activated receptor  $\alpha$  (*PPAR $\alpha$* ) gene is associated with variations in serum lipid levels, **189**
- Prader-Willi syndrome**, Prader-Willi syndrome and a deletion/duplication within the 15q11-q13 region, **202**
- Relationship between clinical and genetic diagnosis of Prader-Willi syndrome, **926**
- Prader-Willi-like phenotype**, Deletion of the *SIM1* gene (6q16.2) in a patient with a Prader-Willi-like phenotype, **594**
- predictive testing**, Predictive testing in the context of pregnancy: experience in Huntington's disease and autosomal dominant cerebellar ataxia, **522**
- predictive value**, Calculating predictive values for the large repeat alleles at the SCA8 locus in patients with ataxia, **935**
- predisposition**, Genetic variants of NHEJ DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, **900**
- predisposition rearrangements**, Bar code screening on combed DNA for large rearrangements of the *BRCA1* and *BRCA2* genes in French breast cancer families, **817**
- pre-eclampsia**, Paternal contribution to the risk for pre-eclampsia, **44**
- preimplantation genetic diagnosis**, Preimplantation genetic diagnosis in clinical practice, **6**
- prelingual non-syndromic hearing loss**, Q829X, a novel mutation in the gene encoding otoferlin (*OTOF*), is frequently found in Spanish patients with prelingual non-syndromic hearing loss, **502**
- premature ovarian failure (POF)**, *FOXL2* mutation screening in a large panel of POF patients and XX males, **e43**
- premature ovarian failure**, Evolution and expression of *FOXL2*, **916**
- prenatal diagnosis**, Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss, **449**
- Genetic testing and genetic counselling in hypertrophic cardiomyopathy: the French experience, **741**
- Preimplantation genetic diagnosis in clinical practice, **6**
- presymptomatic diagnosis**, Genetic testing and genetic counselling in hypertrophic cardiomyopathy: the French experience, **741**
- presymptomatic gene carriers**, Behavioural complaints in participants who underwent predictive testing for Huntington's disease, **857**
- primary infertility**, Klinefelter-like phenotype and primary infertility in a male with a paracentric Xq inversion, **e28**
- primary lymphoedema**, Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with *FOXC2* mutations or linkage to 16q24, **478**
- proactive genetic counselling**, Comparison of genetic services with and without genetic registers: access and attitudes to genetic counselling services among relatives of genetic clinic patients, **e85**
- Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes about genetic counselling among probands referred to three genetic clinics, **e84**
- probable silent polymorphism**, Frequency of mutations in the early growth response 2 gene associated with peripheral demyelinating neuropathies, **e81**
- procollagen**, A single amino acid substitution (D1441Y) in the carboxyl-terminal propeptide of the  $\alpha 1(I)$  chain of type I collagen results in a lethal variant of osteogenesis imperfecta with features of dense bone diseases, **23**
- promoter –269T>G**, The intron 14 2140+5G>A variant in the low density lipoprotein receptor gene has no effect on plasma cholesterol levels, **e57**
- prospective**, Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience, **546**
- protein kinase A**, *PRKAR1A*, one of the Carney complex genes, and its locus (17q22-24) are rarely altered in pituitary tumours outside the Carney complex, **e78**
- Proteus syndrome**, Germline mutation of the tumour suppressor *PTEN* in Proteus syndrome, **937**



- proto-oncogenes**, The null oncogene hypothesis and protection from cancer, **12**
- PSEN1 mutation**, Spastic paraparesis and atypical dementia caused by *PSEN1* mutation (P264L), responsible for Alzheimer's disease, **e2**
- psoriasis**, Streptococcal infection distinguishes different types of psoriasis, **767**
- psychological well being**, The use of audiotapes in consultations with women from high risk breast cancer families: a randomised trial, **697**
