

## Contents

Feasibility of neonatal screening for Duchenne muscular dystrophy R SKINNER, A E H EMERY, G SCHEUERBRANDT, AND J SYME *page 1*

Effect of exercise on serum creatine kinase in carriers of Duchenne muscular dystrophy R F GAINES, S M PUESCHEL, E A SASSAMAN, AND J L DRISCOLL *page 4*

An epidemiological and genetic study of facial clefting in France. I Epidemiology and frequency in relatives C BONAÏTI, M L BRIARD, J FEINGOLD, B PAVY, J PSAUME, G MIGNE-TUFFERAUD, AND J KAPLAN *page 8*

Survivors of neuroblastoma and ganglioneuroma and their families S BUNDEY AND K EVANS *page 16*

Apnoea following suxamethonium: the genetic study of four generations of a family A A MORGAN *page 22*

Genetic counselling in haemophilia by discriminant analysis 1975-1980 E S BARROW, C H MILLER, H M REISNER, AND J B GRAHAM *page 26*

Genetic aspects of fibrodysplasia ossificans progressiva J M CONNOR AND D A P EVANS *page 35*

A new camptodactyly syndrome M BARAITSER *page 40*

Fragile (X)(q27) sites in a pedigree with female carriers showing mild to severe mental retardation G C WEBB, J L HALLIDAY, D B PITT, C G JUDGE, AND M LEVERSHA *page 44*

Cytogenetic and histological studies of testicular biopsies from subfertile men with chromosome anomaly M J W FAED, M A LAMONT, AND K BAXBY *page 49*

Chromosome distribution studies in XXY karyotypes M KIRSCH-VOLDERS, L HENS, H VAN DEN BERGHE, B SCHOLBERG, AND C SUSANNE *page 57*

Clinical Genetics Society. Abstracts of scientific papers presented on 9 and 10 April 1981 at Sheffield *page 63*

### Case reports

De novo interstitial deletion in the long arm of chromosome 9: a new chromosome syndrome K L YING, C J R CURRY, K B RAJANI, S H KASSEL, AND R S SPARKES *page 68*

A malformed baby with two separate de novo translocations W E CHEWINGS, T P COCKS, R J M GARDNER, AND J E CLARKSON *page 70*

A second patient with partial deletion of the short arm of chromosome 3: karyotype 46,XY,del(3)(p25) M C HIGGIN-BOTTOM, J T MASCARELLO, H HASSIN, AND W K MCCORD *page 71*

Chronic renal disease, myotonic dystrophy, and gonadoblastoma in XY gonadal dysgenesis J L SIMPSON, R S K CHAGANTI, J MOURADIAN, AND J GERMAN *page 73*

### Short reports

A severely retarded male with deletion of chromosomes 15 (pter→q13) and 10 (q26→qter) A SMITH AND G DEN DULK *page 77*

Alobar holoprosencephaly and otocephaly in a female infant with a normal karyotype and placental villitis A R GABA, G J ANDERSON, D L VANDYKE, AND J L CHASON *page 78*

Correspondence *page 79*

Announcements *page 80*

ASTM CODEN: JMDGAE (19) 1-80 (1982)

