Molecular Diagnosis & Therapy

Additional Information for Authors

PLEASE READ THIS INFORMATION IN CONJUNCTION WITH THE JOURNAL INSTRUCTIONS FOR

AUTHORS (found at https://www.springer.com/adis/journal/40291)

Title Page

Title/subtitle — if using a subtitle, please separate this from the main title with a colon. Titles and subtitles of manuscripts reporting the results of original research should describe the intervention/methodology/setting, rather than describe the study results.

Running heading – a running heading (short version of the title), of up to 100 characters, should be provided.

Abstract – for narrative reviews, abstracts should be unstructured (i.e. no headings). For original research articles, abstracts should be structured (e.g. following the guidance of the CONSORT statement for manuscripts reporting the results of randomized clinical trials, the PRISMA Statement for systematic reviews, with or without a meta-analysis, and the STROBE statement for observational studies).

Plain language summaries (PLSs) - PLSs are not mandatory. If provided, they should be up to 250 words in length and placed after the abstract of the article under the heading 'Plain Language Summary'.

Keywords – a list of keywords is not required.

Key points – two to three short bullet points should be provided summarizing the key findings and implications of the paper. These should be presented in non-technical language and not repeat verbatim text found in the abstract. They should be placed beneath the abstract under the heading of 'Key Points'.

Declarations – a section entitled 'Declarations' should be provided that contains the following subsections (if any of these sections is not applicable, state "Not applicable" under the subsection heading):

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- VI. Consent for publication if an identifiable clinical photo of a patient is published, or if an article contains patient data that could be identifiable, a statement should be included to confirm that consent of patients was obtained to publish their data.
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Acknowledgements – this section should be used to acknowledge the assistance of individuals who do not meet the criteria for authorship but who have made a substantial contribution to the manuscript/study. Acknowledgment of any medical writing support should include the nature of the support, the name of the medical writer and their employer, and the funding sources for the support. You must obtain written permission from any individual you acknowledge.

Text

Please use double-spaced text, page numbering (starting with the title page) and line numbering.

Headings – the headings of the sections/subsections should the numbered using the decimal system (e.g. 1; 2, 2.1, 2.2, 2.2.1, 2.2.2; 3; 4; etc.), starting with the Introduction and finishing with the Conclusions.

Language - the journal does not have a preference for British vs US English, but whichever style is used, it must be consistently applied throughout the manuscript.

Nomenclature

Please adhere to the HUGO Gene Nomenclature Committee (HGNC) guidelines for using human gene names and symbols, the guidelines can be accessed at http://www.genenames.org/about/guidelines

This web site also has links to the correct nomenclature to use for other species.

Please use the standard descriptions for sequence variants from HGVS variant nomenclature, guidelines are to be found at http://www.hgvs.org/mutnomen/

Please use the standard descriptions for phenotypes from HPO terminology, guidelines are to be found at http://www.human-phenotype-ontology.org

Given that human molecular diagnostics is based on sharing data on genes, variants and phenotypes, it is suggested by The Human Variome Project that authors consider submitting their data to an open access public variant database. This will help to standardize published data; hopefully it might identify and prevent errors, whilst also ensuring that valuable data is not lost.

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References

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