

Contents

Original articles

A simple and efficient method for microdissection and microFISH J J M Engelen, J C M Albrechts, G J H Hamers, J P M Geraedts	265
Localisation of the gene for glycogen storage disease type 1c by homozygosity mapping to 11q C D Fenske, S Jeffery, J L Weber, R S Houlston, J V Leonard, P J Lee	269
Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene deletion syndrome? J J Jonsson, A Renieri, P G Gallagher, C E Kashtan, E M Cherniske, M Bruttini, M Piccini, F Vitelli, A Ballabio, B R Pober	273
Molecular studies in Finnish patients with familial juvenile nephronophthisis exclude a founder effect and support a common mutation causing mechanism S Ala-Mello, E-M Sankila, O Koskimies, A de la Chapelle, H Kääriäinen	279
Genetic localisation of mental retardation with spastic diplegia to the pericentromeric region of the X chromosome: X inactivation in female carriers F Martínez, M Tomás, J M Millán, A Fernández, F Palau, F Prieto	284
Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott-Rallison syndrome D T Bonthron, N Dunlop, D G D Barr, A A El Sanousi, L I Al-Gazali	288
Myotonia congenita in northern Finland: an epidemiological and genetic study P Baumann, V V Myllylä, J Leisti	293
Chromosome mapping of Rett syndrome: a likely candidate region on the telomere of Xq F Xiang, Z Zhang, A Clarke, P Joseluiz, N Sakkubai, B Sarojini, C D Delozier-Blanchet, I Hansmann, L Edström, M Anvret	297
Population genetics of hyperphenylalaninaemia resulting from phenylalanine hydroxylase deficiency in Portugal I Rivera, P Leandro, U Lichter-Konecki, I Tavares de Almeida, M C Lechner	301
Evidence for anticipation in autosomal dominant limb-girdle muscular dystrophy M C Speer, J M Gilchrist, J M Stajich, P C Gaskell, C A Westbrook, S K Horrigan, L Bartoloni, L H Yamaoka, W K Scott, M A Pericak-Vance	305
Clinical practice in medical genetics	
Contracting for clinical genetic services: the Welsh model H E Hughes, J K Alderman, M Krawczak, C Rogers	309
Syndrome of the month	
The dystonias P R Jarman, T T Warner	314
Short reports	
Trisomy 2q11.2→q21.1 resulting from an unbalanced insertion in two generations I A Glass, P Stormer, P T S P Oei, E Hacking, P D Cotter	319
Familial neurofibromatosis type 1 associated with an overgrowth syndrome resembling Weaver syndrome C J van Asperen, W C G Overweg-Plandsoen, M H Cnossen, D A van Tijn, R C M Hennekam	323
Simultaneous adrenocortical carcinoma and ganglioneuroblastoma in a child with Turner syndrome and germline p53 mutation E K Pivnick, W L Furman, G V N Velagaleti, J J Jenkins, N A Chase, R C Ribeiro	328
Two adult females with a distinct familial mental retardation syndrome: non-progressive neurological symptoms with ataxia and hypotonia, similar facial appearance, hypergonadotrophic hypogonadism, and retinal dystrophy J-P Fryns, C Van Lingen, K Devriendt, E Legius, P Raus	333
Keratosis follicularis spinulosa decalvans: confirmation of linkage to Xp22.13-p22.2 M E M Porteous, L Strain, L J Logie, R M Herd, E C Benton	336
Absence of PAX2 gene mutations in patients with primary familial vesicoureteric reflux K-L Choi, L A McNoe, M C French, P J Guilford, M R Eccles	338
Severe testotoxicosis phenotype associated with Asp578→Tyr mutation of the lutrophin/choriogonadotrophin receptor gene J Müller, B Gondos, S Kosugi, T Mori, A Shenker	340
Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation K Nagai, M Nagao, M Nagao, S Yanai, K Minagawa, Y Takahashi, Y Takekoshi, A Ishizaka, Y Matsuzono, O Kobayashi, T Itagaki	342
Abstracts	
Medical genetics: advances in brief	345
Letters to the Editor	
Craniosynostosis and chromosome 22q11 deletion J C S Dean, D C de Silva, W Reardon	346
Spectrum of clinical features associated with interstitial chromosome 22q11 deletions M Di Rocco, A Buocompagni, P Picco, S Vignola, C Borrone, G Gimelli	346
Clinical features of chromosome 22q11 deletion A Hunter	346
Reply to letters regarding clinical features of chromosome 22q11 deletion A Ryan, J A Goodship, D I Wilson, on behalf of the European Consortium	347
Absence of a del(22q11) in a patient with the 3C (craniocerebellocardiac) syndrome J M Saraiva, E Matoso, I Marques	347
A mother with VCFS and unilateral dysplastic kidney and her fetus with multicystic dysplastic kidneys: additional evidence to support the association of renal malformations and VCFS P M Czarnecki, D L Van Dyke, S Vats, G L Feldman	348
New overgrowth syndrome and FGFR3 dosage effect M M Cohen Jr, G Neri	348
Reply M Partington, G Turner	348
Robinow syndrome M Van Steensel	349
Reply M A Sabry, E A R Ismail, N Al-Torki, S Farah	350
Book review	
Human Cytogenetic Cancer Markers	350
Notice	
Notice to contributors	352