Journal of Human Genetics

ABOUT THE JOURNAL

Aims and Scope

The *Journal of Human Genetics* is an international journal publishing articles on human genetics, including medical genetics and human genome analysis. It covers all aspects of human genetics, including molecular genetics, clinical genetics, behavioral genetics, immunogenetics, pharmacogenomics, population genetics, functional genomics, epigenetics, genetic counseling and gene therapy.

Articles on the following areas are especially welcome: genetic factors of monogenic and complex disorders, genome-wide association studies, genetic epidemiology, cancer genetics, personal genomics, genotype-phenotype relationships and genome diversity.

- Medical genetics
- Human genome analysis
- Gene cloning and mapping
- Linkage and association analyses
- Rare variants in diseases and phenotypes
- Mutational analysis
- Susceptibility genes to multifactorial disorders
- Human evolution
- Cancer genetics
- Gene therapy
- Genetic and functional analysis of animal models of disease or behaviour
- Genetic polymorphisms of biologically important genes
- Novel mutations found in patients with hereditary diseases
- Novel mutations found in cancer cells
- Population genetics
- Human diseases with epigenetic or chromatin dysregulation
- Human epigenome analysis
- Noncoding RNAs
- Developmental Origins of Health and Disease
- Prenatal diagnosis
- Molecular cytogenetics and genomic structural variation

Journal Details

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To qualify as a contributing author, one must meet all of the following criteria:

1) Conceived and/or designed the work that led to the submission, acquired data, and/or played an important role in interpreting the results.
2) Drafted or revised the manuscript.
3) Approved the final version.
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- have a validation mechanism for registration data
- be managed by a not-for-profit organization

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2) the International Standard Randomized Controlled Trial Number Registry (www.controlled-trials.com);
3) the Cochrane Renal Group Registry (www.cochrane-renal.org);
4) and the European Clinical Trials Database (https://eudract.ema.europa.eu/).

The trial registry number must be included in the manuscript and provided on submission.

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Nucleotide and protein sequences
Microarray gene expression data
Genome-wide association data

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Falsification is the practice of altering research data with the intention of giving a false impression. This includes, but is not limited to, manipulating images, removing outliers or "inconvenient" results, or changing, adding or omitting data points. Fabrication is the practice of inventing data or results and recording and/or reporting them in the research record. Data falsification and fabrication call into question the integrity and credibility of data and the data record, and as such, they are among the most serious issues in scientific ethics.

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- make inquiries of other titles believed to be affected;
- forward concerns to the author's employer or person responsible for research governance at the author's institution;
- refer the matter to other authorities or regulatory bodies (for example, the Office of Research Integrity in the US or the General Medical Council in the UK); or
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Digital image enhancement is acceptable practice, although it can result in the presentation of unrepresentative data as well as in the

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Images submitted with a manuscript for review should be minimally processed (for instance, to add arrows to a micrograph). Authors should retain their unprocessed data and metadata files, as editors may request them to aid in manuscript evaluation. If unprocessed data is unavailable, manuscript evaluation may be stalled until the issue is resolved.

A certain degree of image processing is acceptable for publication, but the final image must correctly represent the original data and conform to community standards. The guidelines below will aid in accurate data presentation at the image processing level:

- Authors should list all image acquisition tools and image processing software packages used. Authors should document key image-gathering settings and processing manipulations in the Methods section.
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- the source of the cell line, including when and from where it was obtained,
- whether the cell line has recently been authenticated and by what method, and
- whether the cell line has recently been tested for mycoplasma contamination.

Further information is available from the International Cell Line Authentication Committee (ICLAC). We recommend that authors check the NCBI database for misidentification and contamination of human cell lines.

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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

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- DDBJ via SAKURA: http://sakura.ddbj.nig.ac.jp/
- EMBL via WEBIN: http://www.ebi.ac.uk/embl/Submission/webin.html
- GenBank via BankIt: http://www.ncbi.nlm.nih.gov/BankIt/ or by stand-alone submission tool Sequin: http://www.ncbi.nlm.nih.gov/Sequin/

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- 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.jpURL: http://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: http://www.ebi.ac.uk
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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.
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