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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- Results
- Discussion
- Acknowledgements
- Conflict of Interest
- Software Availability (for Software Reports)
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Examples:

Journal article, up to six authors:

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Journal article, more than six authors:

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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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- 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

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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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