

Notice to contributors

The *Journal of Medical Genetics* publishes original research relevant to medical genetics, along with reviews, annotations, and editorials on important and topical subjects. It also acts as a forum for discussion, debate, and information exchange through its Letters to the Editor column, conference reports, and notices. The journal particularly encourages submissions on the molecular basis of human disease, the clinical manifestations of genetic disorders, applications of molecular genetics to medical practice, and the systematic evaluation of such applications. The journal attempts to handle the review process and publication as expeditiously as possible. Accelerated publication is available where warranted by scientific urgency and recommended by reviewers. Submissions are accepted only on the understanding that they have not been and will not be published elsewhere, and are subject to editorial revision. They should be sent to:

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Manuscripts from North America may be sent directly to the North American Editor Manuscripts that do not conform to the following instructions may be returned for modification before being reviewed.

- **All submissions** must be typewritten on one side of the paper only, using double spacing and ample margins. A total of three copies of articles and letters should be supplied, together with three copies of any tables or figures. The pages should be numbered. All abbreviations must be defined.

- Manuscripts must be accompanied by:

- **Covering Letter** Each author must sign the covering letter as evidence of consent to publication and all authors will be required to transfer copyright of their articles to the journal before publication.

- **Consent** All identifiable photographs of patients should be accompanied by written permission for use. Pedigrees, particularly those containing information on people who may be presymptomatic carriers of later onset disease, should, as far as possible, be anonymised.

- **Ethical approval** The critical assessment of submitted papers will include ethical considerations; documentary evidence (where relevant) of ethical committee review and approval will be very helpful.

- **Title sheet** stating the title of the paper, as well as a short title, the authors, their departments and institutions, and the name and postal address (+ Tel/Fax numbers) of the corresponding author.

- **Referees** Authors are welcome to suggest people particularly competent to act as referees; although the journal may not use the suggested referees on that occasion, they will be added to our database. Conversely, authors' wishes on those whom they would prefer not to referee their work will be respected.

Categories of contributions

1. Original papers represent a substantial body of laboratory or clinical work. The study should be presented in sections, namely:

- **Abstract** No more than 250 words, summarising the problem being considered in the study, how the study was performed, the salient results and the principal conclusions of the study. *Structured abstracts* (in the format used by eg the *British Medical Journal* and the *Journal of the American Medical Association*) have been shown to be more useful to readers, and their use is encouraged wherever possible.
- **Keywords** (maximum of four) These should be given beneath the abstract.

- **Introduction** Brief description of the background that led to the study.
- **Methods** Details relevant to the conduct of the study. Statistical methods should be clearly explained at the end of this section.

- **Results** Work should be reported in SI units. Undue repetition in text and tables should be avoided. Comment on validity and significance of results is appropriate but broader discussion of their implications should be placed in the next section.
- **Discussion** The nature and findings of the study are placed in context of other relevant, published data. Subheadings that aid clarity of presentation are encouraged.

- **Data access** Reference should be made to availability of detailed data, either through public databases or otherwise, and to availability of materials used for reported investigations. It is generally expected that

genomic and similar data should be lodged in appropriate public databases at or before the time of publication. Authors are encouraged to make DNA or cell lines available to other workers.

- **Acknowledgments and affiliations** People with direct involvement in the study but not included in authorship may be acknowledged. The source of financial support and industry affiliations of all those involved should be stated.

- **References** In accordance with the Vancouver agreement these are cited by the numerical system and listed in the order cited in the text, not in alphabetical order by authors' names. (In the text, the reference number should be given between square brackets on the line, not superscript.) All authors should be listed. Journal titles are abbreviated in accordance with the style of Index Medicus. See references in the journal.

- **Figures** should be kept to a minimum and should be numbered consecutively in Arabic numerals. Legends should be typed on a separate sheet.

- **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 above the table.

- **Illustrations** Colour illustrations can be accepted; however, authors are asked to pay part of the cost.

- **Nomenclature** Current standard international nomenclature should be adhered to.

Chromosomes: *ISCN 1995. An International System for Human Cytogenetic Nomenclature*, Mitelman F (ed); S Karger, Basel, 1995

Genes: **McAlpine P. In: The Genetics Nomenclature Guide (Human).** *Trends in Genetics Supplement*, March 1995. For latest references, instructions, and list of approved symbols see URL <http://www.gene.ucl.ac.uk/nomenclature/> (or contact nome@galton.ucl.ac.uk)

Enzymes: Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry. New York: Academic Press, 1992 Information also available on URL <http://expasy.hcuge.ch/sprot/enzyme.html>

2. Short reports

A brief communication presenting laboratory or clinical work, collected case reports, or single case reports. Reports of single mutations at loci which have already been documented will be published only if they are of unusual clinical or biological interest. The format can be identical to Original papers (see above) but in many circumstances the main body of the text may be better presented without division into sections. Short reports are intended to occupy no more than 2 printed pages; equivalent to about 1000 words, 2 tables/figures, and about 15 references. Brevity and clarity are always likely to enhance the chance of a manuscript being accepted for publication.

3. Review articles

Authors are welcome to discuss possible topics for review directly with the Editor.

4. Syndrome of the month

These reviews are commissioned by special editors, to whom suggestions can be addressed.

5. Hypothesis articles

Contributions which present an interesting theory, discussed in relation to published data, are welcome.

6. Diagnostic advances

A limited number of brief notes, occupying no more than one printed page, with novel technical information whose dissemination is likely to be of importance to those involved in running clinical and laboratory genetic services.

7. Letters

These are welcome on any relevant topic and will be published rapidly. Those relating to or responding to previously published items in the journal will be shown to those authors, where appropriate.

Proofs

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. For commercial reprints, contact Sheila Rowe. Tel: 0171 383 6450. Fax: 0171 383 6556. **Disk version** Manuscripts are published directly from disk. Final revised versions of papers will therefore be needed as hard copy and on disk. Instructions will be sent to authors on invitation to revise or on acceptance.

A complimentary copy of the journal will be sent to the corresponding author on publication.