

Journal of

MEDICAL GENETICS

Review article537 Genes and epilepsy *R M Gardiner***Original articles**545 The clinical features of osteogenesis imperfecta resulting from a non-functional carboxy terminal pro α 1(I) propeptide of type I procollagen and a severe deficiency of normal type I collagen in tissues *W G Cole, P E Campbell, J G Rogers, J F Bateman*552 Adult polycystic kidney disease: knowledge, experience, and attitudes to prenatal diagnosis *K A Hodgkinson, L Kerzin-Storarr, E A Watters, R Harris*559 Genetic and environmental factors in hypospadias *C Stoll, Y Alembik, M P Roth, B Dott*564 Genetic analysis of treated and untreated phenylketonuria in one family *L A Tyfield, A L Meredith, M J Osborn, R Primavesi, T L Chambers, J B Holton, P S Harper*569 Repeated freezing and thawing of peripheral blood and DNA in suspension: effects on DNA yield and integrity *K S Ross, N E Hailes, K F Kelly***Syndrome of the month**571 Sotos syndrome *T R P Cole, H E Hughes***Case reports**577 Occurrence of the α thalassaemia-mental retardation syndrome (non-deletional type) in an Australian male *M P Harvey, A Kearney, A Smith, R J Trent*582 Chromosome imbalance, normal phenotype, and imprinting *L Bortotto, E Piovon, R Furlan, H Rivera, O Zuffardi*588 Partial monosomy for chromosome 22 in a patient with del(22)(pter→q13.1::q13.33→qter) *D R Romain, J Goldsmith, H Cairney, L M Columbano-Green, R H Smythe, R G Parfitt*590 Alstrom's syndrome: further evidence of autosomal recessive inheritance and endocrinological dysfunction *S J Charles, A T Moore, J R W Yates, T Green, P Clark***Dysmorphology report**593 Another case of microcephaly, facial clefting, and preaxial polydactyly *S L Marles, A E Chudley***Conference report**595 Symposium on genomic imprinting, Manchester *A Clarke*

597-600 Letters to the Editor • Book reviews • Notices