

TABLE OF CONTENTS

1	SCOPE
1	PREPARATION OF MANUSCRIPTS
1	Presubmission Inquiries
1	Preparation of submissions
1	Cover Page
1	Title page
1	Conflict of interest notification page
2	Abstract and keywords
2	Text
2	Introduction
2	Materials (or patients) and methods
2	Results and discussion
2	Acknowledgements
3	References
3	FIGURES AND ARTWORK
3	GUIDELINES
3	Color figures
3	Color charge
3	Color on the web
3	Black and white images
4	Graphs, histograms and statistics
4	Tables
4	ENGLISH LANGUAGE SUPPORT
4	SUPPLEMENTARY INFORMATION
4	SUBMISSION OF MANUSCRIPTS
4	Peer review process and reviewer recommendations
4	License to Publish
5	Manuscript deposition service
5	Advance Online Publication
5	Proofs
5	Offprints
5	Open Access and self-archiving
5	EDITORIAL POLICIES
5	Conflict of interest
5	Clinical trials
6	Peer review policy
6	Anonymity
6	Selection of peer reviewers
6	Authorship
6	Copyright
7	Copyright permissions
7	Plagiarism and fabrication
7	Duplicate publication
7	Image manipulation
7	Correction and retraction policy
7	Research Data Policy
7	Reporting Guidelines
8	Gene Nomenclature
8	BIOETHICS
8	Human and other animal experiments
8	Genetics disorders
8	Biosecurity policy
8	COMMUNICATION
8	Correspondence with the journal
8	Communication with the media
9	Communication between scientists
9	BUSINESS CONTACT

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, and ethical, legal and social implications of genetics. Innovation and influence on research and practice, as well as quality, methodology and impact on the 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

Robert D. Steiner, MD, FAAP,

FACMG

University of Wisconsin School
of Medicine and Public Health

geneticsinmedicine@acmg.net

MANAGING EDITOR

Jan Higgins, PhD, ELS

American College of Medical Genetics
and Genomics (ACMG)

7101 Wisconsin Avenue, Suite 1101

Bethesda, MD 20814

Tel: +1 (240) 204-9650

geneticsinmedicine@acmg.net

EDITORIAL ASSISTANT/ SOCIAL MEDIA MANAGER

Katie Murphy

American College of Medical Genetics
and Genomics (ACMG)

7101 Wisconsin Avenue, Suite 1101

Bethesda, MD 20814

Tel: +1 (240) 204-9856

geneticsinmedicine@acmg.net

PREPARATION OF MANUSCRIPTS

Genetics in Medicine publishes:

- Comments
- Systematic review articles
- Reviews
- Articles
- Brief Communication
- Education reports
- Correspondence

Reviews and Comments are usually solicited by the Editor. If you have an article that is suitable for GIM but does not readily fit into one of the article types above, please contact the Editorial Office at geneticsinmedicine@acmg.net.

Case reports/single patient/family gene-disease association reports: *Genetics in Medicine* rarely publishes case reports or reports of gene-disease associations in a single patient/family; exceptional examples may be appropriate for consideration if they are likely to be of considerable interest to a broad audience.

PRESUBMISSION INQUIRIES

Presubmission inquiries are provided as a service to authors (e.g. to see if a manuscript is likely to be of interest to the journal) and are not mandatory nor even necessarily recommended. *Genetics in Medicine* cannot guarantee the rapidity of response to inquiries, but will strive to answer within two business days. Please submit presubmission inquiries through our submission system at <https://www.editorialmanager.com/gim> and select the article type "presubmission inquiry". Contact us directly if the response is delayed. Presubmission inquiries are a means of soliciting informal feedback on whether a manuscript falls within the scope of the journal and is likely to meet the editors' standards. Presubmission inquiry does not take the place of formal manuscript review. The process, by nature, is imperfect. Editors are being asked to review only a very small sample of a manuscript to determine its appropriateness for the journal. In addition, with multiple inquiries coming in daily and a subset of the manuscripts never being submitted despite positive responses, editors don't have the luxury of spending significant time on presubmission inquiries. Authors should take the response as a recommendation and nothing more. A positive response is no guarantee that a manuscript will be sent out for review or accepted for publication. A negative response does not guarantee rejection if the full manuscript is submitted. Finally, we do not accept appeals to presubmission inquiry responses. Authors who wish to submit despite a negative response should follow the journal's author instructions for submission.

Minimum necessary for presubmission inquiry:

Entire manuscripts should NOT be sent as presubmission, but rather submitted as specified in the journal's instruction for authors. The cover letter from the corresponding author requesting a presubmission inquiry should include:

- Manuscript title and abstract
- Full manuscript author list and affiliations
- Significant real or potential author conflicts of interest
- Brief introduction to the manuscript and statement of interest to the journal
- Brief description of relevant findings and their implications

PREPARATION OF SUBMISSIONS

Manuscripts must be submitted in clear, concise English with:

- double-spaced text including references, figure legends, and tables
- 1-inch margin on all sides
- no justified margins
- no line numbers
- page numbers

Assemble the manuscript file in the following order: (1) cover letter, (2) title page, (3) abstract page and key words, (4) text, (5) acknowledgments, (6) references (7) figure legends. Tables and figures should be uploaded as separate files in the submission system, using the appropriate file category and should come after the manuscript file and be in sequential order.

COVER LETTER

Genetics in Medicine requires that most submissions be accompanied by a brief (one to two pages) cover letter and encourages the inclusion of one if not required. The cover letter is your opportunity to briefly explain to the editors why the manuscript is sufficiently novel and important to warrant publication in GIM.

In your cover letter, we also appreciate a brief explanation of why the manuscript is a good "fit" for GIM and why our readership, in particular, would be interested in it or would find it useful. Remember that GIM's readership is typically focused on clinical issues (including patient care, the clinical laboratory and clinically-oriented research). Thus, pointing out how our readers would find your work of use is valuable.

-
-

Calling our attention to similar work that has appeared in GIM previously (by your own group or others) can demonstrate that the topic of the manuscript is of relevance to our readers if it carries such previously published work further or sheds new light on those general topics.

In the interests of transparency and to help readers form their own judgments of potential bias, authors must declare whether or not there are any competing financial interests in relation to the work described. This information must be included in the cover letter.

The corresponding author is responsible for alerting the editorial office, in the cover letter, if the manuscript includes subjects about which a previous report has been published or about a manuscript that is submitted to, under review by, in press at, or to be submitted to or published in another journal in the future. Contributions being prepared for or submitted to *Genetics in Medicine* can be posted on recognized preprint servers (such as ArXiv), and on collaborative web-sites such as wikis or the author's blog. The website and URL must be identified to the editor in the cover letter.

