

Contents

**Review article**

- Spotch locus mouse mutants: models for neural tube defects and Waardenburg syndrome type I in humans *C E Moase, D G Trasler* 145

**Annotation**

- Family history of breast cancer: how important is it? *D T Bishop* 152

**Original articles**

- Family history and risk of breast cancer *R S Houlston, E McCarter, S Parbhoo, J H Scurr, J Slack* 154

- Epidemiology of breast cancer in families in Iceland *H Tulinius, H Sigvaldason, G Ólafsdóttir, L Tryggvadóttir* 158

- Prenatal prediction of spinal muscular atrophy *R J Daniels, G K Suthers, K E Morrison, N H Thomas, M J Francis, C G Mathew, S Loughlin, A Heiberg, D Wood, V Dubowitz, K E Davies* 165

- Prenatal prediction of Werdnig-Hoffmann disease using linked polymorphic DNA probes *J Melki, S Abdelhak, P Burlet, V Raclin, J Kaplan, R Spiegel, S Gilgenkrantz, N Philip, M-L Chauvet, Y Dumez, M-L Briard, J Frézal, A Munnich* 171

- An intrachromosomal insertion causing 5q22 deletion and familial adenomatous polyposis coli in two generations *I Cross, J Delhanty, P Chapman, L V Bowles, D Griffin, J Wolstenholme, M Bradburn, J Brown, C Wood, A Gunn, J Burn* 175

- Prenatal diagnosis and presymptomatic detection of neurofibromatosis type 1 *M Upadhyaya, A Fryer, J MacMillan, W Broadhead, S M Huson, P S Harper* 180

- Clinical variability of type 1 neurofibromatosis: is there a neurofibromatosis-Noonan syndrome? *H J Stern, H M Saal, J S Lee, P R Fain, D E Goldgar, K N Rosenbaum, D F Barker* 184

- Absence of linkage of Noonan syndrome to the neurofibromatosis type 1 locus *M Sharland, R Taylor, M A Patton, S Jeffery* 188

- Analysis of quantitative PCR for the diagnosis of deletion and duplication carriers in the dystrophin gene *S Abbs, M Bobrow* 191

**Case reports**

- Distal spinal muscular atrophy with vocal cord paralysis *C Pridmore, M Baraitser, E M Brett, A E Harding* 197

- Unusual occurrence of cervical myelopathy in a case of Stickler's syndrome *S Noël, D Balériaux, N Telerman-Toppet* 200

**Abstracts**

- Abstracts of the annual scientific meeting of the Association of Clinical Cytogeneticists held on 3 to 5 July 1991 at Earnshaw Hall, Sheffield 203

- Medical genetics: advances in brief 214

**Letters to the Editor**

- A new form of infantile spinal muscular atrophy *A García-Alix, J I Rodriguez, J Quero* 215

- MASA syndrome (a form of complicated spastic paraplegia) and X linked hydrocephalus: variable expression of the same mutation at Xq28? Call for families *C Schrander-Stumpel, J P Frys, J J Cassiman, E Legius, A Spaepen, C J Höweler* 215

- Book reviews 216

- Notices 216