**British Medical Association Tavistock Square London WC1**

# CONTENTS

No 1 *FEBRUARY* 1982

Feasibility of neonatal screening for Duchenne muscular dystrophy R SKINNER, A E H EMERY, G SCHEUERBRANDT, AND J SYME .. .. .	1
Effect of exercise on serum creatine kinase in carriers of Duchenne muscular dystrophy R F GAINES, S M PUESCHEL, E A SASSAMAN, AND J L DRISCOLL .. .. .	4
An epidemiological and genetic study of facial clefting in France. I Epidemiology and frequency in relatives C BONAITI, M L BRIARD, J FEINGOLD, B PAVY, J PSAUME, G MIGNE-TUFFERAUD, AND J KAPLAN .. .. .	8
Survivors of neuroblastoma and ganglioneuroma and their families S BUNDEY AND K EVANS .. .. .	16
Apnoea following suxamethonium: the genetic study of four generations of a family A A MORGAN .. .. .	22
Genetic counselling in haemophilia by discriminant analysis 1975-1980 E S BARROW, C H MILLER, H M REISNER, AND J B GRAHAM .. .. .	26
Genetic aspects of fibrodysplasia ossificans progressiva J M CONNOR AND D A P EVANS .. .. .	35
A new camptodactyly syndrome M BARAITSER .. .. .	40
Fragile (X)(q27) sites in a pedigree with female carriers showing mild to severe mental retardation G C WEBB, J L HALLIDAY, D B PITT, C G JUDGE, AND M LEVERSHA .. .. .	44
Cytogenetic and histological studies of testicular biopsies from subfertile men with chromosome anomaly M J W FAED, M A LAMONT, AND K BAXBY .. .. .	49
Chromosome distribution studies in XXY karyotypes M KIRSCH-VOLDERS, L HENS, H VAN DEN BERGHE, B SCHOLBERG, AND C SUSANNE .. .. .	57
Clinical Genetics Society. Abstracts of scientific papers presented on 9 and 10 April 1981 at Sheffield .. .. .	63
<b>Case reports</b>	
De novo interstitial deletion in the long arm of chromosome 9: a new chromosome syndrome K L YING, C J R CURRY, K B RAJANI, S H KASSEL, AND R S SPARKES .. .. .	68
A malformed baby with two separate de novo translocations W E CHEWINGS, T P COCKS, R J M GARDNER, AND J E CLARKSON .. .. .	70
A second patient with partial deletion of the short arm of chromosome 3: karyotype 46,XY, del(3)(p25) M C HIGGINBOTTOM, J T MASCARELLO, H HASSIN, AND W K MCCORD .. .. .	71
Chronic renal disease, myotonic dystrophy, and gonadoblastoma in XY gonadal dysgenesis J L SIMPSON, R S K CHAGANTI, J MOURADIAN, AND J GERMAN .. .. .	73
<b>Short reports</b>	
A severely retarded male with deletion of chromosomes 15 (pter→q13) and 10 (q26→qter) A SMITH AND G DEN DULK .. .. .	77
Alobar holoprosencephaly and otocephaly in a female infant with a normal karyotype and placental villitis A R GABA, G J ANDERSON, D L VANDYKE, AND J L CHASON .. .. .	78
Correspondence .. .. .	79
Announcements .. .. .	80

No 2 *APRIL* 1982

Haematological and obstetric aspects of antenatal diagnosis of $\beta$ -thalassaemia: experience with 200 cases A CAO, M FURBETTA, A ANGIUS, A XIMENES, C ROSATELLI, T TUVERI, M T SCALAS, A M FALCHI, G ANGIONI, AND F CAMINITI .. .. .	81
A family study of Charcot-Marie-Tooth disease A P BROOKS AND A E H EMERY .. .. .	88
The high frequency of juvenile Huntington's chorea in South Africa M R HAYDEN, J M MACGREGOR, D S SAFFER, AND P H BEIGHTON .. .. .	94
Serum gonadotrophins in Down's syndrome W A CAMPBELL, J LOWTHER, I MCKENZIE, AND W H PRICE .. .. .	98
Bone demineralisation in patients with Turner's syndrome M A SMITH, J WILSON, AND W H PRICE .. .. .	100
A new variant of spondylometaphyseal dysplasia with autosomal dominant mode of inheritance J M GARCÍA-CASTRO, C M ISALES-FORSYTHE, AND P DÍAZ DE GARAU .. .. .	104

Autosomal dominant asymmetrical radial dysplasia, dysmorphic facies, and conductive hearing loss (facioauricular radial dysplasia) A E HARDING, C M HALL, AND M BARAITSER . . . . .	110
Roberts syndrome: clinical and cytogenetic aspects N P MANN, J FITZSIMMONS, AND P COOKE . . . . .	116
Family studies on the chromosomal location of the retinoblastoma gene (Rb-1) J E N MORTEN, D G HARNDEN, AND S BUNDEY . . . . .	120
Chiasma derived genetic maps and recombination fractions: chromosome 13 with reference to the proposed 13q14 retinoblastoma locus R W PALMER AND M A HULTÉN . . . . .	125
Autosomal recessive Klippel-Feil syndrome E OLIVEIRA DA SILVA . . . . .	130
Apparent enhanced response to the induction of sister chromatid exchange by mitomycin C in myotonic dystrophy VIJAYALAXMI, A E H EMERY, AND H J EVANS . . . . .	135
Marshall/Stickler syndrome M BARAITSER . . . . .	139