NLM Research Reporting Guidelines and Initiatives and EQUATOR list many guidelines to promote high quality of published research. The editors at GIM are encouraging authors to seek out appropriate guidelines for their studies, to fill in checklists if available, upload those checklists with your manuscript at submission, and to detail in the cover letter what guidelines were followed in producing your manuscript.

Title page

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

Conflict of interest notification page

Authors must disclose, on a separate page, any commercial association that might pose, create or create the appearance of 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.

Abstract and keywords

References should not be cited in the abstract. Please be concise (200 words maximum in the abstract). 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 communications, 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 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.

Abbreviations must be defined at first mention in the text, table, and each figure.

Terminology

Please use the following terms:

genome sequencing instead of whole genome sequencing
 exome sequencing instead of whole exome sequencing.
 pathogenic variant instead of mutation
 secondary finding instead of incidental finding

For more information please refer to: [Jarvik GP, Evans JP. Mastering genomic terminology. Genet Med. 2016;19:491.](#)

Introduction

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 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.

Results and discussion

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 Style* SI conversion units table at http://www.amamanualofstyle.com/oso/public/jama/si_conversion_table.html for calculating SI units from conventional units.

The Discussion should begin with a very succinct summary of the major conclusions of the paper and then go on to focus on the interpretation and 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 provide some indication of the direction future research should, or could, take.

Article type	Description	Abstract	Word count ^a	Display items ^b	References (max.)
Article	This category is self-explanatory. Introduction, Materials and Methods, Results, and Discussion sections are required.	Yes, structured, als 200 words maximum	4,000	5	40
Brief Communication	Reports of important and relevant clinical observations that inform the practice of Clinical Medical Genetics. Introduction, Materials and Methods, Results, and Discussion sections are required.	Yes, structured, 200 words maximum	2,000	2	20
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 gim@acmg.net with any manuscript topic that may be appropriate as a review.	Yes, unstructured, 200 words maximum	5,000	5	75
Systematic review articles	<p>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.</p> <p>We refer all authors to the PRISMA website: http://prisma-statement.org/ and require that all submitted reviews in this category have the PRISMA flow diagram (http://prisma-statement.org/documents/PRISMA%202009%20flow%20diagram.doc) as Figure 1 of the paper. Please ensure you submit the correct checklist with your article. There are several to choose from here: http://prisma-statement.org/</p>	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 editorial office at geneticsinmedicine@acmg.net for further details.
Comment	Authoritative opinions or discussions of current topics important to the field and to readers of <i>Genetics in Medicine</i> . These are generally commissioned; however, the Editor will also consider unsolicited editorials.	No	1,500 words	0	10
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
Correspondence	Responses to published papers.	No	1,000	0	10

^aExcluding abstract, tables, figures, references, and online-only material

^bThis includes 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 count as more than one display item.

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. If 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

Accuracy of reference data is the author's responsibility. Verify all entries against original sources, especially journal titles, inclusive page numbers, publication dates, accents, diacritical marks, and spelling in languages other than English. List the references in the numerical order in which they appear in the manuscript. Reference citations will appear as superscript numbers in the text. Only list references that appear within the text; supplemental references should appear only in the supplemental material. List all authors if there are six or fewer; if there are more than six authors, list the first three and then et al. Avoid using abstracts as references. Personal communications, unpublished observations, and submitted manuscripts are not legitimate references: they must be cited in the text as "(unpublished data)." Journal titles should be abbreviated according to PubMed. If it is unclear how a reference should be formatted, please consult the *American Medical Association Manual of Style: A Guide for Authors and Editors*, 10th edition, pages 39-79. *Genetics in Medicine* will accept manuscripts generated using the Reference Manager and Endnote JAMA template. Sample reference styles are as follows:

Journal article: Pomponio RJ, Hymes J, Reynolds TR, et al. Mutations in the human biotinidase gene that cause profound biotinidase deficiency in symptomatic children: molecular, biochemical, and clinical analysis. *Pediatr Res*. 1997;42:840-848.

Book: Gelehrter TD, Collins FS, Ginsburg D, editors. *Principles of medical genetics*, 2nd ed. Baltimore: Williams & Wilkins; 1998.

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, 24-27 April 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. <http://www.curie.fr/curie/sml/bca>. Accessed 30 January 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/\(September\)](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.

Work submitted to a publication but not accepted, personal communications, preprints, and unpublished observations are not legitimate references: They must be cited in the text as "(unpublished data)" or "(personal communication)," as appropriate. Be prepared to submit letters of authorization from the sources if the manuscript is accepted for publication.

Preprint: See above. Refer to preprints in the body of the text as (unpublished data).

To cite a paper published in *Genetics in Medicine*, use the following abbreviation: *Genet Med*.

FIGURES AND ARTWORK GUIDELINES

Figures and images should be labeled sequentially, numbered, and cited in the text. Figure legends should be brief, specific and appear on a separate manuscript page after the References section. Refer to (and cite) figures specifically in the text of the paper. Figures should not be embedded within the text. If a table or figure has been published before, the authors must obtain written permission to reproduce the material in both print and electronic formats from the copyright owner and submit it with the manuscript. This follows for quotes, illustrations and other materials taken from previously published works not in the public domain. The original source should be cited in the figure caption or table footnote. Do not use three-dimensional histograms when the addition of the third dimension gives no extra information. Scale markers should be used in images taken on a microscope and indicate the type of stain used. Please note that red and green must not be used together in a figure as some readers cannot perceive a difference between them. Detailed guidelines for submitting artwork

can be found by downloading the artwork guidelines PDF.

It is preferred that you submit production quality artwork with your initial online submission. Then, if your article is accepted, we will not require the artwork to be resubmitted following the peer-review process. We can review articles that do not contain production quality artwork, but if your article is accepted, the artwork quality will need to be improved prior to publication.

Color figures

Color figures for production must be supplied in the following format:

Minimum resolutions:

Halftone images	300 dpi (dots per inch)
Color images	300 dpi saved as CMYK
Images containing text	400 dpi
Line art	1,000 dpi

Sizes:

Figure width - single image	86 mm (should be able to fit into a single column of the printed journal)
Figure width - multi-part image	178 mm (should be able to fit into a double column of the printed journal)
Text size	8 point (should be readable after reduction - avoid large type or thick lines)
Line width	Between 0.5 and 1 point

Color charges apply for figures printed in color: \$500 for the first color figure and \$250 for each subsequent figure. Current ACMG members who are first or senior/ corresponding authors are exempt, as are authors who have opted for Open Access. This exemption does not include student members. Authors may choose to have their figures in color in the HTML online-only version of the manuscript and in black and white in the PDF/print version of the manuscript (gratis) by providing both color and greyscale versions of each, and opting out of color charges at submission and on the Color Artwork form. The Color Artwork is available to download upon resubmission.

