Journal of Medical Genetics February 1980 Vol 17 No 1

Contents

Review article: Haems and chlorophylls: comparison of function and formation G A F HENDRY AND O T G JONES page 1

An overall genetic risk assessment for radiological protection purposes P OFTEDAL AND A G SEARLE page 15

Angiokeratoma corporis diffusum with features of a mucopolysaccharidosis D I MCCALLUM, R F MACADAM, AND A W JOHNSTON page 21

Inbreeding effects on fetal growth and development P S S RAO AND S G INBARAJ page 27

Sickle cell disease in Sicily e f roth, G schiliro, a russo, s musumeci, e rachmilewitz, v neske, and r nagel page 34

Dominant inheritance in a family with primary atrophic rhinitis J R SIBERT AND R P E BARTON page 39

Sex-influenced expression of Madelung's deformity in a family with dyschondrosteosis J R LICHTENSTEIN, M SUNDARAM, AND R BURDGE page 41

High resolution of a small pericentric inversion of chromosome 11 H AUTIO-HARMAINEN AND A DE LA CHAPELLE page 44

Trisomy 21 mosaicism in two successive generations in a family J C PARKE, F S GRASS, R PIXLEY, AND J DEAL page 48

Dermatoglyphic findings in Poland's syndrome M ATASU page 50

Frontometaphyseal dysplasia: autosomal dominant or X-linked? P BEIGHTON AND H HAMERSMA page 53

Ouestion and answer page 57

Case reports:

Agenesis of the corpus callosum with mosaicism 46,XY/47,XY, extra ring chromosome G ZAMBONI, F BERNARDI, C DANESINO, U MARCELLO DEL MAJNO, G BEGHINI, AND B DALLA BERNARDINA page 59

47,XXX chromosome constitution in a male u bigozzi, g simoni, e montali, l dalprà, f rossella, m piazzini, and a borghi page 62

Complex chromosomal rearrangement leading to partial trisomy 22 I L HANSTEEN, L SCHIRMER, S HESTETUN, AND A BRØGGER page 66

Concurrence of anorexia nervosa and yellow mutant albinism J T KELLY, J ROHDE, C J WITKOP, AND A JOHANNES page 68

Correspondence page 72

Book reviews page 77

ASTM CODEN: JMDGAE (17) 1-80 (1980)

British Medical Association Tavistock Square London WC1

Journal of Medical Genetics February 1980 Vol 17 No 1

Contents

Contents

Review article: Haems and chlorophylls: comparison of function and formation G A F HENDRY AND O T G JONES page 1

An overall genetic risk assessment for radiological protection purposes P OFTEDAL AND A G SEARLE page 15

Angiokeratoma corporis diffusum with features of a mucopolysaccharidosis D I MCCALLUM, R F MACADAM, AND JOHNSTON page 21

Sickle cell disease in Sicily E F ROTH, G SCHILIRO, A RUSSO, S MUSUMECI, E RACHMILEWITZ, V NESKE, AND R NAGEL page 27

Sex-influenced expression of Madelung's deformity in a family with dyschondrosteosis J R LICHTENSTEIN, M SUNDAR AM AND R BURDGE page 41

High resolution of a small pericentric inversion of chromosome 11 H AUTIO-HARMAINEN AND A DE LA CHAPELLE page 34

Trisomy 21 mosaicism in two successive generations in a family J C PARKE, F S GRASS, R PIXLEY, AND J DEAL page

Dermatoglyphic findings in Poland's syndrome M ATASU page 50

Frontometaphyseal dysplasia: autosomal dominant or X-linked? P BEIGHTON AND H HAMERSMA page 53

Question and answer page 57

Case reports:

Agenesis of the corpus callosum with mosaicism 46,XY/47,XY, extra ring chromosome G ZAMBONI, F BERNARDI, C DANESINO, U MARCELLO DEL MAJNO, G BEGHINI, AND B DALLA BERNARDINA page 59

47,XXX chromosome constitution in a male U BIGOZZI, G SIMONI, E MONTALI, L DALPRÀ, F ROSSELLA, M PIAZZINI. a borghi *page* 62

Complex chromosomal rearrangement leading to partial trisomy 22 I L HANSTEEN, L SCHIRMER, S HESTETUN, a brøgger page 66

Concurrence of anorexia nervosa and yellow mutant albinism J T KELLY, J ROHDE, C J WITKOP, AND A JOHANNES page 88.

