JOURNAL OF MEDICAL GENETICS

Diaphragmatic hernia in the south-west of England T. J. DAVID and CORINNE A. ILLINGWORTH 253

Congenital malformations associated with anencephaly and iniencephaly T. J. DAVID and ANGELA NIXON 263

Syndrome designations M. MICHAEL COHEN, Jr. 266

Probable common origin of a hereditary fundus dystrophy (Sorsby's familial pseudoinflammatory macular dystrophy) in an English and Australian family H. KALMUS and D. SEEDBURGH 271

Pfeiffer syndrome: report of a family and review of the literature YEHEZKEL NAVEH and ABRAHAM FRIEDMAN 277

Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome: dominant inheritance and variable expression VICTOR B. PENCHASZADEH and TERESA C. DE NEGROTTI 281

Chronic spinal muscular atrophy of facioscapulohumeral type TETSUO FURUKAWA and YASUO TOYO-KURA 285

Genetic analyses of pyloric stenosis suggesting a specific maternal effect K. K. KIDD and M. A. SPENCE 290

Chromosome survey of total population of mentally subnormal in North-East of Scotland R. M. SPEED, A. W. JOHNSTON, and H. J. EVANS 295

Alkaline phosphatase activity in cultured skin fibroblasts from fibrodysplasia ossificans progressiva LAN M. ARON, and 307

Topographic approach for analysis of palm crease variants HANNA DAR and R, SCHMIDT 310

Further studies on the genetic heterogeneity of cebocephaly G. I. LAZJUK, I. W. LURIE, and M. K. NEDZVED 314

Short communication:

X mapping in man: evidence against direct measurable linkage between ocular albinism and deutan colour blindness W. G. PEARCE and RUTH SANGER 319

Case reports:

Non-disjunction of an unusual X chromosome I. HAYATA, M. OSHIMURA, MICHELLE J. MARINELLO, R. M. BANNERMAN, and A. A. SANDBERG 320

Reciprocal translocation, 4q - ;21p + , giving rise to Down's syndrome M. B. JENKINS and L. BOYD 323

Multiple congenital defects associated with trisomy for long arm of No. 4 M. ISSA, A. M. POTTER, and C. E. BLANK 326

Tetraploidy in a liveborn infant MITCHELL S. GOLBUS, RONALD BACHMAN, SANDRA WILTSE, and BRYAN D. HALL 329

46,XY/46,XY,21q — mosaicism in an infant with neutropenia and properdin deficiency RICHARD L. NEU, JAMES A. STOCKMAN, III, ROGER E. SPITZER, and RUSSELL H. TOMAR 332

Book reviews 335

Announcement 336



ASTM CODEN: JMDGAE (13) 253-336 (1976)