

Contents

Editorial *page* 1

Discordant sex in one of three monozygotic triplets B DALLAPICCOLA, C STOMELO, G FERRANTI, A DI LECCI, AND M PURPURA *page* 6

The estimation of recurrence risks in monogenic disorders using flanking marker loci R M WINTER *page* 12

Investigation of human chromosome polymorphisms by scanning electron microscopy C J HARRISON, E M JACK, T D ALLEN, AND R HARRIS *page* 16

The prevalence of translocations in parents of children with regular trisomy 21: a possible interchromosomal effect? R H LINDENBAUM, M HULTÉN, A MCDERMOTT, AND M SEABRIGHT *page* 24

Reduced penetrance in tuberous sclerosis M BARAITSER AND M A PATTON *page* 29

Increased frequency of lymphocytic mitotic non-disjunction in recurrent spontaneous aborters R C JUBERG, J KNOPS, AND P N MOWREY *page* 32

Profound limb deficiency, thoracic dystrophy, unusual facies, and normal intelligence: a new syndrome S A AL-AWADI, A S TEEBI, T I FARAG, K M NAGUIB, AND M Y EL-KHALIFA *page* 36

Trigonocephaly and the Opitz C syndrome C SARGENT, J BURN, M BARAITSER, AND M E PEMBREY *page* 39

Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases R BERNSTEIN, J ISDALE, M PINTO, J DU TOIT ZAAIJMAN, AND T JENKINS *page* 46

β^+ -thalassaemia in the Po river delta region (northern Italy): genotype and β globin synthesis L DEL SENNO, M PIRASTU, R BARBIERI, F BERNARDI, D BUZZONI, G MARCHETTI, C PERROTTA, C VULLO, Y W KAN, AND F CONCONI *page* 54

A family with three independent autosomal translocations associated with 7q32 \rightarrow 7qter syndrome H N BASS, R S SPARKES, M M LESSNER, M FOX, B PHOENIX, AND J BERNAR *page* 59

Case reports

De novo tandem duplication 9p (p12 \rightarrow p24) with normal GALT activity in red cells T MOTEGI, K WATANABE, N NAKAMURA, T HASEGAWA, AND Y YANAGAWA *page* 64

A complex balanced rearrangement involving four chromosomes in an azoospermic man M T RODRIGUEZ, M J MARTIN, AND J A ABRISQUETA *page* 66

A child with a recombinant of chromosome 8 inherited from her carrier mother I C S BARNES, D KUMAR, AND R J M BELL *page* 67

'Pure' partial trisomy 2q in a male owing to malsegregation of a maternal translocation t(X:2)(p22:3;q32:1) G PLESSIS, J COUTURIER, C TURLEAU, S DESPOISSES, AND J DELAVENNE *page* 70

Partial trisomy 12q24:31 \rightarrow qter E H TAJARA, M VARELLA-GARCIA, AND A C T GUSSON *page* 73

Fraser syndrome presenting as monozygotic twins with bilateral renal agenesis G MORTIMER, H P MCEWAN, AND J R W YATES *page* 76

Familial opposable triphalangeal thumbs associated with duplication of the big toes P MERLOB, M GRUNEBaum, AND S R REISNER *page* 78