- psychomotor delay**, Pure terminal duplication of the short arm of chromosome 19 in a boy with mild microcephaly, **e60**
- PTEN**, Germline mutation of the tumour suppressor *PTEN* in Proteus syndrome, **937**
- ptosis**, Novel autosomal dominant mandibulofacial dysostosis with ptosis: clinical description and exclusion of *TCOF1*, **484**
- PTPN11 mutations**, *PTPN11* mutations in LEOPARD syndrome, **571**
- pure 1q trisomy syndrome**, Silver-Russell phenotype in a patient with pure trisomy 1q32.1-q42.1: further delineation of the pure 1q trisomy syndrome, **582**
- Q829X mutation**, Q829X, a novel mutation in the gene encoding otoferlin (*OTOF*), is frequently found in Spanish patients with prelingual non-syndromic hearing loss, **502**
- questionnaire**, Continuous medical education approaches for clinical genetics: a postal survey of general practitioners, **e69**
- radiological findings**, Dysosteosclerosis: a report of three new cases and evolution of the radiological findings, **603**
- real time RT-PCR**, Identification of cryptic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, **286**
- rearrangements**, Detection of large rearrangements of exons 13 and 22 in the *BRCA1* gene in German families, **e36**
- renal cancer**, Exclusion of *PTEN*, *CTNNB1*, and *PTCH* as candidate genes for Birt-Hogg-Dube syndrome, **e10**
- renal cysts**, Report of a new case of "genitopatellar" syndrome which challenges the importance of absent patellae as a defining feature, **933**
- renal tubular acidosis**, Novel *ATP6V1B1* and *ATP6V0A4* mutations in autosomal recessive distal renal tubular acidosis with new evidence for hearing loss, **796**
- reproductive counselling**, Reproductive counselling for women with myotonic dystrophy, **e15**
- RET proto-oncogene**, Specific haplotypes of the *RET* proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma, **260**
- retinal detachment**, Dilated cardiomyopathy, sudden cardiac death, hypoplastic discs, and retinal detachment: a new autosomal dominant syndrome, **221**
- retinitis pigmentosa**, Distinctive audiometric features between USH2A and USH2B subtypes of Usher syndrome, **281**  
Dominant X linked retinitis pigmentosa is frequently accounted for by truncating mutations in exon ORF15 of the *RPGR* gene, **284**  
In vitro analysis of aminoglycoside therapy for the Arg120stop nonsense mutation in *RP2* patients, **62**
- retinoblastoma**, A T to C mutation in the polypyrimidine tract of the exon 9 splicing site of the *RB1* gene responsible for low penetrance hereditary retinoblastoma, **e21**
- Rett syndrome**, Functional characterisation of *MeCP2* mutations found in male patients with X linked mental retardation, **132**  
*MECP2* gene nucleotide changes and their pathogenicity in males: proceed with caution, **586**
- ring chromosome 4**, Partial deletion of 4p and 4q in a fetus with ring chromosome 4: phenotype and molecular mapping of the breakpoints, **e23**
- ring chromosome 22**, Molecular characterisation of a ring chromosome 22 in a patient with severe language delay: a contribution to the refinement of the subtelomeric 22q deletion syndrome, **e17**
- RNA splicing**, Identification of cryptic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, **286**
- Robinow syndrome**, Robinow syndrome, **305**
- ROR2 gene**, Robinow syndrome, **305**
- RPGR gene**, Dominant X linked retinitis pigmentosa is frequently accounted for by truncating mutations in exon ORF15 of the *RPGR* gene, **284**
- RSK2**, Coffin-Lowry phenotype in a patient with a complex chromosome rearrangement, **e41**  
Coffin-Lowry syndrome: clinical and molecular features, **705**
- Rubinstein-like syndrome**, Molecular studies in 10 cases of Rubinstein-Taybi syndrome, including a mild variant showing a missense mutation in codon 1175 of *CREBBP*, **496**
- Rubinstein-Taybi syndrome**, Molecular analysis of the *CBP* gene in 60 patients with Rubinstein-Taybi syndrome, **415**  
