

## **JIMD – Journal of Inherited Metabolic Disease**

### **Aims and Scope**

The Journal of Inherited Metabolic Disease (JIMD) is the official journal of the Society for the Study of Inborn Errors of Metabolism (SSIEM). By enhancing communication between workers in the field throughout the world, the JIMD aims to improve the management and understanding of inherited metabolic disorders. It publishes results of original research and new or important observations pertaining to any aspect of inherited metabolic disease in humans and higher animals. This includes clinical (medical, dental and veterinary), biochemical, genetic (including cytogenetic, molecular and population genetic), experimental (including cell biological), methodological, theoretical, epidemiological, ethical and counselling aspects. The JIMD also reviews important new developments or controversial issues relating to metabolic disorders and publishes reviews and short reports arising from the Society's annual symposia. A distinction is made between peer-reviewed scientific material that is selected because of its significance for other professionals in the field and non-peer-reviewed material that aims to be important, controversial, interesting or entertaining (“Extras”).

The JIMD exists as two sister publications which are served by a single Editorial Team and a single manuscript submission and review process: the traditional print and online journal JIMD and JIMD Reports, which publishes scientifically sound research findings or clinical observations that warrant communication in the peer-reviewed literature but are of more limited interest to the readers. In addition to full electronic publication, and print publication as book chapters, the abstract of a JIMD Reports publication is also printed in the JIMD, to reach the widest possible readership. All other types of articles are published electronically and in print in the JIMD.

### **Instructions for submission**

#### ***Manuscript submission***

Material submitted to the JIMD (including JIMD Reports) must conform to the uniform requirements for manuscripts submitted to biomedical journals as outlined by the International Committee of Medical Journal Editors (ICMJE); see <http://www.icmje.org/icmje-recommendations.pdf> for details.

#### ***Online Submission***

All scientific contributions for publication in the JIMD (including JIMD Reports) must be submitted by the web-enabled online manuscript submission and review system. As the review process is also fully web-based, this system allows editors to keep review times as short as possible and offers authors the option to track progress of the review of their manuscripts. The online manuscript submission and review system for the JIMD offers easy and straightforward log-in and submission procedures. Please refer to: [www.editorialmanager.com/boli](http://www.editorialmanager.com/boli).

The system supports a wide range of submission file formats for manuscripts (Word, WordPerfect, RTF, TXT and LaTeX) and figures (TIFF, EPS, Microsoft® Office formats and Postscript). PDF is not an acceptable file format.

If you encounter any difficulties while submitting your manuscript online, please contact the responsible Editorial Assistant by clicking on "CONTACT US" from the tool bar.

### ***Manuscript Structure***

The first page should include:

- *Title* of the article
- *Authors' names and institutional affiliations* set out as in a current issue of the JIMD
- Name, email address and full postal address, including postal (ZIP) code, of the author who will be dealing with correspondence and proofs.
- *Word counts* for the text (excluding summary, acknowledgments, references and figure legends) and the summary.
- *Number of figures and tables.*
- Whether a *colour picture* is provided that may be used for the front cover of the issue in which the article appears.

The next page should include:

- A *summary* (= *abstract*) of not more than 250 words (Medline allows a maximum of 4096 characters and will truncate longer abstracts).
- A *concise 1 sentence take-home message* (synopsis) of the article, outlining what the reader learns from the article (this is usually printed on the inside back cover of the JIMD).

Following these pages, authors are required to provide the following, which are detailed above in the section "General Rules."

- Details of the contributions of individual authors
- The name of the corresponding author
- A competing interest statement
- Details of funding
- Details of ethics approval
- A patient consent statement
- Documentation of approval from the Institutional Committee for Care and Use of Laboratory Animals (or comparable committee)
- A list of approximately six keywords

### ***Recommendations for Manuscript Length***

Competition for publication in all scientific journals has become increasingly intense, and the JIMD is no exception. We strongly encourage prospective authors to consider brevity in their presentation, and if needed to avail themselves of the online supplementary material for those Figures and Tables that could be accommodated in that venue. In order for the Editorial Board to accommodate the broadest perspective of submissions, and to maximize the access for prospective authors to both the JIMD and JIMD Reports, the following recommendations for length have been formulated:

- Full articles: Total word count 3000, including 500 words for the Introduction and a maximum of 4 combined figures/tables.
- Reports: Total word count 2250, including 400 words for the introduction and a maximum of 3 total figures/tables. It is expected that more comprehensive reviews will exceed these

limits, but the authors of such reviews are again encouraged to work for brevity and succinctness in presentation. In all instances, literature citations should be reasonable and appropriate for the presentation, but should not exceed 30 citations for full articles and 25 citations for reports. Appropriate use of the cited literature is one way in which prospective authors can constrain the length of their submissions.