ASTM CODEN: JMDGAE (22) 1-80 (1985) ISSN 0022-2593

British Medical Association Tavistock Square London WC1

CONTENTS

No 1 FEBRUARY 1985

Editorial	1
Discordant sex in one of three monozygotic triplets B DALLAPICCOLA, C STOMELO, G FERRANTI, A DI LECCI, AND M PURPURA	9
The estimation of recurrence risks in monogenic disorders using flanking marker loci R M WINTER	12
Investigation of human chromosome polymorphisms by scanning electron microscopy C J HARRISON, E M JACK, T D ALLEN, AND R HARRIS	16
The prevalence of translocations in parents of children with regular trisomy 21: a possible inter-chromosomal effect? R H LINDENBAUM, M HULTÉN, A McDERMOTT, AND M SEABRIGHT	24
Reduced penetrance in tuberous sclerosis M BARAITSER AND M A PATTON	29
Increased frequency of lymphocytic mitotic non-disjunction in recurrent spontaneous aborters R C JUBERG, J KNOPS, AND P N MOWREY	32
Profound limb deficiency, thoracic dystrophy, unusual facies, and normal intelligence: a new syndrome S A AL-AWADI, A S TEEBI, T I FARAG, K M NAGUIB, AND M Y EL-KHALIFA	36
Trigonocephaly and the Opitz C syndrome C SARGENT, J BURN, M BARAITSER, AND M E PEMBREY	39
Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases R BERNSTEIN, J ISDALE, M PINTO, J DU TOIT ZAAIJMAN, AND T JENKINS	46
β^+ -thalassaemia in the Po river delta region (northern Italy): genotype and β globin synthesis L DEL SENNO, M PIRASTU, R BARBIERI, F BERNARDI, D BUZZONI, G MARCHETTI, C PERROTTA, C VULLO, Y W KAN, AND F CONCONI	54
A family with three independent autosomal translocations associated with 7q32 \rightarrow 7qter syndrome H N BASS, R S SPARKES, M M LESSNER, M FOX, B PHOENIX, AND J BERNAR	59
Case reports	
De novo tandem duplication 9p (p12 \rightarrow p24) with normal GALT activity in red cells T MOTEGI, K WATANABE, N NAKAMURA, T HASEGAWA, AND Y YANAGAWA	64
A complex balanced rearrangement involving four chromosomes in an azoospermic man M T RODRIGUEZ, M J MARTIN, AND J A ABRISQUETA	66
A child with a recombinant of chromosome 8 inherited from her carrier mother I C S BARNES, D KUMAR, AND R J M BELL	67
'Pure' partial trisomy 2q in a male owing to malsegregation of a maternal translocation t(X;2)(p22.3;q32.1) G PLESSIS, J COUTURIER, C TURLEAU, S DESPOISSES, AND J DELAVENNE	70
Partial trisomy 12q24.31 \rightarrow qter E H TAJARA, M VARELLA-GARCIA, AND A C T GUSSON	73
Fraser syndrome presenting as monozygotic twins with bilateral renal agenesis G MORTIMER, H P McEWAN, AND J R W YATES	76
Familial opposable triphalangeal thumbs associated with duplication of the big toes P MERLOB, M GRUNEBaum, AND S R REISNER	78

No 2 APRIL 1985

Editorial	81
Frequency and replication status of the fragile X, fra(X)(q27-28), in a pair of monozygotic twins of markedly differing intelligence E TUCKERMAN, T WEBB, AND S E BUNDEY	85
First trimester fetal diagnosis of genetic disorders: clinical evaluation of 250 cases B BRAMBATI, G SIMONI, C DANESINO, A OLDRINI, E FERRAZZI, L ROMITTI, G TERZOLI, F ROSSELLA, M FERRARI, AND M FRACCARO	92
Cell surface abnormality in clones of skin fibroblasts from a carrier of Duchenne muscular dystrophy J HILLIER, G E JONES, H E STATHAM, J A WITKOWSKI, AND V DUBOWITZ	100
A pedigree study of perinatally lethal renal disease A BANKIER, M DE CAMPO, R NEWELL, J G ROGERS, AND D M DANKS	104
Necropsy findings in neonatal asphyxiating thoracic dystrophy S B TURKEL, E H DIEHL, AND J A RICHMOND	112