Molecular studies in 10 cases of Rubinstein-Taybi syndrome, including a mild variant showing a missense mutation in codon 1175 of *CREBBP*, **496**
- Russell-Silver syndrome**, Silver-Russell phenotype in a patient with pure trisomy 1q32.1-q42.1: further delineation of the pure 1q trisomy syndrome, **582**
- Sanfilippo syndrome type B**, Identification and characterisation of mutations underlying Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), **e3**
- sarcoma**, Low rate of *TP53* germline mutations in breast cancer/sarcoma families not fulfilling classical criteria for Li-Fraumeni syndrome, **941**
- SCA8**, Calculating predictive values for the large repeat alleles at the SCA8 locus in patients with ataxia, **935**
- screening**, Molecular screening for Smith-Magenis syndrome among patients with mental retardation of unknown cause, **e59**  
Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience, **546**
- SDHB**, *SDHB* mutation analysis in familial and sporadic pheochromocytoma identifies a novel mutation, **e64**
- second trimester amniocentesis**, Physical and psychomotor development of 1799 children born after second trimester amniocentesis for maternal serum positive triple test screening and normal prenatal karyotype, **e75**
- segregation analysis**, Branchio-oculo-facial syndrome and branchio-otic/branchio-oto-renal syndromes are distinct entities, **71**
- seipin**, Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy, **722**
- seizures**, Towards a suggestive facial dysmorphism in adenylosuccinate lyase deficiency?, **440**
- sequence analysis**, Genetic and functional analysis of the von Hippel-Lindau (VHL) tumour suppressor gene promoter, **463**
- serum lipid levels**, A Val227Ala polymorphism in the peroxisome proliferator activated receptor  $\alpha$  (*PPAR $\alpha$* ) gene is associated with variations in serum lipid levels, **189**
- sex chromosome mosaicism**, A comparative study between infertile males and patients with Turner syndrome to determine the influence of sex chromosome mosaicism and the breakpoints of structurally abnormal Y chromosomes on phenotypic sex, **e80**
- sex ratio**, Hirschsprung disease and *L1CAM*: is the disturbed sex ratio caused by *L1CAM* mutations?, **e11**
- SHOX**, Prevalence of mutations in the short stature homeobox containing gene (*SHOX*) in Madelung deformity of childhood, **758**
- SHOX point mutations**, *SHOX* point mutations and deletions in Leri-Weill dyschondrosteosis, **e33**
- Silver-Russell syndrome**, Characterisation of the growth regulating gene *IMP3*, a candidate for Silver-Russell syndrome, **575**  
Investigation of the *GRB2*, *GRB7*, and *CSH1* genes as candidates for the Silver-Russell syndrome (SRS) on chromosome 17q, **e13**
- SIM1 gene**, Deletion of the *SIM1* gene (6q16.2) in a patient with a Prader-Willi-like phenotype, **594**
- sinus venosus type ASD**, Two novel frameshift mutations in *NKX2.5* result in novel features including visceral inversions and sinus venosus type ASD, **807**
- skewed X inactivation**, High frequency of skewed X inactivation in young breast cancer patients, **30**
- skin**, Molecular changes in skin predict predisposition to breast cancer, **e1**
- skin tumours**, Clinical and genetic studies of Birt-Hogg-Dubé syndrome, **906**
- SMARCA1**, Longevity in Schimke immuno-osseous dysplasia, **922**
- Smith-Lemli-Opitz syndrome**, Smith-Lemli-Opitz syndrome: carrier frequency and spectrum of *DHCR7* mutations in Canada, **e31**
- Smith-Magenis syndrome**, Molecular screening for Smith-Magenis syndrome among patients with mental retardation of unknown cause, **e59**
- SNF2 protein**, Longevity in Schimke immuno-osseous dysplasia, **922**
- somatic mosaicism**, Molecular genetic analysis of the NF2 gene in young patients with unilateral vestibular schwannomas, **315**
- sorting nexin 3**, Sorting nexin 3 (*SNX3*) is disrupted in a patient with a translocation t(6;13)(q21;q12) and microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP) phenotype, **893**
- SOST**, Identification of a 52 kb deletion downstream of the *SOST* gene in patients with van Buchem disease, **91**
