Journal of

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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.

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.
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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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2 Sheffield LJ, Danks DM, Mayne V, Hutchinson LA. Chondrodysplasia punctata—23 cases of a mild and relatively common variety. *J Pediatr* 1976;**89**:916–23.
3 Horswell BB, Holmes AD, Barnett JS, Levant BA. Maxillonasal dysplasia (Binder's syndrome): a critical review and case study. *J Oral Maxillofac Surg* 1987;**45**:114–22.
4 Sheffield LJ, Halliday JL, Danks DM, Rogers JG, Poulos A, Morison N. Clinical, radiological and biochemical classification of chondrodysplasia punctata. *Am J Hum Genet* 1989;**45**: 64A.
5 Sheffield LJ, Halliday JL, Jensen FO, Danks DM. To lump or split types of chondrodysplasia punctata. *Proc Greenwood Genetic Center* (in press).
6 Sheffield LJ, White J, David DJ, Nugent M. Chondrodysplasia punctata (mild type) presenting as Binder's syndrome. *Pathology* 1984;**16**:104–5.
7 Maroteaux P. Brachytelephalangic chondrodysplasia punctata: a possible x-linked recessive form. *Hum Genet* 1989;**82**:167–70.
8 Shaul WL, Emery H, Hall JG. Chondrodysplasia punctata and maternal warfarin use during pregnancy. *Am J Dis Child* 1975;**129**:360–2.

BOOK REVIEW

Prenatal Diagnosis and Prognosis. Ed R Lilford. (Pp 245; £39·50.) London: Butterworth Scientific Ltd. 1990.

The editor's stated aim for this book is to concentrate on the prognosis rather than the diagnosis of disorders and abnormalities detected prenatally, since it is the prognosis for a disorder which influences parents' decision making, and current publications often lack this type of information. The book is highly successful in this regard, providing detailed, authoritative, and clear summaries of what is known about the outcome for malformations and chromosomal abnormalities detected prenatally. The book does not cover metabolic disorders, but includes sections on DNA technology and risk calculation, sampling procedures, and decision making.

The book starts with chapters on antenatal ultrasound diagnosis of craniospinal defects, and extra-craniospinal anomalies, with a separate chapter on renal tract anomalies. The authors take the useful approach of starting with an ultrasound abnormality and then discussing diagnostic possibilities and prognosis. Extensive reviews of published reports are presented and information regarding prognosis summarised in a form which is immediately useful to the clinician 'on the spot' who may not have time to review published reports personally, or may not have rapid access to the relevant ones. The authors give recommendations for further investigation and management, based on their own and reported experience, and several algorithms are included.

A chapter on screening for Down's syndrome leads the reader through the mathematical complexity of calculating composite risks and interpreting the results of multiple screening tests, providing information essential to clinicians setting up an antenatal screening programme or counselling patients. There is then an introductory chapter on chromosomes which provides a clear explanation of terminology and chromosome structure for those unfamiliar with modern cytogenetics. This is followed by an excellent chapter on prenatal diagnosis of chromosome anomalies which covers the identification of pregnancies at high risk, and the interpretation of results, including addressing the particularly difficult problems of identifying confined placental abnormalities and assessing the likely clinical significance of mosaics, de novo structural rearrangements, and supernumerary markers. This is the longest chapter in the book (61 pages) and contains a wealth of useful information, detracted from only by the complexity of the text subheadings.

Three subsequent chapters examine the role of recombinant DNA technology in clinical practice: an introduction to modern genetics; a description of current technology for DNA diagnosis; and an explanation of risk calculation using results from DNA studies. These chapters provide a very well presented outline of this rapidly developing area, essential reading for those not familiar with this aspect of prenatal diagnosis.

Chapters on invasive diagnostic procedures and decision analysis in prenatal diagnosis conclude a book that is highly informative and enjoyable to read. The multi-author text is well written throughout and there are many excellent illustrations and tables as well as algorithms suggesting management schemes. This book usefully fills a gap in the market and should be highly commended to all clinicians involved in any aspect of prenatal diagnosis.

HELEN KINGSTON

NOTICE

Clinical genetics: recent advances

The British Council are holding a one week course on 'Clinical genetics: recent advances' on 23 to 28 March 1992 at the Institute of Medical Genetics for Wales, Cardiff. Further information can be obtained from Courses Department, The British Council, 10 Spring Gardens, London SW1A 2BN. Tel: 071-389 4406/4264/4252.

Association of Clinical Cytogeneticists

The annual scientific meeting of the ACC will take place at Earnshaw Hall, University of Sheffield on 2 to 4 July 1991. The Scientific Sessions will include papers on aspects of clinical cytogenetics, molecular genetics, and the cytogenetics of malignant disorders. The guest lecturer will be Professor M Greaves who will be talking about aetiological mechanisms in leukaemia. For further information contact Mrs I Barnes, Centre for Human Genetics, 117 Manchester Road, Sheffield S10 5DN.