

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*
- 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, 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

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 SUNDARA, 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 49*
- 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, 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*

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 NEVIN AND W P JOHNSTON *page 87*

Concordance rates in twins for anencephaly W H JAMES *page 93*

Excess of cancer deaths in grandparents of patients with retinoblastoma C BONAÏTI-PELLIÉ AND M L BRIARD-GUILLEMOU *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 VÁRADY, 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*

Partial anomalous pulmonary venous drainage in two patients with Turner's syndrome W H PRICE AND R F WILLEY *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 H 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→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 *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

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 BROWNING *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 DENGLE, 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 COWAN, J 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 250*
- 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 261*
- Increase in the amount of fetal lymphocytes in maternal blood during pregnancy M KIRSCH-VOLDERS, E LISSENS-VAN 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, 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 300*
- Segregation of an X ring chromosome in two generations B DALLAPICCOLA, L BRUNI, B BOSCHERINI, A M PASQUINO, 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 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 RAEURN *page 313*
- Case reports:
- Prenatal diagnosis of a de novo non-fluorescent Y chromosome J H PRIEST, A T L CHEN, P M FERNOFF, J A REIDY, AND C WHITSETT *page 314*
- 13q-/r(13) mosaicism N NIKAWA, 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 DORSCH, C CASTEN, M RAUER, AND K SZEGO *page 319*
- Gonadal dysgenesis in a 46,XY female mosaic for double autosomal trisomies 8 and 21 J M SULEWSKI, T P DANG, S WARD, 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

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 *page* 357

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 E R 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 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 Tavistock Square London WC1

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, M R 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 G MILNER *page* 447

Selective IgA deficiency with 18q+ and 18q- karyotypic anomalies R M LEWKONIA, C C LIN, AND R H A HASLAM *page* 453

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 Eⁱ and E^k at cholinesterase locus 1 R T EVANS, J IQBAL, A A DIETZ, T LUBRANO, AND H M RUBINSTEIN *page* 464

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 11q- 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-P 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