

Contents

Annotations

- Low segregation ratios in autosomal recessive disorders *S Bunday, I D Young* 449
Isolation of the defective gene in X linked agammaglobulinaemia *D Vetrie* 452

Original articles

- Population studies of the fragile X: a molecular approach *P A Jacobs, H Bullman, J Macpherson, S Youings, V Rooney, A Watson, N R Dennis* 454
Complications of the naevoid basal cell carcinoma syndrome: results of a population based study *D G R Evans, E J Ladusans, S Rimmer, L D Burnell, N Thakker, P A Farndon* 460
Two new mutations in the dihydropteridine reductase gene in patients with tetrahydrobiopterin deficiency *I Dianzani, D W Howells, A Ponzone, J A Saleeba, P M Smooker, R G H Cotton* 465
A syndrome of insulin resistance resembling leprechaunism in five sibs of consanguineous parents *L I Al-Gazali, M Khalil, K Devadas* 470
RFLP analysis for APP 717 mutations associated with Alzheimer's disease *S R Zeldenrust, J Murrell, M Farlow, B Ghetti, A D Roses, M D Benson* 476
Further investigation of the HEXA gene intron 9 donor splice site mutation frequently found in non-Jewish Tay-Sachs disease patients from the British Isles *E C Landels, P M Green, I H Ellis, A H Fensom, M M Kaback, J Lim-Steele, K Zeiger, N Levy, M Bobrow* 479
Three patients with ring (X) chromosomes and a severe phenotype *N R Dennis, A L Collins, J A Crolla, A E Cockwell, A M Fisher, P A Jacobs* 482
Genetic mapping of dinucleotide repeat polymorphisms and von Hippel-Lindau disease on chromosome 3p25-26 *M A Pericak-Vance, K J Nunes, E Whisenant, D B Loeb, K W Small, J M Stajich, J B Rimmner, L H Yamaoka, D I Smith, H A Drabkin, J M Vance* 487
Osteogenesis imperfecta type III: mutations in the type I collagen structural genes, COL1A1 and COL1A2, are not necessarily responsible *G A Wallis, B Sykes, P H Byers, C G Mathew, D Viljoen, P Beighton* 492
Interaction of incontinentia pigmenti and factor VIII mutations in a female with biased X inactivation, resulting in haemophilia *R Coleman, S A Genet, J I Harper, A O M Wilkie* 497
Frequency of $\Delta F508$ in a Mexican sample of cystic fibrosis patients *L Orozco, M Salcedo, J L Lezana, M Chávez, H Valdez, M Moreno, A Carnevale* 501
A large family with patent ductus arteriosus and unusual face *H R Davidson* 503

Portraits in medical genetics

- Edward Meryon (1809-1880) and muscular dystrophy *A E H Emery, M L H Emery* 506

Short reports

- A new restriction fragment length polymorphism at the DXS101 locus allows carrier detection in a family with X linked agammaglobulinaemia *A Sweatman, R Lovering, H Middleton-Price, A Jones, G Morgan, R Levinsky, C Kinnon* 512
Cerebellar ataxia and ectodermal dysplasia in brothers *M Baraitser, W Reardon, A McShane, J Wilson* 515
Deletion 9p and sex reversal *C P Bennett, Z Docherty, S A Robb, P Ramani, J R Hawkins, D Grant* 518
Holoprosencephaly and sacral agenesis in a fetus with a terminal deletion 7q36→7qter *N Morichon-Delvallez, A-L Delezoide, M Vekemans* 521
Severe intrauterine growth retardation, blepharophimosis, and cylindrical nose with midline groove: a new syndrome? *C E M de Die-Smulders, R P Droog, M van Dijk, J P Fryns* 525
Congenital nystagmus cosegregating with a balanced 7;15 translocation *M A Patton, S Jeffery, N Lee, C Hogg* 526
Duplication of chromosome 15 in the region 15q11-13 in a patient with developmental delay and ataxia with similarities to Angelman syndrome *J Clayton-Smith, T Webb, X J Cheng, M E Pembrey, S Malcolm* 529

Abstracts

- Medical genetics: advances in brief 532

Letters to the Editor

- Molecular diagnosis of myotonic dystrophy *R M Winter* 533
Reply *G K Suthers, K E Davies, S M Huson* 533
Weyers' ulnar ray/oligodactyly syndrome *M S Lungarotti, A Calabro* 533
The contribution of genetic factors to the pathogenesis of type I (insulin dependent) diabetes mellitus *F J Grundbacher* 533

Book reviews

536

Notices

536