Human Genome Variation
Guide for Authors

About the Journal ............................................................... 1
Article Type Specifications .................................................. 2
Preparation of Articles ......................................................... 2
How to Submit ..................................................................... 6
Post-Acceptance ................................................................. 6
Costs .................................................................................. 7
Editorial Policies ................................................................. 7
Further information ............................................................ 11

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 article; these 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.

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 Nature Publishing Group'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.

Journal Details
Editor-in-Chief:
Katsushi Tokunaga
Professor, Department of Human Genetics
Graduate School of Medicine
The University of Tokyo

Editorial office:
Human Genome Variation Editorial Office
Nature Publishing Group
Chiyoda Building
2-37 Ichigayatamachi, Shinjuku-ku
Tokyo 162-0843 Japan
Email: hgv@nature.com

Impact factor:
Human Genome Variation is in the process of applying for listing by Thomson Reuters for an Impact Factor.

Abstracted in:
Google Scholar
ARTICLE TYPE SPECIFICATIONS

<table>
<thead>
<tr>
<th>Article Description</th>
<th>Word Limit</th>
<th>Tables/Figures</th>
<th>References</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Article</strong></td>
<td>Article: 5,000 words max including abstract (150-200 words) but excluding references and figure captions.</td>
<td>Max of 6</td>
<td>Max of 50. Please use as recent as possible.</td>
</tr>
<tr>
<td><strong>Review Article</strong></td>
<td>Article: 5,000 words max including abstract (150-200 words) but excluding references and figure captions.</td>
<td>Max of 8</td>
<td>Max of 100</td>
</tr>
<tr>
<td><strong>Data Report</strong></td>
<td>Article: 1,500 words max including abstract (70 words) excluding references, figures and tables.</td>
<td>Max of 2</td>
<td>Max of 20</td>
</tr>
<tr>
<td><strong>Editorial</strong> (by Editor invitation only)</td>
<td>1,000 words</td>
<td>Max of 2</td>
<td>Max of 5</td>
</tr>
</tbody>
</table>

PREPARATION OF ARTICLES

Please note that original articles must contain the following components. Please see below for further details.

- Title page
- Abstract
- Introduction
- Materials and Methods
- Results
- Discussion
- Acknowledgements
- Conflict of Interest
- References
- Figure legends
- Tables
- Figures
Cover Letter: The uploaded covering letter must state the material is original, has not been previously published and has not been submitted for publication elsewhere while under consideration. If the manuscript has been previously considered for publication in another journal, please include the previous reviewer comments, to help expedite the decision by the Editorial team. A Conflict of Interest statement should also be included.

Title Page: The title page should bear the title of the paper, the full names of all the authors and their affiliations, together with the name, full postal address, telephone and e-mail address of the author to whom correspondence are to be sent (this information is also asked for on the electronic submission form).

- The title should be brief, informative, of 150 characters or less and should not make a statement or conclusion.
- The running title should consist of no more than 50 letters and spaces. It should be as brief as possible, convey the essential message of the paper and contain no abbreviations.
- Authors should disclose the sources of any support for the work, received in the form of grants and/or equipment and drugs.
- If authors regard it as essential to indicate that two or more co-authors are equal in status, they may be identified by an asterisk symbol with the caption ‘These authors contributed equally to this work’ immediately under the address list.

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: nomenclature@ebi.ac.uk; see also www.gene.ucl.ac.uk/nomenclature. Approved mouse symbols are provided by The Jackson Laboratory, e-mail: nomenclature@informatics.jax.org; see also www.informatics.jax.org/mgihome/nomenclature.

Avoid listing multiple names of genes (or proteins) separated by a slash, as in ‘OCT4/POU5F1’, as this is ambiguous (it could mean a ratio, a complex, alternative names or different subunits). Use one name throughout and include the other at first mention: ‘OCT4 (also known as POU5F1)’.