Please note, for ACMG members claiming a color fee waiver, color figures in the printed journal are at the managing editor's discretion.

Color on the web

Authors who wish their articles to have FREE color figures on the web (only available in the HTML, full text version of manuscripts) must supply separate files in the following format. These files should be submitted as supplementary information and authors are asked to mention that they would like color figures on the web in their submission letter.

For single images:

Width	500 pixels (authors should select "constrain proportions," or equivalent instructions, to allow the application to set the correct height automatically.)
Resolution	125 dpi (dots per inch)
Format	JPEG for photographs GIF for line drawings or charts
Filenaming	Please save image with .jpg or .gif extension to ensure it can be read by all platforms and graphics packages.

For multi-part images:

Width	900 pixels (authors should select "constrain proportions," or equivalent instructions, to allow the application to set the correct height automatically.)
Resolution	125 dpi (dots per inch)
Format	JPEG for photographs GIF for line drawings or charts
Filenaming	Please save image with .jpg or .gif extension to ensure it can be read by all platforms and graphics packages.

Black and white images

- Image resolution of at least 300 dpi at publication size
- Images should be scanned at a minimum of 300 dpi
- During software manipulation of images, care should be taken that resolution is maintained
- Images may be rotated or scaled, but this must be the same in the x and

- y dimensions
- Contrast and brightness can be adjusted, but this must be uniform across the entire image, and must not result in the loss of any feature, band or spot. The background should still be visible
- If lanes are removed, and once separate parts of an image are joined together, a black, white or grey line should indicate clearly where the image was cut
- If black borders are drawn around the image, the lines should correspond to all edges where the image was cut
- Protein molecular weights or DNA fragment sizes should be indicated for all figure panels showing gel electrophoresis

Graphs, histograms and statistics

- Error bars must be described in the figure legend
- Axes on graphs should extend to zero, except for log axes
- Statistical analyses (including error bars and *p* values) should only be shown for independently repeated experiments, and must not be shown for replicates of a single experiment
- The number of times an experiment was repeated (*N*) must be stated in the legend

Tables

Tables must be cited in the text. Each table must begin on a separate page, double-spaced. The table number should be Arabic followed by a period and a brief informative title. Use the same type as used for the text. Supply a brief heading for each column. Indicate footnotes to tables with lower case, superscript, italic letters. Do not use vertical lines in the table. Use horizontal lines above and below the column headings and at the bottom of the table only. Use extra space to delineate sections within the table. Create tables using the table creating and editing feature of your word processing software (e.g., Word, WordPerfect). Excel or comparable spreadsheet programs can be used for large tables that will be published online-only. Please note that tables cannot be sub-divided into Table 1A, 1B etc. Each table will be counted as one display item.

Excel Spreadsheet Conversion Errors

Excel spreadsheets, when used with default settings, can convert gene names to dates and floating point numbers (see references: "Mistaken Identifiers: Gene name errors can be introduced inadvertently when using Excel in bioinformatics" Zeeberg BR *et al.* BMC Bioinformatics 2004;5:80 DOI: 10.1186/1471-2105-5-80 and "Gene name errors are widespread in the scientific literature" by Ziemann M *et al.* Genome Biology 2016;17:177 DOI: 10.1186/s13059-016-1044-7). Zeeberg *et al.* details several options on how to avoid these errors.

We therefore ask the authors to check carefully any Excel Spreadsheets being submitted as files, both at review and at publication (especially for supplementary files), for these type of errors.

English language support

For editors and reviewers to accurately assess the work presented in your manuscript you need to ensure the English language is of sufficient quality to be understood. If you need help with writing in English, you should consider:

- Asking a colleague who is a native English speaker to review your manuscript for clarity.
- Visiting the [English language tutorial](#) which covers the common mistakes when writing in English.
- Using a professional language editing service where editors will improve the English to ensure that your meaning is clear and identify problems that require your review. Two such services are provided by our affiliates [Nature Research Editing Service](#) and [American Journal Experts](#).

English language editing services

- [Nature Research Editing Service](#)
- [American Journal Experts](#)
- [Inter-Biotech](#)
- [SPI Professional Editing Services](#)
- [Write Science Right](#)

Please note that the use of a language editing service is not a requirement for publication in this journal and does not imply or guarantee that the article will be selected for peer review or accepted.

If your manuscript is accepted, it will be checked by our copyeditors for spelling and formal style before publication.

SUPPLEMENTARY INFORMATION

Supplementary information is material directly relevant to the conclusion of an article that cannot be included in the printed version owing to space or format constraints. It is posted on the journal's web site exactly as submitted by the author(s) and linked to

the article when the article is published. It may consist of data files, graphics, movies, or extensive tables.

The printed article must be complete and self-explanatory without the supplementary information. Supplementary information enhances a reader's understanding of the paper but should not be essential to that understanding.

Supplementary information must be supplied to the editorial office in its final form for peer review.

Supplementary information should be cited in the text as "Figure S1, S2, etc." "Table S1, S2, etc." or "Supplementary Materials and Methods."

Please supply the supplementary information via Editorial Manager, the electronic manuscript submission and tracking system, in an acceptable file format (see below).

Accepted file formats

- Quick Time files (.mov)
- Graphical image files (.gif)
- HTML files (.html)
- MPEG movie files (.mpg)
- JPEG image files (.jpg)
- Sound files (.wav)
- Plain ASCII text (.txt)
- Acrobat files (.pdf)
- MS Word documents (.doc)
- Postscript files (.ps)
- PowerPoint files (.ppt)
- MS Excel spreadsheet documents (.xls) Please read below for formatting of Excel files and Gene Names

We cannot accept TeX or LaTeX.

File sizes must be as small as possible, so that they can be downloaded quickly. Images should not exceed 640 x 480 pixels (9 x 6.8 inches at 72 pixels per inch) but we would recommend 480 x 360 pixels as the maximum frame size for movies. We would also recommend a frame rate of 15 frames per second. If applicable to the presentation of the supplementary information, use a 256 color palette. Please consider the use of the lower specification for all of these points if the supplementary information can still be represented clearly. Our recommended maximum data rate is 150 KB/s.

Please note: We do not allow the resupplying of Supplementary Information files for style reasons after a paper has been exported in production, unless there is a serious error that affects the science and, if by not replacing, it would lead to a formal correction once the paper has been published. In these cases, we would make an exception and replace the file; however, there are very few instances where a Supplementary Information file would be corrected post publication.

