Subject index

1 translocation, Transmission of an unbalanced (Y;1) translocation in Britanny, France, e32
10p13-14 deletion, Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic congenital heart defects, e16
11q23, Prevalence of SDHB, SDHC, and SDHD germline mutations in clinic patients with head and neck parangangiomas, 178
15q11-13 deletions for Willy syndrome and a deletion/duplication within the 15q11-13 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 (Harbouyan syndrome); CDPD1 maps to 20p13, 114
22q11 deletions, A novel atypical 22q11.2 deletion in father and son, e62
22q11 deletion syndrome, A novel atypical 22q11.2 deletion in father and son, e62
22q11 deletion, Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic congenital 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, ectodactyly, prognathism (MMEP) phenotype, 893
7510T>C mutation, Methylated inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial RNA(gene) gene, e62
7-dehydrocholesterol, Smith-Lemli-Opitz syndrome: carrier frequency and spectrum of DHC97 mutations in Canada, e31
8994 polymorphism, The frequency of mtDNA 8994 polymorphism in early newborns and the 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 2q53 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 and 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
acral callosal syndrome, De novo GLI3 mutation in acral callosal syndrome: broadening the phenotypic spectrum of GLI defects and overlap with murine models, 804
acrofacial dysostosis, Acrofacial dysostosis in a patient with the TSC2-PTEN contiguous gene syndrome, 136
acro-renal-ocular syndrome, Okhirio syndrome and acro-renal-ocular syndrome: clinical overlap, expansion of the phenotype, and absence of PAX2 mutations in two new families, 68
ACVR1L1 gene, Identification of 13 new mutations in the ACVR1L1 gene in a group of 52 unselected Italian patients affected by hereditary haemorrhagic telangiectasia, e39
adrenosynaptic lysine deficiency, Towards a suggestive facial dysmorphism in adrenosynaptic lysine 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 spondyloepiphysial 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, discontinuation of the contiguous gene deletion syndrome in Xq22.3 (ATS-M), 399
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, Aminoglycoside therapy for the Arg120stop nonsense mutation in RP2 patients, 62
aneuploidy, Cytogenetic analysis of spermatozoa from males aged 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, e27
apollipoprotein E, Apolipoprotein E4 is only a weak predictor of dementia and cognitive decline in the general population, 639
arthromelia, SNP 5103XV 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 (ASPA) 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 SCAB locus in patients with ataxia, 935
Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuroopharyngeal syndrome (CCFNS) and Marinesco-Sjogren 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 2q53 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
autocrine, 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, Spondyloepiphysial dysplasia with multiple dislocations (Hall type); three further cases and evidence of autosomal dominant inheritance, 668
autosomal dominant polycystic kidney disease, mutation of the PKD1 transcript and PCKR, 422
Omphalocoele in three generations with autosomal dominant transmission, 184
autosomal recessive inheritance, Infante 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, 74
autosomal recessive retinitis pigmentosa, Novel homozygous mutation in the alpha subunit of the rod cGMP gated channel (CNAG41) in two Spanish sisters affected with autosomal recessive retinitis pigmentosa, e66
autosome translocations, Transmission of an unbalanced (Y;1) translocation in Britain, France, e55
autogyozing mapping, Mapping of a novel locus for achromatop sia (ACHMM4) to 1p and identification of a germline mutation in the alpha subunit of cone transducin (GNAT2), e56
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-Dube syndrome, e106
Birt-Hogg-Dube syndrome, Exclusion of PTEN, CTNNB1, and BIRT-HOGG-DUBÉ syndrome, e10
Birt-Hogg-Dube syndrome, Clinical and genetic studies of Birt-Hogg-Dube syndrome, e106
blepharophimosis-ptosis-epicanthus inversus syndrome (BPIES), 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, Dysostosesclerosis: a report of three new cases and evolution of the radiological findings, 603
Book reviews, Bahou WF. Genetics for haematologists: the bone dysplasia, 422
BRCA1, A breast cancer family from Spain with germline mutations in both the BRCA1 and BRCA2 genes, e44
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
breast cancer, A cryptic deletion of 2q25 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, 697
breast cancer risk Ashkenazi breast and ovarian cancer families, 611
Detection of large rearrangements of exons 13 and 22 in the BRCA1 gene in German families, e36
ATM mutations in Finnish breast cancer patients, 192
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 breast cancer in young women, 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 predisposition to breast cancer, 91
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 families in Moroccan 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, A candidate gene analysis in the candidate Möbius syndrome genes POG and GATA2 on chromosome 3 and EGR2 on chromosome 10, e30
carbohydrate deficient glycoprotein syndrome, Protein losing enteropathy-hepatic fibrosis syndrome in Saguayan-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib, 849
carloxy-terminal propeptide (C-propeptide), A single amino acid substitution (D141Y1) in the carboxyl-terminal propeptide of the proX(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 eurachromatic variant with no clinical manifestation?, 769
Cardiac sodium channel gene SCN5A, 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 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 DHCGR7 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 cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-Sjögren syndrome, 838

