

JOURNAL OF MEDICAL GENETICS

Editor: C. A. CLARKE

Assistant Editors: CEDRIC CARTER and JOHN L. HAMERTON

Review: The 'Law of Ancestral Heredity' and the Mendelian-Ancestrarian Controversy in England, 1880-1906	1
P. FROGGATT and N. C. NEVIN	
Annotation: Phenylketonuria	37
BARBARA E. CLAYTON	
An Inherited Kidney Disease of Mice Resembling Human Nephronophthisis	41
MARY F. LYON and E. V. HULSE	
Chromosome Surveys in Penal Institutions and Approved Schools	49
PATRICIA A. JACOBS, WILLIAM H. PRICE, SHIRLEY RICHMOND, and R. A. W. RATCLIFF	
Chromosome Studies in 101 Mentally Handicapped Jamaican Children	59
MARIGOLD J. THORBURN and PATRICIA A. MARTIN	
Congenital Cleft Lip: A Genetic Study of 496 Propositi	65
CHARLES M. WOOLF	
Familial Incidence of Cerebrovascular Disease	84
JOHN MARSHALL	
Genetic-Epidemiological Studies in Progressive Muscular Dystrophy	90
JANINA PROT	
Gene Deletion and Duplication Effects on Phenotype and Gamma Globulin Levels ..	97
NORREN L. RUDD and PAUL H. LAMARCHE	
Asphyxiating Thoracic Chondrodystrophy: Association with Renal Disease and Evidence for Possible Heterozygous Expression	107
M. H. K. SHOKRIB, C. S. HOUSTON, and C. F. AWEN	
Concordance of Amyotrophic Lateral Sclerosis in a Pair of Dizygous Twins of Consanguineous Parents	113
J. DUMON, J. MACKEN, and TH. DE BARSY	
Case Reports:	
Presumptive 46,XX/46,XY/47,XXY Mosaicism in a Hermaphrodite	117
AMALA CHAUDHURI, SUBIR K. CHATTERJEE, and SUSANTA SARKAR	
Trisomy of Chromosome 16 in a Neonate, 47,XY,?16+	123
ANGELA I. TAYLOR	
Correspondence	126
Fourth International Congress of Human Genetics: Announcement	126
Book Reviews	127

BROWN UNIVERSITY
LIBRARY
MARCH 1971

J Med Genet: first published as on 1 March 1971. Downloaded from <http://jmg.bmj.com/> on May 11, 2021 by guest. Protected by copyright.

CONTENTS

No. 1. MARCH 1971

Review: The 'Law of Ancestral Heredity' and the Mendelian-Ancestrian Controversy in England, 1889-1906. P. FROGGATT AND N. C. NEVIN	1
Annotation: Phenylketonuria. BARBARA E. CLAYTON	37
An Inherited Kidney Disease of Mice Resembling Human Nephronophthisis. MARY F. LYON AND E. V. HULSE	41
Chromosome Surveys in Penal Institutions and Approved Schools. PATRICIA A. JACOBS, WILLIAM H. PRICE, SHIRLEY RICHMOND, AND R. A. W. RATCLIFF	49
Chromosome Studies in 101 Mentally Handicapped Jamaican Children. MARIGOLD J. THORBURN AND PATRICIA A. MARTIN	59
Congenital Cleft Lip: A Genetic Study of 496 Propositi. CHARLES M. WOOLF	65
Familial Incidence of Cerebrovascular Disease. JOHN MARSHALL	84
Genetic-Epidemiological Studies in Progressive Muscular Dystrophy. JANINA PROT	90
Gene Deletion and Duplication Effects on Phenotype and Gamma Globulin Levels. NOREEN L. RUDD AND PAUL H. LAMARCHE	97
Asphyxiating Thoracic Chondrodystrophy: Association with Renal Disease and Evidence for Possible Heterozygous Expression. M. H. K. SHOKEIR, C. S. HOUSTON, AND C. F. AWEN	107
Concordance of Amyotrophic Lateral Sclerosis in a Pair of Dizygous Twins of Consanguineous Parents. J. DUMON, J. MACKEN, AND TH. DE BARSY	113
Case Reports:	
Presumptive 46,XX/46,XY/47,XXY Mosaicism in a Hermaphrodite. AMALA CHAUDHURI, SUBIR K. CHATTERJEE, AND SUSANTA SARKAR	117
Trisomy of Chromosome 16 in a Neonate, 47,XY,?16+. ANGELA I. TAYLOR	123
Correspondence	126
Fourth International Congress of Human Genetics: Announcement	126
Book Reviews	127