Results: The description of results should not simply reiterate data that appear in tables and figures and, likewise, the same data should not be displayed in both tables and figures. The results section should be concise and follow a logical sequence. If the paper describes a complex series of experiments, it is permissible to explain the protocol/experimental design before presenting the results. Do not discuss the results or draw any conclusions in this section. This section may be divided into subheadings to assist the reader. Large datasets or other cumbersome data pertinent to the manuscript may be submitted as supplementary information.

Discussion: Do not recapitulate the results, but discuss their significance against the background of existing knowledge, and identify clearly those aspects that are novel. The final paragraph should highlight the main conclusion(s), and provide some indication of the direction future research should take. This section may be divided into subheadings to assist the reader. Results and Discussion may be combined.

Acknowledgements: These should be brief, and should include sources of financial support, material (e.g. novel compounds, strains, etc.) not available commercially, personal assistance, advice from colleagues and gifts.

Conflict of Interest: Authors must declare whether or not there are any competing financial interests in relation to the work described. This information must be included at this stage and will be published as part of the paper. Conflict of Interest should be noted in the cover letter and in the paper. Please see the Conflict of Interest documentation in the Editorial Policy section for detailed information.
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).” Similarly, references to a general website instead of a specific page or document should not be included in the reference list, but instead inserted in the text in parentheses, e.g., “(http://who.org)”. If there are references that are cited only in a figure or table, place them at the end of the list. In other words, number all references in the main text first, then any that appear only in figures, followed by any that appear only in tables.

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.


Book publishers. In publishers’ names, omit “Company”, “Inc.”, “Verlag”, first names and any initials (e.g., “Wiley”, not “John Wiley”, “Norton”, not “WW Norton”). Include the city, state (for all cities except New York), and country (including “USA” but not “UK” for London—in other words, New York and London are the only cities that do not need to be further described.

Online publication. In general, the order is author (or authoring entity, e.g., “US Food and Drug Administration”), title of page or document, date (at least the year), URL (always include “http://”), access date (if relevant).

Examples:

Journal article, up to six authors:

Journal article, more than six authors:

Journal article, e-pub ahead of print:

Journal article, in press [note that the year is not included for “in press” references] :

Abstract/supplement:

Letter:

Book (complete):

Book (chapter in book):

Book (with volume and edition information):

Meeting:

**Online (journal):**

**Online (dated report):**

**Online (dynamic Web page):**

**Thesis:**

**Package inserts and prescribing information:**

**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-dimensional histograms is strongly discouraged when the addition of the third dimension gives no extra information. 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.

**Tables:** These should be labelled sequentially and cited within the text. Each table should be presented on its own page, numbered and titled. Reference to table footnotes should be made by means of Arabic numerals. Tables should not duplicate the content of the text. They should consist of at least two columns; columns should always have headings. Authors should ensure that the data in the tables are consistent with those cited in the relevant places in the text, totals add up correctly, and percentages have been calculated correctly. Unlike figures or images, tables may be embedded into the word processing software if necessary, or supplied as separate electronic files.

**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:
- Include a text summary (no more than 50 words) to describe the contents of each file.
- Identify the types of files (file formats) submitted.
- Include the text “Supplementary information is available at (journal name)’s website” at the end of the article and before the references.

**House Style**
- All pages and lines are to be numbered. To add page numbers in MS Word, go to Insert then Page Numbers. To add line numbers go to File, Page Setup, then click the Layout tab. In the Apply to box, select Whole document, click Line Numbers then select the Add line numbering check box, followed by Continuous.
- Use a coarse hatching pattern rather than shading for tints in graphs.
- 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.
- Statistical methods: For normally distributed data, mean (SD) is the preferred summary statistic. Relative risks should be expressed as odds ratios with 95% confidence interval. To compare two methods for measuring a variable the method of Bland & Altman (1986, Lancet 1, 307–310) should be used; for this, calculation of P only is not appropriate.
- Units: Use metric units (SI units) as fully as possible. Preferably give measurements of energy in kilojoules 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:
- DDBJ via SAKURA: http://sakura.ddbj.nig.ac.jp/
- EMBL via WEBIN: www.ebi.ac.uk/embl/Submission/webin.html