Novel mutations in the γ-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
collagenopathy resulting from a novel mutation in the type II collagen gene, Characterised by high resolution multicolour banding and region specific FISH probes, 718
cystic fibrosis gene (CFTR) gene W846X mutation, Analysis of locus heterogeneity and phenotypic variation, 196
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

GG repeat A single base alteration in the CGG repeat region of the 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 cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-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 cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-Sjögren syndrome, 838

CD4, Promoter polymorphisms in the CD4 receptor gene and their potential association with the severity of chronic periodontitis, 844

CD36 deficiency, Identification of cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-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 cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-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 cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-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 cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-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 cystic splice site, exon skipping, and novel point mutations in type I CD36 deficiency, 286

CD Ib, Prototypic fibroblast fibrosarcoma in Sageney-Lac-St-Jean, a congenital disorder of glycosylation type Ib, e49
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, e67
centromere specific multicolour FISH, Maternal uniparental disomy 12 in a healthy girl with a 47,XX,-der(12)[p11→q11]/46,XX karyotype, e59
cataract syndrome, Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataract-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinseso-Sjögren syndrome, 838

CD14, Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis, 844
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
ectodactyly, Sorting Nexin 3 (SNX3) is disrupted in a patient with a translocation t(6;13)(q21.x12) and microcephaly, microphthalmia, syndactyly, and percutaneous needle biopsy, 235
education, Education improves general practitioner (GP) management of familial breast/ovarian cancer: findings from a cluster randomised controlled trial, 779
ECC syndrome, Analysis of the p63 gene in classical ECC syndrome, related syndromes, and non-syndromic orofacial clefts, 559
the p63 gene in ECC and other syndromes, 377
EGR2 mutation, Frequency of mutations in the early growth response 2 gene associated with peripheral demyelinating neuropathies, 681
elastin vascularopathy, 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 immunodeficiency, 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 statistical 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, 49
Evolution and expression of FOXL2, 916
externalising behaviour, Behavioural phenotype of Bardet-Biedl syndrome, 76
facial dysmorphism, Towards a suggestive facial dysmorphism in adenylsuccinate lyase deficiency?, 440
facial nerve paralysis, Mutation analysis in the candidate Móbius syndrome genes POGT and GATA2 on chromosome 3 and EGR2 on chromosome 10, 330
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 testing hereditary 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, 671
familial hypercholesterolaemia, CYSL27S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, 674
familial juvenile hyperuricaemic nephropathy, Mutations of the UMOR gene are responsible for myelocytic kidney disease 2 and familial juvenile hyperuricaemic nephropathy, 882
familiality, 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, 34
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
FB1N1, Sensitivity of conformation sensitive gel electrophoresis in detecting mutations in Marfan syndrome and related conditions, 34
FB1N1 mutation detection, Twelve novel FB1N1 mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of FB1N1 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
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
fibrocalculous pancreatic diabetes, Mutations in the pancreatic secretory trypsin inhibitor gene (PSTI1/PNPK1) rather than the cathepsin tryptase gene (CTSS) 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, 58
fingers, Multifocal glomus tumours of the fingers in two patients with neurofibromatosis type 1, 457
Finnish, Four novel mutations in the OFD1 (Crox15) gene in Finnish patients with oral-facial-digital syndrome type 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, 60
Sublenticular rearrangements detected by FISH in three of 33 families with idiopathic mental retardation and minor physical anomalies, 635
fluorescent genotyping, Automated fluorescent genotyping detects 10% of cryptic subtelomeric rearrangements in idiopathic syndromic mental retardation, 266
FMRI, A single base alteration in the CGG repeat region of FMRI: possible effects on gene expression and protein, 436
Mosaicism for FMRI and FMRI2 deletion: a new case, 200
FMRI2, Mosaicism for FMRI1 and FMRI2 deletion: a new case, 200
folate, Inositol and folate resistant neural tube defects, 45
Maternal MTHFR genotypes contribute 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 contribution of a specific NCL2 alleles, 825
founder mutation, Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the HGO gene, 40
FOXL2, Analysis of the phenotypic abnormalities in lymphoedemadistichiasis syndrome in 74 patients with FOXL2 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, 443
fragile X syndrome, A study based school of children with learning disability indicates poor levels of genetic investigation, 419
A single base alteration in the CGG repeat region of FMRI: possible effects on gene expression and phenotype, 196
Mosaicism for FMRI and FMRI2 deletion: a new case, 200
framesshift mutation, Two novel frameshift mutations in NXX2.25 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, 58
FRN, Correlation between a specific Wilms tumour suppressor gene (WT1) mutation and the histological findings in Wilms tumour (WT), 833
fructose intolerance, Molecular analysis of the aldolase B gene in patients with hereditary fructose intolerance from Spain, 586
γ-crystallin, Novel mutations in the γ-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 oligonucleotide arrays, 436
mutation, CYSL27S (FH-Kairouan) and D245N (FH-Tozeur) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, 674
general practitioner, Education improves general practitioner (GP) management of familial breast/ovarian cancer: findings from a cluster randomised controlled trial, 779
general practitioners, Continental medical education approaches for clinical genetics: a postal survey of general practitioners, 699
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
Subject index 5o f1 3
HOX genes, Severe digital abnormalities in a patient heterozygous for both 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(12;22)(q31;22) associated with mesomelic dysplasia, e34