No. 2. JUNE 1971

Genetic Control of Nortriptyline Kinetics in Man: A Study of Relatives of Propositi with High Plasma Concentrations. MARIE ÅSBERG, DAVID A. PRICE EVANS, AND FOLKE SJÖQVIST	129
The Genetic Basis of Variation in Factor 8 Levels Among Haemophiliacs. D. F. ROBERTS	136
Ahaptoglobinaemia and Predisposition to Iron-Deficiency Anaemia. ELIANE S. AZEVÊDO, SALMA ASSEMAN, MÔNICA MARIA MARTINS DE SOUZA, AND MARIA EUTÁLIA ALMEIDA SANTANA	140
Genetics of Childhood Spinal Muscular Atrophy. ELIZABETH J. WINSOR, E. GORDON MURPHY, MARGARET W. THOMPSON, AND T. EDWARD REED	143
Double Heterozygosity for Glucose-6-Phosphate Dehydrogenase Deficiency. T. K. CHAN AND M. C. S. LAI	149
Genetic Studies on Hypospadias in Males. YOU C. CHEN AND PAUL V. WOOLLEY, JR.	153
Virilizing Adrenal Carcinoma in Two Sibs. MOHSEN MAHLOUJJI, HOSSAIN RONAGHY, AND WERNER DUTZ	160
Chromosome Studies in Selected Spontaneous Abortions: Polyploidy in Man. D. H. CARR	164
Fertility in Balanced Heterozygotes for a Familial Centric Fusion Translocation, t(DqDq). J. A. WILSON	175
Brief Note:	
47,XX,13+ with Snodgrass Phenotype II: Are Different Chromosomes Associated with Two Clinical Varieties of D-trisomy? RICHARD L. NEU, SALMA REGINA ASSEMAN, AND LYTT I. GARDNER	179
Chromosomal Aberrations Induced by T Strain Mycoplasmas. R. B. KUNDSIN, M. AMPOLA, S. STREETER, AND P. NEURATH	181
A Family with Balanced Translocation, t(5p-;Gp+). BEVERLY J. WHITE, LISA C. VAN DE WATER, AND JOE-HIN TJIO	188
Mental Retardation, Unusual Facies, and Abnormal Nails Associated with a Group-G Ring Chromosome: A Case Report on Two Unrelated Cases. VICTOR DUBOWITZ, PATRICIA COOKE, DOREEN COLVER, AND FRANK HARRIS	195
A Familial Variant of Chromosome 9. CATHERINE G. PALMER AND JANE SCHRODER	202

Case Reports:

Di Guglielmo Syndrome in a t(DqDq) Heterozygote. B. DALLAPICCOLA AND P. MALACARNE ..	209
A Case of XYY Down's Syndrome Confirmed by Autoradiography. RENATA LAXOVA, JANET A. MC-KEOWN, P. SALDAÑA, AND J. A. D. TIMOTHY ..	215
49,XXXXY Chromosomal Anomaly in a Neonate. ALBERTO HAYEK, VINCENT RICCARDI, LEONARD ATKINS, AND HARDY HENDREN ..	220
Ring 13 Chromosome with Normal Haptoglobin Inheritance. JOSEPH G. HOLLOWELL, L. GAYLE LITTLEFIELD, AYUT DHARMKRONG-AT, GORDON M. FOLGER, CLARK W. HEATH, JR, AND GERALD E. BLOOM ..	222
A Group-C Ring Chromosome in a Mentally Deficient Male. A. J. THERKELSEN, BODIL MØLLER, AND K. HENNINGSEN ..	227
A Child with a Ring G Chromosome (46,XX,Gr). N. C. NEVIN, B. MACLAVERTY, AND W. A. B. CAMPBELL ..	231
A Complex Chromosomal Rearrangement with Formation of a Ring 4. MARTIN BOBROW, LILIAN F. JONES, AND G. CLARKE ..	235
The Use of Quinacrine Fluorescence in the Identification of B and E Group Chromosomes Involved in Structural Abnormalities. M. BOBROW AND P. L. PEARSON ..	240
Additional G-like Chromosome in a Malformed Boy. M. FRACCARO, MAJ HULTÉN, S. D. JAYAKAR, AGNETA LINDSJÖ, J. LINDSTEN, AND L. TIEPOLO ..	244
Correspondence ..	250
Book Reviews ..	251
Symposium on Sickle Cell Disease: Announcement ..	255

