Notice to Contributors

Papers, which should be in duplicate and in the Vancouver style, should be sent to the Editor, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR. A stamped addressed postcard should be enclosed for return to author as acknowledgement of receipt of MS. Overseas authors should enclose an international reply paid coupon. Submission of a paper will be held to imply that it contains original work which has not been previously published. 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* with particularly instructive clinical or genetic features: to be not longer than 1000 words of text, two or at most three figures, one table (if necessary), and eight references. *Short reports* of unusual cases: to be not longer than 500 words of point form description with a clinical photograph and partial karyotype, if appropriate, and no more than two or three references. Single case reports will usually only be considered in one of these forms. *Review articles* will generally be by invitation, but suggestions from authors wishing to prepare a review article will be welcome. *Annotations*, *Hypotheses*, *Preliminary communications*, and *Technical notes* will also be considered, as will *Short communications* giving information on new translocations, chromosome identification by banding techniques, and second and third findings of important haemoglobins. Contributions to the *Correspondence* and *Question and answer* columns will be welcomed. Publication of papers thought to be of special importance may be expedited.

All contributions should be accompanied by a summary 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. Twenty-five free reprints will be supplied and further 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. Pedigrees should use squares and circles. Generations should be numbered with Roman and individuals with Arabic numerals; members belonging to the same generation should be horizontally aligned.

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

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

Nomenclature. Authors should refer to the following publications.

(1) Chromosomes: ISCN. An international system for human cytogenetic nomenclature (1978). *Birth Defects* 1978; **XIV**:No 8. Also in *Cytogenet Cell Genet* 1978;**21**: 309–404.

(2) Dermatoglyphs: Penrose LS. Memorandum on dermatoglyphic nomenclature. *Birth Defects* 1968;**4**:No 3.

(3) Enzymes: WHO Scientific Group. Standardization of procedures for the study of glucose-6-phosphate dehydrogenase. *WHO Tech Rep Ser* 1967;No 366.

(4) Blood coagulation: International Committee of Haemostasis and Thrombosis (Graham JB *et al*). A genetic nomenclature for human blood coagulation. *Thromb Haemostas* 1973;**30**:2–11.

(5) Loci: Conventional nomenclature should be used, with lower case lettering as appropriate (for example, Race RR, Sanger R. *Blood groups in man*. 6th ed. Oxford, London: Blackwell, 1975; and Giblett ER. *Genetic markers in human blood*. Oxford, London: Blackwell, 1969).

(6) Genes: Shows TB *et al*. International system for human gene nomenclature (1979). *Cytogenet Cell Genet* 1979;**25**:96–116.

SI units. The units in which the authors' work was measured should be cited first followed by either the SI units or the traditional units. This does not apply to tables, but here a conversion factor should be added as a footnote.

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Announcements

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THE FIRST INTERNATIONAL SYMPOSIUM ON REPRODUCTIVE MEDICINE

The First International Symposium on Reproductive Medicine will be held on 4–6 October 1984 in Houston, Texas, USA. For further information contact: *Europe*: G Frajese, Member of Steering Committee, Clinica Medica 5, University of Rome Medical School, Via di Porta Pinciana 34, 00187 Roma, Italy. *USA*: E Steinberger, Member of Steering Committee, Department of Reproductive Medicine and Biology, University of Texas Medical School, PO Box 20708, Houston, Texas 77025, USA.

SPECIAL METHODS IN HUMAN CYTOGENETICS

A laboratory course on recent advances in cytogenetics methods will be offered by the Laboratory of Genetics, Department of Pathology, Memorial Sloan-Kettering Cancer Center, New York City, during the period 6–10 October 1983. The following techniques will be covered by means of lectures and laboratory exercises. (1) Handling of germ cells for meiotic chromosome studies. (2) Sperm penetration assay and sperm cytogenetics using the Humster system. (3) High resolution chromosome analysis of meiotic, somatic, and tumour cells. (4) Mapping of single copy genes by in situ hybridisation. (5) Tumour cytogenetics, including clonal assays. Enrolment will be limited to 20 candidates with previous experience in human cytogenetics. Course fee is $1000. Apply as soon as possible with a brief statement of background and experience in cytogenetics and area(s) of special interest to Dr R S K Chaganti, Memorial Sloan-Kettering Cancer Center, 1275 York Avenue, New York, NY 10021, USA. (Telephone 212/794-7100.)