H+−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

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, 225

idiopathic mental retardation, Subtelomeric rearrangements detected by FISH in three of 33 families with idiopathic mental retardation and minor physical anomalies, e55

IGF2, Characterisation of the growth regulating gene IGF3, 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

Ins2, Current status of human chromosome 14, 81

Integri 1B gene polymorphism, Association of an Integri 1B gene polymorphism (−511) with Parkinson’s disease in Finnish patients, 400

in situ hybridisation, Cyto genetic analysis of spermatozoa from males aged between 47 and 71 years, e63

inborn errors, Protein losing enteropathy-hepatitis 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

inflammatory bowel disease, Transient neonatal diabetes, a disorder of imprinting, 872

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

klinefelter syndrome, Klinefelter-like phenotype and primary infertility in a male with a paracentric X 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?, e71

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 language delay: a severe deficit in the refinement of the subtelomeric 22q deletion syndrome, e17

late infantile onset neuronal ceroid lipofuscinosis (CLN2, LINCL), Identification of novel CLN2 mutations shows Canadian specific CLN2 alleles, 822

Laurence-Moon-Bardet-Biedl syndrome, Behavioural phenotype of Bardet-Biedl syndrome, e76

LDL receptor, CYS127S (FH-Kairouan) and D245N (FH-Touez) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, e74

learning disability, A school based study of children with learning difficulties: evidence for autosomal recessive inheritance, 575

learning disability, High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH), 822

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-Weil dyschondrosteosis, SHOX point mutations and deletions in Leri-Weil dyschondrosteosis, e35

leukaemia, Hereditary duplication of proximal chromosome 1q (q11q22) in a patient with 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 NEJ1 DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, 900

limb, Thrombocytopaenia-absent radius syndrome: a clinical genetic study, 876

limb development, The fibrillin-1 gene (FBLN1) is disrupted in a t(11;12)(q22;q21) associated with a complex type of spondyloactyly, 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

linkage analysis, A genome wide scan for familial high myopia suggests a novel locus on chromosome 7q36, e118

linkage disequilibrium, An MLH1 haplotype is over-represented on chromosomes carrying an HNPPC predisposing mutation in MLH1, e323

ischaemic heart disease, Identification of cryptic splice site, exon skipping, and novel point mutations in type 1 CDS8 deficiency, 286

isochromosome 15, Severe phenotype in Angelman syndrome resulting from paternally 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

klinefelter syndrome, Klinefelter-like phenotype and primary infertility in a male with a paracentric X 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?, e71

