## CONTENTS

**No 1  FEBRUARY 1986**

<table>
<thead>
<tr>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Editorial</td>
<td>1</td>
</tr>
<tr>
<td>Review article: Gene mapping and chromosome 19</td>
<td>2</td>
</tr>
<tr>
<td>Syndrome of the month: Dubowitz syndrome</td>
<td>11</td>
</tr>
<tr>
<td>Meiotic recombination between two polymorphic restriction sites within \ the \ $\beta$ globin gene cluster</td>
<td>14</td>
</tr>
<tr>
<td>Evidence against the structural gene encoding type II collagen (COL2A1) as the mutant locus in \ achondroplasia</td>
<td>19</td>
</tr>
<tr>
<td>X linked hydrocephalus: a survey of a 20 year period in Victoria, Australia</td>
<td>23</td>
</tr>
<tr>
<td>Autosomal recessive or sex linked recessive: a counselling dilemma</td>
<td>32</td>
</tr>
<tr>
<td>A computer programme to calculate risk in X linked disorders using multiple marker loci</td>
<td>35</td>
</tr>
<tr>
<td>A computer programme for estimation of genetic risk in X linked disorders, combining pedigree and DNA probe data with other conditional information</td>
<td>40</td>
</tr>
<tr>
<td>Prenatal diagnosis of bullous ichthyosiform erythroderma: detection of tonofilament clumps in fetal epidermal and amniotic fluid cells</td>
<td>46</td>
</tr>
<tr>
<td>Early prenatal investigation of a pregnancy at risk of adenosine deaminase deficiency using chorionic villi</td>
<td>52</td>
</tr>
<tr>
<td>Linkage analysis of peripheral neurofibromatosis (Von Recklinghausen disease) and chromosome 19 markers linked to myotonic dystrophy</td>
<td>55</td>
</tr>
<tr>
<td>Genetic analysis of malformations causing perinatal mortality</td>
<td>58</td>
</tr>
<tr>
<td>The lethal multiple congenital anomaly syndrome of polydactyly, sex reversal, renal hypoplasia, and unilobular lungs</td>
<td>64</td>
</tr>
<tr>
<td>Sensitivity to ultraviolet radiation in a dominantly inherited form of xeroderma pigmentosum</td>
<td>72</td>
</tr>
<tr>
<td>Abstracts of the annual scientific meeting of the Association of Clinical Cytogeneticists held at the University of Newcastle upon Tyne on 3 and 4 July 1985</td>
<td>79</td>
</tr>
<tr>
<td>Case reports</td>
<td></td>
</tr>
<tr>
<td>A case of Fryns syndrome</td>
<td>82</td>
</tr>
<tr>
<td>Umbilical cord hernia in a child with autosomal recessive chondrodysplasia punctata</td>
<td>84</td>
</tr>
<tr>
<td>Trisomy 14 mosaicism in a 2 year old girl</td>
<td>86</td>
</tr>
<tr>
<td>Short reports</td>
<td></td>
</tr>
<tr>
<td>First trimester diagnosis from chorionic villi of a der(15),t(9;15)(q33;q14)mat identified by DA/DAPI staining</td>
<td>89</td>
</tr>
<tr>
<td>A three way translocation in mother and daughter</td>
<td>90</td>
</tr>
<tr>
<td>Familial inv(1)(p36:3q12) associated with sterility</td>
<td>90</td>
</tr>
<tr>
<td>Complex translocation involving chromosomes Y, 1, and 3 resulting in deletion of segment 3q23→q25</td>
<td>91</td>
</tr>
<tr>
<td>Prune belly appearance in a Turner subject</td>
<td>92</td>
</tr>
<tr>
<td>Book reviews</td>
<td>94</td>
</tr>
<tr>
<td>Announcement</td>
<td>96</td>
</tr>
</tbody>
</table>
No 2 APRIL 1986

Annotation: Molecular euphoria R HARRIS
The Carter Lecture for 1985: Of mice and men, metals and mutations D M DANKS
Gene mapping and medical genetics: Chromosome 1 in relation to human disease S POVEY AND J M PARRINGTON