Correspondence page 72

Book reviews page 77

ASTM CODEN: JMDGAE (17) 1–80 (1980)

British Medical Association Tavistock Square London WC1 **British Medical Association Tavistock Square London WC1**

lished

Contents

Contents

Contents

Contents

Contents

Contents

Contents

Contents

Contents

Amniocentesis in the West Midlands: report on 1000 births T webb, J H EDWARDS, A H CAMERON, J M CRAWLEY, M HULTEN D I RUSHTON, AND R A THOMPSON page 81

Risk of recurrence after two children with central nervous system malformations in an area of high incidence N C NEVING AND W P JOHNSTON page 87 AND W P JOHNSTON page 87

Concordance rates in twins for an encephaly W H JAMES page 93

Excess of cancer deaths in grandparents of patients with retinoblastoma C Bonaiti-Pellié and M L Briard-Guillemoting page 95

A family and population study of the genetic polymorphism of debrisoquine oxidation in a white British population D A PRICE EVANS, A MAHGOUB, T P SLOAN, J R IDLE, AND R L SMITH page 102

No sex difference in mutation rates of Duchenne muscular dystrophy N YASUDA AND K KONDÔ page 106

Parental factors associated with rigidity in Huntington's disease C J BRACKENRIDGE page 112

Familial cerebellar ataxia presenting with down beat nystagmus G D SCHOTT page 115

Syndrome of polydactyly, cleft lip/palate or lingual lump, and psychomotor retardation in endogamic gypsies V VARADI, L SZABÓ, AND Z PAPP page 119

Recessive aplasia cutis congenita of the limbs N FREIRE-MAIA, M PINHEIRO, AND C C ORTEGA page 123

An epidemiological study of facial clefting in Manitoba J WELCH AND A G W HUNTER page 127 Excess of cancer deaths in grandparents of patients with retinoblastoma C BONAÏTI-PELLIÉ AND M L BRIARD-GUILLEMOT_

An epidemiological study of facial clefting in Manitoba J WELCH AND A G W HUNTER page 127

Partial anomalous pulmonary venous drainage in two patients with Turner's syndrome W H PRICE AND R F WILL page 133

Diaphragmatic hernia in Avon T J DAVID, V M PARKER, AND C A ILLINGWORTH page 135

Monozygotic twins discordant for Wiedemann-Beckwith syndrome and the implications for genetic counselling A C BERRY, E M BELTON, AND C CHANTLER page 136

A family study of bladder exstrophy E IVES, R COFFEY, AND C O CARTER page 139

Dermatoglyphs and chromosome mosaicism in parents of children with trisomy 18 M HOLMES-SIEDLE, S KERR, R LINDENBAUM, AND M BOBROW page 142

Case reports:

Dic(21:21) in a Down's syndrome child with an unusual chromosome 9 variant in the mother J M BERG, H A GARDNER. R J M GARDNER, E G GOH, V D MARKOVIĆ, N E SIMPSON, AND R G WORTON page 144

Ring chromosome 10: $46.XX.r(10)(p15\rightarrow q26)$ r tsukino, n tsuda, t dezawa, t ishii, and m koike page 148

Ring chromosome 10: 46,XX,r(10)(p15 \rightarrow q26) r tsukino, n tsuda, t dezawa, t ishii, and m koike page 148

Hypomelanosis of Ito with triphalangeal thumbs m k kukolich, b w althaus, m v r freeman, and r c lewandowski $\stackrel{>}{\leftarrow}$ page 151

A case of Klinefelter's syndrome with 47,Xi(Xq)Y karyotype G PONZIO, M DEMARCHI, G GALLONE, D FONZO, AND A O CARBONARA page 152

Announcements page 155

Papers presented at the Clinical Genetics Society meeting, 15–16 November 1979 page 156

Correspondence page 157

Book reviews page 159

ASTM CODEN: JMDGAE (17) 81–160 (1980)

British Medical Association Tavistock Square London WC1

British Medical Association Tavistock Square London WC1

19 δ

Contents

Screening of thalassaemia carriers in intermediate schools in Latium: results of four years' work E SILVESTRONI, I BIANCO, B GRAZIANI, C CARBONI, M VALENTE, M LERONE, AND S U D'ARCA page 161