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 language delay: a severe deficit in the refinement of the subtelomeric 22q deletion syndrome, e17

late infantile onset neuronal ceroid lipofuscinosis (CLN2, LINCL), Identification of novel CLN2 mutations shows Canadian specific CLN2 alleles, 822

Laurence-Moon-Bardet-Biedl syndrome, Behavioural phenotype of Bardet-Biedl syndrome, e76

LDL receptor, CYS127S (FH-Kairouan) and D245N (FH-Touez) mutations in the LDL receptor gene in Tunisian families with familial hypercholesterolaemia, e74

learning disability, A school based study of children with learning difficulties: evidence for autosomal recessive inheritance, 575

High throughput screening of human subtelomeric DNA for copy number changes using multiplex amenable probe hybridisation (MAPH), 822

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-Weil dyschondrosteosis, SHOX point mutations and deletions in Leri-Weil dyschondrosteosis, e35

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 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 NEJ1 DNA ligase IV can affect the risk of developing multiple myeloma, a tumour characterised by aberrant class switch recombination, 900

limb, Thrombocytopaenia-absent radius syndrome: a clinical genetic study, 876

limb development, The fibrillin-1 gene (FBLN1) is disrupted in a t(11;12)(q22;q21) associated with a complex type of spondyloactyly, 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, e118

A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter, 567

Identification of candidate lung cancer susceptibility genes in mice using oligonucleotide arrays, 644

Linkage of otosclerosis to a second locus (OTSC3) on human chromosome 6p21.3-22.3, 473

linkage disequilibrium, An MLH1 haplotype is over-represented on chromosomes carrying an HNPPC predisposing mutation in MLH1, e323

Subject index 7 of 13

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 families, 125

lipodystrophy, Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy, 722

lipopolysaccharide, Promoter polymorphisms in the CD14 receptor and their potential association with the severity of chronic periodontitis, 844

locus heterogeneity, A genome wide scan for familial high myopia suggests loci on chromosome 7q36, 418

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, 513

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 expanded R1 gene responsible for low penetrance hereditary retinoblastoma, e21

lymphangiomatosis, 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 overstated, 335

Moddeling deformity, Prevalence of mutations in the short stature homeobox containing gene (SHOX) in Moddeling deformity of childhood, 758

magnetic resonance imaging, Corpus callosum hypoplasia and associated brain anomalies in Nijmegen breakage syndrome, 625

male breast cancer, Novel BRCA2 mutation in a Polish family with hamartoma and two male breast cancers, e35

male infertility, A new case of Yo 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 mandibulo- faciоskeletal syndrome 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, 10p, Xp15

Marfan syndrome, Sensitivity of conformation sensitive gel electro- phoresis 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

Marinесsco-Sjögren syndrome, Linkage to 18qter differentiates two clonally overlapping syndromes: congenital cataracts- facial dysmorphism neuropathy (CCFDN) syndrome and Marinесsco-Sjögren syndrome, e38

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

MCA/HR, 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

MDH1 mutations, Naturally occurring mutations and functional polymorphisms in multidrug resistance 1 gene: correlation with microsatellite instability and lymphoid infiltration in colorectal cancers, 340

Mitochondrial DNA, Leber hereditary optic neuropathy, 162

Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, e27

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 Catalanian families with colorectal cancer, e29

missense mutation, De novo GL3 mutation in acrocallosal syndrome: broadening the phenotypic spectrum of GL3 defects and overlap with murine models, e80

mitochondrial DNA, Leber hereditary optic neuropathy, 162

Spinocerebellar ataxia and the A3243G and A8344G mtDNA mutations, e27

mitochondrial RNA (tRNA<sup>Met</sup>)<sub>UCCN</sub> gene Maternally inherited non- syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial RNA (tRNA<sup>Met</sup>)<sub>UCCN</sub> gene, e82

