

Contents

Original articles

The association of Angelman's syndrome with deletions within 15q11-13 M PEMBREY, S J FENNELL, J VAN DEN BERGHE, M FITCHETT, D SUMMERS, L BUTLER, C CLARKE, M GRIFFITHS, E THOMPSON, M SUPER, AND M BARAITSER *page 73*

Carrier detection and early diagnosis of Wilson's disease by restriction fragment length polymorphism analysis A FIGUS, R LAMPIS, M DEVOTO, M S RISTALDI, A IDEO, S DE VIRGILIS, A M NURCHI, A CORRIAS, R CORDA, M E LAI, A TOCCO, A DEPLANO, A SOLINAS, L ZANCAN, W-H LEE, A CAO, M PIRASTU, AND A BALESTRIERI *page 78*

Prenatal exclusion testing for Huntington's disease: a problem of too much information F A MILLAN, A CURTIS, M MENNIE, S HOLLOWAY, M BOXER, M J W FAED, J W CRAWFORD, W A LISTON, AND D J H BROCK *page 83*

Familial supravalvular aortic stenosis: a genetic study F CHIARELLA, F D BRICARELLI, G LUPI, P BELLOTTI, S DOMENICUCCI, AND C VECCHIO *page 86*

Spondyloenchondrodysplasia H MENGER, K KRUSE, AND J SPRANGER *page 93*

Interstitial deletion of distal 13q associated with Hirschsprung's disease M A LAMONT, M FITCHETT, AND N R DENNIS *page 100*

Hereditary distal muscular atrophy with vocal cord paralysis and sensorineural hearing loss: a dominant form of spinal muscular atrophy? E BOLTSHAUSER, W LANG, T SPILLMANN, AND E HOF *page 105*

Knowledge and perceptions of haemoglobinopathy carrier screening among general practitioners in Cardiff D SHICKLE AND A MAY *page 109*

Rhodanese isozymes in three subjects with Leber's optic neuropathy D B WHITEHOUSE, C J M POOLE, P R N KIND, AND D A HOPKINSON *page 113*

Portraits in medical genetics Joseph Adams (1756-1818) A E H EMERY *page 116*

Syndrome of the month Stickler's syndrome I K TEMPLE *page 119*

Case reports

Interstitial deletion of the long arm of chromosome 2 with normal levels of isocitrate dehydrogenase I A GLASS, C A SWINDLEHURST, D A AITKEN, W McCREA, AND E BOYD *page 127*

A terminal deletion (14)(q31.1) in a child with microcephaly, narrow palate, gingival hypertrophy, protuberant ears, and mild mental retardation F-S YEN, P E PODRUCH, AND B WEISSKOPF *page 130*

Familial distal trisomy 8(q24.13→qter) D R ROMAIN, R A BLOXHAM, L M COLUMBANO-GREEN, C J CHAPMAN, R G PARFITT, R H SMYTHE, AND H CAIRNEY *page 133*

Is there an autosomal recessive form of the split hand and split foot malformation? J ZLOTOGORA AND N NUBANI *page 138*

Conference report European Society for Human Genetics. Satellite meeting on meiotic microspreading A C CHANDLEY AND Y RUMPLER *page 141*

Book reviews *page 143*

Announcements *page 144*

ASTM CODEN: JMDGAE (26) 73-144 (1989) ISSN 0022-2593

British Medical Association Tavistock Square London WC1