Case reports

Parental consanguinity and the Majewski syndrome I L BLACK, J FITZSIMMONS, E FITZSIMMONS, AND A J THOMAS . . . . .	141
Down's syndrome phenotype and autosomal gene inactivation in a child with presumed (X;21) de novo translocation K TAYSI, R S SPARKES, T J O'BRIEN, AND D R DENGLER . . . . .	144
A case of inverted insertion assessed by R and G banding M A DE ARCE, E LAW, L MARTIN, AND J G MASTERTON . . . . .	148
Familial pericentric inversion of chromosome 11 detected prenatally M H RUSSELL, P MILLER, AND A KILLAM . . . . .	151
Two pericentric inversions, inv(2)(p11q13) and inv(5)(p13q13), in a patient referred for psychiatric problems D R ROMAIN, C J CHAPMAN, L COLUMBANO-GREEN, R H SMYTHE, AND O GEBBIE . . . . .	153
Partial deletion of the long arm of chromosome 4: a clinical syndrome A LIPSON, J COLLIS, AND C GREEN . . . . .	155
Correspondence . . . . .	158
Book reviews . . . . .	159

No 3 JUNE 1982

Review article The mutation rate to Huntington's chorea M SHAW AND A CARO . . . . .	161
Sensitivity to ionising radiation of lymphocytes from Huntington's chorea patients compared to controls D MCGOVERN AND T WEBB . . . . .	168
Isozyme patterns and protein profiles in neuromuscular disorders Y H EDWARDS, T D TIPLER, J A MORGAN-HUGHES, J S NEERUNJUN, AND D A HOPKINSON . . . . .	175
$\delta\beta$ (F)-thalassaemia in Sardinia A CAO, M A MELIS, R GALANELLO, A ANGIUS, M FURBETTA, P GIORDANO, AND L F BERNINI . . . . .	184
Familial polyposis coli and its extracolonic manifestations S B COHEN . . . . .	193
Alteration of NADH-diaphorase and cytochrome <i>b<sub>5</sub></i> reductase activities of erythrocytes, platelets, and leucocytes in hereditary methaemoglobinaemia with and without mental retardation M TAKESHITA, T MATSUKI, K TANISHIMA, T YUBISUI, Y YONEYAMA, K KURATA, N HARA, AND T IGARASHI . . . . .	204
Two first cousins with spondyloepiphyseal dysplasia tarda (X linked recessive form), one also with poikiloderma atrophicans vasculare progressing to lymphocytic lymphoma W A BRANFORD, G W BEVERIDGE, AND R WYNNE-DAVIES . . . . .	210
Familial occurrence of a syndrome with branchial dysplasia, mental deficiency, club feet, and inguinal herniae J C LAMBERT, N AYRAUD, J MARTIN, R MARIANI, M FERRARI, AND M DONZEAU . . . . .	214
Nomogram for estimating specific consanguinity risks R CRUZ-COKE . . . . .	216
Leser-Trelat sign in mother and daughter with breast cancer H T LYNCH, R M FUSARO, J A PESTER, AND J F LYNCH . . . . .	218

Case reports

Inherited partial X chromosome duplication in a mentally retarded male K BRØNDUM NIELSEN AND F LANGKJÆR . . . . .	222
A complex chromosome rearrangement resulting in trisomy 15q22→qter P N HOWARD-PEEBLES, P R SCARBROUGH, J SHARPE, W H FINLEY, AND S C FINLEY . . . . .	224
Familial pericentric inversion of chromosome 13 resulting in duplication 13q22→qter M HABEDANK . . . . .	227
Two Robertsonian translocations in a boy with mental retardation E LIEBER AND P SHAH . . . . .	229

Autoimmune chronic active hepatitis in Down's syndrome	A J MCCULLOCH, P G INCE, AND P KENDALL-TAYLOR	232
An adult female with spondyloepiphyseal dysplasia tarda	J M CONNOR, D A P EVANS, AND I B SARDHARWALLA	234
Book reviews		237