SUBMISSION OF MANUSCRIPTS

The first thing you need to do, if you have not already done so, is register for an account here <http://www.editorialmanager.com/gim/>. Then follow the step-by-step submission process. For a full author tutorial on using Editorial Manager click here: https://www.editorialmanager.com/homepage/DOCS/Author_Tutorial.doc.

Specific questions can be addressed to the Editorial Assistant, Katie Murphy, at geneticsinmedicine@acmg.net.

Peer review process and reviewer recommendations

All manuscripts will be assessed by the Editor-in-Chief, and, if appropriate will be sent to a section editor and two or more external reviewers. Manuscripts that are not reviewed will be returned to the authors. Authors MUST provide a list of potential reviewers who are especially qualified to referee the work and would not have a conflict of interest. Please include the reviewers' names, e-mail addresses, institutional affiliations, and academic degrees on the online submission form. The editors are, however, not obligated to use those reviewers. Manuscripts will be reviewed for content, originality, importance to the field, appropriateness of statistical analysis, and derivation of conclusions. Authors should note that manuscripts may be returned after initial review by the editorial office if the paper is deemed unlikely to be reviewed favorably by virtue of insufficient or general interest for *GIM*'s readership. This rapid rejection process enables the author to promptly submit for publication elsewhere. If sent for review, the outcome may be acceptance with or without revisions, non-acceptance with an opportunity to make revisions for resubmission, or rejection. Manuscripts being revised by the authors will be held by the editorial office for a maximum of 180 days, after which a new submission is required. Please contact the editorial office if you are not able to make this deadline. If a revision is requested, please include a tracked Word file of your edits.

License to Publish

The corresponding author must complete and sign a License to Publish form upon submission. A copy of the standard License to Publish form can be found at <https://www.nature.com/documents/aj-ltp.pdf>. We also offer Open Access. For

helping on deciding whether open access is right for you, and which open access license you prefer please go here: <https://creativecommons.org/choose/>. If authors choose to publish their article as Open Access content, an Open Access License to Publish form can be found at <https://www.nature.com/documents/aj-ltpoa-cbysand.pdf>. The Open Access License to Publish form is only needed if you choose to publish an Open Access paper. If you are a US, UK, or Canadian government employee, please find the forms [here](#).

ACMG does not require authors of original research papers to assign copyright of their published contributions. Authors grant ACMG an exclusive license to publish, in return for which they can re-use their papers in their future printed work. Springer Nature's [author license page](#) provides details of the policy and a sample form.

Authors are encouraged to submit their version of the accepted, peer-reviewed manuscript to their funding body's archive, for public release six months after publication. In addition, authors are encouraged to archive their version of the manuscript in their institution's repositories (as well as on their personal web sites), also six months after the original publication. Authors should cite the publication reference and doi number on any deposited version, and provide a link from it to the published article on the Springer Nature website. This policy complements the policies of the US National Institutes of Health, the Wellcome Trust and other research funding bodies around the world. Springer Nature recognizes the efforts of funding bodies to increase access of the research they fund, and strongly encourages authors to participate in such efforts.

Manuscript deposition service

A number of research funding agencies now require or request authors to submit accepted articles (after peer review and acceptance, but before the final published article) to a repository that is accessible online by all without charge. As a service, Springer Nature and *Genetics in Medicine* can help authors fulfill these mandates by archiving your manuscript in PubMed Central and/or UK PubMed Central on your behalf. Authors can select this option and provide details on their funding agencies during submission. A list of funders that Springer Nature deposits on behalf of is available here: http://www.nature.com/authors/author_resources/deposition.html

Advance Online Publication

Genetics in Medicine publishes most content online ahead of print as Advance Online Publication. This will be the final version of the manuscript and will subsequently appear, unchanged, in print.

Proofs

Corresponding authors will receive electronic page proofs to check the copy-edited and typeset article before publication. An e-mail will be sent to the corresponding author with a URL link where proofs and support documents (e.g., an offprint order form) will be available.

It is the author's responsibility to ensure that there are no errors in the proofs. Changes that have been made to conform to journal style will stand if they do not alter the authors' meaning. Only the most critical changes to the accuracy of the content will be made. Changes that are stylistic or are a reworking of previously accepted material will be disallowed. The publisher reserves the right to deny any changes that do not affect the accuracy of the content.

Proofs must be returned within 48 hours of receipt. Failure to do so may result in a delay to publication. Extensive corrections cannot be made at this stage.

Offprints

Offprints may be ordered using the order form accompanying the proofs.

Open Access and self-archiving

Self-archiving and manuscript deposition (green open access)

Authors of original research articles are encouraged to submit the author's version of the accepted paper (the unedited manuscript) to a repository for public release six months after publication. Springer Nature also offers a free, opt-in Manuscript Deposition Service for original research articles in order to help authors fulfill funder and institutional mandates.

[Learn more about self-archiving and manuscript deposition.](#)

Open access publication (gold open access)

Upon submission, authors of original research articles can opt to pay an article processing charge (APC) to allow their article to be published open access immediately upon publication, if it is accepted. Open access articles are published under Creative Commons licenses, which allow authors to retain copyright to their work while making it open to readers.

To facilitate self-archiving Springer Nature deposits open access articles in PubMed Central, Europe PubMed Central, and PubMed Central Canada on publication. Authors are also permitted to post the final, published PDF of their article on a website, institutional repository or other free public server immediately on publication.

Visit our [open research site](#) for further information about:

- [Creative Commons licensing](#)
- [Creative Commons license options and APCs for Genetics in Medicine](#)

- [APC payment FAQs](#)
- [Help in identifying funding for APCs](#)
- [Site license price adjustments for hybrid journals](#)
- [Editorial process for OA publication in hybrid journals](#)
- [Self-archiving and deposition](#)

Find out more about open access publishing at Springer Nature:

- [What is open access?](#)
- [Open access at Springer Nature](#)
- [Springer Nature journals offering an open access publishing option](#)

Compliance with open access mandates

Springer Nature's open access journals allow authors to comply with all funders' open access policies worldwide. Authors may need to take specific actions to achieve compliance with funder and institutional open access mandates.

[Learn more about open access compliance.](#)

EDITORIAL POLICIES

Genetics in Medicine is committed to maintaining high standards for the integrity of the published scientific record. Authors should take note and adhere to the journal editorial policies noted below. The journal will investigate any instances of suspected scientific fraud, image manipulation, plagiarism, duplicate publication, undocumented conflicts of interest, and other cases that compromise research ethics or the journal's scientific integrity. Depending on the investigation, the journal may opt to publish corrections, or in serious cases of scientific misconduct, request that the authors retract their paper or impose a retraction on the paper. In such matters, the journal is likely to consult the guidelines at COPE, CSE, and ICMJE.