- Sotos syndrome**, Dysmorphism, variable overgrowth, normal bone age, and severe developmental delay: a "Sotos-like" syndrome?, **148**
- spastic paraparesis**, Spastic paraparesis and atypical dementia caused by *PSEN1* mutation (P264L), responsible for Alzheimer's disease, **e2**
- spastic paraplegia**, Genetic mapping of a susceptibility locus for disc herniation and spastic paraplegia on 6q23.3-q24.1, **387**
- spastin gene mutation**, *Spastin* gene mutation in Japanese with hereditary spastic paraplegia, **e46**
- spermatozoa**, Cytogenetic analysis of spermatozoa from males aged between 47 and 71 years, **e63**
- SPG4**, Autosomal dominant (AD) pure spastic paraplegia (HSP) linked to locus SPG4 affects almost exclusively males in a large pedigree, **e77**

- Spastin* gene mutation in Japanese with hereditary spastic paraplegia, **e46**
- spinocerebellar ataxia**, Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, **e22**
- spinocerebellar ataxia type 2**, Comorbid *VHL* and *SCA2* mutations in a large kindred: confounding diagnosis of neurological dysfunction caused by CNS *VHL* vascular tumours versus *SCA2* atrophic neurodegeneration, **e37**
- spondyloepimetaphyseal dysplasia with multiple dislocations (Hall type)**, Spondyloepimetaphyseal dysplasia with multiple dislocations (Hall type): three further cases and evidence of autosomal dominant inheritance, **666**
- SREBP**, Identification of mutations in the gene encoding sterol regulatory element binding protein (SREBP)-2 in hypercholesterolaemic subjects, **271**
- streptococcal infection**, Streptococcal infection distinguishes different types of psoriasis, **767**
- subcortical cysts**, A common ancestral haplotype in carrier chromosomes from different ethnic backgrounds in vacuolating megalencephalic leucoencephalopathy with subcortical cysts, **54**
- subtelomeric**, Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience, **546**
- subtelomeric 22q deletion**, Molecular characterisation of a ring chromosome 22 in a patient with severe language delay: a contribution to the refinement of the subtelomeric 22q deletion syndrome, **e17**
- subtelomeric deletions**, An aetiological study of 25 mentally retarded adults with autism, **205**
- subtelomeric DNA**, High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH), **790**
- subtelomeric rearrangements**, Automated fluorescent genotyping detects 10% of cryptic subtelomeric rearrangements in idiopathic syndromic mental retardation, **266**
- Subtelomeric rearrangements detected by FISH in three of 33 families with idiopathic mental retardation and minor physical anomalies, **e53**
- succinate dehydrogenase**, Prevalence of *SDHB*, *SDHC*, and *SDHD* germline mutations in clinic patients with head and neck paragangliomas, **178**
- sudden cardiac death**, Dilated cardiomyopathy, sudden cardiac death, hypoplastic discs, and retinal detachment: a new autosomal dominant syndrome, **221**
- sudden death**, SNP S1103Y in the cardiac sodium channel gene *SCN5A* is associated with cardiac arrhythmias and sudden death in a white family, **913**
- supernumerary marker chromosome (15)**, A patient with a supernumerary marker chromosome (15), Angelman syndrome, and uniparental disomy resulting from paternal meiosis II non-disjunction, **e9**
- supernumerary marker chromosome (SMC)**, Supernumerary marker chromosomes (SMC) and uniparental disomy (UPD): coincidence or consequence?, **775**
- suppressor**, Expression of wild type and mutant *TSC2*, but not *TSC1*, causes an increase in the G1 fraction of the cell cycle in HEK293 cells, **676**
- supravalvular aortic stenosis**, Cardiovascular manifestations in 75 patients with Williams syndrome, **554**
- survival**, Survival in trisomy 13 and trisomy 18 cases ascertained from population based registers, **e54**
- Sutherland-Haas syndrome**, A new MRXS locus maps to the X chromosome pericentromeric region: a new syndrome or narrow definition of Sutherland-Haas genetic locus?, **276**
