

# Human Genome Variation

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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.

#### Journal Details

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Issei Imoto  
Division of Molecular Genetics  
Aichi Cancer Center Research Institute Preventive Medicine

##### Editorial office:

*Human Genome Variation* Editorial Office  
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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:

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## ARTICLE TYPE SPECIFICATIONS

Article Description	Word Limit	Tables/ Figures	References
<p><b>Article</b> Studies that are of high scientific quality and that are of interest to the diverse readership of the journal. Manuscripts should include an abstract and appropriate experimental details to support the conclusions. Articles should be no more than 5000 words excluding references and figure legends and should not normally include more than six display items (tables and/or figures). They should include title, abstract, introduction, materials and methods, results and discussion sections.</p>	<p>Article: <b>5,000</b> words max including abstract (150-200 words) but excluding references and figure captions.</p>	<p>Max of <b>6</b></p>	<p>Max of <b>50</b>. Please use as recent as possible.</p>
<p><b>Review Article</b> Review Articles are normally solicited by the editors; however, we also welcome timely, unsolicited Review Articles. Authors with proposals for Review Articles should present information concerning the proposed content and authors to the editors prior to submission.</p>	<p>Article: <b>5,000</b> words max including abstract (150-200 words) but excluding References and figure captions.</p>	<p>Max of <b>8</b></p>	<p>Max of <b>100</b></p>
<p><b>Data Report</b> 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.</p>	<p>Article: <b>1,500</b> words max including unstructured abstract. References, figures and tables.</p>	<p>Max of <b>2</b></p>	<p>Max of <b>20</b></p>
<p><b>Software Report</b> Short papers describing newly developed software, application tools or databases designed to support studies and investigations of human genome variations and epigenetics. The scope of this article type includes genetic association study, genome analysis, sequence analysis, gene expression analysis, epigenetics, population genetics, cancer genomics. Suggested length of the paper is 1200 words or less including unstructured abstract and 'Software availability statement' in the end of the manuscript.</p>	<p>Article: <b>1,200</b> words max including unstructured abstract and software availability statement. References, figures and tables.</p>	<p>Max of <b>3</b></p>	<p>Max of <b>20</b></p>
<p><b>Editorial</b> (by Editor invitation only) Proposals for Editorial may be submitted; however, authors should only send an outline of the proposed paper for initial consideration.</p>	<p>1,000 words</p>	<p>Max of <b>2</b></p>	<p>Max of <b>5</b></p>

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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
- Software Availability (for Software Reports)
- References
- Figure legends
- Tables
- Figures

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- 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.

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**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.

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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](http://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](http://www.informatics.jax.org/mgihome/nomen).

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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.

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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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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.

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Miller W, Flynn P, McCullough J, Balfour HH Jr., Goldman A, Haake R *et al.* Cytomegalovirus infection after bone marrow transplantation: an association with acute graft-v-host disease. *Blood* 1986; **67**: 1162–1167.

**Journal article, e-pub ahead of print:**

Bonin M III, Pursche S, Bergeman T, Leopold T, Illmer T, Ehninger G *et al.* F-ara-A pharmacokinetics during reduced-intensity conditioning therapy with fludarabine and busulfan. *Bone Marrow Transplant* 2007; e-pub ahead of print 8 January 2007; doi:10.1038/sj.bmt.1705565.

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

Gallardo RL, Juneja HS, Gardner FH. Normal human marrow stromal cells induce clonal growth of human malignant T-lymphoblasts. *Int J Cell Cloning* (in press).

*Abstract/supplement:*

Syrjala KL, Abrams JR, Storer B, Heiman JR. Prospective risk factors for five-year sexuality late effects in men and women after haematopoietic cell transplantation. *Bone Marrow Transplant* 2006; **37**(Suppl 1): S4 (abstract 107).

*Letter:*

Caocci G, Pisu S. Overcoming scientific barriers and human prudence [letter]. *Bone Marrow Transplant* 2006; **38**: 829–830.

*Book (complete):*

Atkinson K, Champlin R, Ritz J, Fibbe W, Ljungman P, Brenner MK (eds). *Clinical Bone Marrow and Blood Stem Cell Transplantation*. Cambridge University Press: Cambridge, UK, 2004.