Number of authors: The number of authors needs to be limited to a maximum of 20; additional authors may be included as consortium, listed as individual contributors at the end of the manuscript and recognized in PubMed.

### ***Covering letter***

Submit a covering letter and use it to explain why your paper should be published in The Journal of Inherited Metabolic Disease.

### **General Rules**

It is a condition of acceptance that all articles have not been and will not be published elsewhere in substantially the same form. The submitting author must have circulated the article and secured final approval of the version to be peer-reviewed from all co-authors prior to article submission. This includes confirmation of

- absence of previous similar or simultaneous publications,
- their inspection of the manuscript,
- their substantial contribution to the work (all authors should have been involved in (a) conception and design, or analysis and interpretation of data, and (b) drafting the article or revising it critically for important intellectual content), and
- their agreement to submission.

It should be noted that these conditions are later confirmed in writing by the corresponding author in a copyright transfer form at the time of acceptance. Publication elsewhere, at any time, of a similar article perhaps only differing in some aspects of data, especially if the JIMD article is not cross-referenced, may justify formal retraction at a later date.

Supplementary (internet-only) material may be published for all articles; we encourage or request deposition of raw data when this appears appropriate.

The following information will be required at the time of online manuscript submission and is required on the page following the details listed in the section “Manuscript Structure” (below):

- *Details of the contributions of individual authors*, making clear who has contributed pertinent aspects of the planning, conduct, and reporting of the work described in the article.
- *Name of one author who serves as guarantor* for the article, accepts full responsibility for the work and/or the conduct of the study, had access to the data, and controlled the decision to publish.
- *A competing interest statement*, i.e., either a statement describing the interests of all authors or a declaration that they have nothing to declare, based on the “Competing Interests Questions” outlined below.
- *Details of funding* for all research studies including a statement that “The author(s) confirm(s) independence from the sponsors; the content of the article has not been influenced by the sponsors”
- *Details of ethics approval* or a statement that it was not required for all research studies

- A *patient consent statement* for all articles or other material that contain personal information about a patient; proof that informed consent was obtained must be available upon request
- If vertebrate animals have been utilized, documentation of *approval from the Institutional Committee for Care and Use of Laboratory Animals* (or comparable committee).
- A list of approximately six keywords; this is of particular importance for recognition of the manuscript after publication by some search engines.

## **Statements, permissions, and signatures**

### ***Authors and contributors***

The number of authors needs to be limited to a maximum of 20; additional authors may be included as consortium, listed as individual contributors at the end of the manuscript and recognized in PubMed.

### ***Declaration of interests and competing Interests***

Conflict of interest exists when an author (or the author's institution), reviewer or editor has financial or personal relationships that inappropriately influence (bias) his or her actions (such relationships are also known as dual commitments, competing interests or competing loyalties). These relationships vary from those with negligible potential to those with great potential to influence judgment, and not all relationships represent true conflict of interest. The potential for conflict of interest can exist whether or not an individual believes that the relationship affects his or her scientific judgment. Financial relationships (such as employment, consultancies, stock ownership, honoraria, or paid expert testimony) are the most easily identifiable conflicts of interest and the most likely to undermine the credibility of the journal, the authors and science itself. However, conflicts can occur for other reasons, such as personal relationships, academic competition and intellectual passion.

*All authors (co-authors) of articles, reports, reviews, editorials and other material submitted to the JIMD (including JIMD Reports) as well as reviewers of manuscripts must answer the following questions:*

1. Have you in the past five years accepted the following from an organisation that may in any way gain or lose financially from the results of your study or the conclusions of your review, editorial or letter:
  - Reimbursement for attending a symposium?
  - A fee for speaking or for organising education?
  - Funds for research or for a member of staff?
  - A fee for consulting?
2. Have you in the past five years been employed by an organisation that may in any way gain or lose financially from the results of your study or the conclusions of your review, editorial or letter? Do you hold any stocks or shares in such an organisation?
3. Have you acted as an expert witness on the subject of your study, review, editorial or letter?
4. Do you have any other competing financial interests?

Authors who have answered "yes" to any of these questions may have a competing interest which should be declared at the time of submission of the article (review, editorial or other

material) and which will be published in JIMD.

