

**Review article**

- Intracellular inclusions, pathological markers in diseases caused by expanded polyglutamine tracts?  
*D C Rubinstein, A Wyettnebach, 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 Nethwang, 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 Slegtenhorst, S Verhoef, A Tempelaars, L Bakker, Q Wang, M Wessels, R Bakker, M Nellist, D Lindhout, D Halley, A van den Ouwehand* 285
- Identification of a single ancestral CYP1B1 mutation in Slovak Gypsies (Roms) affected with primary congenital glaucoma  
*M Plášilová, I Stolov, 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, P Kopp, P M Kroisel, I Tului, B Brambati, M Adnolfi* 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 Cibis, A Headrick Gettel, R Rinaldi, D J Harris, R A White* 316
- Association of a lymphotxin  $\alpha$  gene polymorphism and atopy in Italian families  
*E Trabetti, C Patuzzo, G Malerba, R Galavotti, L C Martinati, A 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 Carrío, 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 Vuillaume, 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 Tonby, 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 Kytolä, 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 Fermont, J Kachaner, D Sidi, J Amiel, S Lyonnet, A Munrich* 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**