No 4 *AUGUST* 1982

A genetic register for Huntington's chorea in South Wales	P S HARPER, A TYLER, S SMITH, P JONES, R G NEWCOMBE, AND V MCBROOM	241
A three generation family study of cleft lip with or without cleft palate	C O CARTER, K EVANS, R COFFEY, AND J A FRASER ROBERTS, A BUCK, M FRASER ROBERTS	246
Measurement of erythrocyte membrane elasticity as a diagnostic aid in Duchenne muscular dystrophy	G B NASH AND S J WYARD	262
Linkage analysis of five pedigrees affected with typical autosomal dominant retinitis pigmentosa	L L FIELD, J R HECKENLIVELY, R S SPARKES, C A GARCIA, C FARSON, D ZEDALIS, M C SPARKES, M CRIST, S TIDEMAN, AND M A SPENCE	266
Screening for latent acute intermittent porphyria: the value of measuring both leucocyte-aminolaevulinic acid synthase and erythrocyte uroporphyrinogen-l-synthase activities	K E L MCCOLL, M R MOORE, G G THOMPSON, AND A GOLDBERG	271
The incidence of Down's syndrome in Nigeria	A A ADEYOKUNNU	277
A family study of craniosynostosis, with probable recognition of a distinct syndrome	C O CARTER, K TILL, V FRASER, AND R COFFEY	280
An unusual form of familial acrocephalosyndactyly	I D YOUNG AND P S HARPER	286
Pfeiffer's type of acrocephalosyndactyly in two families	J VANĚK AND F LOŠAN	289
Familial Poland anomaly	T J DAVID	293
Human chromosomal heteromorphisms in American blacks. VI. Higher incidence of longer Y owing to non-fluorescent (nf) segment	R S VERMA, M EVANS-MCCALLA, AND H DOSIK	297

Case reports

The Johanson-Blizzard syndrome	M BARAITSER AND S V HODGSON	302
Pericentric inversion of chromosome 1 in an azoospermic man	A TÓTH, M GAÁL, G SÁRA, AND J LÁSZLÓ	303
X long arm deletion with oligomenorrhoea	K MIJIN, E STOLEVIĆ, S ADŽIĆ, Z LAĆA, AND S MARKOVIĆ	305
Cd banding studies in a homologous Robertsonian 13;13 translocation	D R ROMAIN, L COLUMBANO-GREEN, J SULLIVAN, R H SMYTHE, O GEBBIE, R PARFITT, AND C CHAPMAN	306

Short reports

Micromelia, polysyndactyly, multiple malformations, and fragile bones in a stillborn child	B F CARPENTER AND A G W HUNTER	311
Pericentric inversion of chromosome 11 in one of two similar retarded brothers	S L EINFELD AND A SMITH	312
Two successive partial trisomies for opposite halves of chromosome 22 in a mother with a balanced translocation	R P BENDEL, S BALDINGER, C MILLARD, AND D C ARTHUR	313
Adjacent 2 translocation involving 13q and 21q	W W WOOLF, C L BRADSHAW, H E HOYME, K L JONES, AND O W JONES	314
Correspondence		316
Announcements		319

No 5 *OCTOBER* 1982

Aetiology and interrelationship of some common skeletal deformities	R WYNNE-DAVIES, A LITTLEJOHN, AND J GORMLEY	321
A family study of isolated cleft palate	C O CARTER, K EVANS, R COFFEY, AND J A FRASER ROBERTS, A BUCK, M FRASER ROBERTS	329
Keratoconus posticus circumscriptus, cleft lip and palate, genitourinary abnormalities, short stature, and mental retardation in sibs	I D YOUNG, W G MACRAE, H E HUGHES, AND J S CRAWFORD	332

Hereditary twenty-nail dystrophy in a Sicilian family L PAVONE, S LI VOLTI, B GUARNERI, M LA ROSA, G SORGE, G INCORPORA, AND F MOLLIKA .. .. .	337
Clinical evidence for heterogeneity in myotonic dystrophy S BUNDEY .. .. .	341
DHT-receptor in cultured human fibroblasts: binding study in a family with androgen insensitivity (complete testicular feminisation) E DONTI, I NICOLETTI, P FILIPPONI, G VENTI, V BOCCHINI, AND F SANTEUSANIO .. .. .	349
The effect of minocycline on potassium leakage from red cells: a study of the genetics and relationship to vestibular adverse reactions B G LANNIGAN AND D A P EVANS .. .. .	354
Fraser syndrome presenting as bilateral renal agenesis in three sibs J BURN AND R P MARWOOD .. .. .	360
Two families with the Li-Fraumeni cancer family syndrome A D J PEARSON, A W CRAFT, J M RATCLIFFE, J M BIRCH, P MORRIS-JONES, AND D F ROBERTS .. .. .	362
Translocation 21q22q in an infertile human male A C CHANDLEY, T B HARGREAVE, AND J M FLETCHER .. .. .	366
Clinical Genetics Society. Abstracts of scientific papers presented on 5 and 6 November 1981 at the Institute of Child Health, London .. .. .	370