- syndrome**, Thrombocytopenia-absent radius syndrome: a clinical genetic study, **876**
- syndromes**, Familial adult renal neoplasia, **1**
- synpolydactyly**, The fibulin-1 gene (*FBLN1*) is disrupted in a t(12;22) associated with a complex type of synpolydactyly, **98**
- synpolydactyly (SPD)**, Severe digital abnormalities in a patient heterozygous for both a novel missense mutation in *HOXD13* and a polyalanine tract expansion in *HOXA13*, **852**
- T cell immunodeficiency**, Longevity in Schimke immuno-osseous dysplasia, **922**
- T gene**, Human *T* and risk for neural tube defects, **e14**
- T lymphoblastic lymphoma**, Hereditary duplication of proximal chromosome 1q (q11q22) in a patient with T lymphoblastic lymphoma/leukaemia: a family study using G banding and comparative genomic hybridisation, **e79**
- talipes equinovarus**, *DTDST* mutations are not a frequent cause of idiopathic talipes equinovarus (club foot), **e20**
- TCOF1**, Screening of *TCOF1* in patients from different populations: confirmation of mutational hot spots and identification of a novel missense mutation that suggests an important functional domain in the protein treacle, **493**
- telomere**, "Molecular rulers" for calibrating phenotypic effects of telomere imbalance, **734**
- teratogenic effects**, Antiepileptic drug therapy during pregnancy: the neurologist's perspective, **248**
- Assessing epidemiological evidence for the teratogenic effects of anticonvulsant medications, **243**
- The teratogenicity of anticonvulsant drugs: a progress report, **245**
- test results**, Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test results, **e26**
- tetralogy of Fallot**, A novel atypical 22q11.2 distal deletion in father and son, **e62**
- therapy**, In vitro analysis of aminoglycoside therapy for the Arg120stop nonsense mutation in RP2 patients, **62**
- three generations**, Omphalocele in three generations with autosomal dominant transmission, **184**
- thrombocytopenia**, Thrombocytopenia-absent radius syndrome: a clinical genetic study, **876**
- TM4SF2 gene**, A novel 2 bp deletion in the *TM4SF2* gene is associated with MRX58, **430**
- TNDM**, Transient neonatal diabetes, a disorder of imprinting, **872**
- TNFRSF11A**, Familial expansile osteolysis in a large Spanish kindred resulting from an insertion mutation in the *TNFRSF11A* gene, **e67**
- TP53**, Low rate of *TP53* germline mutations in breast cancer/sarcoma families not fulfilling classical criteria for Li-Fraumeni syndrome, **941**
- training**, Management of women with a family history of breast cancer in the North West Region of England: training for implementing a vision of the future, **531**
- transient neonatal diabetes**, Transient neonatal diabetes, a disorder of imprinting, **872**
- translocation**, Abnormal expression of the *KLF8* (*ZNF741*) gene in a female patient with an X;autosome translocation t(X;21)(p11.2;q22.3) and non-syndromic mental retardation, **113**
- Sorting nexin 3 (*SNX3*) is disrupted in a patient with a translocation t(6;13)(q21;q12) and microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP) phenotype, **893**
- translocation 2q35**, A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2;8 translocation, **391**
- transmission disequilibrium test**, Association between markers in chromosomal region 17q23 and young onset hypertension: a TDT study, **42**
- Treacher Collins syndrome**, Screening of *TCOF1* in patients from different populations: confirmation of mutational hot spots and identification of a novel missense mutation that suggests an important functional domain in the protein treacle, **493**
- treacle protein**, Screening of *TCOF1* in patients from different populations: confirmation of mutational hot spots and identification of a novel missense mutation that suggests an important functional domain in the protein treacle, **493**
- trisomy 13**, Survival in trisomy 13 and trisomy 18 cases ascertained from population based registers, **e54**
- trisomy 18**, Survival in trisomy 13 and trisomy 18 cases ascertained from population based registers, **e54**