Syndrome of the month: The orofaciiodigital (OFD) syndromes M BARAITSER
Familial partial lipodystrophy: two types of an X linked dominant syndrome, lethal in the hemizygous state J KÖBERLING AND M G DUNNIGAN
Partial lipatrophy with insulin resistant diabetes and hyperlipidaemia (Dunnigan syndrome) J BURN AND M BARAITSER
Familial polyposi coli: growth characteristics of karyotypically variable cultured fibroblasts, response to epidermal growth factor and the tumour promoter 12-0-tetradecanoyl phorbol-13-acetate S H RIDER, H A MAZZULLO, M B DAVIS, AND J D A DELHANTY
A new ε globin HincII variant fragment length polymorphism in the south African Negroid family M RAMSAY, J A THOMSON, AND T JENKINS

Abnormal haemoglobin among pregnant women from Mozambique M C WILLCOX, J LILJESTRAND, AND S BERGSTROM
The relationship between the acetylator and the sparteine hydroxylation polymorphisms D HARMER, D A P EVANS, L C EZE, M JOLLY, AND E J WHIBLEY
Familial calcification of the basal ganglia with cerebrospinal fluid pleocytosis L MEHTA, J Q TROUNCE, J R MOORE, AND I D YOUNG
A Noonan-like short stature syndrome with sparse hair M BARAITSER AND M A PATTON

Abstracts of the meeting of the Clinical Genetics Society held at the Clinical Research Centre, Northwick Park Hospital, London on 8 and 9 November 1985

Case reports
Duchenne muscular dystrophy in a female with a translocation involving Xp21 N C NEVIN, A E HUGHES, M CALWELL, AND J H K LIM
An additional case of Smith-Lemli-Opitz syndrome in a 46,XY infant with female external genitalia P R SCARBOURGH, K HUDDELENTON, AND S C FINLEY
Possible Waardenburg syndrome with gastrointestinal anomalies J NUTMAN, R STEINHERZ, Y SIVAN, AND R M GOODMAN
Multifocal meningiomas in a patient with a constitutional ring chromosome 22 T ARINAMI, I KONDO, H HAMAGUCHI, AND S NAKAJIMA
Prenatal diagnosis and follow up of a child with a complex chromosome rearrangement M H BOGART, C L BRADSHAW, O W JONES, AND J E SCHANBERGER
Primary myelodysplastic syndrome with complex chromosomal rearrangements in a patient with Klinefelter's syndrome S M N ABIDI, M GRIFFITHS, D G OSCIER, G J MUFTI, AND T J HAMBLIN
Partial trisomy 6p and partial trisomy 22 resulting from 3:1 meiotic disjunction of maternal (6p;22q) translocation P R SCARBOURGH, A J CARROLL, S C FINLEY, AND K HAMERICK

Correspondence
Announcements

No 3 JUNE 1986

Gene mapping and medical genetics: Molecular genetics of human chromosome 4 J F GUSELLA, T C GILLIAM, M E MACDONALD, S V CHENG, AND R E TANZI
Syndrome of the month: Walker-Warburg syndrome (Warburg syndrome, HARD±E syndrome) D DONNAI AND P A FARDON
Deletion of the steroid 21-hydroxylase and complement C4 genes in congenital adrenal hyperplasia G RUMSBY, M C CARROLL, R R PORTER, D B GRANT, AND M HJELM
DNA polymorphic haplotypes on the short arm of chromosome 11 and the inheritance of type I diabetes mellitus G A A FERNIS, G A HITMAN, R TREMBATH, L WILLIAMS, A TARN, E A GALE, AND D J GALTON
Exclusion of close linkage between the parathyroid hormone gene and a mutant gene locus causing idiopathichypoparathyroidism J SCHMIDTKE, K KRUSE, B PAPE, AND G SIPPELL

J Med Genet: first published as on 1 February 1986. Downloaded from http://jmg.bmj.com on 1 November 1994 by guest. Protected by copyright.
The estimation of risks from the induction of recessive mutations after exposure to ionising radiation

A G Searle and J H Edwards

Risk of dominant mutation in older fathers: evidence from osteogenesis imperfecta

A D Carothers, S J McAllion, and C R Paterson

Freeman-Sheldon syndrome: a disorder of congenital myopathic origin?

