

Contents

Original articles

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