An autosomal dominant syndrome with 'acromegaloid' features and thickened oral mucosa H E HUGHES, P J McALPINE, D W COX, AND S PHILIPPS	119
Kabuki make-up syndrome (Niikawa-Kuroki syndrome) associated with congenital heart disease S OHDO, H MADOKORO, T SONODA, T NISHIGUCHI, K KAWAGUCHI, AND K HAYAKAWA	126
The frequency of lactase phenotypes in Aymara children E BALANZA AND G TABOADA	128
The clinical features of the Cohen syndrome: further case reports C NORTH, M A PATTON, M BARAITSER, AND R M WINTER	131
Dermatoglyphs in children with mitral valve prolapse J S H TAY, W C L YIP, H K YAP, B W LEE, H B WONG, AND S O CHAY	135
Abstracts of the meeting of the Clinical Genetics Society held on 8, 9, and 10 November 1984 at the Royal Free Hospital, London	137
Case reports	
De novo paracentric inversion in an X chromosome H M HERR, S J HORTON, AND C I SCOTT JR	140
Monosomy 13q32.3→qter: report of two cases H RIVERA, S A GONZÁLEZ-FLORES, F RIVAS, J SÁNCHEZ- CORONA, M MOLLER, AND J M CANTÚ	142
Congenital diaphragmatic hernia in half sibs A H LIPSON AND G WILLIAMS	145
A case of suspected teratogenic holoprosencephaly M STABILE, A BIANCO, SIANNUZZI, M C BUONOCORE, AND V VENTRUTO	147
Tracheo-oesophageal anomalies in the Goldenhar anomalad A MENDELBERG, I ARIEL, P MOGLE, AND I ARAD	149
Severe pseudoachondroplasia with parental consanguinity I D YOUNG AND J R MOORE	150
Short reports	
A case of malignant spinal cord ependymoma in association with a duplication of part of the long arm of chromosome 12 B G R NEVILLE, A C BERRY, AND Y STODDART	154
Interstitial deletion of chromosome 2 S MARKOVIĆ, M KRSTIĆ, V ŠULOVIĆ, Z RADOJKOVIĆ, AND S ADŽIĆ	154
Familial occurrence of a pseudodicentric chromosome 21 I HANCKE AND K MILLER	155
Extra euchromatic band in the qh region of chromosome 9 Z DOCHERTY AND M A HULTÉN	156
Book reviews	158
Announcements	160

No 3 JUNE 1985

Editorial	161
Review article: Erroneous theories of sex determination U MITTWOCH	164
Hypothesis: An hypothesis regarding the origin of aneuploidy in man: indirect evidence from an experimental model M H KAUFMAN	171
Linkage analysis of a DNA polymorphism proximal to the Duchenne and Becker muscular dystrophy loci on the short arm of the X chromosome C S BROWN, P L PEARSON, N S T THOMAS, M SARFAZAI, P S HARPER, AND D J SHAW	179
Polymorphism of DNA sequence in the human pro $\alpha 2(I)$ collagen gene A F GROBLER-RABIE, D K BREBNER, S VANDENPLAS, G WALLIS, R DALGLEISH, R E KAUFMAN, A J BESTER, C G P MATHEW, AND C D BOYD	182
Exclusion of the $\alpha 1(II)$ cartilage collagen gene as the mutant locus in type IA osteogenesis imperfecta B SYKES, R SMITH, S VIPOND, C PATERSON, K CHEAH, AND E SOLOMON	187
Seckel syndrome: an overdiagnosed syndrome E THOMPSON AND M PEMBREY	192
A new brachydactyly syndrome with similarities to Julia Bell types B and E P PITT AND I WILLIAMS	202
Clinical and haematological evaluation of β thalassaemia intermedia with increased Hb F and Hb A ₂ in heterozygotes: β thalassaemia intermedia I C ALTAY AND A GURGEY	205
Clinical and haematological evaluation of β thalassaemia intermedia characterised by unusually low Hb F and increased Hb A ₂ : β thalassaemia intermedia II A GURGEY, S KAYIN, E KANSU, AND C ALTAY	213

Case reports

Extra Yq and partial monosomy 12p due to a Y;12 translocation in a boy with features of the 12p deletion syndrome	E ORYE, M CRAEN, G LAUREYS, R VAN COSTER, AND B VAN MELE	222
Interstitial deletion of the long arm of chromosome 11	J M KLEP-DE PATER, H F DE FRANCE, AND J B BIJLSMA	224
Interstitial deletion 2q24-3: case report with high resolution banding	J BERNAR, R S SPARKES, AND S ALLENSWORTH	226
Hyperinsulinaemic hypoglycaemia in an infant with mosaic trisomy 13	V S SMITH AND G P GIACOIA	228
Complex cardiac malformation in a case of trisomy 9	T WILLIAMS, I ZARDAWI, R QUAIFE, AND I D YOUNG	230
Phenotypic delineation of ring chromosome 15 and Russell-Silver syndromes	G N WILSON, S ESAUDER, M BUSH, AND I Z BEITINS	231
Book reviews		237