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
- EMBL: EMBL Nucleotide Sequence Submissions, European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge CB10 1SD, U.K.; telephone: +44-1223-494400; fax: +44-1223-494472; e-mail: datasubs@ebi.ac.uk URL: www.ebi.ac.uk
- GenBank: National Center for Biotechnology Information, National Library of Medicine, Bidg. 38A, Rm 8N-803, Bethesda, Maryland 20894, USA; telephone: +1-301-496-2475; fax: +1-301-480-9241; e-mail: info@ncbi.nlm.nih.gov URL: www.ncbi.nlm.nih.gov

**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.

**Language Editing**
Authors who are not native speakers of English sometimes receive negative comments from referees or editors about the language and grammar usage in their manuscripts, which can contribute to a paper being rejected.

To reduce the possibility of such problems, we strongly encourage such authors to take at least one of the following steps:
- Have your manuscript reviewed for clarity by a colleague whose native language is English.
- Review the tips for technical writing here: http://www.nature.com/authors/author_resources/how_write.html.
- Use an English language editing service such as Nature Publishing Group Language Editing. An editor will improve the English to ensure that your meaning is clear and to identify problems that require your review.
Please note that the use of a language editing service is at the author's own expense and does not guarantee that the article will be selected for peer review or accepted.

HOW TO SUBMIT

Pre-submission Enquiries
Please submit via our online manuscript submission system or via e-mail to HGV editorial office.

Online Submission
We only accept manuscript submission via our online manuscript submission system. Before submitting a manuscript, authors are encouraged to consult both our Editorial Policies and the Submission Instructions for our online manuscript submission system. If you have not already done so, please register for an account with our online manuscript system. You will be able to monitor the status of your manuscript online throughout the Editorial process.

Submission of Revisions
Authors submitting a revised manuscript after review are asked to include the following:
(1) A rebuttal letter, indicating point-by-point how you have addressed the comments raised by the reviewers. If you disagree with any of the points raised, please provide adequate justification in your letter.
(2) A marked-up version of the manuscript that highlights changes made in response to the reviewers' comments in order to aid the Editors and reviewers. Do not use track changes or comments.
(3) A 'clean' (non-highlighted) version of the revised manuscript.

POST-ACCEPTANCE

Once a manuscript is accepted, the corresponding author must complete and sign a Licence to Publish form and Article Processing Charge Payment form on behalf of all authors and return it to the Editorial office. Failure to promptly return the form will result in delay of publication.

Nature Publishing Group does not require authors of original research papers to assign copyright of their published contributions. Authors grant NPG an exclusive licence to publish, in return for which they can re-use their papers in their future printed work. NPG's author licence page provides details of the policy.

Publication
Publishing Open Access will mean the paper is freely accessible online immediately upon publication. By paying this charge authors are permitted to post the final, published PDF of their article on a website, institutional repository or other free public server, immediately on publication.

Open Access articles can be published under one of three Creative Commons licences, at the free choice of the authors. Authors should note that some funders require papers to be published under a specific licence and so should check the funder mandate to ensure compliance:

With regards to payment, usual credit terms are 30 days from receipt of invoice. Failure to pay your invoice within the stated credit term may result in the Open Access status of the paper being rescinded. You may also be subject to such penalties as restrictions on your ability to publish with Nature Publishing Group in the future, involvement of a third party debt collection agency and legal proceedings. For further information on Open Access please see the FAQs page.

Open Access Waiver
Please note that some institutions have enacted Open Access policies that conflict with our own. If any corresponding or contributing authors are from these institutions, you will need to provide a waiver from the institution of every affected author, which can be obtained from the institution. This waiver should be submitted at the same time as the Open Access Licence to Publish form.

Self-Archiving
Authors are also encouraged to submit their version of the accepted, peer reviewed manuscript to their funding body's archive for public release six months after print 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 NPG website.