J Vaněk, J Janda, V Amblérová, and F Lošan

Type II syndactyly or synpolydactyly

P Merlob and M Grunebaum

Mental retardation associated with congenital heart disease, blepharophimosis, blepharoptosis, and hypoplastic teeth

S Ohdo, H Madokoro, T Sonoda, and K Hayakawa

Red cell genetic abnormalities in Peninsular Arabs: sickle haemoglobin, G6PD deficiency, and α and β thalassaemias

J M White, M Byrne, R Richards, T Buchanan, E Katoulis, and K Weerasinghe

Bone marrow and peripheral blood globin chain synthesis in sickle cell β+ thalassaemia

F F Costa and M A Zago

Case reports

Trisomy 18 in a 13 year old girl

L Mehta, R S Shannon, D P DUCKETT, and I D Young

Prenatal detection of monosomy 18p and trisomy 18q mosaicism with unexpected fetal phenotype

S D Sutton and M A C Ridler

Partial monosomy 13q and partial trisomy 18p: case report with necropsy findings

D Beneck, M A Greco, S R WOLMAN, L E MCMORROW, V Jansen, and J Cason

18q+, the progeny of a balanced translocation (t;1;18)mat: case report with necropsy findings

A Hindi, D Beneck, M A Greco, and S R WOLMAN

Is reprogramming a human teratogen?

R M Pauli and B J Pettersen

Tel Hashomer camptodactyly syndrome: report of a case with myopathic features

M A Patton, K D McDERMOT, B D LAKE, and M Baraitser

A male infant with the Catel-Manzke syndrome and dislocatable knees

E M Thompson, R M Winter, and M J H Williams

Outcome after prenatal detection of a sporadic, unstable translocation t(5;21)

A O Martin, I BENUCK, H S TRAISMAN, M S SWANSON, N TRAKAS, K LAING, B J ROINSKY, J BEAIRD, E S TRAISMAN, S ELIAS, and J L SIMPSON

Short report

A fragile site 10q25 in human sperm chromosomes

R H Martin

Addendum

M SARFARAZI and H WILLIAMS

Correspondence

.

Book reviews

.

Announcements

.

No 4 AUGUST 1986

Editorial

Syndrome of the month: The Van der Woude syndrome (dominantly inherited lip pits and clefts)

A Schinzel and M Kläusler

Cystic fibrosis carrier detection using a linked gene probe

M Farrall, P Scambler, K W Klinger, K Davies, C Worrall, R Williamson, and B Wainwright

Application of three intragenic DNA polymorphisms for carrier detection in haemophilia B

J M Connor, A F Pettigrew, C Shiach, I M HANN, G D O LOWE, and C D FORBES

Phenotype-karyotype correlation in patients trisomic for various segments of chromosome 13

S A Tharapel, R C LEWANDOWSKI, A T Tharapel, and R S Wilroy

Risk estimation in autosomal dominant disorders with reduced penetrance

A E H Emery

Genetic heterogeneity in Gaucher disease

J Zlotogora, R ZAIZOV, C KLIBANSKY, Y MATOTH, G BACH, and T COHEN

Differentiation of heterozygotes in recessive albinism

D F Roberts, J G R Kromberg, and T Jenkins

The birth prevalence rates for the skeletal dysplasias

I M ORIOLI, E E CASTILLA, and J G Barbosa-Neto

Aetiological factors in hypospadias

E CALZOLARI, M R CONTIERO, E RONCARATI, P L MATTIUZ, and S VOLPATO

Ratio of crown-rump distance to total length in preterm and term infants

P Merlob, Y Sivan, and S H Reisner

A paracentric inversion of 7q illustrating a possible interchromosomal effect

J L Watt, K Ward, D A COUZIN, G S Stephen, and A Hill
Autosomal dominant thoracolaryngopelvic dysplasia: Barnes syndrome  J BURN, C HALL, D MARSDEN, AND D J MATTHEW