- trisomy 19p**, Pure terminal duplication of the short arm of chromosome 19 in a boy with mild microcephaly, **e60**
- tropical calcific pancreatitis**, Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis, **347**
- truncating mutation**, Heterozygous truncating mutation in the human homeobox gene *GSH2* has no discernable phenotypic effect, **686**
- TSC2-PKD1 contiguous gene syndrome**, Acrofacial dysostosis in a patient with the *TSC2-PKD1* contiguous gene syndrome, **136**
- tuberous sclerosis complex**, Expression of wild type and mutant *TSC2*, but not *TSC1*, causes an increase in the G1 fraction of the cell cycle in HEK293 cells, **676**
- tumour necrosis factor- $\alpha$** , Association of polymorphisms and allelic combinations in the tumour necrosis factor- $\alpha$ -complement MHC region with coronary artery disease, **46**
- tumour suppressor gene**, *PRKAR1A*, one of the Carney complex genes, and its locus (17q22-24) are rarely altered in pituitary tumours outside the Carney complex, **e78**
- tumour suppressor genes**, The null oncogene hypothesis and protection from cancer, **12**
- Tunisia**, CYS127S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, **e74**
- Turner syndrome**, A comparative study between infertile males and patients with Turner syndrome to determine the influence of sex chromosome mosaicism and the breakpoints of structurally abnormal Y chromosomes on phenotypic sex, **e80**
- Phenotypic effects of mosaicism for a 47,XXX cell line in Turner syndrome, **217**
- type I collagen**, Testing for osteogenesis imperfecta in cases of suspected non-accidental injury, **382**
- ulcerative colitis**, Association of Crohn's disease and ulcerative colitis with haplotypes of the *MLH1* gene in Italian inflammatory bowel disease patients, **332**



- ultrasonography**, Prenatal detection of cystic fibrosis by ultrasonography: a retrospective study of more than 346 000 pregnancies, **443**
- unbalanced (Y;1) translocation**, Transmission of an unbalanced (Y;1) translocation in Brittany, France, **e52**
- Unified Huntington's Disease Rating Scale (UHDRA)**, Behavioural complaints in participants who underwent predictive testing for Huntington's disease, **857**
- uniparental disomies**, Automated fluorescent genotyping detects 10% of cryptic subtelomeric rearrangements in idiopathic syndromic mental retardation, **266**
- uniparental disomy**, A patient with a supernumerary marker chromosome (15), Angelman syndrome, and uniparental disomy resulting from paternal meiosis II non-disjunction, **e9**  
Maternal uniparental disomy 12 in a healthy girl with a 47,XX,+der(12):(p11→q11:)/46,XX karyotype, **519**  
Severe phenotype in Angelman syndrome resulting from paternal isochromosome 15, **e4**
- uniparental disomy (UPD)**, Supernumerary marker chromosomes (SMC) and uniparental disomy (UPD): coincidence or consequence?, **775**
- urogenital anomalies**, Correlation between a specific Wilms tumour suppressor gene (*WT1*) mutation and the histological findings in Wilms tumour (WT), **e83**
- uromodulin**, Mutations of the *UMOD* gene are responsible for medullary cystic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, **882**
- USH2A and USH2B subtypes**, Distinctive audiometric features between USH2A and USH2B subtypes of Usher syndrome, **281**
- Usher syndrome**, Distinctive audiometric features between USH2A and USH2B subtypes of Usher syndrome, **281**
- vacuolating megalencephalic leucoencephalopathy**, A common ancestral haplotype in carrier chromosomes from different ethnic backgrounds in vacuolating megalencephalic leucoencephalopathy with subcortical cysts, **54**
- Val227Ala polymorphism**, A Val227Ala polymorphism in the peroxisome proliferator activated receptor  $\alpha$  (*PPAR $\alpha$* ) gene is associated with variations in serum lipid levels, **189**
- van Buchem disease**, Identification of a 52 kb deletion downstream of the *SOST* gene in patients with van Buchem disease, **91**
