

Journal of Medical Genetics
 August 1997 Vol 34 No 8
 Contents

Original articles

- Localisation of a gene causing endocrine neoplasia to a 4 cM region on chromosome 1p35-p36** C Williamson, A A J Pannett, J T Pang, C Wooding, M McCarthy, M N Sheppard, J Monson, R N Clayton, R V Thakker 617
- RDS/peripherin gene mutations are frequent causes of central retinal dystrophies** S Kohl, M Christ-Adler, E Apfelstedt-Sylla, U Kellner, A Eckstein, E Zrenner, B Wissinger 620
- The effect of FMR1 CGG repeat interruptions on mutation frequency as measured by sperm typing** C B Kunst, E P Leeftang, J C Iber, N Arnheim, S T Warren 627
- Craniosynostosis associated with FGFR3 pro250arg mutation results in a range of clinical presentations including unsutural sporadic craniosynostosis** W Reardon, D Wilkes, P Rutland, L J Pulleyn, S Malcolm, J C S Dean, R D Evans, B M Jones, R Hayward, C M Hall, N C Nevin, M Baraitser, R M Winter 632
- Developmental enamel defects in tuberous sclerosis: a clinical genetic marker?** N Flanagan, W J O'Connor, B McCartan, S Miller, J McMenamin, R Watson 637
- Cryptic terminal rearrangement of chromosome 22q13.32 detected by FISH in two unrelated patients** K F Doheny, H E McDermid, K Harum, G H Thomas, G V Raymond 640
- De Lange syndrome: subjective and objective comparison of the classical and mild phenotypes** J E Allanson, R C M Hennekam, M Ireland 645
- Counselling dilemmas associated with the molecular characterisation of two Angelman syndrome families** H L Gilbert, J L Buxton, C T J Chan, T McKay, S Cottrell, S Ramsden, R M Winter, M E Pembrey, S Malcolm 651

Syndrome of the month

- Waardenburg syndrome** A P Read, V E Newton 656

Short reports

- Neurogenic chronic idiopathic intestinal pseudo-obstruction, patent ductus arteriosus, and thrombocytopenia segregating as an X linked recessive disorder** D R FitzPatrick, L Strain, A E Thomas, D G D Barr, A Todd, N M Smith, W G Scobie 666
- Hydrocephalus and Hirschsprung's disease in a patient with a mutation of L1CAM** N Okamoto, Y Wada, M Goto 670
- Assessment of French patients with LPL deficiency for French Canadian mutations** L Foubert, J L De Gennes, J P Lagarde, E Ehrenborg, A Raisonier, J P Girardet, M R Hayden, P Benlian 672
- "Cutis tricolor": congenital hyper- and hypopigmented macules associated with a sporadic multisystem birth defect: an unusual example of twin spotting?** R Happle, G Barbi, D Eckert, I Kennerknecht 676
- Prenatal diagnosis of 22q11 deletions: a series of five cases with congenital heart defects** F L Raymond, J M Simpson, C M Mackie, G K Sharland 679
- Phenotypic expression of the fibroblast growth factor receptor 3 (FGFR3) mutation P250R in a large craniosynostosis family** A Golla, P Lichtner, S von Gernet, A Winterpacht, J Fairley, J Murken, S Schuffenhauer 683
- Genetic heterogeneity in Schwartz-Jampel syndrome: two families with neonatal Schwartz-Jampel syndrome do not map to human chromosome 1p34-p36.1** K A Brown, L I Al-Gazali, L M Moynihan, N J Lench, A F Markham, R F Mueller 685
- Monozygotic twins discordant for Aicardi syndrome** T Costa, W Greer, G Rysiecki, J R Buncic, P N Ray 688
- Interstitial deletion, del(4)(q12q21.1), owing to de novo unbalanced translocation in a 2 year old girl: further evidence that the piebald trait maps to proximal 4q12** A Schinzel, C P Braegger, L Brecevic, F Dutly, F Binkert 692
- Familial four breakpoint complex chromosomal rearrangement as a cause of monosomy 9p22→pter and trisomy 10p11.2→pter and 11q21 analysed by dual and triple colour FISH** P Stankiewicz, E Kostyk, E Bocian, H Stańczak, J Parczewska, E Piątkowska, T Mazurczak, J J Pietrzyk 696

Abstracts

- Medical genetics: advances in brief** 700

Letters to the Editor

- Juvenile onset Huntington's disease in an Omani child with asymptomatic, at risk parents** E M Scrimgeour, R L Koul, P R Chand, J K J Tharakan, C A Frew 701
- Mirror hands and feet** M A M van Steensel 701
- A study of brothers with Klinefelter syndrome** C G Woods, J Noble, A R Falconer 702

Book reviews

- The Child with Multiple Birth Defects • Subcellular Biochemistry • Cultural and Ethnic Diversity. A Guide for Genetics Professionals • Genetic Disorders among Arab Populations** 702