Journal of

MEDICAL GENETICS

Notice to Contributors

Papers, which should be in triplicate and in the Vancouver style (*Br Med J* 1982;**284**:1766–70), should be sent to the Editor, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR. Papers from the USA can be submitted to the North American Editor, Dr P M Conneally, Department of Medical Genetics, James Whitcomb Riley Hospital for Children RR129, Indiana University Medical Center, Indianapolis, Indiana 46223, USA. Submission of a paper will be held to imply that it contains original work which has not been previously published. The signature of each author is required on the covering letter. Permission to republish must be obtained from the Editor. Identifiable photographs of patients must be accompanied by written consent.

Papers should conform to one of the following categories.
Original contributions on clinical or laboratory aspects of medical genetics in man and on related animal studies.
Case reports or family reports with particularly instructive clinical or genetic features: to be no longer than 1000 words, with no more than three figures, one table, and eight references.
Short reports: to be no longer than 500 words with a clinical photograph and partial karyotype, if appropriate, and no more than three references.
Review articles will generally be by invitation, but suggestions from authors wishing to prepare a review article will be welcomed.
Short communications and *Technical notes* will also be considered.
Letters to the Editor in relation to papers and to other relevant topics will be welcomed.

Publication of papers thought to be of special importance may be expedited.

SI units should be used. All contributions should be accompanied by an abstract or structured abstract giving the main results and conclusions. Typescripts should be double spaced with wide margins. One page proof will be sent to the author submitting the paper and alterations on the proof, apart from printer's errors, are not permitted. Reprints may be ordered when the proof is returned.

Figures should be kept to a minimum and should be numbered consecutively in Arabic numerals. Legends should be typed on a separate sheet. Photographs should be on glossy paper and diagrams should be drawn on stout white paper. Photographs of karyotypes do not reproduce well. Chromosomes should be cut out and stuck onto stout paper. Any lettering should be indicated on a separate transparent overlay. Colour printing can be undertaken.

Tables should not be included in the body of the text, but should be typed on separate pages and numbered with Arabic numerals. A legend should be provided.

References should conform precisely to the style current in this Journal. Authors are responsible for the *accuracy* and *completeness* of their references as these will not be checked by the Editorial Office.

Some notes on nomenclature can be found in *J Med Genet* 1991; **28**:72.

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anomalies, lower thoracic vertebral scoliosis, two sacral segments (partial sacral agenesis), abnormal pelvis with abnormal right ischium and pubis and overlapping of pubic bones, and questionable dislocated left hip. There was no bone in the presacral appendage. Both parents had normal skeletal surveys.

In addition to this new patient, a patient reported previously as having VATER association[3] may represent another example of a disorganisation-like gene. The 31 week old male had multiple *v*ertebral anomalies, imperforate *a*nus, distal *t*racheo-*e*sophageal atresia with partial proximal oesophageal atresia, bilateral *r*enal dysplasia, single umbilical artery, bilateral cleft lip and palate, and dysplastic ears. Although VATER 'association'[4] might describe this patient (and would also characterise our patient), the presence of additional unusual anomalies (agenesis of the bladder, urethra, and penis with rudimentary scrotum, sacral caudal skin appendage, and right sided 'lobster claw' foot) suggests that the diagnosis of disorganisation homologue is more accurate.

I agree with Winter and Donnai[1] that patients with "extra limbs, appendages, or hamartomatous structures, in association with polydactyly or partial duplication/reduction of limbs and apparently distinct malformations, such as urogenital, body wall, and craniofacial abnormalities" may be the result of a disorganisation-like gene, especially when such patients are atypical examples of their diagnoses. Although this new patient lacks duplicated digits or limbs, several malformations (phallus-like sacral structure, rudimentary perineum, left sided foot-like appendage, absent right sided radius and thumb, ectopic renal, adrenal, and thymic tissues) resemble patients with the disorganisation-like complex.

Although the partial sacral agenesis, absent kidneys, abnormal testes, imperforate anus, and shortened lower segment and lower extremities could be attributed to the caudal dysplasia sequence (caudal regression syndrome),[5] the type and extent of non-caudal anomalies suggest a more widespread condition. These entities are not mutually exclusive. If there is indeed a single gene disorder in humans resembling the mouse mutant disorganisation, then the caudal dysplasia and Potter oligohydramnios *sequences* may occur as part of that disorganisation-like *syndrome*.[6] Similarly, VATER *association*, which describes many features of our patient and that of Dusmet *et al*,[3] may also be found within the broader context of that *syndrome*.

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1 Winter RM, Donnai D. A possible human homologue for the mouse mutant disorganisation. *J Med Genet* 1989;**26**:417–20.
2 Donnai D, Winter RM. Disorganisation: a model for 'early amnion rupture'? *J Med Genet* 1989;**26**:421–5.
3 Dusmet M, Fete F, Crusi A, Cox JN. VATER association: report of a case with three unreported malformations. *J Med Genet* 1988;**25**:57–60.
4 Weaver DD, Mapstone CL, Yu PL. The VATER association. Analysis of 46 patients. *Am J Dis Child* 1986; **140**:225–9.
5 Jones KL. *Smith's recognizable patterns of human malformation*. Philadelphia: Saunders, 1988:575.
6 Spranger J, Benirschke K, Hall JG, *et al*. Errors of morphogenesis: concepts and terms. *J Pediatr* 1982;**100**:160–5.

BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add £2 per item for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank, or by credit card (MASTERCARD, VISA or AMERICAN EXPRESS) stating card number, expiry date, and your full name.

Genomic Imprinting. Ed M Monk, A Surani. (Pp 155; £60.00.) Cambridge: The Company of Biologists. 1990.

Genomic imprinting is attracting increasing attention as a possible explanation for some of the unusual or non-classical inheritance patterns seen in human genetic disease. Therefore this collection of papers from an international symposium on genomic imprinting held in Manchester in April 1990 is both topical and potentially of great interest to many clinicians and scientists working in medical genetics. The range of topics covered is extremly wide including aspects of genomic imprinting in plants, yeast, insects, and mammals. Only two of the 18 papers are directly concerned with clinical genetics: Angus Clarke (Cardiff) and Judith Hall (Vancouver) each discuss the relevance of genomic imprinting to human genetic disease. Although there is some overlap between papers, together both papers document the clinical disorders in which there is strong evidence for parental genome effects, and also speculate on those disorders which appear to be candidates for imprinting but for which so far there is little evidence. The recent experimental evidence in support of the Laird hypothesis for fragile X is encouraging, but in many human genetic diseases the molecular evidence for imprinting is less than the number of hypotheses and models proposed. Such models are based on extrapolations from processes studied in invertebrates and small mammals, such as position effect variegation in *Drosophila*, and transgenes, gene methylation, and imprinted regions in the mouse genome. All of these (and other relevant) topics are the subject of individual contributions so that this volume represents a convenient starting point from which to explore many of the diverse genetic phenomena which are encompassed in the term 'genomic imprinting'.

Inevitably, as with all symposia proceedings, this volume does not provide a completely comprehensive and coordinated account of the subject. Nevertheless, the distinguished contributors cover a wide area to a high standard. I found this collection of papers informative and provocative and would recommend it to the many clinicians and scientists with an interest in this rapidly advancing field.

E R MAHER

Chromosome Banding. A T Sumner. (Pp 434; £60.00.) Glasgow: Harper Collins. 1990.

Chromosome banding refers to the patterns of bands which may be

induced along the length of chromosomes and the methods used to produce them. This comprehensive review by one of the modern pioneers in this field documents the important theoretical implications and practical applications of a branch of science which goes back for almost a century, but which has undergone explosive growth over the last 20 years.

After a brief historical survey, chapters on classification and chromosome structure are followed by 10 chapters which explore different types of banding from the ubiquitous G banding through to the use of immunocytochemistry or restriction endonucleases to produce banding patterns. In each case, methods, mechanisms, and applications are considered with several chapters ending with useful concluding remarks. Although the modern era of banding has been centred on human chromosomes, work with animal and plant chromosomes is included wherever relevant methods have been applied. The last three chapters deal with the surprising degree of chromosomal polymorphism found in many species, the types of evolutionary change deduced from banding patterns, and the implications of banding for genome organisation.

Although methods are dealt with in some detail, this book is not a techniques manual. Rather, its strength lies in the consideration of the mechanisms underlying different methods, and the broader implications which banding patterns have for chromosome structure and function, and for gene regulation. The book should therefore be of interest to those involved in the wider fields of cell, molecular, and evolutionary biology as well as to geneticists.

The author's patient and reflective style presents comparative data clearly; nor are ambiguities and contradictions glossed over. A comprehensive bibliography includes references from 1990 and the book contains many beautiful illustrations.

The gulf between our understanding of gene structure and function at the molecular level and the structure, function, and organisation of chromatin at the whole chromosome level should prove a fertile field for investigation for which this volume provides an excellent stimulus.

JOHN BARBER

NOTICES

Recent trends in medical biotechnology: an update

An International Seminar on 'Recent trends in medical biotechnology: an update' is being organised by the Departments of Pathology and Genetics (University of Madras) and Departments of Biotechnology (Anna University, Madras, and All India Institute of Medical Sciences, New Delhi) on 4 to 7 February 1992 in Madras. Apart from lectures by outstanding scientists from India and abroad, demonstrations of techniques and scientific exhibitions of products are also planned. Participants are invited to present posters. For information write to Dr (Mrs) M Madhavan, Department of Pathology, Dr A L Mudaliar Postgraduate Institute of Basic Medical Sciences, University of Madras, Taramani, Madras 600 113, India.

Genetics Services Provision—An International Perspective

This satellite meeting to the International Congress of Human Genetics will be held on 4 October 1991 at the Pan American Health Organization in Washington, DC. For further information contact Karen Greendale, New York State Department of Health, Wadsworth Center for Laboratories and Research, Room E-275, Empire State Plaza, PO Box 509, Albany, NY 12201-0509, USA. Fax (518) 474-8590.