Carrier detection in Duchenne muscular dystrophy J s fitzsimmons, J I mclachlan, w g reeves, D w marriott, a m J woolfson, and J mayhew page 165

Antiproteases and Down's syndrome in an Australian population H MCPHEE, R ANANTHAKRISHNAN, AND L I TAFT page 170

The syndromes of Marshall and Weaver N FITCH page 174

Haemoglobin Lepore Boston-Washington in Sicily: clinical, haematological, and biosynthetic studies G SCHILIRO, S MUSUMECI, G PIZZARELLI, A FISCHER, M A ROMEO, AND G RUSSO page 179

Haemoglobin K Woolwich: a study of the family of a homozygote R CABANNES, P AMEGNIZIN, A SANGARE, D ARNE, R CASEY, AND H LEHMANN page 183

Alkaline phosphatase activity of normal and cystic fibrosis fibroblasts D A AITKEN AND A HOOGEVEEN page 187

G6PD (Dublin): chronic non-spherocytic haemolytic anaemia resulting from glucose-6-phosphate dehydrogenase deficiency in an Irish kindred s R MCCANN, A M SMITHWICK, I J TEMPERLEY, AND K TIPTON page 191

Dermatoglyphs in duplication of the thumb H SHIONO AND T OGINO page 194

Huntington's disease in two New Britain families E M SCRIMGEOUR page 197

A family study of spina bifida and anencephalus in Belfast, Northern Ireland (1964–1968) N C NEVIN AND W P JOHNSTON page 203

Ichthyosis, hepatosplenomegaly, and cerebellar degeneration in a sibship P S HARPER, R MARKS, P J DYKES, AND I D YOUNG page 212

Gonadal dysgenesis in a patient with an X;3 translocation: case report and review N J CARPENTER, B SAY, AND D BROWN-ING page 216

Familial pericentric inversion 19 D K JORDAN, K TAYSI, AND N L BLACKWELL page 222

Occasional abstract: Growing embryos in vitro, with special reference to congenital abnormalities in the offspring of mothers with diabetes mellitus C A CLARKE page 226

Case reports:

Tetrasomy 9p: confirmation by enzyme analysis s J MOEDJONO, B F CRANDALL, AND R S SPARKES page 227

A severely retarded 18-year-old boy with tertiary partial trisomy 14 A SMITH, G DEN DULK, AND G ELLIOTT page 230

Partial trisomy of the short arm of chromosome 8 resulting from balanced maternal translocation L A Jones, D R DENGLER, K TAYSI, G D SHACKELFORD, AND A F HARTMANN page 232

Aarskog's syndrome with Hirschsprung's disease, midgut malrotation, and dental anomalies D D HASSINGER, J J MULVIHILL, AND J B CHANDLER page 235

A digitopalatal syndrome with associated anomalies of the heart, face, and skeleton R E STEVENSON, H A TAYLOR, O M BURTON, AND H B HEARN page 238

Correspondence page 243

Announcements page 244

ASTM CODEN: JMDGAE (17) 161-244 (1980)

British Medical Association Tavistock Square London WC1

Contents

Incidence of Duchenne muscular dystrophy in New South Wales and the Australian Capital Territory J COWAJ MACDESSI, A STARK, AND G MORGAN page 245

Pitfalls of genetic counselling in Pfeiffer's syndrome M BARAITSER, M BOWEN-BRAVERY, AND P SALDANA-GARCIA page 2.

Menkes X linked disease: heterozygous phenotype in uncloned fibroblast cultures N HORN page 257

Menkes X linked disease: two clonal cell populations in heterozygotes N HORN, P MOOY, AND V M MCGUIRE page 262

Increase in the amount of fetal lymphocytes in maternal blood during pregnancy M KIRSCH-VOLDERS, E LISSENS-VA ASSCHE, AND C SUSANNE page 267

Estimates of the likelihood that a Down's syndrome child of unknown genotype is a consequence of an inherited translocation s G ALBRIGHT AND E B HOOK page 273

A family with diaphyseal aclasis and peripheral dysostosis A P BROOKS AND R WYNNE-DAVIES page 277