MLH1, An MLH1 haplotype is over-represented on chromosomes carrying an HNPPC 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, Germine MSH2 and MLH1 mutational spectrum in HNPPC 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 PG7 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 map to the X chromosome pericentric region: a new syndrome or narrow definition of Sutherland-Haan genetic locus?, 76

mtDNA, Childhood onset mitochondrial myopathy and lactic acidosiсs caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene, e81

The frequency of mtDNA 8994 polymorphism and detection of the NARP 8993 mutation, 204

MTTHFR, Human T and risk for neural tube defects, e14

MTTHFR gene, Changes in frequencies of heterozygous thermolabile 5,10-methylenetetrahydrofolate reductase gene in fetuses with neural tube defects, 366

MTTHFR genotype, Maternal MTTHFR genotype contributes to the risk of non-syndromic cleft lip and palate, e38

mucopolysaccharidosis type IIIB, Identification and characterisation of mutations underlying Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB), e3
Novel mutations in the γ-crystallin genes cause autosomal dominant congenital glaucoma.

Organisation of the pericentromeric region of chromosome 15: at least four partial gene copies are amplified in patients with a proximal duplication of 15q.

Promoter polymorphisms in the CD14 receptor gene and their potential association with the severity of chronic periodontitis.

Specific haplotypes of the RET proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma.

PCR, Analysis of the p63 gene in classical EEC syndrome, related genetic syndromes, and non-syndromic congenital glaucoma.

DNA for large rearrangements of the BRCA1 and BRCA2 genes in French breast cancer families.

A breast cancer family from Spain with germline mutations in paraganglia, paraganglioma.

Mutations in the pancreatic secretory trypsin inhibitor gene (PSTI/SPINK1) are significantly associated with tropical calcific pancreatitis.

Phosphomannose isomerase, Phenotypic sex, SDHB mutation analysis in familial and sporadic phaeochromocytoma.

Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes to genetic counselling among probands referred to three genetic clinics.

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 breast cancer susceptibility.

Absence of 22q11 deletions in 211 patients with developmental delay analysed using PCR.

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.

Linkage analysis and molecular screening for HNPCC in the UK show a low frequency of mutations, 857.

Phosphomannose isomerase, Protein losing enteropathy-hepatocytic fibrosis syndrome in Saguenay-Lac-St-Jean, Quebec is a congenital disorder of glycosylation type Ib.

A Val227Ala polymorphism in the peroxisome proliferator-activated receptor α (PPARα) gene is associated with variations in serum lipid levels, 189.

Preimplantation genetic diagnosis, Preimplantation genetic diagnosis in clinical practice.

Preimplantation genetic diagnosis, Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes to genetic counselling among probands referred to three genetic clinics.

Prostate-specific antigen, Genetic testing, Prostate cancer.

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 breast cancer susceptibility.

Absence of 22q11 deletions in 211 patients with developmental delay analysed using PCR.

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.

Linkage analysis and molecular screening for HNPCC in the UK show a low frequency of mutations, 857.

Phosphomannose isomerase, Protein losing enteropathy-hepatocytic fibrosis syndrome in Saguenay-Lac-St-Jean, Quebec is a congenital disorder of glycosylation type Ib.

A Val227Ala polymorphism in the peroxisome proliferator-activated receptor α (PPARα) gene is associated with variations in serum lipid levels, 189.

Preimplantation genetic diagnosis, Preimplantation genetic diagnosis in clinical practice.

Preimplantation genetic diagnosis, Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes to genetic counselling among probands referred to three genetic clinics.

Prostate-specific antigen, Genetic testing, Prostate cancer.
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 (UHDRS), 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 van Buchem disease, 91
Val227Ala polymorphism, A Val227Ala polymorphism in the A locus for isolated cataract on human Xp, e7
vestibular schwannoma, Molecular genetic analysis of the NF2 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, e58
Correlation between a specific Wilms tumour suppressor gene (WT1) mutation and the histological findings in Wilms tumour (WT), e53
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 S’ non-coding region of the connexin32 gene: possible pitfalls in the diagnosis of X linked Charcot-Marie-Tooth neuropathy (CMTX), e55
X linked mental retardation, A new MRXS locus maps to the Xq22.3 (ATS-MR), e13
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, Apor syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in patients with Xq22.3 (A1S-MR), e35
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, e92
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, 597
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