XERODERMA PIGMENTOSUM REGISTRY

Xeroderma pigmentosum (XP) is a rare, autosomal recessive disease (frequency, one to four per million) in which affected persons exhibit sun sensitivity, cutaneous pigmentary abnormalities, and a high incidence of skin cancer. In addition, some patients with XP have neurological abnormalities such as mental deterioration or sensorineural deafness. Many of these symptoms may be manifestations of repeated environmental insults on a defective DNA repair system; cells from most patients with XP have been shown to have deficient excision repair of thymine dimers in ultraviolet (UV) radiation damaged DNA. The XP patients without this defect ('XP variants') appear to have another, more subtle, deficiency in DNA repair. Sun sensitivity and defective DNA repair is associated with mutagenesis and increased killing of XP cells following UV irradiation. XP is thus the best human model currently available for study of environmental-genetic interaction.

Using cell fusion techniques, seven different complementation groups of XP (named A, B, C, etc) have been reported in addition to the variant group. At the molecular level, XP has proved to be far more complex than anticipated. The precise molecule that is defective has not yet been identified despite extensive research. Characteristic clinical and epidemiological features exist among these complementation groups; group C, for example, is the most common group in the United States and in Europe, but is rare in Japan, where group A predominates. Neurological defects have been found in two groups, A and D, but differ in age of onset between them. Melanoma is common in American and European patients, but is extremely rare in Japan. Furthermore, XP cells in vitro are also abnormally sensitive to a number of chemical carcinogens, such as acetoxyacetoaminofluorene and nitroquinoline oxide, suggesting that XP patients might be expected to develop abnormally large numbers of internal neoplasms as well as skin cancers, but to date only a very few such tumours have been reported.

There is a large number of unanswered questions about XP that relate to disease symptoms and clinical course, as well as to fundamental issues in carcinogenesis. In order to obtain answers to these questions, many of which are largely amenable to epidemiological analysis, the Xeroderma Pigmentosum Registry has been formed.

The Registry is a data gathering organisation and there is no intent to assume management of individual patients. Cooperation of physicians who know of patients with XP is essential for the Registry to accomplish its mission. Physicians who are aware of such patients should write to: Xeroderma Pigmentosum Registry, C/o Dr W Clark Lambert, Department of Pathology, Room C520, UMDNJ-New Jersey Medical School, 100 Bergen Street, Newark, New Jersey 07103, USA. (Telephone (201) 456-5722 or 456-4841.)

12TH ANNUAL MEETING OF EUROPEAN WORKING GROUP FOR CYSTIC FIBROSIS

The 12th Annual Meeting of the European Working

Group for Cystic Fibrosis will be held in Athens, Greece, on 3–4 October 1983. This meeting will be preceded by the Annual Meeting of the International Cystic Fibrosis (Mucoviscidosis) Association in the same venue on 1–2 October 1983. Further information on these two meetings is obtainable from Ron Tucker, Executive Director, Cystic Fibrosis Research Trust, Alexandra House, 5 Blyth Road, Bromley, Kent BR1 3RS. (Telephone 01-464 7211.)

THE XX CONGRESS OF THE INTERNATIONAL SOCIETY OF HEMATOLOGY

The XX Congress of the International Society of Hematology will be held in Buenos Aires, Argentina, from 1–7 September 1984. Spanish and English will be the official languages of the Congress. The scientific programme will be centred on fundamental issues of hematology and will be developed through different kinds of activities, such as plenary sessions, symposia, workshops, and poster discussion sessions. Included in the Congress week will be a 2-day educational programme. The Secretariat of the Congress will gladly deliver more detailed information on request. All inquiries about the Congress should be addressed to: XX Congress of the International Society of Hematology, Viamonte 2008, 1056 Buenos Aires, Argentina.