# *Human Genome Variation*

## Guide for Authors



## 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* ***i***s 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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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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Examples:

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Belkaid Y, Rouse BT. Natural regulatory T cells in infectious disease. *Nat Immunol* 2005; **6**: 353–360.

*Journal article, more than six authors:*

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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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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*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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[Nucleotide and protein sequences]
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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.

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