

Information for Authors

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Aims and Scope

AJHG provides a record of research and review relating to heredity in humans and to the application of genetic principles in medicine and public policy, as well as in related areas of molecular and cell biology. Topics explored by *AJHG* include behavioral genetics, biochemical genetics, clinical genetics, cytogenetics, dysmorphology, gene therapy, genetic counseling, genetic epidemiology, genomics, immunogenetics, molecular genetics, neurogenetics, and population genetics.

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All submissions are initially evaluated in depth by the scientific editors. Papers that do not conform to the general criteria for publication will be returned to the authors without detailed review, typically within three to five days. Otherwise, manuscripts will be sent to at least two reviewers who have agreed in advance to assess the paper rapidly. The editors will make every effort to reach decisions on these papers within four weeks of the submission date. If revisions are a condition of publication, generally four weeks are allowed for revisions and only one revised version of the paper is considered. Evaluations of conceptual advance and significance are made based on literature available on the day of the final decision, not the day of submission. Accepted papers will be published within two months of acceptance. Any major changes after acceptance are subject to review and may delay publication.

Fast-Track Review

If circumstances so warrant, a fast-track review of a paper may be requested in the cover letter. At the editor's discretion, a 48-hour fast-track review will be undertaken to expedite manuscripts that deserve rapid review and publication.

Editorial Policies

Manuscripts are considered with the understanding that no part of the work has been published previously in print or electronic format and that the paper is not under consideration by another publication or electronic medium. All in-press or submitted works pertinent to the manuscript under consideration by *AJHG* (including those cited in the manuscript under consideration) must accompany the submission. Related manuscripts that have been submitted elsewhere during the period of revision must accompany revised manuscripts. Failure to provide copies of related manuscripts under consideration elsewhere may delay the review process and may be grounds for rejection.

Authorship

The *AJHG* ascribes to the authorship and contributorship guidelines defined by the International Committee of Medical Journal Editors (www.ICMJE.org).

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The corresponding author is responsible for ensuring that all appropriate contributors are listed as authors and that all authors have agreed to the manuscript's content and its submission to *AJHG*.

Conflict of Interest

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If the manuscript reports results of studies of either human subjects or materials obtained from human subjects, state in the Material and Methods section that the study was approved by the appropriate institutional review board (IRB) and/or that appropriate informed consent was obtained from human subjects. Copies of signed consent forms from identifiable persons appearing in any photographs submitted with the manuscript must be faxed to the *AJHG* office (fax: 617-525-4569) or included in the submission file at the time of submission. To avoid violation of HIPAA regulations, the consent form should state that subjects understand that their identity (but not their medical records) will be divulged to the *AJHG* office by the transmission of the signed consent form. Alternatively, a letter from a member of the institution's IRB can be sent confirming that signed consent forms have been obtained from the

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An implicit term and condition of publishing in *AJHG* is that authors be willing to distribute any materials and protocols used in the published experiments to qualified researchers for their own use. Materials include but are not limited to cells, DNA, antibodies, reagents, organisms, and mouse strains, or if necessary the relevant ES cells. These materials must be made available with minimal restrictions and in a timely manner, but it is acceptable to request reasonable payment to cover the cost of provision and transport of materials. If there are restrictions to the availability of any materials, data, or information, these must be disclosed in the cover letter and the Material and Methods section of the manuscript at the time of submission.

Nucleic acid and protein sequences, single-nucleotide polymorphisms (SNPs), copy number variants (CNVs), microarray data, and macromolecular structures determined by X-ray crystallography (along with structure factors) must be deposited in the appropriate public database and must be accessible without restriction from the date of publication. The URL of the databases used must be included in the Web Resources section of the manuscript. All entry names and/or accession numbers must be included in the Material and Methods section. Microarray data should be MIAME compliant (for guidelines see <http://www.mged.org/Workgroups/MIAME/miame.html>).

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Please provide a figure or table that summarizes the full results of your genome-wide scan.

