

Review article

Intracellular inclusions, pathological markers in diseases caused by expanded polyglutamine tracts?
D C Rubinsztein, A Wyttenbach, J Rankin 265

Original articles

Systematic characterisation of disease associated balanced chromosome rearrangements by FISH: cytogenetically and genetically anchored YACs identify microdeletions and candidate regions for mental retardation genes
J Wirth, H-G Nothwang, S van der Maarel, C Menzel, G Borck, I Lopez-Pajares, K Brøndum-Nielsen, N Tommerup, M Bugge, H-H Ropers, T Haaf 271

A molecular and FISH analysis of structurally abnormal Y chromosomes in patients with Turner syndrome
D O Robinson, P Dalton, P A Jacobs, K Mosse, M M Power, D H Skuse, J A Crolla 279

Mutational spectrum of the TSC1 gene in a cohort of 225 tuberous sclerosis complex patients: no evidence for genotype-phenotype correlation
M van Slechtenhorst, S Verhoef, A Tempelaars, L Bakker, Q Wang, M Wessels, R Bakker, M Nellist, D Lindhout, D Halley, A van den Ouweland 285

Identification of a single ancestral CYP11B1 mutation in Slovak Gypsies (Roms) affected with primary congenital glaucoma
M Pláčilová, I Stoilov, M Sarfarazi, L Kádasi, E Feráková, V Ferák 290

Evidence for a functional repeat polymorphism in the promoter of the human NRAMP1 gene that correlates with autoimmune versus infectious disease susceptibility
S Searle, J M Blackwell 295

Rapid detection of chromosome aneuploidies by quantitative fluorescence PCR: first application on 247 chorionic villus samples
B Pertl, S Kopp, P M Kroisel, L Tului, B Brambati, M Adinolfi 300

High frequency of BRCA1/2 germline mutations in 42 Belgian families with a small number of symptomatic subjects
G Goelen, E Teugels, M Bonduelle, B Neyns, J De Grève 304

The accuracy of diagnoses as reported in families with cancer: a retrospective study
F S Douglas, L C O'Dair, M Robinson, D G R Evans, S A Lynch 309

Identification and quantification of somatic mosaicism for a point mutation in a Duchenne muscular dystrophy family
T A Smith, S C Yau, M Bobrow, S J Abbs 313

ERG phenotype of a dystrophin mutation in heterozygous female carriers of Duchenne muscular dystrophy
K M Fitzgerald, G W Gibis, A Headrick Gettel, R Rinaldi, D J Harris, R A White 316

Association of a lymphotoxin α gene polymorphism and atopy in Italian families
E Trabetti, C Patuzzo, G Malerba, R Galavotti, L C Martinati, A L Boner, P F Pignatti 323

Short reports

47,XX,UPD(7)mat,+r(7)pat/46,XX,UPD(7)mat mosaicism in a girl with Silver-Russell syndrome (SRS): possible exclusion of the putative SRS gene from a 7p13-q11 region
O Miyoshi, T Kondoh, H Taneda, K Otsuka, T Matsumoto, N Niikawa 326

Unusual fan shaped ossification in a female fetus with radiological features of boomerang dysplasia
S Odent, P Loget, B Le Marec, A-L Delezoide, P Maroteaux 330

Trisomy/tetrasomy 21 mosaicism in CVS: interpretation of cytogenetic discrepancies between placental and fetal chromosome complements
A Soler, E Margarit, A Carrió, D Costa, R Queralt, F Ballesta 333

Directly inherited partial trisomy of chromosome 6p identified in a father and daughter by chromosome microdissection
M B Delatycki, L Voullaire, D Francis, V Petrovic, A Robertson, L M Webber, H R Slater 335

Ectopic NORs on human chromosomes 4qter and 8q11: rare chromosomal variants detected in two families
M Guttenbach, T Haaf, C Steinlein, J Caesar, A Schinzel, M Schmid 339

Congenital variant Rett syndrome in a girl with terminal deletion of chromosome 3p
J Wahlström, A Uller, T Johannesson, D Holmqvist, C Darnfors, M Vujic, B Tonny, B Hagberg, T Martinsson 343

Letters to the Editor

Breakpoint mapping by FISH in a Sotos patient with a constitutional translocation t(3;6)
K Kok, A Mosselaar, H Faber, T Dijkhuizen, T G Draaijers, A Y van der Veen, C H C M Buys, C T R M Schrander-Stumpel 346

Familial testicular cancer: lack of evidence for trinucleotide repeat expansions and association with PKD1 in one family
B T Teh, K Linblad, B Nord, S Kytälä, M Schalling, C Larsson, E Rapley, P Biggs, R Huddart, M Stratton, S Hii, D Nicol 348

Tricuspid atresia and conotruncal malformations in five families
D Bonnet, L Fermon, J Kachaner, D Sidi, J Amiel, S Lyonnet, A Munnich 349

Clinical governance and genetic medicine. Specialist genetic centres and the Confidential Enquiry into Counselling for Genetic Disorders by non-geneticists (CEGEN)
R Harris, H J Harris 352

Notice to contributors

36
4
Vol 36 No 4 Pages 265-352

JOURNAL OF MEDICAL GENETICS

April 1999

JMG
JOURNAL OF MEDICAL GENETICS