No. 3. SEPTEMBER 1971

Polycystic Disease of Kidneys and Liver Presenting in Childhood. HELEN BLYTH AND BARBARA G. OCKENDEN ..	257
Polycystic Kidneys Associated with Malformations of the Brain, Polydactyly, and Other Birth Defects in Newborn Sibs. K. FRIED, E. LIBAN, M. LURIE, S. FRIEDMAN, AND S. H. REISNER ..	285
X-linked Spondyloepiphyseal Dysplasia Tarda: Clinical and Linkage Data. R. M. BANNERMAN, G. B. INGALL, AND J. F. MOHN ..	291
Haemoglobin Osu-Christiansborg: A New β -chain Variant of Haemoglobin A (β 52 (D3) Aspartic Acid \rightarrow Asparagine) in Combination with Haemoglobin S. F. I. D. KONOTY-AHULU, JUDITH L. KINDERLERER, H. LEHMANN, AND B. RINGELHANN ..	302
Differentiation of Two Genetically Specific Types of Depression by the Response to Antidepressant Drugs. C. M. B. PARE AND J. W. MACK ..	306
Mutation Rate in Duchenne Muscular Dystrophy. H. K. GOSWAMI AND B. D. CHAURASIA ..	310
Familial Total Anomalous Pulmonary Venous Return. J. E. PAZ AND E. E. CASTILLA ..	312
Distribution of ABO Blood Groups, G6PD Deficiency, and Abnormal Haemoglobins in Leprosy. N. SAHA, H. B. WONG, B. BANERJEE, AND M. O. WONG ..	315
Rh Immunization following Incompatible Blood Transfusion and a Possible Long-term Complication of Anti-D Immunoglobulin Therapy. M. E. J. BEARD, J. PEMBERTON, J. BLAGDON, AND W. J. JENKINS ..	317
Review: Suppression of the Immune Response. JOHN BRADLEY AND C. J. ELSON ..	321
Group G Deletion Syndromes. ROBERT P. KELCH, MARGERY FRANKLIN, AND ROY D. SCHMICKEL ..	341
The Inheritance of a Structural Anomaly of One Chromosome No. 16 in a Kindred (46,16-,C+). JUAN CHEMKE AND ARTHUR ROBINSON ..	346
The 13q - Deletion Syndrome. ELIZABETH GRACE, J. DRENNAN, DOREEN COLVER, AND R. R. GORDON ..	351
Somatic Stigmata of Turner's Syndrome in a Patient with 46,XXq-. MAUREEN BOCIAN, EVA KRMPOTIC, KATARINA SZEGO, AND IRA M. ROSENTHAL ..	358
Case Reports:	
Klinefelter's Syndrome and G Trisomy. B. ERDTMANN, A. A. GOMES DE FREITAS, R. P. DE SOUZA, AND F. M. SALZANO ..	364
A Case of Cri-du-Chat Associated with Cataracts and Transmitted from a Mother with a 4/5 Translocation. HAROLD GROTSKY, LILLIAN Y. F. HSU, AND KURT HIRSCHHORN ..	369
A Prisoner with an Unusual Karyotype (46,XY,Dq-). J. KAHN AND FRED A. REED ..	372
Partial Deletion of a Group-F (19-20) Chromosome in a Physically Handicapped Male Patient. PAUL GENEST, MARC BOUCHARD, AND JACQUELINE POTY ..	374
An Extra Small Metacentric Autosome in a Mentally Retarded Boy with Multiple Malformations. SALVADOR ARMENDARES, LEONOR BUENTELLO, AND FABIO SALAMANCA ..	378
An Extra Small Metacentric Chromosome in Association with Multiple Congenital Abnormalities. W. H. FINLEY, S. C. FINLEY, AND D. MONSKY ..	381
Trisomy D/Trisomy E Mosaicism in an Infant Male. RICHARD J. WARREN AND JOHN I. KEITH ..	384

X Chromosome Long Arm Deletion in a Patient with Down's Syndrome. FRED W. LUTHARDT AND CATHERINE G. PALMER	387
Trisomy 18 in One of Fraternal Twins. DENNIS GERTZER AND GERALD NATHENSON	392
Book Reviews	395

