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 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 ***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 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

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- Abstract
- Introduction
- Materials and Methods
- Results
- Discussion
- Acknowledgements

- Conflict of Interest
- References
- Figure legends
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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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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).

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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.

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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:

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- **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.

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