No 4 AUGUST 1985

Editorial

Review article: Molecular genetics of the human X chromosome	K E DAVIES	241
A population study of adult onset limb-girdle muscular dystrophy	J R W YATES AND A E H EMERY	243
A community study of severe mental retardation in the West Midlands and the importance of the fragile X chromosome in its aetiology	S BUNDEY, T P WEBB, A THAKE, AND J TODD	250
Hereditary sideroblastic anaemia and ataxia: an X linked recessive disorder	R A PAGON, T D BIRD, J C DETTER, AND I PIERCE	259
The inheritance of primary lymphoedema	R F DALE	267
Zygoty determination in newborn twins using DNA variants	C DEROM, E BAKKER, R VLIETINCK, R DEROM, H VAN DEN BERGHE, M THIERY, AND P PEARSON	274
A familial pericentric inversion of chromosome 22 with a recombinant subject illustrating a 'pure' partial monosomy syndrome	J L WATT, I A OLSON, A W JOHNSTON, H S ROSS, D A COUZIN, AND G S STEPHEN	279
Survey on haemoglobin variants, β thalassaemia, glucose-6-phosphate dehydrogenase deficiency, and haptoglobin types in Turks from Western Thrace	M AKSOY, A KUTLAR, F KUTLAR, G DINÇOL, S ERDEM, AND S BAŞTESBIHÇI	283
Admission of Hb S heterozygotes to a general hospital is relatively reduced in malarial areas	B COLOMBO AND L FELICETTI	288
Evaluation of haematological findings in 50 Bahraini patients with sickle cell disease and in some of their parents	M A BUHAZZA, A B BIKHAZI, AND F P KHOURI	291
Cornelia de Lange syndrome in several members of the same family	D KUMAR, C E BLANK, AND B L GRIFFITHS	296
High incidence of Meckel's syndrome in Gujarati Indians	I D YOUNG, A B RICKETT, AND M CLARKE	301
Gene deletion in an Italian haemophilia B subject	F BERNARDI, L DEL SENNO, R BARBIERI, D BUZZONI, R GAMBARI, G MARCHETTI, F CONCONI, F PANICUCCI, M POSITANO, AND S PITRUZZELLO	305
Two brothers with Martsof's syndrome	J M SÁNCHEZ, C BARREIRO, AND H FREILIJ	308
Familial pulmonary valve stenosis, atrial septal defect, and unique electrocardiogram abnormalities	A A CIUFFO, E CUNNINGHAM, AND T A TRAILL	311
Case reports		
Complex translocation in a boy with trichorhinophalangeal syndrome	J M SÁNCHEZ, J D LABARTA, T C DE NEGROTTI, AND A M MIGLIORINI	314
Interstitial deletion of chromosome 7p detected antenatally	K MARKS, L HILL, R G CHITHAM, AND W L WHITEHOUSE	319
Correspondence		319
Book reviews		324
Announcements		324