Robinow syndrome without mesomelic 'brachymelia': a report of five cases  M D BAIN, R M WINTER, AND J BURN

Microcephaly, short stature, and developmental delay associated with a chemotactic defect and transient hypogammaglobulinaemia in two brothers  B SAY, N BARBER, G C MILLER, AND S E GROGG

Megacystis-microcolon-intestinal hypoperistalsis syndrome: confirmation of autosomal recessive inheritance  R M WINTER AND S A S KNOWLES

Case reports
Monozygotic twins concordant for congenital short femur  J M CONNOR, P S RAE, AND R A C CONNOR
Poland syndrome associated with 'morning glory' syndrome (coloboma of the optic disc)  D T PIŠTELJ, D VRANJEŠEVIĆ, S APOSTOLSKI, AND D D PIŠTELJ
Interstitial deletion of chromosome 4q diagnosed prenatally  J M CAMPBELL, J WILLIAMS, AND G BATCUP

Short reports
A case of de novo, double, balanced translocations (distal 9p to 3p, distal 18q to 3q)  J E HART
Pregnancy wastage associated with paracentric inversion of chromosome 13  B G BATEMAN, R NEU, W C NUNLEY, AND T E KELLY
A pericentric inversion duplication of the subcentromeric region of chromosome 12q  A P ALBERT, J HOLDSWORTH, AND C MASSYN
Partial trisomy 1q25→qter  E J TAWN, M B R ROBERTS, R G GHAZALA, AND E M T PYTA
Secondary amenorrhoea and 47,XX,i(Xq) karyotype  T E KELLY, J W WILKS, AND H E WYANDT
Necropsy findings in a child with FG syndrome  E M THOMPSON, B N HARDING, B D LAKE, AND S C SMITH

Conference report: Report on meeting on molecular studies of inherited cancer syndromes  D G HARNDEN
Correspondence
Book reviews
Announcements

No 5 OCTOBER 1986

Editorial
Syndrome of the month: Williams syndrome  J BURN
The frequency of the fragile X chromosome among schoolchildren in Coventry  T P WEBB, S BUNDEY, A THAKE, AND J TODD

Twelve families with fragile X(q27)  T WEBB, A THAKE, AND J TODD
Replication status of fragile X(q27-3) in 13 female heterozygotes  E TUCKERMAN, T WEBB, AND A THAKE
Mutations linked to the pro a2(I) collagen gene are responsible for several cases of osteogenesis imperfecta type I  G WALLIS, P BEIGHTON, C BOYD, AND C G MATHEW
A model system for the analysis of gene exclusion: cystic fibrosis and chromosome 19  B WAINWRIGHT, M FARROLL, E WATSON, AND R WILLIAMSON
A new strategy for mapping the human genome  D J SHAW
Pseudoachondroplasia: clinical diagnosis at different ages and comparison of autosomal dominant and recessive types. A review of 32 patients (26 kindreds)  R WYNNE-DAVIES, C M HALL, AND I D YOUNG

Severe mental retardation in six generations of a large South African family carrying a translocation t(6;10)(q27;q25-2)  J BRUSNIČK, K M M VAN HEERDEN, G DE JONG, A S CRONJE, AND A E RETIEF
Segregation of a t(3;20) translocation through three generations resulting in unbalanced karyotypes in six persons  K B NIELSEN, N TOMMERUP, B JESPERSEN, P NYGAARD, AND L KLEIF
The effects of severe mixed environmental pollution on human chromosomes  A KATSAKONI, S NAKOU, I ANTONIADOU-KOUMATOU, AND G B CÔTÉ

Pitfalls in prenatal diagnosis of β thalassaemia  C ROSATELLI, L MACCIIONI, M T SCALEA, AND A CAO
Microtia with mental atresia and conductive deafness: mild and severe manifestations within the same sibship  P STRISCIOGLIO, A BALLABIO, AND G PARENTI

