

MLCNY



51027000216546

Journal of

MEDICAL GENETICS

Original articles

- 577 Chromosome in situ suppression hybridisation in clinical cytogenetics *M A Hulten, C P Gould, A S H Goldman, J J Waters*
- 583 Confirmation of linkage of hyperkalaemic periodic paralysis to chromosome 17 *M C Koch, K Ricker, M Otto, T Grimm, E P Hoffman, R Rüdell, K Bender, B Zoll, P S Harper, F Lehmann-Horn*
- 587 The human genes for complement components 6 (C6) and 9 (C9) are closely linked on chromosome 5 *S Rogne, O Myklebost, J H Olving, H T Kyrkjebø, R Jonassen, B Olaisen, T Gedde-Dahl Jr*
- 591 Use of probes for ZFY, SRY, and the Y pseudoautosomal boundary in XX males, XX true hermaphrodites, and an XY female *E T Pereira, J C Cabral de Almeida, A C Y R G Gunha, M Patton, R Taylor, S Jeffery*
- 596 Velocardiofacial (Shprintzen) syndrome: an important syndrome for the dysmorphologist to recognise *A H Lipson, D Yuille, M Angel, P G Thompson, J C Vandervoord, E J Beckenham*

605 Do familial neural tube defects breed true? *E Drainer, H M May, J L Tolmie*

609 49,XXXXY syndrome: behavioural and developmental profiles *C A Lomelino, A L Reiss*

613 Birth distribution in cystic fibrosis in Saguenay-Lac-St-Jean, Quebec, Canada *J Daigneault, G Aubin, F Simard, M De Braekeleer*

615 High ⁶⁴Cu uptake and retention values in two clinically atypical Menkes patients *T Tonnesen, C Garrett, A-M Gerdes*

Case reports

619 Microcephaly-cardiomyopathy: a new autosomal recessive phenotype? *I M Winship, D L Viljoen, P M Leary, M M De Moor*

622 Sibs with mental retardation, supraorbital sclerosis, and metaphyseal dysplasia: frontometaphyseal dysplasia, craniometaphyseal dysplasia, or a new syndrome? *W Reardon, C M Hall, M J Dillon, M Baraitser*

627 Smith-Magenis syndrome: a new contiguous gene syndrome. Report of three new cases *A Moncla, M O Livet, M Auger, J F Mattei, M G Mattei, F Giraud*

633 A new recessive syndrome of unusual facies and multiple structural abnormalities *Y Thakker, D Donnai*

636 Postaxial acrofacial dysostosis (Miller) syndrome: a new case *J Vigneron, M Stricker, P Vert, J M Rousselot, M Levy*

639 A de novo translocation t(3;17)(q26.3;q23.1) in a child with Cornelia de Lange syndrome *M Ireland, C English, I Cross, W T Houlshby, J Burn*

641 Mosaic partial trisomy 17q2 *P A King, A Ghosh, M Tang*

Letters to the Editor

644 The frequency of mental retardation in hypochondroplasia *R Wynne-Davies, M A Patton*

644 Unusual inheritance of Becker type muscular dystrophy *R Lisker, O Mutchinick, L Ruz*

644 Haematometra in the Langer-Giedion syndrome *M W Partington, J Rae, M J Payne*

645 Two additional patients representing the possible human homologue for the mouse mutant disorganisation (Ds) *A E Lin*

647-648 Book reviews • Notices