

Information for Authors

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The entire contents of the *AJHG* are published online, at <http://www.ajhg.org>. The full text is available to subscribers, as is online-only material, including video clips and archival databases. Six months after publication, the electronic edition of the *AJHG* is freely accessible to the general public.

Aims and Scope

The American Journal of Human Genetics 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 the *AJHG* include behavioral genetics, biochemical genetics, clinical genetics, cytogenetics, dysmorphology, gene therapy, genetic counseling, genetic epidemiology, genomics, immunogenetics, molecular genetics, neurogenetics, and population genetics.

The *AJHG* welcomes submissions of articles and reports on timely subjects concerning all aspects of human genetics, including studies of model organisms that are of direct relevance to human genetics. Manuscripts should be written in a manner accessible to investigators representing diverse backgrounds in human genetics. Descriptions of new statistical methods of general interest to the genetics community are welcome. New methods should be compared to existing methods using real data and/or simulations with parameters (e.g. haplotype frequencies, effect sizes) that are based on a real data example (e.g. marker/haplotype data from the HapMap project). All novel computer programs must be made publicly available by the time the manuscript is published and a URL for the website must be included in the Web Resources section of the manuscript. Letters commenting on material previously published in the *AJHG* are also welcome.

The *AJHG* does not publish reports of either single mutations or mutational surveys of previously identified loci unless they have unusual significance and substantial insight. Descriptions of new linkage assignments will be considered only if they are of special interest. Reports of negative data will not normally be considered.

Editorial Process

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 3-5 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 4 weeks of the submission date. If revisions are a condition of publication, we generally allow 4 weeks for revisions and consider only one revised version of the paper. Evaluations of conceptual advance and significance are made based on the literature available on the day of the final decision, not the day of submission. Accepted papers will be published within 3 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 24- to 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 the paper is not under consideration by another publication or electronic medium. All in-press or submitted works that are pertinent to the manuscript under consideration by the 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. Under no circumstances will any paper be considered that contains any data that have been submitted for publication elsewhere.

Authorship

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 the *AJHG*. In a case where we become aware of an authorship dispute, authorship must be approved in writing by all of the parties.

Conflict of Interest

The *AJHG* requires all authors to disclose any financial conflict of interest that might be construed to influence the results or interpretation of their manuscript. Authors must declare any such conflict in the cover letter accompanying the manuscript and in the Acknowledgments section of the manuscript itself. The

corresponding author will be asked to sign a form on behalf of all the authors regarding potential conflicts of interest at the time of acceptance. As a guideline, any affiliation associated with a payment or financial benefit exceeding \$10,000 p.a. or 5% ownership of a company or research funding by a company with related interests would constitute a conflict that must be declared. This policy applies to all submitted research manuscripts and review material. Examples of statement language include: AUTHOR is an employee and shareholder of COMPANY; AUTHOR is a founder of COMPANY and a member of its scientific advisory board; This work was supported in part by a grant from COMPANY.

Studies Involving Humans and Animals

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 at the time of submission (fax: 617-525-4569). 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 your IRB can be sent to us confirming that signed consent forms have been obtained from the individuals pictured in the manuscript, without naming the individuals. Your manuscript cannot be processed until we receive these consent forms or a letter from your IRB.

All experiments on live vertebrates or higher invertebrates must be performed in accordance with relevant institutional and national guidelines and regulations. In the manuscript, a statement identifying the committee approving the experiments and confirming that all experiments conform to the relevant regulatory standards must be included in the Experimental Procedures section. The editors reserve the right to seek comments from reviewers or additional information from authors on any cases in which concerns arise.

Distribution of Materials and Data

One of the terms and conditions of publishing in the *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 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 maintenance 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>).

Newly described SNPs should be submitted to an appropriate database such as dbSNP (<http://www.ncbi.nlm.nih.gov/SNP/>) prior to submission of revised manuscripts. The identification numbers should be used to describe the SNPs in the manuscript.

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

In addition to the information that must be deposited in public databases as detailed above, authors are encouraged to contribute additional information to the appropriate databases. Authors are also encouraged to deposit materials used in their studies to the appropriate repositories for distribution to researchers.

Clinical Trials

All clinical trials must be registered in a public trials registry. See <http://jama.ama-assn.org/cgi/content/full/292/11/1363> for guidelines.

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All meta-analyses and systematic reviews of observational studies should comply with MOOSE guidelines (<http://jama.ama-assn.org/cgi/content/full/283/15/2008>).

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Please provide a URL for the website upon which the novel computer program described in your manuscript will be made publicly available.