No 5 OCTOBER 1985

Review article: The human Y chromosome	P GOODFELLOW, S DARLING, AND J WOLFE	329
Annotation: Classroom teaching in genetics and birth defects: the Nottingham experience	J S FITZSIMMONS	345
Chronic proximal spinal muscular atrophy of childhood and adolescence: problems of classification and genetic counselling	I HAUSMANOWA-PETRUSEWICZ, J ZAREMBA, AND J BORKOWSKA	350
Genetics and biochemical variability of variants of 21 hydroxylase deficiency	M T GORDON, D I CONWAY, D C ANDERSON, AND R HARRIS	354
The genetic control of phenformin 4-hydroxylation	R R SHAH, D A P EVANS, N S OATES, J R IDLE, AND R L SMITH	361
A family study of hidradenitis suppurativa	J S FITZSIMMONS AND P R GUILBERT	367
Osmotic fragility test in heterozygotes for α and β thalassaemia	L MACCIONI AND A CAO	374
The first observation of Hb D Punjab β^0 thalassaemia in an English family with 22 cases of unsuspected β^0 thalassaemia minor among its members	S WORTHINGTON AND H LEHMANN	377
Phenotypic variation in LADD syndrome	E THOMPSON, M PEMBREY, AND J M GRAHAM	382
Three new cases of oculodentodigital (ODD) syndrome: development of the facial phenotype	M A PATTON AND K M LAURENCE	386
Familial absence of the pectoralis major, serratus anterior, and latissimus dorsi muscles	T J DAVID AND R M WINTER	390
Abstracts of the meeting of the Clinical Genetics Society held on 24 and 25 April 1985 at the University of Birmingham		393
Case reports		
Reciprocal translocation between chromosomes 8 and 9 in atypical chronic myeloid leukaemia	D G OSCIER, G J MUFTI, A GARDINER, AND T J HAMBLIN	398
Partial 2p deletion in a girl with a complex chromosome rearrangement involving chromosomes 2, 6, 11, and 21	R S YOUNG, M A MEDRANO, AND K L HANSEN	401
Sacrococcygeal teratoma and normal alphafetoprotein concentration in amniotic fluid	M SZABÓ, P VARGA, A ZALATNAI, J HIDVÉGI, Z TÓTH, AND Z PAPP	405
The Nager acrofacial dysostosis syndrome with the tetralogy of Fallot	E THOMPSON, R CADBURY, AND M BARAITSER	408
Correspondence		411
Obituary		416

No 6 DECEMBER 1985

Editorial		417
Sir Cyril Clarke		418
Biographical details of contributors to special issue		419
Prenatal diagnosis of the common haemoglobin disorders	D J WEATHERALL, J M OLD, S L THEIN, J S WAINSCOT, AND J B CLEGG	422
Marcella O'Grady Boveri (1865–1950) and the chromosome theory of cancer	V A MCKUSICK	431
Application of an intragenic genomic probe to genetic counselling for haemophilia B in the west of Scotland	J M CONNOR, A F PETTIGREW, I M HANN, C D FORBES, G D O LOWE, AND N A AFFARA	441
Genetic linkage between Huntington's disease and the DNA polymorphism G8 in South Wales families	P S HARPER, S YOUNGMAN, M A ANDERSON, M SARFARAZI, O QUARRELL, R TANZI, D SHAW, P WALLACE, P M CONNEALLY, AND J F GUSELLA	447
Immunogenetic studies in families with rheumatoid arthritis and autoimmune thyroid disease	P A SANDERS, D M GRENNAN, P A DYER, G G DE LANGE, AND R HARRIS	451
Should women at high risk of neural tube defect have an amniocentesis?	K M LAURENCE, G ELDER, K T EVANS, B M HIBBARD, M HOOLE, AND C J ROBERTS	457
Prenatal diagnosis of ornithine carbamoyl transferase deficiency using a gene specific probe	M E PEMBREY, J M OLD, J V LEONARD, C H RODECK, R WARREN, AND K E DAVIES	462

Collagen genes and proteins in osteogenesis imperfecta	F M POPE, A C NICHOLLS, J MCPHEAT, P TALMUD, AND R OWEN	4
The acetylator phenotypes of Saudi Arabian diabetics	D A PRICE EVANS, S PATERSON, P FRANCISCO, AND G ALVAREZ	47
Familial complex autosomal translocations involving chromosomes 7, 8, and 9 exhibiting male and female transmission with segregation and recombination	S WALKER, P J HOWARD, AND D HUNTER	48
HLA antigens in psoriasis and psoriatic arthritis	J C WOODROW AND A ILCHYSYN	49
Book reviews	49
Index	50