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### SCOPE

*Genetics in Medicine*, the official journal of the American College of Medical Genetics and Genomics, aims to enhance the knowledge and practice of medical genetics. We publish original articles that are relevant to the broad aspects of medical genetics, including manuscripts in the areas of clinical genetics, biochemical genetics, cytogenetics, molecular genetics, public health genetics, genetic epidemiology, genetics counseling, genetic education, clinical informatics, ethical, legal and social implications of genetics and genetics legacies. Innovation, the quality of study design and methodology, and the ability to disseminate knowledge in training or delivery of genetic medicine will be considered during the review process. *Genetics in Medicine* does not publish case reports. Submitted manuscripts should not contain previously published material.

### EDITOR-IN-CHIEF

James P. Evans, MD, PhD  
University of North Carolina  
Department of Genetics  
5095 Genetic Medicine Building  
Campus Box 7264  
Chapel Hill, NC 27599-7264

### MANAGING EDITOR

Jan Higgins, PhD, ELS  
American College of Medical Genetics and Genomics  
7220 Wisconsin Avenue, Suite 300  
Bethesda, MD 20814  
Fax: 888-706-2009  
jim@acmg.net

### PREPARATION OF MANUSCRIPTS

*Genetics in Medicine* publishes:

- Commentaries
- Systematic review articles
- Reviews
- Original research articles
- Brief reports
- Education reports
- Letters to the editor

Reviews, Commentaries, Genetic Legacy and Special articles are usually solicited by the Editor.

*Genetics in Medicine* does not publish individual case reports.

### DOCUMENTS FROM ACMG COMMITTEES

Articles submitted from ACMG committees are to be <4,500 words, 45 references and 5 figures/tables. Word limits exclude abstract, tables, figures, references and online-only material. Display items include tables and figures. Each printed page counts as one display item, therefore if any one table or figure takes up more than one printed page it will be rounded up to the next full page for counting purposes. For further information, please contact Jan Higgins, Managing Editor, at jim@acmg.net.

### PREPARATION OF SUBMISSIONS

Manuscripts must be submitted in clear, concise English. They should be double-spaced including references, figure legends, and tables, on one side of the page only. Leave 1-inch margins on all sides. Do not use justified margins or line numbers. Number every page. Assemble the manuscript file in the following order: (1) title page, (2) abstract page and key words, (3) text, (4) acknowledgments, (5) references (6) figure legends. Tables and figures are uploaded as separate files in the system, using the appropriate file category. They should come after the manuscript file and be in sequential order.

### Title page

The first page of the manuscript should include, in the following order: (1) title of paper, (2) short running title, (3) authors' full name and academic degrees (no more than two), (4) authors' primary affiliations, and (5) a specific mailing address, telephone number, fax number, and e-mail address to whom correspondence concerning the manuscript should be sent.

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Authors must disclose, on a separate page, any commercial association that might pose or create a conflict of interest with the information presented in any submitted manuscript. Such associations include consultancies, stock ownership, or other equity interests, patent licensing arrangements and payments for conducting or publicizing a study described in the manuscript. All sources of funds supporting the work must be disclosed. Additional detail, if necessary, may be provided in a separate statement to the editors. This information will not influence the editorial decision. See p6 for more information.

### Abstract and keywords

References should not be cited in the abstract. Please be concise (200 words maximum). Do not use unusual abbreviations. However, if an acronym or abbreviation appears more than twice in the abstract, spell out on first use and use the abbreviation thereafter. For original articles, brief reports, and systematic reviews, the abstract must be structured to the journal style to include a Purpose (the rationale for the study), Methods (a brief description of methods), Results (presentation of significant results) and Conclusion (a succinct statement of data interpretation). For all other types of manuscript submissions, an unstructured abstract is acceptable. Then, skip a line under the abstract and provide five key words, including those in the title, to be used as indexing terms.

**Text**

The text should be organized in Sections in the following order (a) INTRODUCTION, (b) MATERIALS AND METHODS, (c) RESULTS, and (d) DISCUSSION. Section headings should be CAPITALIZED and bolded and aligned on the left margin. Subheadings should be used where appropriate and should be in Title Case, bold face, and also aligned on the left margin.

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The Introduction should assume that the reader is knowledgeable in the field and should therefore be as brief as possible but can include a short historical review where desirable.

**Materials (or patients) and methods**

This section should be brief, but contain sufficient detail, so that all experimental procedures can be reproduced, and include references. Methods, however, that have been published in detail elsewhere should not be described in detail. Authors should provide the name of the manufacturer and their location for any specifically named medical equipment and instruments, and all drugs should be identified by their pharmaceutical names, and by their trade name if relevant.