Negro α-thalassaemia: genetic studies in homozygous sickle cell disease G R SERJEANT, K P MASON, AND B E SERJEANT page 281

Haemoglobin E trait and probable α-thalassaemia in a black American family: a family study A F EL-SHIRBINY, of PARKHURST, R E BETTIGOLE, AND K D TOURBAF page 285

Recessively inherited growth hormone deficiency in a family from Iraq M D C DONALDSON, S M TUCKER, AND D B GRANT page 288

Female phenotype and multiple abnormalities in sibs with a Y chromosome and partial X chromosome duplication; H-Y antigen and Xg blood group findings R BERNSTEIN, T JENKINS, B DAWSON, J WAGNER, G DEWALD, G C KOO, AND S S WACHTEL page 291

Dermatoglyphs in carriers of a balanced 15;21 translocation a RODEWALD, M ZANKL, H ZANKL, AND K D ZANG page 36

Segregation of an X ring chromosome in two generations B DALLAPICCOLA, L BRUNI, B BOSCHERINI, A M PASQUING L CHESSA, AND P VIGNETTI page 306

Pericentric inversion (13) with two different recombinants in the same family E M WILLIAMSON, J F MILLER, AND M SEABRIGHT page 309

Review of 'The provision of services for the prenatal diagnosis of fetal abnormality in the United Kingdom'. Report of the Clinical Genetics Society Working Party. Supplement No 3 to the Bulletin of the Eugenics Society, November 1978.

J A RAEBURN page 313

Case reports:

Prenatal diagnosis of a de novo non-fluorescent Y chromosome J H PRIEST, A T L CHEN, P M FERNHOFF, J A REIDY, AN C WHITSETT page 314

13q-/r(13) mosaicism n niikawa, t tamura, f tomiyasu, and t kajii page 316

45,X/46,XY/47,XY, +21 mosaicism in a hypogonadal phenotypic male M sparagana, P W K wong, T R dorsci $\frac{G}{T}$ C casten, M rauer, and K szego page 319

Gonadal dysgenesis in a 46,XY female mosaic for double autosomal trisomies 8 and 21 JM SULEWSKI, T P DANG, S WARI AND R L LADDA page 321

Correspondence page 324

Announcements page 328

ASTM CODEN: JMDGAE (17) 245-328 (1980)

British Medical Association Tavistock Square London WC1

jmag.brogi.comayon Agopsi 19, 2024 by guest-Protected by copy

Contents

Genetic aspects of hereditary motor and sensory neuropathy (types I and II) A E HARDING AND P K THOMAS page 329

Linkage and association between HLA and 21-hydroxylase deficiency P T KLOUDA, R HARRIS, AND D A PRICE page 337

Genetic inheritance of susceptibility to tinea imbricata D RAVINE, K J TURNER, AND M P ALPERS page 342

Ellis-van Creveld syndrome: report of 15 cases in an inbred kindred E OLIVEIRA DA SILVA, D JANOVITZ, AND S CAVALCANTI DE ALBUQUERQUE page 349

α-thalassaemia in Sardinian infants R GALANELLO, G DIANA, M FURBETTA, A ANGIUS, M A MELIS, C ROSATELLI, AND A CAO

Origin of the additional chromosome in Down's syndrome: a study of 20 families D F ROBERTS AND M H CALLOW page 363

Maternal age and origin of non-disjunction in trisomy 21 J F MATTEI, S AYME, M G MATTEI, AND F GIRAUD page 368

Clinical manifestations of familial 13;18 translocation W A BLATTNER, M L KISTENMACHER, S TSAI, H H PUNNETT, AND ER GIBLETT page 373

New C band markers of human chromosomes: C band position variants R B PHILLIPS page 380

Occasional abstract: Recent views on genetic factors in retinoblastoma s BUNDEY page 386

Technical note: Culture of bloody amniotic fluid for chromosome analysis: an improved method N M GREGSON AND M JOHNSON page 388

Case reports:

A liveborn case of 49,XXXY,+18 N B KARDON, A L BERGER, M ELICE, J G DAVIS, AND E C JENKINS page 389

Proximal femoral focal deficiency associated with the Robin anomalad E R GRAVISS, P A MONTELEONE, L R WAMPLER, M J SILBERSTEIN, AND A E BRODEUR page 390

