



NLM 00599512 0

Contents

Editorial *page* 385

Syndrome of the month: Williams syndrome J BURN *page* 389

The frequency of the fragile X chromosome among schoolchildren in Coventry T P WEBB, S BUNDEY, A THAKE, AND J TODD *page* 396

Twelve families with fragile X(q27) T WEBB, A THAKE, AND J TODD *page* 400

Replication status of fragile X(q27-3) in 13 female heterozygotes E TUCKERMAN, T WEBB, AND A THAKE *page* 407

Mutations linked to the pro $\alpha 2(I)$ collagen gene are responsible for several cases of osteogenesis imperfecta type I G WALLIS, P BEIGHTON, C BOYD, AND C G MATHW *page* 411

A model system for the analysis of gene exclusion: cystic fibrosis and chromosome 19 B WAINWRIGHT, M FARRALL, E WATSON, AND R WILLIAMSON *page* 417

A new strategy for mapping the human genome D J SHAW *page* 421

Pseudoachondroplasia: clinical diagnosis at different ages and comparison of autosomal dominant and recessive types. A review of 32 patients (26 kindreds) R WYNNE-DAVIES, C M HALL, AND I D YOUNG *page* 425

Severe mental retardation in six generations of a large South African family carrying a translocation t(6;f0)(q27;q25-2) J BRUSNICKÝ, K M M VAN HEERDEN, G DE JONG, A S CRONJÉ, AND A E RETIEF *page* 435

Segregation of a t(3;20) translocation through three generations resulting in unbalanced karyotypes in six persons K B NIELSEN, N TOMMERUP, B JESPERSEN, P NYGAARD, AND L KLEIF *page* 446

The effects of severe mixed environmental pollution on human chromosomes A KATSANTONI, S NAKOU, I ANTONIADOU-KOUMATOU, AND G B CÔTÉ *page* 452

Pitfalls in prenatal diagnosis of β thalassaemia C ROSATELLI, I MACCIONI, M T SCALAS, AND A CAO *page* 456

Microtia with meatal atresia and conductive deafness: mild and severe manifestations within the same sibship P STRISCIUGLIO, A BALLABIO, AND G PARENTI *page* 459

Partial trisomy 7 (q32→qter) syndrome in two children D A COUZIN, N HAITES, J L WATT, AND A W JOHNSTON *page* 461

Three children with partial trisomy 1q and partial monosomy 3p G T MCCARTHY, C N FEAR, AND A C BERRY *page* 466

Abstracts of the meeting of the Clinical Genetics Society held at the University of Wales College of Medicine, Cardiff, on 18 and 19 April 1986 *page* 468

Case reports

A terminal deletion of the long arm of chromosome 4 [46.XX,dcl(4)(q33)] in an infant with phenotypic features of Williams syndrome R D JEFFERSON, J BURN, K L GAUNT, S HUNTER, AND E V DAVISON *page* 474

A de novo X:13 translocation with abnormal phenotype s V HODGSON, J C K BARBER, A DOWIE, AND V DUBOWITZ *page* 477

Terminal deletion of the long arm of chromosome 10 H CURTIS, R T HOWELL, AND C COPE *page* 478

Announcement *page* 480

ASTM COI

0022-2593

Britis

Tavistock Square London WC1