Conflict of interest

In the interests of transparency and to help readers form their own judgments of potential bias, authors must declare whether or not there are any competing financial interests in relation to the work described. This information must be included in their cover letter and in the conflict of interest section of their manuscript. In cases where the authors declare a competing financial interest, a statement to that effect is published as part of the article. If no such conflict exists, the statement will simply read that the authors have nothing to disclose.

For the purposes of this statement, competing interests are defined as those of a financial nature that, through their potential influence on behavior or content, or from perception of such potential influences, could undermine the objectivity, integrity or perceived value of a publication. They can include any of the following:

Funding: Research support (including salaries, equipment, supplies, reimbursement for attending symposia, and other expenses) by organizations that may gain or lose financially through this publication. The role of the funding body in the design of the study, collection and analysis of data and decision to publish should be stated.

Employment: Recent (while engaged in the research project), present or anticipated employment by any organization that may gain or lose financially through this publication.

Personal financial interests: Stocks or shares in companies that may gain or lose financially through publication; consultation fees or other forms of remuneration from organizations that may gain or lose financially; patents or patent applications whose value may be affected by publication.

The statement must contain an explicit and unambiguous statement describing any potential conflict of interest, or lack thereof, for any of the authors as it relates to the subject of the report.

Examples include "Dr. Smith receives compensation as a consultant for XYZ Company," "Dr. Jones and Dr. Smith have financial holdings in ABC Company," or "Dr. Jones owns a patent on the diagnostic device described in this report." These statements acknowledging or denying conflicts of interest must be included in the manuscript under the heading 'Conflict of Interest'. The conflict of interest disclosure appears in the cover letter, in the manuscript submission process and before the References section in the manuscript.

Following the conflict of interest declaration in 'Disclosure/Conflict of Interest,' there must be a listing for each author, detailing the names of organizations, institutions, companies, and individuals, including intermediaries such as sub-contractors or conference organizers, from whom they have received compensation for professional services in any of the previous three years, or from whom they anticipate receiving such compensation in the near future, whether or not these affiliations appear to have any relevance to the topic covered in the submission. Neither the precise amount received from each entity nor the aggregate income from these sources needs to be provided. Professional services include any activities for which the individual is, has been, or will be compensated with cash, royalties, fees, stock or stock options in exchange for work performed, advice or counsel provided, or for other services related to the author's professional knowledge and skills. This would include, but not necessarily be limited to, the identification of organizations from

which the author received contracts or in which he or she holds an equity stake if professional services were provided in conjunction with the transaction.

The authors are expected to disclose any other financial holdings or considerations, such as stocks, bonds or donations of supplies or equipment that a reasonable person could construe as possibly influencing the objectivity of the report. When there is uncertainty about what should be listed, it is best to disclose all holdings or affiliations to ensure that there is no question about intent to withhold information. Authors should communicate with the editorial office (geneticsinmedicine@acmg.net) if they have questions about this policy.

Examples of declarations are:

Conflict of interest: The authors declare no conflict of interest.

Conflict of interest: Dr. Caron's work has been funded by the NIH. He has received compensation as a member of the scientific advisory board of Acadia Pharmaceutical and owns stock in the company. He also has consulted for Lundbeck and received compensation. Dr. Rothman and Dr. Jensen declare no potential conflict of interest.

If subsequent to publication it is learned that relevant information was not disclosed, a corrigendum describing the infraction will be published in the journal and linked to the article in PubMed. In addition, the published article may be retracted with a statement describing the nondisclosure and restrictions may be placed on future publications in the journal, solely at the discretion of the editor and College.

Clinical trials

As defined by the International Committee of Medical Journal Editors (ICMJE), a clinical trial is any research project that prospectively assigns human subjects to intervention and comparison groups to study the cause-and-effect relationship between a medical intervention and a health outcome. A medical intervention is any intervention used to modify a health outcome and includes but is not limited to drugs, surgical procedures, devices, behavioral treatments, and process-of-care changes. A trial must have at least one prospectively assigned concurrent control or comparison group in order to trigger the requirement for registration. Nonrandomized trials are not exempt from the registration requirement if they meet the above criteria.

When reporting experiments on human subjects, indicate whether the procedures were in accordance with the ethical standards of the responsible committee on human experimentation (institutional or regional) or with the Helsinki Declaration of 1975 (as revised in 1983). Include Institutional Review Board or Animal Care and Use Committee approvals. Please see the section of *Genetics in Medicine's* Guide to Authors on bioethics for more details on patient anonymity and informed consent in experiments on human subjects.

All clinical trials must be registered in a public registry prior to submission. *Genetics in Medicine* subscribes to the trials registration policy of the ICMJE (www.ICMJE.org) and considers only trials that have been appropriately registered before submission, regardless of when the trial closed to enrollment. Acceptable registries must meet the following ICMJE requirements: the registry must be accessible to the public at no charge. It must be open to all prospective registrants and managed by a not-for-profit organization. There must be a mechanism to ensure the validity of the registration data, and the registry should be electronically searchable. Trial registration with missing fields or fields that contain uninformative terminology is inadequate.

Examples of registries that meet these criteria include (1) the registry sponsored by the United States National Library of Medicine (<http://www.clinicaltrials.gov>); (2) the International Standard Randomized Controlled Trial Number Registry (<http://www.controlled-trials.com>); and (3) the European Clinical Trials Database (<https://eudract.ema.europa.eu/index.html>).

Springer Nature endorses the toolkits and guidelines produced by the following bodies:

Committee on Publication Ethics: <http://publicationethics.org/>

Good Publication Practice: <http://www.gpp-guidelines.org/>

Medical Publishing Insights and Practices Initiative: <http://www.mpip-initiative.org/>

Peer review policy

All contributions that are selected for peer review are sent to independent reviewers. The identity of reviewers is confidential and manuscripts are considered private information. Papers may be rejected without external review at the Editor's discretion. Authors must suggest or recommend for exclusion reviewers at the time of submissions.

Anonymity

Reviewers' identities are not released to authors, except when reviewers specifically ask to be identified. If reviewers wish to reveal their identities while the manuscript is under consideration, this should be done through the Editor. Should a reviewer contact an author directly, we ask authors to inform the Editor as soon as possible. We prohibit any attempt by authors to confront reviewers or determine their identities. Our own policy is to neither confirm nor deny any speculation about reviewers' identities, and we encourage reviewers to adopt a similar policy. In accordance with ICMJE guidelines, reviewer comments should not be published or otherwise publicized without permission of the reviewer, author and editor.