This policy complements the policies of the US National Institutes of Health, the Wellcome Trust and other research funding bodies around the world. NPG recognises the efforts of funding bodies to increase access of the research they fund, and strongly encourages authors to participate in such efforts.

Proofs
The corresponding author will receive an e-mail containing a URL linking to the proofing site. Proof corrections must be returned within 48 hours of receipt. Failure to do so may result in delayed publication. Extensive corrections cannot be made at this stage.

COSTS

Article Processing Charge
Authors whose papers are accepted for publication in Human Genome Variation are required to pay an article-processing charge (APC). Depending on the type of Creative Commons license and type of contents, the following charge will apply (plus VAT where applicable):

<table>
<thead>
<tr>
<th>Creative Commons License</th>
<th>Articles &amp; Review Article</th>
<th>Data Report</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC-NC-SA/CC-NC-ND</td>
<td>£2,650 / $3,975 / €3,000 / ¥424,000 / RMB 27,900</td>
<td>£1,000 / $1,500 / €1,140 / ¥160,000 / RMB 10,500</td>
</tr>
<tr>
<td>CC-BY</td>
<td>£2,900 / $4,400 / €3,300 / ¥467,000 / RMB 30,700</td>
<td>£1,100 / $1,650 / €1,250 / ¥176,000 / RMB 11,550</td>
</tr>
</tbody>
</table>

The Creative Commons Attribution 3.0 Unported License (CC-BY) grants greater end user rights including commercial reuse, papers published under this license will be charged at a premium APC. For more information on this licence please see the press release.

EDITORIAL POLICIES

A manuscript will be considered for publication on the understanding that all named authors have agreed to its submission and that if accepted it will not be later published in the same or similar form in any language without the consent of the publishers.

Duplicate Publication
Papers must be original and not published or submitted for publication elsewhere. This rule also applies to non-English language publications. NPG allows and encourages prior publication on recognized community preprint servers for review by other scientists before formal submission to a journal. The details of the preprint server concerned and any accession numbers should be included in the cover letter accompanying manuscript submission. This policy does not extend to preprints available to the media or that are otherwise publicized outside the scientific community before or during the submission and consideration process.

Permissions
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. A quote will be supplied upon acceptance of your paper.

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, behavioural 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, please 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.

All clinical trials must be registered in a public registry prior to submission. *Human Genome Variation* follows the trials registration policy of the ICMJE ([www.icmje.org](http://www.icmje.org)) and considers only trials that have been appropriately registered before submission, regardless of when the trial closed to enrolment. Acceptable registries must meet the following ICMJE requirements:

- be publicly available, searchable, and open to all prospective registrants
- have a validation mechanism for registration data
- be managed by a not-for-profit organization.

The trial registry number for eligible papers will be collected during the submission process.

Nature Publishing Group endorses the toolkits and guidelines produced by the following bodies:

**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 after the acknowledgements of their manuscript.

*Human Genome Variation* requires authors of all submitted original research papers to declare any Conflict of Interest (COI) in relation to the submitted work, following the guidelines and detailed regulations set by the Japan Society of Human Genetics (JSHG) in 2012.

Authors submitting their manuscripts using the journal’s online manuscript tracking system are required to make their declaration as part of this process and to specify the competing interests in cases where they exist.

**Criteria for COI disclosure**

1. Received honoraria of 1,000,000 yen or more (in one year) for lectures, article contributions or such activities to support promotional activities
2. Received 1,000,000 yen or more (in one year) for Employment, supervising or advisory position
3. Given stock or stock options worth 5% or more of the total share, or generating profit of 1,000,000 yen or more (in one year).
4. Received patent royalties or licensing fees of 1,000,000 yen or more (in one year)
5. Received research funding of 2,000,000 yen or more (in one year)
6. Outside research activities, given travels, gifts or any other benefit worth 50,000 yen or more in one year.

