

MLCNY



51027000216547

*Journal of*

# MEDICAL GENETICS

**Original articles**

- 649 Molecular and cytogenetic studies of the Prader-Willi syndrome *R J Trent, F Volpato, A Smith, R Lindeman, M-K Wong, G Warne, E Haan*
- 655 Genetic counselling in facioscapulohumeral muscular dystrophy *P W Lunt, P S Harper*
- 665 A closely linked DNA marker for facioscapulohumeral disease on chromosome 4q *M Upadhyaya, P W Lunt, M Sarfarazi, W Broadhead, J Daniels, M Owen, P S Harper*
- 672 Sex ratio of the mutation frequencies in haemophilia A: coagulation assays and RFLP analysis *A H J T Bröcker-Vriends, F R Rosendaal, J C van Houwelingen, E Bakker, G J B van Ommen, J J P van de Kamp, E Briët*
- 681 Linkage analysis in adenomatous polyposis coli: the use of four closely linked DNA probes in 20 UK families *M B Cachon-Gonzalez, J D A Delhanty, J Burn, K Tsioupra, M B Davis, J Attwood, P Chapman*
- 686 Silent mutations in the phenylalanine hydroxylase gene as an aid to the diagnosis of phenylketonuria *L Kalaydjieva, B Dworniczak, C Aulehla-Scholz, M Devoto, G Romeo, M Sturhmann, V Kucinskas, V Yurgelyavicius, J Horst*

**Case reports**

- 691 A male with type I orofacioidigital syndrome *J Goodship, J Platt, R Smith, J Burn*
- 695 Miller syndrome (postaxial acrofacial dysostosis): further evidence for autosomal recessive inheritance and expansion of the phenotype *A L Ogilvy-Stuart, A C Parsons*
- 701 A new form of autosomal dominant arthrogyriposis *M M R Lai, M A Tettenborn, J G Hall, L J Smith, A C Berry*
- 704 Cerebrocostomandibular syndrome in four sibs, two pairs of twins *V Drossou-Agakidou, A Andreou, V Soubassi-Griva, M Pandouraki*
- 708 Encephalopathy with intracerebral calcification, white matter lesions, growth hormone deficiency, microcephaly, and retinal degeneration: two sibs confirming a probably distinct entity *C G Bönemann, P Meinecke, H Reich*
- 712 Disorganisation: a possible cause of apparent conjoint twinning *M A Petzel, R P Erickson*
- 715 A cystic fibrosis patient homozygous for the nonsense mutation R553X *J Bal, M Sturhmann, M Schloesser, J Schmidtke, J Reiss*

**Short report**

- 718 An animal model for maternal phenylketonuria *C Roux, F Rey, S Lyonnet, S Nizard, N Mulliez, A Munnich*

**Conference report**

- 720 Social and genetic implications of customary consanguineous marriage among British Pakistanis. Report of a meeting held at the Ciba Foundation on 15 January 1991 *B Modell*

**Letters to the Editor**

- 724 The non-deletion type of  $\alpha$  thalassaemia/mental retardation: a recognisable dysmorphic syndrome with X linked inheritance *A O M Wilkie, M E Pembrey, R J Gibbons, D R Higgs, M E M Porteous, J Burn, R M Winter*
- 724 Leiomyosarcoma in a patient with trisomy 8 mosaicism *W M Molenaar, B De Jong, E Van den Berg*
- 725 Further evidence for the location of the BPES gene at 3q2 *C E M de Die-Smulders, J J M Engelen, J M Donk, J P Fryns*

725-728 Book reviews