Selection of peer reviewers

Reviewer selection is critical to the publication process, and we base our choices on many factors, including expertise, reputation, and specific recommendations.

Authorship

Requirements for all categories of articles largely conform to the "Uniform Requirements for Manuscripts Submitted to Biomedical Journals," developed by the ICMJE. A manuscript will be considered for publication with the understanding that:

all named authors have agreed to its submission

it is not currently being considered for publication by another journal

if the paper is accepted, it will not subsequently be published in the same or similar form in any language without the consent of publisher

Each author must have contributed sufficiently to the intellectual content of the submission. The corresponding author should list all authors and their contributions to the work. Any changes to the author list after submission, such as a change in the order of the authors, or the deletion or addition of authors, will follow the guidelines as set out by COPE (<http://publicationethics.org/>). The corresponding author must confirm that he or she has had full access to the data in the study and final responsibility for the decision to submit for publication. The ICMJE recommends that authorship be based on the following 4 criteria:

1. Substantial contributions to the conception or design or the work; or the acquisition, analysis, or interpretation of data for the work; AND
2. Drafting the work or revising it critically for important intellectual content; AND
3. Final approval of the version to be published; AND
4. Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

In addition to being accountable for the parts of the work he or she has done, an author should be able to identify which co-authors are responsible for specific other parts of the work. In addition, authors should have confidence in the integrity of the contributions of their co-authors.

When a large multi-author group has conducted the work, the group ideally should decide who will be an author before the work is started and confirm who is an author before submitting the manuscript for publication. All members of the group named as authors should meet all four criteria for authorship, including approval of the final manuscript, and they should be able to take public responsibility for the work and should have full confidence in the accuracy and integrity of the work of other group authors. They will also be expected as individuals to complete conflict-of-interest statements.

Some large multi-author groups designate authorship by a group name, with or without the names of individuals. When submitting a manuscript authored by a group, the corresponding author should specify the group name if one exists, and clearly identify the group members who can take credit and responsibility for the work as authors. The byline of the article identifies who is directly responsible for the manuscript, and MEDLINE lists as authors whichever names appear on the byline. If the byline includes a group name, MEDLINE will list the names of individual group members who are authors or who are collaborators, sometimes called non-author contributors, if there is a note associated with the byline clearly stating that the individual names are elsewhere in the paper and whether those names are authors or collaborators.

Acquisition of funding, collection of data, or general supervision of the research group alone does not constitute authorship.

All persons designated as authors should qualify for authorship, and all those who qualify should be listed.

Each author should have participated sufficiently in the work to take public responsibility for appropriate portions of the content.

Contributions by individuals who made direct contributions to the work but do not meet all of the above criteria should be noted in the Acknowledgments section of the manuscript.

Medical writers and industry employees can be contributors. Their roles, affiliations, and potential conflicts of interest should be included in the author list or noted in the Acknowledgments and/or Contributors section concurrent with their contribution to the work submitted. Signed statements from any medical writers or editors declaring that they have given permission to be named as an author, as a contributor, or in the Acknowledgments section is also required. Failure to acknowledge these contributors can be considered inappropriate, which conflicts with *Genetics in Medicine's* editorial policy.

Copyright

Authors are not required to grant the copyright of their articles to the American College of Medical Genetics and Genomics, Springer Nature, or *Genetics in*

GENETICS IN MEDICINE

Medicine. Instead, authors provide the American College of Medical Genetics and Genomics and *Genetics in Medicine* with an exclusive license. Authors are free to reuse their own papers in any future published work and on their own website or institutional repository

Copyright permissions

If a table or figure has been published previously, the authors must obtain written permission from the copyright owner to reproduce the material in both print and electronic formats and submit such permission with the manuscript. This is applicable to quotes, illustrations and other materials taken from previously published works not in the public domain. The original source should be cited in the figure caption or table footnote. Color figures can be reproduced if necessary, but the authors may be expected to contribute toward the cost of publication. A quote will be supplied upon acceptance of the paper.

Plagiarism and fabrication

Plagiarism is when an author attempts to pass off someone else's work as his or her own. Duplicate publication, sometimes called self-plagiarism, occurs when an author reuses substantial parts of his or her own published work without providing the appropriate references. Plagiarism without dishonest intent is relatively frequent, for example, when an author reuses parts of an introduction from an earlier paper.

CrossCheck is a multi-publisher initiative to screen published and submitted content for originality. *Genetics in Medicine* uses CrossCheck to detect instances of overlapping and similar text in accepted submitted manuscripts. To find out more about CrossCheck visit www.crossref.org/crosscheck.html.

If plagiarism is found, the journal will contact the author and, in some cases, the author's institute and funding agencies to resolve the issue.

Duplicate publication

Papers must be original and not previously published or submitted for publication elsewhere. This rule also applies to non-English language publications. The corresponding author is responsible for alerting the editorial office, in the cover letter, if the manuscript includes subjects about which a previous report has been published or about a manuscript that is submitted to, under review by, in press at, or to be submitted to or published in another journal in the future. All such work must be referred to and referenced in the new paper and a copy of the manuscript and/or cohort should be included with the submission as a supplemental file.

Image manipulation

Images submitted with a manuscript for review should be minimally processed. Authors should retain their unprocessed data and metafiles, as Editors may require them to aid in manuscript evaluation. If unprocessed data is available, manuscript evaluation may be stalled until the issue is resolved.

A certain degree of image processing is acceptable for publication (and for some experiments, fields and techniques is unavoidable), but the final image must correctly represent the original data and conform to community standards. The guidelines below will aid in accurate data presentation at the image processing level.

Authors should list all image acquisition tools and image processing software packages used. Authors should document key image-gathering settings and processing manipulations in the Materials and Methods section.

Images gathered at different times or from different locations should not be combined into a single image, unless it is stated that the resultant image is a product of time-averaged data or a time-lapse sequence. If juxtaposing images is essential, the borders should be clearly demarcated in the figure and described in the legend.

Touch-up tools, such as cloning and healing tools in Photoshop, or any feature that deliberately obscures manipulations are to be avoided.

Processing (such as changing brightness and contrast) is appropriate only when it is applied equally across the entire image and is applied equally to controls. Contrast should not be adjusted so that data disappears.

Excessive manipulations, such as processing to emphasize one region in the image at the expense of others (for example, through the use of a biased choice of threshold settings), is inappropriate, as is emphasizing experimental data relative to the control.