If the self-reported COI includes any of the above listed situation, authors should add COI statement to the end of the manuscript main text, and before the acknowledgement or the list of references. Please note that the disclosure is required only for the relationship that the author had within one year before the date of submission.

Referees are also requested to indicate any potential conflict they might have reviewing a particular paper.

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.

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 heading, there must be a listing for each author, detailing the professional services relevant to 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.

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.

**Sharing data sets**
A condition of publication is that authors are required to make materials, data and associated protocols promptly available to
others without preconditions. Data sets must be made freely available to readers from the date of publication, and must be
provided to editors and peer reviewers at submission, for the purposes of evaluating the manuscript.

For the following types of data set, submission to a community-endorsed, public repository is mandatory. Accession numbers
must be provided in the paper. Examples of appropriate public repositories are listed below.

[Nucleotide and protein sequences]
DNA and RNA sequences: DDBJ, Genbank, or European Nucleotide Archive(ENA).
DNA sequencing data (traces for capillary electrophoresis and short reads for next-generation sequencing): DDBJ Sequence
Read Archive, NCBI Sequence Read Archive(SRA), or EBI Sequence Read Archive(ERA). Deep sequencing data: deposit in
GEO or ArrayExpress upon submission to the journal. Accession numbers must be provided in the published manuscript. This
policy includes even short stretches of novel sequence information such as epitopes, functional domains, genetic markers, or
haplotypes. Short novel sequences must include surrounding sequence information to provide context. The sequences of all
RNAi, antisense and morpholino probes must be included in the paper or deposited in a public database, with the accession
number quoted. When an unpublished library is included in the paper, at minimum the sequences of the probes central to the
conclusions of the paper must be presented.

Protein sequences: deposit in Protein DataBank, UniProt.

[Microarray gene expression data]
MIAME-compliant microarray data: deposit in GEO or MGED web site specifying microarray standards.

[Genome-wide association data]
We encourage authors to deposit data from genome-wide association studies in *Human Genome Variation* Database, dbGap, or
EGA (European Genome-phenome Archive) upon submission to the journal.

**Pre- and Post-Submissions**
Authors are welcome to post pre-submission versions or the original submitted version of the manuscript on a personal blog, a
collaborative wiki or an institution-hosted repository recognized preprint server (such as ArXiv) at any time (but not
subsequent pre-accept versions that evolve due to the Editorial process).

The published version — copyedited and in the individual NPG journal format — may not be posted on any website or
preprint server.

For content published under a creative commons license, authors can replace the submitted version with the final published
version at publication as long as a publication reference and URL to the published version on the journal website are provided.

The editors also reserve the right to reject a paper even after it has been accepted if it becomes apparent that there are serious
problems with the scientific content or with violations of our publishing policy.

**Peer Review**
Manuscripts sent out for peer review are evaluated by at least one independent reviewer (often two or more). Authors are
welcome to suggest independent reviewers to evaluate their manuscript, as well as request individuals or laboratories. All
recommendations are considered, but it is at the Editor’s discretion their choice of reviewers. To expedite the review process,
only papers that seem most likely to meet Editorial criteria are sent for external review. Papers judged by the editors to be of
insufficient general interest or otherwise inappropriate are rejected promptly without external review. The editors then make a
decision based on the reviewers' evaluations:

- **Accept**, with or without Editorial revisions.
- **Revise**, with the author addressing concerns raised by the reviewers before a final decision is reached.
• **Reject**, but indicate to the authors that further work might justify a resubmission.
• **Reject outright**, typically on grounds of specialist interest, lack of novelty, insufficient conceptual advance or major technical and/or interpretational problems.

Selecting Peer Reviewers
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 @ npg 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**
**Confidentiality and pre-publicity**
**Plagiarism and fabrication**
FURTHER INFORMATION

For inquiries related to submission requirements, please contact the Editorial office. For inquiries related to advertising, subscriptions, permissions, papers in production or publishing a supplement, please contact the publisher’s office.