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Please provide a URL and title for the website upon which the novel computer program described in the manuscript will be made publicly available, in a Web Resources section within the manuscript.

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Presubmission Inquiries

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EES identifies the status as “received by editor” until an initial editorial decision is made. If the manuscript is sent for review, EES will update the status to “under review.” When reviews are complete, the status will say, “reviews completed.” If further information is needed about the status of a submitted manuscript, please email ajhg@ajhg.net.

Manuscript Formats

A brief description of each article type is provided below. Please refer to "Preparation and Online Submission of Research Manuscripts" for detailed information about the organization of research articles and their submission.

Articles

Articles are full-length, original research articles and should include the following sections in the main text: Abstract, Introduction, Material and Methods (or Subjects and Methods), Results, and Discussion.

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Reports are short (approximately four printed pages, at about 900 words per page) scientific correspondence with an Abstract but without further subdivision of the manuscript.

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Reviews are to be between 7 and 12 printed pages (approximately 900 words per page) and should include the following sections: Introduction, body, Conclusions, Acknowledgements, Web Resources (as necessary), and References.

The body of the text may be divided with bold headings and further subdivided with italic headings. The Conclusions section should contain speculation regarding the future of the reviewed topic. Figures and tables may be included. Please note that proper copyright permissions are required for display items previously published in any form.

Once a topic is agreed upon by authors and editors, authors are encouraged to submit a short Abstract (around 250 words) outlining the Review in advance of the full submission.

Preparation and Online Submission of Research Manuscripts

Submission

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Text Specifications

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Nonstandard abbreviations should be defined when first used in the text. Use of abbreviations should be kept at a minimum.

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MIM numbers should be listed in the text, but not in the Abstract, after the first mention of all disorders and genes. Example: adrenal hypoplasia congenita (AHC [MIM 300200]). In the Web Resources section, only the URL for the OMIM database, and not any individual MIM numbers, should be noted. OMIM's preferred website listing is as follows: Online Mendelian Inheritance in Man (OMIM), <http://www.omim.org/>.

All mutations should be described at both the DNA and the protein level and as outlined in "Mutation Nomenclature Extensions and Suggestions to Describe Complex Mutations: A Discussion" (den Dunnen JT and Antonarakis SE [2000]. *Hum.Mutat.* 15:7–12). This can be found at <http://www.hgvs.org/mutnomen/>.

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Manuscript Organization

AJHG manuscripts generally contain the following sections, in this order: Title Page, including title, authors, affiliations, contact information, and additional footnotes (if any); Abstract; Main Text (which, in articles, should be separated into Introduction, Material and Methods [or Subjects and Methods], Results, and Discussion); Supplemental Data description, if any (note: this section should not include supplemental figures or tables; it should merely list how many and what types of files are included in the Supplemental Data); Acknowledgments; Appendices (if any); Web Resources (if any); References; Figure Titles and Legends; Tables (with a title and legend for each). These sections (Title through Tables) should be provided as one document.

Each figure should be uploaded individually as a separate file. (See "Figure Organization, Formats, and Style.")

All Supplemental Data (aside from very large tables of data or Excel files) should be compiled into a Word document and uploaded as a single file.

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Titles may occupy no more than three lines of type. Each line should contain no more than 54 characters, *including spaces*. The title should convey the conceptual significance of the paper to a broad readership.

Authors and Affiliations

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The Abstract consists of a single paragraph of fewer than 250 words. It should clearly convey the conceptual advance and significance of the work to a broad readership. In particular, the Abstract should contain a brief background of the question, a description of the results without extensive experimental detail, and a brief summarization of the significance of the findings. References should not be cited in the Abstract.

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The Introduction should be succinct, with no subheadings, and should present the background information necessary to provide a biological context for the results.

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This section should be titled “Supplemental Data” and should briefly list what types of data are included in the Supplemental Data. For example, “Supplemental Data include four figures and two tables.”

Acknowledgments

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Detailed results of statistical analyses may be presented as an Appendix. Appendices may contain subheadings.