Positive and negative controls, as well as molecular size markers, should be included on each gel and blot—either in the main figure or an expanded data supplementary figure. The display of cropped gels and blots in the main paper is encouraged if it improves the clarity and conciseness of the presentation. In such cases, the cropping must be mentioned in the figure legend.

Vertically sliced gels that juxtapose lanes that were not contiguous in the experiment must have a clear separation or a black line delineating the boundary between the gels.

Cropped gels in the paper must retain important bands.

Cropped blots in the body of the paper should retain at least six band widths above and below the band.

High-contrast gels and blots are discouraged, as overexposure may mask additional bands. Authors should strive for exposures with gray backgrounds. Immunoblots should be surrounded by a black line to indicate the borders of the blot, if the

Guide for Authors

background is faint. For quantitative comparisons, appropriate reagents, controls and imaging methods with linear signal ranges should be used.

Microscopy adjustments should be applied to the entire image. Threshold manipulation, expansion or contraction of signal ranges and the altering of high signals should be avoided. If pseudocoloring and nonlinear adjustment (for example, "gamma changes") are used, this must be disclosed. Adjustments of individual color channels are sometimes necessary on merged images, but this should be noted in the figure legend.

We encourage inclusion of the following with the final revised version of the manuscript for publication:

In the Materials and Methods section, specify the type of equipment (microscopes/objective lenses, cameras, detectors, filter model and batch number) and acquisition software used. Although we appreciate that there is some variation between instruments, equipment settings for critical measurements should also be listed.

The display lookup table (LUT) and the quantitative map between the LUT and the bitmap should be provided, especially when rainbow pseudocolor is used. It should be stated if the LUT is linear and covers the full range of the data.

Processing software should be named and manipulations indicated (such as type of deconvolution, three-dimensional reconstructions, surface and volume rendering, "gamma changes," filtering, thresholding and projection).

Authors should state the measured resolution at which an image was acquired and any downstream processing or averaging that enhances the resolution of the image.

Correction and retraction policy

We recognize our responsibility to correct errors. Content published online (as Advance Online Publication - AOP) or in a print issue is final and cannot be amended. The online and print versions are both part of the published record; therefore, the original version must be preserved and changes to the paper should be made as a formal correction. If an error is noticed in an AOP article, a correction should accompany the article when it publishes in print. An HTML (or full-text) version of the correction will also be created and linked to the original article. If the error is found in an article after print publication, the correction will be published online and in the next available print issue. We communicate only with the Corresponding Author regarding all issues involving corrections and retractions.

Retraction. Notification of invalid results. All coauthors must sign a retraction specifying the error and stating briefly how the conclusions are affected.

Decisions about corrections are made by the Editor (sometimes with advice of peer reviewers) and this sometimes involves author consultation. Requests to make corrections that do not affect the paper in a significant way or impair the reader's understanding of the contribution (a spelling mistake or grammatical error, for example) are not considered.

In cases where coauthors disagree about a correction, the Editor will take advice from independent peer reviewers and impose the appropriate correction, noting the dissenting author(s) in the text of the published version.

Research Data Policy

We strongly encourage that all datasets on which the conclusions of the paper rely should be available to readers. We encourage authors to ensure that their datasets are either deposited in publicly available repositories (where available and appropriate) or presented in the main manuscript or additional supporting files whenever possible. Variants with clinical assertions regarding pathogenicity should be deposited in ClinVar. Where an appropriate repository does not exist, the information must be made available to referees at submission and to readers promptly upon request. Any restrictions on material availability or other relevant information must be disclosed in the manuscript's Methods section and should include details of how materials and information may be obtained.

Please see the journal's guidelines on Research Data policy [here](#).

Reporting Guidelines

NLM Research Reporting Guidelines and Initiatives and EQUATOR list many guidelines to promote high quality of published research. The editors at GIM are encouraging authors to seek out appropriate guidelines for their studies, to fill in checklists if available, upload those checklists with your manuscript at submission, and to detail in the cover letter what guidelines were followed in producing your manuscript. We will be asking our editors and reviewers to look over these checklists, when included, as part of the review process. Adhering to published guidelines will be seen as a positive factor when your manuscript is reviewed by both the editorial staff and external reviewers.

Based on the type of studies that GIM publishes, here is a partial list of guidelines that may apply. However, this is not an exhaustive list. We encourage potential authors to seek established guidelines and checklists that pertain to their research and use those that they feel may enhance the validity of their work:

- Consolidated Health Economic Evaluation Reporting Standards (CHEERS)

Statement

- Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology
- ACMG policy statement: updated recommendations regarding analysis and reporting of secondary findings in clinical genome-scale sequencing
- American College of Medical Genetics standards and guidelines for interpretation and reporting of postnatal constitutional copy number variants
- Strengthening the Reporting of Genetic Association Studies (STREGA): An Extension of the STROBE Statement.
- Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors
- Nomenclature of gene variations and guidelines on variation databases
- An introduction to standardized clinical nomenclature for dysmorphic features: the Elements of Morphology project
- Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA).
- The HuGENet™ HuGE Review Handbook, version 1.0. Guidelines for systematic review and meta-analysis of gene disease association studies
- Publication of population data for forensic purposes
- Reporting genetic results in research studies: summary and recommendations of an NHLBI working group
- Gene expression-based prognostic signatures in lung cancer: ready for clinical use?
- REporting recommendations for tumour MARKer prognostic studies (REMARK)
- Strengthening the reporting of Genetic Risk Prediction Studies: the GRIPS Statement
- Standards for Reporting Implementation Studies (StaRI) Statement

The only mandatory checklist that GIM imposes on all original research articles was instigated at the request of the NIH: Transparency of reporting and the reproducibility of published results, focusing on elements of methodological information that are usually poorly reported. Authors MUST supply that checklist at the revision stage. The Reporting Checklist will be provided in the initial decision letter, but may also be found [here](#).

Gene Nomenclature

The genetic notation and symbols approved by the Human Gene Mapping Workshop (HGMW) should be used. Approved gene symbols and nomenclature can be obtained from the HUGO Gene Nomenclature Committee. Mendelian Inheritance in Man (MIM) nomenclature and instructions for a specific entry should be used when appropriate. Use ISCN nomenclature for cytogenetic notation (Mitelman F[ed]. ISCN 1995. An international system for human cytogenetic nomenclature. S. Karger, Basel, 1995). Human gene names and loci are given in italicized capital letters and Arabic numbers. Protein products are not italicized. Mouse nomenclature should use the International Committee on Standardized Genetic Nomenclature for Mice. Authors should express all measurements in International System of Units (SI) with conventional units in parenthesis. Conversion tables are available in the "American Medical Association Manual of Style: A Guide for Authors and Editors," 10th edition, pages 795.