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AJHG facilitates author posting in connection with the voluntary posting request of the NIH (referred to as the NIH “Public Access Policy,” see <http://publicaccess.nih.gov/>) by submitting all peer-reviewed authors’ manuscripts directly to PubMed Central six months after publication. This ensures that you will have responded fully to the NIH policy and also the policies of other research-funding organizations, such as Wellcome Trust and Howard Hughes Medical Institute.

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Prepublication Publicity

Work intended for submission to the *AJHG*, currently under consideration at the *AJHG*, or in press at the *AJHG* may not be discussed with the media before publication. Providing preprints, granting interviews, discussing data with members of the media, or participating in press conferences in advance of publication without prior approval from the *AJHG* editorial office may be grounds for rejection. With regards to the scientific literature, papers in press in the *AJHG* should not be discussed in reviews elsewhere prior to the date of publication. Authors are welcome to present and discuss material intended for publication in the *AJHG* at scientific meetings; however, we do require that they refrain from discussing the findings with members of the press beyond those included in the formal presentation. In addition, we encourage meeting organizers to contact our editorial office in advance of a meeting if they have any questions regarding this policy. Any questions or issues regarding prepublication publicity should be directed to the Press Officer of Cell Press, Cathleen Genova (cgenova@cell.com).

Interaction with Members of the Press for Papers in Press

Authors are free to talk with the press starting on the Friday of the week before online publication and to release information provided that its use is embargoed until noon Eastern Time on the day of online publication. We also allow authors to discuss their work in press with other scientific journals for purposes of coverage in review material. Please refer any inquiries from other journals to us.

If your press office wishes to issue a press release, they should contact us in advance for final embargo information. Any questions or issues regarding interaction with the press should be directed to the Press Officer of Cell Press, Cathleen Genova (cgenova@cell.com).

Presubmission and Status Inquiries

Presubmission Inquiries

If you would like editorial input on whether your paper might be a strong candidate for consideration at the *AJHG*, you can submit a presubmission inquiry to ajhg@ajhg.net. This should include an abstract plus a brief description of the results and an explanation of the interest and significance to the broad readership of the *AJHG*. We try to respond to inquiries within 2–5 days.

Status Inquiries

Editorial Manager identifies the status as “with editor” until a decision is made. If you would like more information about the status of your submitted manuscript, please email ajhg@ajhg.net.

Manuscript Formats

Articles, Reports, Letters to the Editor, and Errata are all handled through our online submission system, Editorial Manager, at <http://www.edmgr.com/ajhg>. 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: Abstract, Introduction, Material and Methods (or Subjects and Methods), Results, and Discussion.

Reports

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. This category is designed for timely publication of original findings that may substantially impact their field.

Letters to the Editor

Letters to the Editor are brief commentaries (less than one printed page, at about 900 words per page) concerning manuscripts published in the *AJHG* within the past four months. They may also present new data. Any letter concerning a previously published manuscript will first be sent for external review to determine whether it meets an appropriate level of significance for publication in the *AJHG*. If the letter achieves this standing, it will then be sent to the corresponding author of the pertinent manuscript, who will be given an opportunity to submit a

response. This response will be provided to the corresponding author of the Letter to the Editor. After considering the response, the authors may choose to withdraw their letter or to proceed with publication without further revision.

Preparation and Online Submission of Research Manuscripts

Overview

The *AJHG* encourages authors to submit manuscripts via our online submission system, Editorial Manager (<http://www.edmgr.com/ajhg>). An author tutorial regarding online submission is available at the Editorial Manager website. Authors may contact the editorial office (ajhg@ajhg.net or 617-525-4770) for assistance. If online submission is not possible, a copy of the manuscript may be sent to Editor, *AJHG*, NRB 160A, 77 Avenue Louis Pasteur, Boston, MA 02115, USA. Please include a CD with electronic files of text and figures in the acceptable formats (see below).

Editorial Manager will prompt you to upload the individual components of your manuscript (cover letter, text, figures, supplemental data, etc.) as separate files. Upon completion of this step, the website will build a composite PDF file of your entire manuscript that will contain links for the editors and reviewers to download the individual high-resolution files of each component. Please note that the version of the PDF file that is accessible to reviewers does not contain the cover letter; any information therein will remain confidential.

Please be aware that Editorial Manager will send all communications about the paper (including the request for final approval and the confirmation of submission) to the individual designated as corresponding author during the submission process, or if no name is designated, to the individual whose account is used to submit the manuscript. If you wish to specify a different author for editorial correspondence after submission please contact the Editorial Assistant at ajhg@ajhg.net.