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Article in a book

2. King, S.M. (2003). Dynein motors: Structure, mechanochemistry and regulation. In *Molecular Motors*, M. Schliwa, ed. (Weinheim, Germany: Wiley-VCH Verlag GmbH), pp. 45–78.

Entire book

3. Cowan, W.M., Jessell, T.M., and Zipursky, S.L. (1997). *Molecular and Cellular Approaches to Neural Development* (New York: Oxford University Press).

Online reference

4. Rothwarf, D.M., and Karin, M. (1999). The NF- κ B pathway: a paradigm in information transfer from membrane to nucleus. *Science's STKE*, http://www.stke.org/cgi/content/full/OC_sigtrans;1999/5/rel

Computer program

5. Hubbard, S.J. and Thornton, J.M. (1993). NACCESS (computer program). Department of Biochemistry and Molecular Biology, University College London.

- Software may also be cited in text; for an in-text citation, include the name and location of the manufacturer in parentheses.

Dissertation/Thesis

6. Smith, J.P. (1985). DNA sequences. PhD thesis, Massachusetts Institute of Technology, Cambridge, Massachusetts.

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(M. Adams et al., 1997, Soc. Neurosci., abstract)

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Figures may not exceed one page.

Figure titles may not contain parenthetical information, reference citations, or footnotes.

All reference citations within a figure must also be included in the figure legend.

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Tables

Include tables in the submitted manuscript as a separate section. Tables should not be saved as figures, i.e. as .jpg or .tif files. All tables intended for print should be incorporated into the end of the manuscript Word file. Tables should not be uploaded individually.

When creating a table, please use the Microsoft Word table function, and please do not place an Excel table into a Word document. Tables not created with the Microsoft Word table function will be sent back for revision. Do not submit a table in PDF format.

Word tables should not be tab or space delineated and should not include color.

Do not use paragraph returns to separate data within a cell.

Tables should include a title, and footnotes and/or legends should be concise.

Table titles may not contain parenthetical information, reference citations, or footnote citations.

Use superscripted lowercase letters (beginning with "a") for footnotes in tables. Do not use numbers or symbols.

Tables must be numbered as Table 1, Table 2, Table 3, etc., rather than as Table 1a, Table 1b, Table 1c, etc.

If bold or italic font is used within a table to indicate some feature of the data, an explanation of its meaning must be given in the table legend.

If a referenced paper or study (e.g., "Author et al.") is mentioned within a table, it must be included in the References list and must be followed by its appropriate citation number within the table.

All abbreviations within a table must be defined in the table legend or footnotes.

Permissions

Please provide proof of permission to include any work cited as "personal communication." This may be in the form of an email communication, letter, or other appropriate form of permission.

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Supplemental Data Organization

Supplemental Data files are restricted to (1) figures that cannot be rendered in print with enough detail to be informative, (2) tables that have too many columns and/or rows to fit across two printed pages, and (3) tables and figures that would substantially lengthen the print version of the manuscript. Additional text will not be allowed as Supplemental Data.

Supplemental Data should be provided with the original submission. Please follow the digital figure guidelines below for preparing figures. In general, supplemental files (movies, databases, tables, etc.) must each be less than 10 MB. All figures and tables should have titles and legends.

Every attempt should be made to submit the Supplemental Data in a composite Word file.

Supplemental Movies and Excel Spreadsheets

Supplemental movies may be submitted through EES as .mov, .avi, .mpeg, or .gif files. By choosing the submission item "Supplemental Movies and Spreadsheets," the PDF builder will embed links within the

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If you have supplemental movies or spreadsheets over 10 MB in size, we ask that you send the files on CD to our offices at *AJHG*, Baylor College of Medicine, One Baylor Plaza, Rm 904E, BCM0225, Houston, TX 77030, USA.

Figure Organization, Format, and Style

Figures must conform to the following guidelines or authors will be asked to revise them. For each figure in your article, please ensure that high resolution digital files are uploaded individually. Please be sure that you embed all fonts if the file type allows for it. Color figures should be created at a resolution of 300 dpi, while grayscale figures should be at 500 dpi. For figures that have multiple panels, the labels should be set in uppercase letters in Helvetica or Arial font.