The penta-X syndrome A MONHEIT, U FRANCKE, B SAUNDERS, AND K L JONES page 392

A case of 47,XY,+der(15),t(3:15)(p25:q11)pat presenting as partial 3p trisomy syndrome with multiple joint con-

A case of 47,XY, +der(15),t(3;15)(p25;q11)pat presenting as partial 3p trisomy syndrome with multiple joint contractures J H HERSH, R M GREENSTEIN, J C PERKINS, AND P C REARDON page 396

De novo duplication 1q32-q42: variability of phenotypic features in partial 1q trisomics M S LUNGAROTTI, A FALORNI A CALABRO, F PASSALACQUA, AND B DALLAPICCOLA page 398

Papers presented at the Clinical Genetics Society meeting, 24-25 April 1980 page 403

Correspondence page 404

Book reviews page 406

Announcement page 408

ASTM CODEN: JMDGAE (17) 329-408 (1980)

British Medical Association Tayistock Square London WC1

Journal of Medical Genetics December 1980 Vol 17 No 6

Contents

Editorial page 409

Obituary page 410

The grandchildren of patients with pyloric stenosis c o carter, k evans, and J warren page 411

Genetic and clinical patterns of heritable cerebellar ataxias in adults. I Genetic analyses K KONDO AND I SOBUE page 416

Autosomal recessive peripheral sensory neuropathy in 3 non-Ashkenazi Jewish families I TAMARI, R M GOODMAN, I SAROVA, M HERTZ, R ADAR, AND T ZVIBACH page 424

The Cohen syndrome: clinical and endocrinological studies of two new cases P BALESTRAZZI, L CORRINI, G VILLANI, M P BOLLA F CASA, AND S BERNASCONI page 430

Down syndrome and maternal age in South Glamorgan I D YOUNG, E M WILLIAMS, AND R G NEWCOMBE page 433

X; Y translocation in an adolescent mentally normal phenotypic male with features of hypogonadism R BERNSTEIN, MR PINTO M ALMEIDA, S M SOLARSH, J MECK, AND T JENKINS page 437

A familial polymorphic variant of chromosome 5 M SEABRIGHT, N M GREGSON, AND M JOHNSON page 444

Location of the gene for 21-hydroxylase deficiency V PUCHOLT, J S FITZSIMMONS, K GELSTHORPE, M A REYNOLDS, AND R D

Cytogenetic and clinical studies in gonadal dysgenesis with 46,X,Xt(qter->p221::p223->qter) karyotype: review and phenotype/karyotype correlations M FERRARO, A DE CAPOA, C MOSTACCI, F PELLICCIA, P ZULLI, M A BALDINI, AND Q DI NISIO page 457

A family segregating for E_1^I and E_2^K at cholinesterase locus 1 RT EVANS, JIOBAL, A A DIETZ, T LUBRANO, AND H M RUBINSTEIN

Sequential staining of euchromatic and heterochromatic regions of the human Y chromosome v J GOYANES page 468

A cryopreservative procedure for storing cultivated and uncultivated amniotic fluid cells in liquid nitrogen S PENTZ AND H HÖRLER page 472

Question and answer page 476

Case reports:

Agenesis of the lung associated with a chromosome abnormality (46,XX,2p+) B SAY, N J CARPENTER, G GIACOIA, AND S JEGATHESAN page 477

A probable case of the homozygous condition of the aniridia gene s v hodgson and k e saunders page 478

Craniosynostosis and syndactyly: expanding the 11 q - chromosomal deletion phenotype B M LIPPE, R S SPARKES, B FASS, AND L NEIDENGARD page 480

Interstitial deletion in the long arms of chromosome 1: 46,XY,del(1)(pter→q22::q25→qter) c estévez de Pablo, J M GARCÍA SAGREDO, M T FERRO, P FERRANDO, AND C SAN ROMÁN page 483

Interstitial deletion of the long arm of chromosome 5 in a deformed boy: 46,XY,del(5)(q13q15) c stoll, J-M LEVY, AND M-1 ROTH page 486

Neurological and neuropathological findings in ring chromosome 4 R S K YOUNG AND E L ZALNERAITIS page 487

Correspondence page 491

Announcement page 492

Index page 493

ASTM CODEN: JMDGAE (17) 409-505 (1980)

British Medical Association Tavistock Square London WC1