Partial trisomy 7 (q32→qter) syndrome in two children  D A COUZIN, N HAITES, J L WATT, AND A W JOHNSTON
Three children with partial trisomy 1q and partial monosomy 3p  G T Mccarthy, C N Fear, and A C Berry .......................................................... 466

Abstracts of the meeting of the Clinical Genetics Society held at the University of Wales College of Medicine, Cardiff, on 18 and 19 April 1986 ........................................ 468

Case reports
A terminal deletion of the long arm of chromosome 4 [46,XX,del(4)(q33)] in an infant with phenotypic features of Williams syndrome R D Jefferson, J Burn, K L Gaunt, S Hunter, and E V Davison ............................................... 474

A de novo X;13 translocation with abnormal phenotype S V Hodgson, J C K Barber, A Dowie, and V Dubowitz .......................................................... 477

Terminal deletion of the long arm of chromosome 10 H Curtis, R T Howell, and C Cope .......................................................... 478

Announcement ........................................................................................................ 480

No 6 DECEMBER 1986

Editorial
Conference report: The elusive muscular dystrophy gene. Fifth Muscular Dystrophy Group Workshop on the X chromosome and muscular dystrophies, April 1986 K E Davies .................................................. 481

Muscular dystrophy in girls with X; autosome translocations Y Boyd, V Buckle, S Holt, E Munro, D Hunter, and J Craig .......................................................... 482

Paternal inheritance of translocation chromosomes in a t(X;21) patient with X linked muscular dystrophy V M Kean, H L Macleod, M W Thompson, P N Ray, C Verellen-Dumoulin, and R G Worton .......................................................... 484

Duchenne muscular dystrophy in one of monozygotic twin girls J Burn, S Povey, Y Boyd, E A Munro, L West, K Harper, and D Thomas .................................................. 484

Duchenne muscular dystrophy with adrenal insufficiency and glycerol kinase deficiency: high resolution cytogenetic analysis with molecular, biochemical, and clinical studies A Clarke, S H Roberts, N S T Thomas, A Whitfield, J Williams, and P S Harper .......................................................... 491

Molecular deletion analysis in Duchenne muscular dystrophy N S T Thomas, P N Ray, R G Worton, and P S Harper .......................................................... 491


The population genetics of Duchenne: natural and artificial selection in Duchenne muscular dystrophy J H Edwards ................................................................... 495

Localisation of Xp21 meiotic exchange points in Duchenne muscular dystrophy families C J Bertelson, J A Bartley, A P Monaco, C Colletti-Feen, F Ichukiewicz, and L M Kunkel .......................................................... 501


Linkage analysis of polymorphisms within the DNA fragment XJ cloned from the breakpoint of an X;21 translocation associated with X linked muscular dystrophy M W Thompson, P N Ray, B Belfall, C Duff, C Logan, I Ross, and R G Worton ......................... 505

Prenatal diagnosis of Duchenne muscular dystrophy by DNA analysis J M Old and K E Davies ................................................................... 505

Carrier detection and prenatal diagnosis in X linked muscular dystrophy using restriction fragment length polymorphisms M Lindlof, H Kääriäinen, K E Davies, and A de la Chapelle ................................................................... 506


A register based system for gene tracking in Duchenne muscular dystrophy A P Read, L Kerzinski-Storrar, R C Mountford, R G Elles, and R Harris .......................................................... 516

Emery-Dreifuss muscular dystrophy: localisation to Xq27.3—qter confirmed by linkage to the factor VIII gene J R W Yates, N A Affara, D M Jamieson, M A Ferguson-Smith, I Hausmanowa-Petrusewicz, J Zaremba, J Borkowska, A W Johnston, and K Kelly .......................................................... 518

X linked muscular dystrophy with contractures A W Johnston and E McKay ................................................................... 518

Localisation of the gene for Emery-Dreifuss muscular dystrophy to the distal long arm of the X chromosome N S T Thomas, H Williams, L J Elsas, L C Hopkins, M Sarfarazi, and P S Harper .......................................................... 521

Index ......................................................................................................................... 530