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ABOUT THE JOURNAL

Aims and Scope

The *European Journal of Human Genetics* is the official journal of the European Society of Human Genetics, publishing high-quality, original research papers, short reports and reviews in the rapidly expanding field of human genetics and genomics. It covers molecular, clinical and cytogenetics, interfacing between advanced biomedical research and the clinician, and bridging the great diversity of facilities, resources and viewpoints in the genetics community.

Key areas include:

- Monogenetic and multifactorial disorders
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- Medical genomics
- Gene mapping and functional studies
- Genotype-phenotype correlations
- Genetic variation and genome diversity
- Statistical and computational genetics
- Bioinformatics
- Advances in diagnostics
- Therapy and prevention
- Animal models
- Genetic services
- Community genetics

This journal also publishes invited editorials and commentaries, announcements of societal and other European activities, and special issues of general interest for the human genetics community.

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Editor-in-Chief:
Dr Alisdair McNeill, University of Sheffield, UK

Editorial office: ejhg@sheffield.ac.uk

Frequency: 12 issues a year

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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.
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It is difficult to specify a threshold at which a financial interest becomes significant, but note that many US universities require faculty members to disclose interests exceeding $10,000 or 5% equity in a company. Any such figure is arbitrary, so we offer as one possible practical alternative guideline: "Declare all interests that could embarrass you were they to become publicly known after your work was published." We do not consider diversified mutual funds or investment trusts to constitute a competing financial interest.

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- **Competing interests.**
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- have a validation mechanism for registration data
- be managed by a not-for-profit organization

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3. the Cochrane Renal Group Registry
4. the European Clinical Trials Database

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NOMENCLATURE

Gene Nomenclature

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. Approved human gene symbols are provided by HUGO Gene Nomenclature Committee (HGNC). Approved mouse symbols are provided by The Jackson Laboratory. For proposed gene names that are not already approved, please submit the gene symbols to the appropriate nomenclature committees as soon as possible, as these must be deposited and approved before publication of an article

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For sequence variants, both in nucleic acids and proteins, the *EJHG* supports the nomenclature recommendations of the Human Variome Project (Cotton RG et al.2007. Nat Genet.39:433 www.nature.com/npg/journal/v39/n4/full/ng2024.html)

Variant descriptions should follow current recommendations of the Human Genome Variation Society (HGVS) and their accuracy is the responsibility of authors. Please visit varnomen.hgvs.org/ for the latest nomenclature updates, for examples of acceptable nomenclature, guidance concerning reference sequences, or if you have further questions.

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Authors are required to submit all data described to a well-respected, long-lasting public repository. Authors must confirm the status of database submission in their cover letter and should note (in e.g. Materials and Methods section) the database(s), to which they have submitted the data, plus provide the URL and accession number(s). To allow accurate review of the manuscript, when data have been submitted to a repository working with controlled access (e.g. the European Genotype Archive) the authors should add a copy of the data submitted.

Recommended repositories include:

- sequences: EMBL, DDBJ, GenBank (incl. Trace Archives (incl. SRA), European Nucleotide Archive (ENA), dbSNP, European Variation Archive (EVA), dbVar, DGVa)
- variants and phenotypes: ClinVar/ClinGen, GV shared LOVD, Decipher
- association studies: dbGaP, EGA (European Genotyping Archive)
- micro-array data: ArrayExpress, GEO (Gene Expression Omnibus)
- proteins: ProteinDataBank, Biological Magnetic Resonance Databank
- protein interactions: DIP (Database of Interacting Proteins), IntAct Molecular Interaction database
- proteomics/metabolomics: PRIDE, MetaboLights
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For all manuscripts that include details, images, or videos relating to an individual person, written informed consent for the publication of these details must be obtained from that person (or their parent or legal guardian in the case of children under 18). If the person has died, consent must be sought from their next of kin. The manuscript must include a statement that written informed consent for publication was obtained.

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If there is suspicion of misconduct, the journal will carry out an investigation following COPE guidelines. Following an investigation, if the allegation raises valid concerns, the author will be contacted and given an opportunity to address the issue. If misconduct is established beyond reasonable doubt, this may result in the Editor implementing one of the following measures:

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In cases where co-authors disagree about a correction or retraction, the editors will take advice from independent peer-reviewers and impose the appropriate measure, noting the dissenting author(s) in the text of the published version.

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