- varicose veins**, Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with *FOXC2* mutations or linkage to 16q24, **478**
- vestibular schwannoma**, Molecular genetic analysis of the NF2 gene in young patients with unilateral vestibular schwannomas, **315**
- VHL gene**, Germline mutations in the von Hippel-Lindau (VHL) gene in patients from Poland: disease presentation in patients with deletions of the entire *VHL* gene, **e38**
- VHL tumour suppressor gene promoter**, Genetic and functional analysis of the von Hippel-Lindau (VHL) tumour suppressor gene promoter, **463**
- visceral inversus**, Two novel frameshift mutations in *NKX2.5* result in novel features including visceral inversus and sinus venosus type ASD, **807**
- vitreoretinopathy**, Vitreoretinopathy with phalangeal epiphyseal dysplasia, a type II collagenopathy resulting from a novel mutation in the C-propeptide region of the molecule, **661**
- von Hippel-Lindau disease**, Comorbid *VHL* and *SCA2* mutations in a large kindred: confounding diagnosis of neurological dysfunction caused by CNS VHL vascular tumours versus *SCA2* atrophic neurodegeneration, **e37**
- Genetic and functional analysis of the von Hippel-Lindau (VHL) tumour suppressor gene promoter, **463**
- Germline mutations in the von Hippel-Lindau (VHL) gene in patients from Poland: disease presentation in patients with deletions of the entire *VHL* gene, **e38**
- W846X mutation**, Relationship between genotype and phenotype for the *CFTR* gene W846X mutation, **e32**
- Williams syndrome**, Cardiovascular manifestations in 75 patients with Williams syndrome, **554**
- Wilms tumour**, Correlation between a specific Wilms tumour suppressor gene (*WT1*) mutation and the histological findings in Wilms tumour (WT), **e83**
- Wnt pathway**, Genetic characterisation of patients with multiple colonic polyps, **297**
- WT1**, A necropsy case of Denys-Drash syndrome with a *WT1* mutation in exon 7, **e48**  
Correlation between a specific Wilms tumour suppressor gene (*WT1*) mutation and the histological findings in Wilms tumour (WT), **e83**
- X chromosome**, A locus for isolated cataract on human Xp, **105**  
High frequency of skewed X inactivation in young breast cancer patients, **30**
- X chromosome inversion**, Klinefelter-like phenotype and primary infertility in a male with a paracentric Xq inversion, **e28**
- X linked Charcot-Marie-Tooth neuropathy**, Allelic variants in the 5' non-coding region of the connexin32 gene: possible pitfalls in the diagnosis of X linked Charcot-Marie-Tooth neuropathy (CMTX), **e58**
- X linked mental retardation**, A new MRXS locus maps to the X chromosome pericentromeric region: a new syndrome or narrow definition of Sutherland-Haas genetic locus?, **276**  
A novel 2 bp deletion in the *TM4SF2* gene is associated with MRXS, **430**  
Abnormal expression of the *KLF8* (*ZNF741*) gene in a female patient with an X;autosome translocation t(X;21)(p11.2;q22.3) and non-syndromic mental retardation, **113**  
Functional characterisation of *MeCP2* mutations found in male patients with X linked mental retardation, **132**
- Xq22.3**, Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR), **359**
- Xq27-q28**, Refined mapping of the gene for otopalatodigital syndrome type I, **e7**
- XX sex reversal**, *FOXL2* mutation screening in a large panel of POF patients and XX males, **e43**
- Y**, Transmission of an unbalanced (Y;1) translocation in Brittany, France, **e52**
- Y chromosome**, A new case of Yq microdeletion transmitted from a normal father to two infertile sons, **e27**  
Localisation of the Y chromosome stature gene to a 700 kb interval in close proximity to the centromere, **507**
- Y chromosome breakpoint**, A comparative study between infertile males and patients with Turner syndrome to determine the influence of sex chromosome mosaicism and the breakpoints of structurally abnormal Y chromosomes on phenotypic sex, **e80**
- young onset**, Association between markers in chromosomal region 17q23 and young onset hypertension: a TDT study, **42**