Standard Human Pedigree Nomenclature: When representing a human pedigree, please refer to Bennet *et al.* "Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors" *J Genet Counsel* (2008) 17:424–433

BIOETHICS

Human and other animal experiments

For original research manuscripts reporting experiments on live vertebrates and/or higher invertebrates, the corresponding author must confirm that all experiments were performed in accordance with relevant guidelines and regulations. The manuscript must include a statement identifying the institutional and/or licensing committee approving the experiments, including any relevant details regarding animal welfare, patient anonymity, drug side effects and informed consent.

For experiments involving human subjects, authors must identify the committee (e.g. Institutional Review Board [IRB]) approving the experiments and include with their submission a statement confirming that informed consent was obtained from all subjects. All human studies must adhere to the principles set out in the Declaration of Helsinki (World Medical Association Declaration of Helsinki, Published online October 19, 2013. doi:10.1001/jama.2013.281053)

It is the author's responsibility to ensure that a patient's anonymity be carefully protected and to verify that any experimental investigation with human subjects reported in the manuscript was performed with informed consent and following all the guidelines for experimental investigation with human subjects required by the institution(s) with which all the authors are affiliated. All identifying details (patient names and/or initials and name of specific hospital unit) should be removed from the text, tables, and/or radiographs or other figures. Patients should be assigned numbers instead. Identifying data should be removed from a manuscript unless important clinically or

epidemiologically. Clinically and epidemiologically significant details include: race, sex, age, occupation, country or region of origin, and/or sexual orientation. Note that the same information must be given for each patient or group.

If any individual data is included, written consent for participation/publication must be obtained from every individual whose data is included. A written statement attesting that the author has received and archived such written patient consent must accompany the manuscript. For research articles, if authors cannot obtain consent to publish individual data they may only provide summary results. Exclusions to this include:

- The patients are de-identified
- The patients are dead
- There has been a waiver granted by the Institutional Review Board (IRB)

If in doubt about whether permission is required to publish, please contact the editorial office at geneticsinmedicine@acmg.org.

If the manuscript contains protected health information (PHI), as defined in the federal Health Insurance Portability and Accountability Act of 1996 and its implementing regulations (HIPAA), a valid HIPAA authorization for participation/publication must have been obtained from every individual whose PHI is included. A written statement attesting that the author has received and archived such HIPAA authorization must accompany the manuscript.

It is preferred that photographs with bars placed over eyes of patients NOT be used in publication, however if they are used, permission from the patient(s) is required. Authors should NOT alter or falsify details in case descriptions to secure anonymity because doing so may introduce false or inaccurate data into the medical literature.

Permissions Form: If identifying details must be retained, the author must attest they have informed consent from the patients (a signed permissions form). This consent should include an opportunity for the patient to read the manuscript to be submitted for publication or waive the right to do so. Permissions must appear in English, in the case of a non-English speaking patient/author the permission should appear first in the native language with the translation to English below and the signature following both. If the patient cannot be located or refuses to consent to publication, the manuscript will NOT be published. In the event that the patient cannot provide consent due to death or legal incompetency (this includes photos of corpses) permission from the power of attorney is needed as well as proof of power of attorney. If the patient is a minor, a legal guardian must provide permission. Previous publication of news coverage does NOT eliminate a patient's right to privacy and does NOT negate the need for patient permission. This informed consent should be indicated in the text of the article (in the Methods section, if appropriate) or in the Acknowledgments at the end of the article. Do not send permissions forms to the journal.

Genetics disorders

Genetic disorders should be accompanied by their corresponding OMIM numbers. For details on OMIM numbers, please refer here <http://www.ncbi.nlm.nih.gov/omim>.

Biosecurity policy

The Editor may also seek advice about submitted papers that raise concerns. These may include, for example, ethical issues or issues of data or materials access. Very occasionally, concerns may also relate to the implications of publishing a paper to the society, including threats to security. In such circumstances, advice will usually be sought simultaneously with the technical peer-review process. As in all publishing decisions, the decision whether to publish is at the discretion of the Editor.

COMMUNICATION

Correspondence with the journal

One author is designated the contact author for matters arising from the published paper (materials requests, technical comments and so on). It is this author's responsibility to inform all coauthors of matters arising during the course of the process and to ensure such matters are dealt with promptly. After acceptance for publication, proofs are e-mailed to this corresponding author, who should circulate the proofs to all coauthors and coordinate corrections among them.

Communication with the media

Material submitted must not be discussed with the media. We reserve the right to halt the consideration or publication of a paper if this condition is broken. If a paper is particularly newsworthy, Springer Nature may send a press release to our list of journalists in advance of publication with an embargo that forbids any coverage of the manuscript, or the findings of the manuscript, until the time and date clearly stated, which will coincide with when the paper is publishing online. Authors whose papers are scheduled for publication may also arrange their own publicity (for instance, through their institution's press offices), but they must strictly adhere to the online publication press embargo and are advised to coordinate their own publicity with Springer Nature's press office.

Communication between scientists

Genetics in Medicine does not wish to hinder communication between scientists. You are free to communicate with other researchers as much as you wish, whether

GENETICS IN MEDICINE

by discussion at scientific meetings or by online collaborative sites such as wikis, but we do not encourage premature publication by discussion with the press (beyond a formal presentation, if at a conference).

Contributions being prepared for or submitted to Genetics in Medicine can be posted on recognized preprint servers (such as ArXiv), and on collaborative web-sites such as wikis or the author's blog. The website and URL must be identified to the editor in the cover letter accompanying submission of the paper, and the content of the paper must not be advertised to the media by virtue of being on the website or preprint server. Material in a contribution submitted to a Nature journal may also have been published as part of a PhD or other academic thesis.

<http://www.nature.com/authors/policies/confidentiality.html>

Guide for Authors

BUSINESS CONTACT

For all business regarding manuscripts and peer review, please contact:
Editorial Office, geneticsinmedicine@acmq.net

ACMG's Privacy Statement: <http://www.acmq.net/PDFLibrary/GIM-Author-Reviewer-Privacy-Information.pdf>

All business regarding journal publication or Springer Nature should be addressed to:

Senior Editor: Elizabeth Yopez, e.yopez@us.nature.com

Production Editor: Steve Coleman, steve.coleman@springernature.com

Alternatively, you can write to: *Genetics in Medicine*, Academic Journals Division, Springer Nature, 233 Spring Street, 3rd Floor, New York, NY 10013, USA