Human Genome Variation
Guide for Authors

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ABOUT THE JOURNAL

Aims and Scope
Human Genome Variation is an online-only, full Open Access journal that contains articles and reports about variation and variability in human genomes and the consequences, implications and future impacts for the study of human genomics.

An important and innovative feature of the journal is the Data Report and Software Report. Data Reports are short reports about human genome variation and variability which describe disease-causing variation and/or their frequencies. In addition, Data Reports can describe and analyse human multifactorial disease associated variations and/or their frequencies. Software Reports are used to report newly developed software, application tools or databases that enhance efficiencies in research of human genome variations.

A further feature of Human Genome Variation will be a curated database of the underlying data from Data Reports, which will grow into an important resource for the genomics community. Human Genome Variation also publishes Articles and Review Articles on the relevant topics in human genome studies. Full Articles will be accompanied by a professionally written Editorial Summary.

The intended audience for Human Genome Variation is researchers, scientists, clinicians, genetic counsellors and those interested in human genomics, from all sectors and from around the world.

Human Genome Variation is committed to providing an efficient service for both authors and readers. A streamlined peer review system, together with the support of an Editorial Board, allows a team of independent editors to make rapid and fair publication decisions. Prompt dissemination of accepted papers to Springer Nature's wide readership and beyond is achieved through a programme of continuous online publication. Published manuscripts are enhanced by innovative web technologies, including interactive browsing and efficient data- and text-mining.

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<td><strong>Data Report</strong>&lt;br&gt;Data Reports are short reports about human genome variation and variability, which describe disease-causing variation and/or their frequencies. In addition, Data Reports can describe, and document human multifactorial disease-associated variations and their frequencies. Data Report authors are asked to check the mutation description information with the mutalyzer name checker (<a href="https://mutalyzer.nl">https://mutalyzer.nl</a>) or relevant description checking system, and make sure that description follows the HGVS nomenclature in advance of submission. Please state in the manuscript cover letter that the checking process was undertaken. This format typically begins with a brief unreferenced abstract (not more than 70 words). The title is limited to 10 words (or 90 characters). The main text is typically no more than 1,500 words, including the abstract and contains no headings. Data Reports normally have no more than 2 display items, although this may be flexible at the discretion of the editor. References are limited to 20.</td>
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- Title page
- Abstract
- Introduction
- Materials and Methods
- Results
- Discussion
- Acknowledgements
- Conflict of Interest
- Software Availability (for Software Reports)
- References
- Figure legends
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Abstract: An abstract of not more than 150–200 words. The abstract should be comprehensible to readers before they have read the paper, and abbreviations and reference citations within the abstract should be avoided.

Introduction: This should give a short, clear account of the background and reasons for undertaking the study. It should not be a review of the literature. The Introduction should assume that the reader is knowledgeable in the field and should therefore be as brief as possible.

Materials and Methods: This section should contain sufficient detail so that all experimental procedures can be repeated by others, in conjunction with cited references. This section may be divided into subheadings to assist the reader. Names of products and manufacturers should be included only if alternative sources are deemed unsatisfactory.

Instruments used, as well as standard techniques and procedures applied throughout the work, should appear in a paragraph at the beginning of the Materials and Methods section. Novel experimental procedures should be described in detail, but published procedures should be referred to by literature citation of the original article and published modifications.

Authors should use approved nomenclature for gene symbols, and use symbols rather than italicized full names (TTN, not titin). Please consult the appropriate nomenclature databases for correct gene names and symbols. A useful resource is LocusLink. Approved human gene symbols are provided by HUGO Gene Nomenclature Committee (HGNC), e-mail: nome@galton.ucl.ac.uk; see also www.gene.ucl.ac.uk/nomenclature. Approved mouse symbols are provided by The Jackson Laboratory, e-mail: nomen@informatics.jax.org; see also www.informatics.jax.org/mgihome/nomen.

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References: Authors are responsible for the accuracy of the references. Only papers directly related to the article should be cited; exhaustive lists of related reading should be avoided. References should be numbered in order of appearance, cited in text using superscript numbers (if the citation falls next to punctuation, the number should be inserted after, not before, the punctuation).

The reference list should be double-spaced, and there should be only one reference per number. Include only published references or those accepted and waiting for publication (listed as ‘in press’ following digital object identifier number) - not personal communications, “submitted” papers, or text notes. (“Personal communication” and “Unpublished data” references should be inserted in the text in parentheses, e.g., “(J. Smith, personal communication).”

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Authors. List all authors up to six. If there are more than six authors, list the first six, then “et al.” (no comma before “et al.”). Do not use “and” before the last author’s name.


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Newspaper:

Press release:

Patent:

Figure Legends: These should be brief, specific and appear on a separate manuscript page after the References section.

Figures: Figures and images should be labelled sequentially and cited in the text. Figures should not be embedded within the text but rather uploaded as separate files. Figure legends should be submitted on a separate sheet with list of text captions to all figures. Detailed guidelines for submitting artwork can be found by downloading our Artwork Guidelines. The use of three-
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- EMBL via WEBIN: www.ebi.ac.uk/embl/Submission/webin.html

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- DDBJ: Center for Information Biology and DNA Data Bank of Japan National Institute of Genetics, Yata, Mishima, Shizuoka 411-8540, JAPAN; telephone: +81-559-81-6853; fax: +81-559-81-6849; e-mail: ddbj@ddbj.nig.ac.jp URL: www.ddbj.nig.ac.jp

Supplementary Information: Supplementary information (SI) is peer reviewed material directly relevant to the conclusion of an article that cannot be included in the article owing to format constraints. The article must be complete and self-explanatory without the SI, which is posted on the journal’s website and linked to the article. SI may consist of data files, graphics, movies or extensive tables. Please see our Artwork Guidelines for information on accepted file types.

