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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
- Development and malformation
- Hereditary cancer
- 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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Dr Alisdair McNeill, University of Sheffield, UK

Editorial office: ejhg@sheffield.ac.uk

Frequency: 12 issues a year

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Article Specification	Specifications
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Complete book:

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Chapter in book:

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Abstracts from the 2020 Annual Scientific Meeting of the British and Irish Hypertension Society (BIHS). *J Hum Hypertens* 34; 2020; 1–20

Website:

Kassambara A. rstatix: pipe-friendly framework for basic statistical tests. 2020. <https://rpkgs.datanovia.com/rstatix/>.

Online Document:

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DIH conducted the meta-regression analyses and contributed to the design of the review protocol, writing the report, arbitrating potentially eligible studies, extracting and analysing data and interpreting results.

NAL contributed to data extraction and provided feedback on the report.

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Researchers should conduct their research – from research proposal to publication – in line with best practices and codes of conduct of relevant professional bodies and/or national and international regulatory bodies.

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Requirements for all categories of articles should conform to the "Uniform Requirements for Manuscripts Submitted to Biomedical Journals," developed by the ICMJE (www.icmje.org).

Each author must have contributed sufficiently to the intellectual content of the submission. The corresponding author should list all authors and their contributions to the work. Any changes to the author list after submission, such as a change in the order of the authors, or the deletion or addition of authors, must be approved by a signed letter from every author. The corresponding author must confirm that he or she has had full access to the data in the study and final responsibility for the decision to submit for publication.

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.
4. Agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Contributions by individuals who made direct contributions to the work but do not meet all of the above criteria should be noted in the Acknowledgments section of the manuscript. Medical writers and industry employees can be contributors. Their roles, affiliations, and potential conflicts of interest should be included in the author list or noted in the Acknowledgments and/or Contributors section concurrent with their contribution to the work submitted. Signed

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In the interests of transparency and to help readers form their own judgments of potential bias, authors must declare whether or not there are any competing financial interests in relation to the work described. The corresponding author is responsible for submitting a competing interests statement on behalf of all authors of the paper. This statement must be included in the cover letter and within the article after the References section listed under 'Competing Interests'.

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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.

The statement included in the submission must contain an explicit and unambiguous description of any potential competing interests, or lack thereof, for any of the authors as it relates to the subject of the report. Examples include

- **Competing interests.**
The authors declare no competing interests.

- **Competing interests.**

Dr Caron's work has been funded by the NIH. He has received compensation as a member of the scientific advisory board of Acadia Pharmaceutical and owns stock in the company. He also has consulted for Lundbeck and received compensation. Dr Rothman and Dr Jensen declare no potential conflict of interest.

Neither the precise amount received from each entity nor the aggregate income from these sources needs to be provided.

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All clinical trials must be registered in a public registry prior to submission and the trial registry number must be included in the manuscript and provided upon submission. The journal follows the trials registration policy of the ICMJE (www.icmje.org) and considers only trials that have been appropriately registered before submission, regardless of when the trial closed to enrolment. Acceptable registries must meet the following ICMJE requirements:

- be publicly available, searchable, and open to all prospective registrants
- 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](http://InternationalStandardRandomizedControlledTrialNumberRegistry)
3. the [Cochrane Renal Group Registry](http://CochraneRenalGroupRegistry)
4. the [European Clinical Trials Database](http://EuropeanClinicalTrialsDatabase)

Randomised Controlled Trials (RCTs) must adhere to the CONSORT statement, (CONsolidated Standards Of Reporting Trials) and submissions must be accompanied by a completed CONSORT checklist (uploaded as a related manuscript file). Further information can be found at www.consort-statement.org.

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](http://HUGOGeneNomenclatureCommittee) (HGNC). Approved mouse symbols are provided by [The Jackson Laboratory](http://TheJacksonLaboratory). 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

Avoid listing multiple names of genes (or proteins) separated by a slash, as in 'Oct4/Pou5f1', as this is ambiguous (it could mean a ratio, a complex, alternative names or different subunits). Use one name throughout and include the other at first mention: 'Oct4 (also known as Pou5f1)'

Sequence variants

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](http://HumanGenomeVariationSociety) (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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Compliance with HGVS nomenclature must be verified using tools such as Mutalyzer (github.com/mutalyzer/mutalyzer/wiki/Mutalyzer_explain.pdf) or VariantValidator (instructions: variantvalidator.org/batch_instructions/), freely available on the web. Errors should be corrected and the file resulting from the final check containing each variant noted in your manuscript must be included in your submission.

Submission of Data to Public Databases

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
- ancestry/forensics: mtDNA (EMPOP), Y-STRs (Y-HRD), autosomal STRs (STRIDER)

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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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Research involving human subjects, human material, or human data must have been performed in accordance with the Declaration of Helsinki and must have been approved by an appropriate ethics committee. A statement detailing this, including the name of the ethics committee and the reference number where appropriate, must appear in all manuscripts reporting such research.

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If human cell lines are used, authors are strongly encouraged to include the following information in their manuscript:

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The *European Journal of Human Genetics* requires authors of papers that are sent for external review to include in their manuscripts relevant details about

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- make inquiries of other titles believed to be affected;
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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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- If the article has already been published online, depending on the nature and severity of the infraction, either an erratum will be published alongside the article or, in severe cases, complete retraction of the article will occur. The reason for the erratum or retraction must be given.
- In either case, the author's institution or funding agency may be informed.

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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