Case reports

A complex rearrangement involving three autosomes in a phenotypically normal male presenting with sterility A JOSEPH AND I M THOMAS .. .. .	375
Yq- in a child with livedo reticularis, snub nose, microcephaly, and profound mental retardation P E PODRUCH, F-S YEN, N D DINNO, AND B WEISSKOPF .. .. .	377
Gross congenital abnormality associated with an apparently balanced chromosomal translocation t(9;17)(q34;q11) H E DOCKERY, H C NEALE, AND P H FITZGERALD .. .. .	380
Report of a new case and clinical delineation of mosaic trisomy 9 syndrome J M SÁNCHEZ, N FIJTMAN, AND A M MIGLIORINI .. .. .	384
The hypertelorism microtia clefting syndrome M BARAITSER .. .. .	387
Congenital universal alopecia, mental deficiency, and microcephaly in two sibs R A PFEIFFER AND J VÖLKLEIN .. .. .	388
Correspondence .. .. .	390
Book reviews .. .. .	395
Announcement .. .. .	400

No 6 DECEMBER 1982

A clinical and genetic study of Hunter's syndrome. 1 Heterogeneity I D YOUNG, P S HARPER, I M ARCHER, AND R G NEWCOMBE .. .. .	401
A clinical and genetic study of Hunter's syndrome. 2 Differences between the mild and severe forms I D YOUNG, P S HARPER, R G NEWCOMBE, AND I M ARCHER .. .. .	408
Spectrum of anomalies in Fanconi anaemia A GLANZ AND F C FRASER .. .. .	412
Genetics of the apolipoprotein-E isoprotein system in man A M CUMMING AND F W ROBERTSON .. .. .	417
Genetics of plasma paroxonase activity L ISELIUS, D A P EVANS, AND J R PLAYFER .. .. .	424
Lymphocyte interphase chromatin in healthy subjects, patients with Down's syndrome, and their parents and sibs K N FEDOROVA AND I E YUDINA .. .. .	427
Studies on the origin of human amniotic fluid cells by immunofluorescent staining of keratin filaments W W CHEN .. .. .	433
Thalassaemia intermedia in a family with $\beta^0$ -thalassaemia and Hb Hasharon M A ZAGO, F F COSTA, AND C BOTTURA .. .. .	437
Parental age and birth order in Chinese children with congenital heart disease J S H TAY, W C L YIP, AND R JOSEPH .. .. .	441
An autosomal dominant syndrome of uveal colobomata, cleft lip and palate, and mental retardation H M KINGSTON, P S HARPER, AND P W JONES .. .. .	444
Studies of a family with incontinentia pigmenti variably expressed in both sexes T W KURCZYNSKI, J S BERNS, AND W E JOHNSON .. .. .	447
An association study of Huntington's disease and HLA H MADSEN, L STAUB NIELSEN, AND S A SØRENSEN .. .. .	452

Odds in genetic counselling	G B CÔTÉ	455
Abstracts of scientific papers presented at the Clinical Genetics Society meeting on 7 and 8 April 1982 at the University of Glasgow		458
<b>Case reports</b>		
Isodicentric X chromosome in a moderately tall patient with gonadal dysgenesis: lack of effect of functional centromere on inactivation pattern	J ROBERTSON, M J W FAED, M A LAMONT, AND A M CROWDER	463
Recurrent spontaneous abortions due to a homologous Robertsonian translocation (14q14q)	R GRACIAS-ESPINAL, S H ROBERTS, D P DUCKETT, AND K M LAURENCE	465
Pregnancy in a patient with 47,XX,i(Xq) karyotype	C R KING AND R N SCHIMKE	467
A homozygote for pericentric inversion of chromosome 4	N J CARPENTER, B SAY, AND N D BARBER	469
The phenotype of ring chromosome 3	G N WILSON, J POOLEY, AND J PARKER	471
Cat-like cry and mental retardation owing to 7q interstitial deletion (7q22→7q32)	D N ABUELO AND T PADRE-MENDOZA	473
<b>Short reports</b>		
Interstitial deletion of the long arm of chromosome 4 in a patient with mental retardation and abnormal phenotype	M DEL VALLE TORRADO, J D LABARTA, AND A M MIGLIORINI	477
Acute transformation of a myeloproliferative state in sideroblastic anaemia with abnormal karyotype	G J MUFTI, T J HAMBLIN, AND M SEABRIGHT	478
Interstitial deletion of the long arm of chromosome 5: 46,XX,del(5)(q13q22)	S OHDO, H MADOKORO, AND K HAYAKAWA	479
Book reviews		480
Index		481