Authors should submit SI files in the FINAL format as they are not edited, typeset or changed, and will appear online exactly as submitted. When submitting SI, authors are required to:

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- Colour should be distinct when being used as an identifying tool.
- At first mention of a manufacturer, the town (and state if USA) and country should be provided.
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- Units: Use metric units (SI units) as fully as possible. Preferably give measurements of energy in kilojoules or Megajoules with kilocalories in parentheses (1 kcal = 4.186kJ). Use % throughout.
- Abbreviations: On first using an abbreviation place it in parentheses after the full item. Very common abbreviations such as FFA, RNA, need not be defined. Note these abbreviations: gram g; litre l; milligram mg; kilogram kg; kilojoule kJ; megajoule MJ; weight wt; seconds s; minutes min; hours h. Do not add s for plural units.

Nucleotide data

New nucleotide data must be deposited in the DDBJ/EMBL/GenBank databases and an accession number obtained before a paper can be accepted for publication. Submission to any one of the three collaborating databanks is sufficient to ensure data entry in all. The accession number should be included in the manuscript, e.g. as a footnote on the title page: The nucleotide sequence data reported are available in the DDBJ/EMBL/GenBank databases under the accession number(s) ----. If requested, the database will withhold release of data until publication. The most convenient method for submitting sequence data is by using the following URLs:

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For special types of submissions (e.g. genomes and bulk submissions), additional submission systems are available at the following sites:

- DDBJ: Center for Information Biology and DNA Data Bank of Japan National Institute of Genetics, Yata, Mishima, Shizuoka 411-8540, JAPAN; telephone: +81-559-81-6853; fax: +81-559-81-6849; e-mail: ddbj@ddbj.nig.ac.jp URL: www.ddbj.nig.ac.jp
HGV Database

*Human Genome Variation* hosts a fully searchable database of genomic variation as documented in the published Data Reports. These will be linked to the journal content and provide an important step towards giving the research community a verified and accessible place to publish, share and further utilize human genomics articles, data and analysis. For this purpose authors of Data Reports are required to fill in the form to register information on the reported genome variation and submit it together with the manuscript. For further information please see Database FAQ page on the journal’s site.

Data Report authors are asked to check the mutation description information with the mutalyzer name checker ([https://mutalyzer.nl](https://mutalyzer.nl)) or relevant description checking system, and make sure that description follows the HGVS nomenclature in advance of submission. Please state in the manuscript cover letter that the checking process was undertaken.

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MIAME-compliant microarray data: deposit in GEO or MGED web site specifying microarray standards.

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Reviewer selection is critical to the publication process, and we base our choice on many factors, based on expertise, reputation, and specific recommendations. A reviewer may decline the invitation to evaluate a manuscript where there is a perceived conflict of interest (financial or otherwise).

Appeals
Even in cases where editors did not invite resubmission, some authors ask the editors to reconsider a rejection decision. These are considered appeals, which, by policy, must take second place to the normal workload. In practice, this means that decisions on appeals often take several weeks. Only one appeal is permitted for each manuscript, and appeals can only take place after peer review.

Decisions are reversed on appeal only if the editors are convinced that the original decision was a serious mistake, not merely a borderline call that could have gone either way. Further consideration may be merited if a referee made substantial errors of fact or showed evidence of bias, but only if a reversal of that referee’s opinion would have changed the original decision. Similarly, disputes on factual issues need not be resolved unless they were critical to the outcome. Thus, after careful consideration of the authors’ points, most appeals are rejected by the editors.

If an appeal merits further consideration, the editors may send the authors’ response or the revised paper to one or more referees, or they may ask one referee to comment on the concerns raised by another referee. On occasion, particularly if the editors feel that additional technical expertise is needed to make a decision, they may obtain advice from an additional referee.

**Correction and Retraction Process**

Once the paper is published online it is considered final and cannot be amended. The online version is part of the published record hence the original version must be preserved and changes to the paper should be made as a formal correction.

Please note the following categories of corrections to peer reviewed content:

- **Erratum.** Notification of an important error made by the journal that affects the publication record or the scientific integrity of the paper, or the reputation of the authors, or of the journal.
- **Corrigendum.** Notification of an important error made by the author that affects the publication record or the scientific integrity of the paper, or the reputation of the authors or the journal.
- **Retraction.** Notification of invalid results. All co-authors 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 peer reviewers' advice) 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 co-authors disagree about a correction, the editors will take advice from independent peer reviewers and impose the appropriate correction, noting the dissenting author(s) in the text of the published version. Please see authors & referees for detailed information about author and referee services and publication policies of the Nature family of journals.

These journals, including *Human Genome Variation*, share a number of common policies including the following:

- Author responsibilities
- Licence agreement and author copyright
- Embargo policy and press releases
- Use of experimental animals and human subjects
- Competing financial interests
- Availability of materials and data
- Digital image integrity and standards
- Biosecurity concerns
- Refutations, complaints and corrections
- Duplicate publication
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- Plagiarism and fabrication

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