Cover Letter

Each submission should be accompanied by a cover letter, which should contain a brief explanation of what was previously known, the conceptual advance provided by the findings, and the significance of the findings to a broad readership. A cover letter may contain suggestions for appropriate reviewers and up to three requests for reviewer exclusions. The cover letter is confidential to the editor and will not be available to reviewers.

Also, indicate in the cover letter whether the first author is a student or postdoctoral fellow and a member of The American Society of Human Genetics (to be eligible for the Cotterman awards, presented at the annual meeting of the ASHG).

General Article Organization and Text Specifications

AJHG manuscripts generally contain the following sections in this order: Title Page including Authors, Affiliations, Contact Information, and Footnotes (if any); Abstract; Text (which, in articles, should be separated into Introduction, Material and Methods, Results, and Discussion); Acknowledgments; References; Figures and Tables, with titles and legends for each; and Supplemental Data. The text (Title through Legends) should be provided as one document, which may also contain the Tables. Figures and Supplemental Data should be provided separately.

For human genes, use genetic notation and symbols approved by the HUGO Gene Nomenclature Committee (HGNC). Approved gene symbols may be obtained, prior to submission, from the HGNC via their Web site (<http://www.genenames.org>) or by contacting HUGO Gene Nomenclature Committee (HGNC), European Bioinformatics Institute (EMBL-EBI), Wellcome Trust Genome Campus, Hinxton, Cambridgeshire CB10 1SA, United Kingdom (fax: 44-1223-494-468; e-mail: hgnc@genenames.org). Nomenclature guidelines are available and the Gene Name Proposal form may be completed on the HGNC Web page (http://www.genenames.org/cgi-bin/hgnc_request.pl). See Wain et al. "Guidelines for Human Gene Nomenclature" (*Genomics* 79:464-470, 2002).

Use ISCN nomenclature for cytogenetic notation (see Shaffer LG and Tommerup N [eds] *ISCN 2005: An International System for Human Cytogenetic Nomenclature*. S. Karger, Basel, [www.karger.com/ISCN]). Human gene names and loci should be written in italicized capital letters and Arabic numerals. Protein product names are not italicized. For human mutation nomenclature, see Antonarakis et al. "Recommendations for a Nomenclature System for Human Gene Mutations" (*Hum Mutat* 11:1-3, 1998) (<http://www3.interscience.wiley.com/cgi-bin/fulltext/5001291/PDFSTART>). Approved mouse nomenclature may be obtained, prior to submission, from the Mouse Genomic Nomenclature Committee (MGNC), telephone: (207) 288-6429; fax: (207) 288-6132; e-mail: nomen@informatics.jax.org; web page: <http://www.informatics.jax.org/mgihome/nomen/>. For other organisms, consult the appropriate authorities.

Pedigrees should be drawn according to published standards (See *AJHG* 1995 56:745-752).

MIM numbers should be listed in the text, but not the abstract, after the first mention of the relevant disorder or gene. Example: adrenal hypoplasia congenita (AHC [MIM 300200]). In the Web Resources section, only the URL for the OMIM database, and not the individual MIM numbers, should be noted. Example: Online Mendelian Inheritance in Man (OMIM), <http://www.ncbi.nlm.nih.gov/Omim/>.

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

Manuscript file types acceptable for submission include Word, LaTeX, RTF, and TXT. Required items differ for each article type and are specified during the submission process.

Please note that the text should be double spaced and pages should be numbered. If using Microsoft Word, insert line numbers (under the Layout option of the Page Setup menu) for ease of reviewer reference. Although abstracts need to be entered as text files separate from the body of the manuscript during the online submission process, they should also be included within the manuscript file.

Manuscripts that do not conform to the format guidelines may be returned to the authors for reformatting.

Preparation of Specific Sections

Title

Titles can 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/Affiliations

Author names should be spelled out rather than set in initials. Authors should be footnoted to corresponding affiliations. Affiliations should contain the following core information: department(s)/subunit(s); institution; city, state/region, postal code; country.

Contact

The contact line should include the email address and phone/fax numbers of the corresponding author. The published corresponding author is responsible for ensuring adherence to all editorial and submission policies and for any communications that may result post-publication. Only one corresponding author may be designated.

Equal Contribution

Joint authorship is permitted only for the first author position, with a maximum of two individuals being given this designation.

Additional Footnotes

Footnotes are only allowed on page 1 of the text (and in tables). They may include a present address for authors or statement of equal contribution to the manuscript.

Abstract

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 summary of the significance of the findings. References should not be cited in the Abstract.