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The Results section should briefly present the experimental data in text, tables or figures. Tables and figures should not be described extensively in the text, either. We will accept data reported using conventional data, but will require all data to also be in SI units. Please refer to the *American Medical Association Manual of*

Article type	Description	Abstract	Word count <sup>a</sup>	Display items <sup>b</sup>	References (max.)
Original research articles	This category is self-explanatory. Introduction, Materials and Methods, Results, and Discussion sections are required.	Yes, structured, 200 words maximum	4,000	5	40
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Reviews	Reviews of current topics in medical genetics. Reviews are generally commissioned; however, the Editor will also consider unsolicited reviews. Please contact the Editorial Office at <a href="mailto:gim@acmg.net">gim@acmg.net</a> with any manuscript topic that may be appropriate as a review.	Yes, unstructured, 200 words maximum	5,000	5	75
Systematic review articles	A systematic review is a literature review focused on answering one or a few questions. The review attempts to identify, appraise, select and synthesize evidence relevant to those questions. A systematic review uses an objective and transparent approach for research synthesis, with the aim of minimizing bias. Systematic evidence reviews can inform clinical practice guidelines and identify gaps in our understanding of genetic issues in medicine and public health.  We refer all authors to the PRISMA website: <a href="http://prisma-statement.org/">http://prisma-statement.org/</a> and require that all submitted reviews in this category have the PRISMA flow diagram ( <a href="http://prisma-statement.org/documents/PRISMA%202009%20flow%20diagram.doc">http://prisma-statement.org/documents/PRISMA%202009%20flow%20diagram.doc</a> ) as Figure 1 of the paper. Please also submit with your article a completed PRISMA checklist ( <a href="http://prisma-statement.org/documents/PRISMA%202009%20checklist.doc">http://prisma-statement.org/documents/PRISMA%202009%20checklist.doc</a> ).	Yes, structured, 200 words maximum	5,000	5	We have no limit on the number of references for the articles meeting inclusion criteria for the review. However we restrict other references to 25. Please email the managing editor at <a href="mailto:gim@acmg.net">gim@acmg.net</a> for further details.
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Education reports	Papers that describe new educational initiatives or approaches to genetics education. A rigorous experimental design and presentation of data to validate the approach are preferred, but are not necessarily required as criteria for publication. Introduction, Materials and Methods, Results, and Discussion sections are required.	Yes, structured, 200 words maximum	4,000	5	40
Genetic Legacy	A brief descriptive article that is a combination of history, biography and autobiography that sum to characterize the nature and impact of being a medical geneticist in the 21st Century. Optimally, the citing and elaboration of past events and associated challenges will attract and inspire those who are considering or have already chosen medical genetics as a career option. These articles are by invite only, however anyone with an interest in submitting a Genetic Legacy article should contact the managing editor at <a href="mailto:gim@acmg.net">gim@acmg.net</a> first.	No	1,500	1 one small photo, figure or table	10
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The Discussion should focus on the interpretation and the significance of the findings with concise objective comments that describe their relation to other work in the area. It should not repeat information in the results. The final paragraph should highlight the main conclusion(s), and provide some indication of the direction future research should take.

## Acknowledgments

Acknowledgments should be brief, and should include all sources of support including sponsorship (e.g., university, charity, commercial organization) and sources of material (e.g., novel drugs) not available commercially, plus substantive contributions by individuals. While expressing appreciation to another scientist for assistance with the research or manuscript, enclose written permission since such an acknowledgment may imply endorsement of the data and conclusions. Acknowledgment of nonscientific assistance (typist, manuscript preparation, and secretarial help) should not be included. However, where a language editing company has given assistance an acknowledgment should be included. Please do not thank the anonymous reviewers.

## References

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**Chapter in a book:** Friedman JM, Dill FJ, Hayden MR. Population genetics. In: Friedman JM, Dill FJ, Hayden MR, editors. NMS genetics. Baltimore: Williams & Wilkins, 1996:105-109.

**Meeting paper:** Kessler S. Transcripts of genetic counseling sessions: exercises in missed opportunities. Paper presented at Talking Human Genetics: Verbal Communication, Knowledge and Genetic Makeup. Hamburg, April 24-27, 1997.

**Thesis/dissertation:** Hellsten E. Positional cloning of the infantile ceroid lipofuscinosis gene [dissertation]. Helsinki: National Public Health Institute, 1995.

**World Wide Web:** Page Institut Curie, 1997. Available at: <http://www.curie.fr/curie/sm/brca>. Accessed January 30, 2000.

**Computer software:** SAS Institute [computer program]. Release 6.12. Cary, NC: SAS Institute, 1997.

**Online database:** OMIM (Online Mendelian inheritance in man). Baltimore: Johns Hopkins University, Center for Medical Genetics, 1996. <http://www3.ncbi.nlm.nih.gov/omim/>(September)

**Work "in press":** Hedrick PW, Black FL. HLA and mate selection: no evidence in South Amerindians. *Am J Hum Genet*. In press.

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- MPEG movie files (.mpg)
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