Original articles
Locationalisation of two candidate genes for mental retardation using a YAC physical map of the Xq21.1–21.2 subbands L Colleaux, M May, J Belouguine, D Lepaslier, C Schwartz, M Fontes 353
Mutation analysis in 24 French patients with glycosogen storage disease type 1a F Chevalier-Perot, D Bazan, A-M Bonardet, N Bruni, G Milhieux, M Mathieu, I Maire 358
Direct detection of 4q35 rearrangements implicated in facioscapulohumeral muscular dystrophy (FSHD) G Deidda, S Cacurri, P Azzaro, L Felicetti 361
Monosomy of distal 4q does not cause facioscapulohumeral muscular dystrophy R Tupler, A Berardinelli, L Barbariero, R Frants, J E Hewitt, G Lanzi, P Maraschio, L Tiepolo 366
Direct molecular diagnosis of CYP21 mutations in congenital adrenal hyperplasia H H Lee, H T Chao, H T Ng, K B Chao 371
FMRI fully expanded mutation with minimal methylation in a high functioning fragile X male Z Wang, A K Taylor, J A Bridge 376
First experiences with genetic counselling based on predictive DNA diagnosis in hereditary gomus tumours (paragangliomas) J C Oosterwijk, J C Jansen, E M van Schoor, A W Oosterhof, P Devilee, E Bakker, M W Zoeteweij, A G L van der Mey 379
A novel deletion at codon 441 on the APC gene associated with ophthalmic lesions (CHRPE) in a South African family J J Grobbelaar, A Ziskind, G de Jong, C J Joubert Oosthuizen, M J Katze 384
Breakpoints in α, β, and satellite III DNA sequences of chromosome 9 result in a variety of pericentric inversions K H Ramesh, R S Verma 395
Small extra ring chromosome derived from chromosome 10p: clinical report and characterisation by FISH E Blennow, E Tillberg 399

Syndrome of the month
Marfan syndrome J R Gray, S J Davies 403

Brief papers
Renal-hepatic-pancreatic dysplasia: an autosomal recessive malformation R Torra, L Alos, J Ramos, X Estivill 409
Argininc-164-tryptophan substitution in connexin32 associated with X linked dominant Charcot-Marie-Tooth disease A Oterino, F I Montón, V M Cabrero, F Pinto, A Gonzalez, N R Lavilla 413
Orocardiodigital syndrome: an oral-facial-digital type II variant associated with atrioventricular canal M C Digilio, B Marino, A Giannotti, B Dallapiccola 416
Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss (CAPOS): a new syndrome P Nicolaides, R E Appleton, A Fryer 419
A boy with developmental delay and a maternally inherited deletion in 15q11q13 M King, C Hardy, B Asenbauer, M Kilpatrick, T Webb 422
Mirror hands and feet: a further case of Laurin-Sandrow syndrome E Hatchwell, N Dennis 426
A new case of fibrochondrogenesis from Spain M I Martinez-Frias, A Garcia, J Cuevas, J L Rodriguez, M Urioste 429

Short reports
Linkage analysis of two Canadian families segregating for X linked spondyloepiphysial dysplasia L E Bernard, D Chiotayat, R Weksberg, M I Van Allen, S Langlois 432
Analysis of GLRA1 in hereditary and sporadic hyperekplexia: a novel mutation in a family cosegregating for hyperekplexia and spastic paraparesis F V Elmslie, S M Hutchings, V Spencer, A Curtis, T Covanius, R M Gardiner, M Rees 435
Acute intermittent porphyria caused by defective splicing of porphobilinogen deaminase RNA: a synonymous codon mutation at −22 bp from the 5’ splice site causes skipping of exon 3 D H Llewellyn, G A Scothe, A J Urquhart, S D Whatley, A G Roberts, P R Harrison, G H Elder 437

Letter to the Editor
Different origins of mutations at the Machado-Joseph locus (MJD1) P Iuggetti, M Zatz, M R Passos Bueno, S K Marie 439

Book review
440

Notices
440