No. 4. DECEMBER 1971

An Analysis Procedure Illustrated on a Triple Linkage of Use for Prenatal Diagnosis of Myotonic Dystrophy. J. H. RENWICK AND D. R. BOLLING	399
Confirmation of Linkage of the Loci for Myotonic Dystrophy and ABH Secretion. J. H. RENWICK, SARAH E. BUNDEY, M. A. FERGUSON-SMITH, AND MARIAN M. IZATT	407
Xg Groups and Sex Abnormalities in People of Northern European Ancestry. RUTH SANGER, PATRICIA TIPPETT, AND JUNE GAVIN	417
The X-linked Blood Group System Xg: Tests on Unrelated People and Families of Northern European Ancestry. RUTH SANGER, PATRICIA TIPPETT, AND JUNE GAVIN	427
On the Distribution of Phenotypes in XXY Males and their Parents. J. H. EDWARDS	434
ABH Secretor Status of the Fetus: a Genetic Marker Identifiable by Amniocentesis. PETER HARPER, WILMA B. BIAS, JUDITH R. HUTCHINSON, AND VICTOR A. MCKUSICK	438
β -Glucuronidase Activity in Fibroblasts Cultured from Persons with and without Cystic Fibrosis. SHIRLEY B. RUSSELL, JAMES D. RUSSELL, AND JOHN W. LITTLEFIELD	441
Annotation: Polymorphism and Protein Evolution. The Neutral Mutation-Random Drift Hypothesis. HARRY HARRIS	444
Individuals at Risk in Families with Genetic Disease. CHARLES SMITH, SUSAN HOLLOWAY, AND ALAN E. H. EMERY	453
Carcinoma of the Breast and Klinefelter's Syndrome. D. G. HARNDEN, N. MACLEAN, AND A. O. LANGLANDS	460
On the Pathogenesis of Favism. ERNESTO SARTORI	462
Abnormal Distribution of ABO Blood Groups in Infantile Pyloric Stenosis. J. A. DODGE	468
Congenital Hypothyroidism and Hyperthyroidism in Monozygotic Twin Girls. PHILIP L. TOWNES AND WILLIAM L. BRADFORD	471
Simple Anonychia: Further Evidence for Autosomal Recessive Inheritance. MOHSEN MAHLOUJJI AND M. AMIDI	478
Review: The Nosology of the Spinal Muscular Atrophies. ALAN E. H. EMERY	481
Monosomy G: Case Report and Review of the Literature. RONALD D. GREENWOOD AND ANNEMARIE SOMMER	496
Hypothesis: Origin of Sex Chromosome Monosomy in Man. PAUL E. POLANI	501
Autosomal Translocation in a Mentally Retarded Male Child with 46,XY,t(2q-;13q+) Complement: Case Report and Review. PAUL GENEST, ROGER LACHANCE, JACQUELINE POTY, AND DAVID JACOB	504
A Family Showing Transmission of a Translocation of t(3porq-;Cq+). JOHN MCHUGH, TREVOR WRIGHT, AND PATRICIA COOKE	509
Case Reports:	
A Patient with 45,X/46,XXq- /46,XXq- dic Karyotype. ALAN C. STEVENSON, JEAN BEDFORD, AND G. MITFORD BARBERTON	513
A Ring-4 Chromosome in a Patient with Normal Intelligence and Short Stature. RAWATMAL B. SURANA, JOHN D. BAILEY, AND PATRICK E. CONEN	517
A Partial D-trisomy/Normal Mosaic Female. G. C. WEBB, O. MARGARET GARSON, MERYL K. ROBSON, AND D. B. PITT	522
A Case of 47,XX,(21q-)+ with some Stigmata of Down's Syndrome and an IQ of 77. RICHARD L. NEU, MARY L. VOORHESS, AND LYTT I. GARDNER	528
A 47,XXq-Y Klinefelter Male. H. SHARAT CHANDRA, G. N. REDDY, JOSHUA PETER, AND G. VENKATACHALAI AH	530
A Case of 48,XYY,21+ in an Infant with Down's Syndrome. RICHARD L. NEU, ALFRED Q. SCHEUER, AND LYTT I. GARDNER	533
An Inherited 1;G Translocation. ALLAN J. EBBIN, MIRIAM G. WILSON, JOSEPH W. TOWNER, AND IRENE FORSMAN	536
Additional Evidence of Gradual Loss of Germ Cells in the Pathogenesis of Streak Ovaries in Turner's Syndrome. LESTER WEISS	540
Book Reviews	545
UK MEDLARS Service: Announcement	547
Index	548