Each figure must be assembled into one file that prints onto one 8 x 11 page. Please do not include separate panels on multiple pages. Micrographs should be provided with a scale bar, if appropriate, instead of magnification.

If references are cited within a figure, they must also be cited within the figure legend (because it is not possible to link within-figure citations to their corresponding references online).

File Naming

Please name files by first author and number of figure, e.g., Smith1.tif, Smith2.tif, etc.

Figure Formats Acceptable for Publication

- TIFF (.tif)
- Encapsulated Postscript (.eps)
- Postscript (.ps)
- Portable Document File (.pdf)
- JPEG (.jpg) – use maximum quality
- PhotoShop (all versions) (.psd)
- Adobe Illustrator (all versions) (.ai) (Note: These files must be sent to the *AJHG* editorial office on CD-ROM via mail rather than uploaded into EES.)

Please note that we cannot accept PowerPoint files. We also cannot accept image files in Canvas, Freehand, Microsoft Word, or Excel.

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Postscript Files (.ps)

There are many different drawing programs, not all of them supported by the software used by *AJHG*. From almost all of these programs, it should be possible to produce a postscript file. When printing, select to print to a (postscript) file, rather than printing by default to a printer. The image must be in portrait orientation. Please be sure to embed all fonts when saving as a postscript file.

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Authors are encouraged to use colors that can be distinguished by color-blind readers. Please submit your figures in RGB or grayscale -- do not convert files to CMYK. This will optimize their appearance online. However, please be aware that we will convert to CMYK for the print version. If possible, embed the ICC profile.

Line Weights

Please make sure that all graph lines, error bars, and outlines are at least ½ point in weight. Lines that are less than ½ point may disappear or be broken during printing.

If you have any questions about digital files, please contact *AJHG* senior managing editor Kerry Evans, at kevans@cell.com.

Submission of Revised Manuscripts

In addition to the sections described above, revised manuscripts must also contain a detailed point by point response to the comments of the reviewers and/or editors. These responses should be compiled into a document titled Response to Reviewers and uploaded individually. This response will be seen by reviewers if the manuscript is sent for re-review. The cover letter is confidential to the editors and should briefly summarize how the revised manuscript addresses these comments. All changes to the manuscript should also be highlighted using the Track Changes feature in Microsoft Word. In general, revised manuscripts will be reconsidered only if resubmitted within four weeks of the date of the original decision.

Checklist for Final Submission

In order for a paper to be accepted and scheduled for publication, the author will be asked to send the following items. Please refer to the letter from the editor for further details and to the additional guidelines below for final figures.

1. Please make sure that your final manuscript includes the following:

- Database accession numbers for new SNPs, gene sequences, protein sequences, copy number variants, microarray data, or structures
- All HUGO-approved gene names as well as all MIM numbers for all relevant disorders or genes
- Includes an explanation of statistical methods in the Material and Methods
- For any figures presenting pooled data, definitions of the measures within the figure legends
- Experimental procedures in sufficient detail
- Author and affiliations lists with superscripted numbers
- Concise titles for figures and tables
- Proper formatting and resolution of figures (see above)
- Proper formatting of tables (see above)

2. Please submit the final version of text and figures through our online editorial system (EES).

3. Please send to us:

On a single CD: any additional supplemental data larger than 10 MB.

Production and Proofs

After final acceptance, the manuscript will be passed to the production team to be copyedited and prepared for printing. Members of ASHG will not be charged page charges or color charges. Nonmember authors will be charged \$450 for the first color figure and \$200 for each additional color figure. In lieu of a per-page fee, non-members will be charged a single fee of \$825 for Articles and \$525 for Reports. Figures may be resized during the production process.

PDF proofs will arrive via email about one week prior to publication and need to be returned with corrections no more than 24 hours after receipt.

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