

Contents

**Original articles**

Localisation of two candidate genes for mental retardation using a YAC physical map of the Xq21.1-21.2 subbands *L Colleaux, M May, J Belougne, D Lepaslier, C Schwartz, M Fontes* 353

Mutation analysis in 24 French patients with glycogen storage disease type 1a *F Chevalier-Porst, D Bozon, A-M Bonardot, N Bruni, G Mithieux, M Mathieu, I Maire* 358

Direct detection of 4q35 rearrangements implicated in facioscapulohumeral muscular dystrophy (FSHD) *G Deidda, S Cacurri, N Piazza, L Felicetti* 361

Monosomy of distal 4q does not cause facioscapulohumeral muscular dystrophy *R Tupler, A Berardinelli, L Barbierato, 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 Choo* 371

FMR1 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 glomus tumours (paragangliomas) *J C Oosterwijk, J C Jansen, E M van Schothorst, A W Oosterhof, P Devilee, E Bakker, M W Zoetewij, 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 Kotze* 384

Cytogenetic and epidemiological findings in Down syndrome, England and Wales 1989 to 1993 *D Mutton, E Alberman, E B Hook for The National Down Syndrome Cytogenetic Register and The Association of Clinical Cytogeneticists* 387

Breakpoints in  $\alpha$ ,  $\beta$ , 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 Alós, J Ramos, X Estivill* 409

Arginine-164-tryptophan substitution in connexin32 associated with X linked dominant Charcot-Marie-Tooth disease *A Oterino, F I Montón, V M Cabrera, 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 Nicolaidis, 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 L Martínez-Frías, A García, J Cuevas, J I Rodríguez, M Urioste* 429

**Short reports**

Linkage analysis of two Canadian families segregating for X linked spondyloepiphyseal dysplasia *L E Bernard, D Chitayat, 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 Covanis, 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 Scobie, 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 Iughetti, M Zatz, M R Passos Bueno, S K Marie* 439

**Book review**

440

**Notices**

440

first published in May 1996. Downloaded from <http://jmg.bmj.com/> on 26 June 2019 by guest. Protected by copyright.