Introduction

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

Material and Methods

The Material and Methods section needs to include sufficient detail so that readers can understand how the experiments were performed, and so that all procedures can be repeated, in conjunction with cited references. This section should also include a description of any statistical methods employed in the study.

Results

This section may be divided with subheadings. Footnotes should not be used and will be transferred to the text.

Discussion

The Discussion should explain the significance of the results and place them into a broader context. It should not be redundant with the Results section. This section may contain subheadings.

Acknowledgments

This section may acknowledge contributions from non-authors, list funding sources, and should include a statement of any conflict of interests. This section should be proofread carefully, as amendments or corrections are unlikely to be considered for publication as Errata. This section may be used to list the contributions of individual authors.

Appendices

Detailed results of statistical analyses may be presented as an Appendix

Web Resources

Please provide accession numbers for data deposited in any genome or locus-specific database. (See Distribution of Materials and Data above for required databases.) The name of the database should be given in the text, and the database is listed along with its URL in a separate section entitled "Web Resources," following the Acknowledgments. URLs are not to be included in the text. For all computer programs, please provide a URL for the website upon which the novel computer program described in your manuscript will be made publicly available.

References

References should include only articles that are published or in press. Unpublished data, submitted manuscripts, abstracts, and personal communications should be cited within the text only. A personal communication should be documented by a letter of permission. Submitted articles should be cited as unpublished data, data not shown, or personal communication. Note: "et al." should only be used after 10 authors. Please use the following style for references:

Article in a periodical: 1. Sondheimer, N., and Lindquist, S. (2000). Rnq1: an epigenetic modifier of protein function in yeast. *Mol. Cell* 5, 163-172.

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.

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Figure Legends

Legends should be included in the submitted manuscript as a separate section. Each figure legend should have a brief title that describes the entire figure without citing specific panels, followed by a description of each panel. For any figures presenting pooled data, the measures should be defined in the figure legends (for example, data are represented as mean +/- SEM).

Tables

When creating a table, please use the Microsoft Word table function and do not place an Excel table in a Word document. Word tables should not be tab or space delineated and should not use color. Tables should include a title, and footnotes and/or legends should be concise. Use superscripted lowercase letters (beginning with 'a') for footnotes in 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. Tables not created with the Microsoft Word table function will need to be revised by the author.

Supplemental Data

Supplemental data 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.

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Supplemental movies may be submitted through Editorial Manager as .mov, .avi, .mpeg, or .gif files. By choosing the submission item "Supplemental Movies and Spreadsheets," the PDF builder will imbed links within the PDF where editors and reviewers will be able to download the files. This also works for Excel files that do not display properly once converted to a PDF. Please note that the size limit for these items is 10 MB per file.

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, NRB 160A, 77 Avenue Louis Pasteur, Boston, MA 02115, USA.

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Figures must conform to the following guidelines or authors will be asked to revise them. For figures that have multiple panels, the labels should be set in uppercase Helvetica or Arial letters and should not contain periods or parentheses. Please be sure to embed all fonts. Each figure must be assembled into one file that prints onto one 8 1/2 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.

Acceptable Image Formats for Online Submission:

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- Portable Document File (.pdf)
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For black and white photographs or micrographs and for any type of graph or drawing in grayscale, the resolution of your file should be a minimum of 500 dpi. For figures that contain color, the resolution should be a minimum of 300 dpi. Please note that figures should meet these resolution numbers at their approximate print sizes.

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We encourage authors to use colors that can be distinguished by color-blind readers. Please submit your figures in RGB or grayscale -- do not convert your 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.

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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.

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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. The cover letter 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 your editor for further details and to the additional guidelines below for final figures.

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- Includes all HUGO-approved gene names as well as all MIM numbers for all relevant disorders or genes
- Includes explanation of statistical methods in the Material and Methods
- For any figures presenting pooled data, definitions of the measures within the figure legends
- Includes experimental procedures of sufficient detail
- Includes author and affiliations lists with superscripted numbers
- Includes concise titles for figures and tables

2. Please submit the final version of text and figures through our online system.

3. Please send to us:

On a single CD:

Any additional supplemental data larger than 10 MB.

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- 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)

Please note that we cannot accept *PowerPoint* files. (See note under Figure Organization, Formats, and Style above).

File Naming

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

Production and Proofs

After final acceptance, the manuscript will be passed to the production team to be copyedited and prepared for printing. Members of the American Society of Human Genetics will not be charged page charges or color charges. Non-member 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 2 weeks prior to publication and need to be returned with corrections no more than 24 hours after receipt.

Contacts at *AJHG*

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Prepublication publicity and press issues: Cathleen Genova, Press Officer,
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