*Book (chapter in book):*

Coccia PF. Hematopoietic cell transplantation for osteopetrosis. In: Blume KG, Forman SJ, Appelbaum FR (eds). *Thomas' Hematopoietic Cell Transplantation*, 3rd edn. Blackwell Publishing: Malden, MA, USA, 2004: 1443–1454.

*Book (with volume and edition information):*

Shadwell, J. The common vampire fish. In: Howlett R, Thomas, A (eds). *Proc 4th Int Symp Transylvanian Fish Soc*, 2nd edn, vol 2. Springer: Berlin, Germany, 2012: 21–29.

*Meeting:*

Brentjens, R, Riviere, I, Frattini, M, Wang, X, Taylor, C, Olszewska, M *et al*. Marked regression of adenopathy following infusion of autologous T cells. Presented at the 13th annual meeting of the American Society of Gene and Cell Therapy, Washington, DC, 17–22 May 2010.

*Online (journal):*

Huynen MMTE, Martens P, Hilderlink HBM. The health impacts of globalisation: a conceptual framework. *Global Health* **1**: 14. <http://www.globalizationandhealth.com/content/1/1/14>.

*Online (dated report):*

Centers for Disease Control and Prevention. Smallpox vaccine and monkeypox. <http://www.cdc.gov/ncidod/monkeypox/pdf/vaccineqa.pdf>. 9 July 2003.

*Online (dynamic Web page):*

National Institutes of Health. Genome-Wide Association Studies (GWAS) (2006). <http://grants.nih.gov/grants/gwas/index.htm>. Accessed 4 January 2007.

*Thesis:*

Gee H. Trends in Infant Growth Rates. Thesis, Princeton University, 1978.

*Package inserts and prescribing information:*

Lamasil [package insert]. Sandoz Pharmaceuticals, 1993.  
Kaletra [prescribing information]. Abbott, 2005.

*Newspaper:*

FDA strengthens warnings on stimulants. *New York Times*, 22 August 2006.

*Press release:*

US Food and Drug Administration. FDA approves updated warfarin (Coumadin) prescribing information. Press release, 16 August 2007.

*Patent:*

Wilson ST, Oak S, Flanigen EM. US patent 4567029 (1986).  
Kuznicki SM, Thrush AK. European patent 0405978A1 (1990).

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- Include the text "Supplementary information is available at (journal name)'s website" at the end of the article and before the references.

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- 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.
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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](http://www.ebi.ac.uk/embl/Submission/webin.html)
- GenBank via BankIt: [www.ncbi.nlm.nih.gov/BankIt/](http://www.ncbi.nlm.nih.gov/BankIt/) or by stand-alone submission tool Sequin: [www.ncbi.nlm.nih.gov/Sequin/](http://www.ncbi.nlm.nih.gov/Sequin/)

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### 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>) 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.

- 1) Classification of pathogenicity for reported variants should be classified according to the ACMG\_AMP Guidelines (Ref. 1) and their modified criteria (see Ref. 2).

#### Reference

1. ACMG\_AMP Guidelines for the Interpretation of Sequence Variants  
Genet Med. 2015; 17(5): 405–424. doi: 10.1038/gim.2015.30
2. Consideration of Cosegregation in the Pathogenicity Classification of Genomic Variants  
Am J Hum Genet 2016; 98(6): 1077-1081. doi: <https://doi.org/10.1016/j.ajhg.2016.04.003>

- 2) Pedigree in the manuscript should be drawn according to the Standardized human pedigree nomenclature (Ref. 3)

#### Reference

3. Standardized human pedigree nomenclature  
J Genet Couns. 2008;17(5):424-33. doi: 10.1007/s10897-008-9169-9.
- 3) Compliance with HGVS nomenclature must be verified using tools such as the Mutalyzer program (<https://mutalyzer.nl/>; den Dunnen, 2016) or VariantValidator (<https://variantvalidator.org/>; Freeman et al., 2018) each of which offers a batch mode to facilitate rapid checking of multiple variant descriptions. (see Ref. 4)

#### Reference

4. Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Hum Mutat.

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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:

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