*Other non-financial interests that authors may like to disclose include:*

- A close relationship with, or a strong antipathy to, a person whose interests may be affected by publication of the article.
- An academic link or rivalry with someone whose interests may be affected by publication of the article.
- Membership in a political party or special interest group whose interests may be affected by publication of the article.
- A deep personal or religious conviction that may have affected what the author wrote and that readers should be aware of when reading the article.

Expert reviewers approached for assessment of submitted articles are also requested to declare conflicts of interest that may impede on their judgement of that article. This specifically includes competing research in the same area that could be negatively affected by publication of the submitted article.

*For additional information see also the ICJME's "Recommendations for the Conduct, Reporting, Editing, and Publication of Scholarly Work in Medical Journals" at <http://www.icmje.org/about-icmje/faqs/icmje-recommendations/>*

## **Ethical guidelines, human and animal rights and consents**

If the work involves the use of human subjects, the author should ensure that the work described has been carried out in accordance with [The Code of Ethics of the World Medical Association](#) (Declaration of Helsinki) for experiments involving humans ([Uniform Requirements for manuscripts submitted to Biomedical journals](#)). Authors should include a statement in the manuscript that informed consent was obtained for experimentation with human subjects. The privacy rights of human subjects must always be observed.

All animal experiments should comply with the [ARRIVE guidelines](#) and should be carried out in accordance with the U.K. Animals (Scientific Procedures) Act, 1986 and associated guidelines, [EU Directive 2010/63/EU for animal experiments](#), or the National Institutes of Health guide for the care and use of Laboratory animals (NIH Publications No. 8023, revised 1978) and the authors should clearly indicate in the manuscript that such guidelines have been followed.

## **Types of article and manuscript requirements**

### **Scientific contributions**

#### ***Full Articles***

The JIMD welcomes scientific contributions for publication as printed full articles in the following categories:

- **Original Articles:** Important manuscripts that may be expected to influence or change clinical or research practice with regard to inherited metabolic disorders. Original articles may include comprehensive studies on disease features in groups of patients, important novel information on a disease or relevant research findings. Case reports are unlikely to be accepted as original papers in print JIMD, unless they describe novel diseases or new aspects of the basic pathomechanism of a disease, supported by novel functional data. The editors may reject submitted manuscripts as original articles but invite revision

or resubmission for publication as Reports in “JIMD Reports”. All authors are invited to provide a colour picture that may be used for the front cover of the issue in which the article appears.

- **Rapid Communications:** Highly competitive and timely manuscripts; please contact the editorial office: editor@jimd.org.
- **Guidelines:** Studies providing a comprehensive, representative analysis of the clinical features, treatment and outcome of inborn errors of metabolism. For detailed information, we refer to the Editorial:  
Zschocke J, Baumgartner MR, Morava E, Patterson MC, Peters V and Rahman S (2016) Recommendations and guidelines in the JIMD: suggested procedures and avoidance of conflicts of interest. *J Inher Metab Dis*, 39:327-330
- **Reviews:** Concise summaries of metabolic pathways, specific disorders, methods, treatment options etc.
- **Metabolic Dissertations:** The JIMD invites all researchers who have completed a Ph.D. or M.D. thesis in the field of inborn errors of metabolism to submit a comprehensive review of the topic of their thesis. The article should not focus solely on the research findings but should cover all relevant information in the respective field. Such reviews preferably (but not necessarily) have a single author (other contributors should be acknowledged) and will be published with a photograph of the investigator.

### ***Images in Metabolic Medicine***

The Editors will consider clear and interesting clinical pictures or other types of images (e.g. laboratory results or observations) submitted with a descriptive paragraph of up to 250 words. Prints, slides or electronic copy are all acceptable. Authors must obtain informed consent for publication of patient-related materials. Case reports or additional information may be added as supplementary material. Images will be fully printed; title and author(s) will be listed in bibliographical databases such as Medline.

### ***Editorials***

The JIMD invites communicating editors and reviewers of articles that have been accepted for publication in the JIMD to provide an editorial that places the article in a broader context. Editorials have no abstract, may be comprised of up to 500 words and should contain no more than two (if any) references. Additional material can be added as supplementary material online. Editorials will be fully printed; title and author(s) will be listed in bibliographical databases such as Medline.

### ***Letters and Clinical/Research Observations***

The JIMD is happy to receive comments on previously published articles in the journal which should reach the editorial office within 4 weeks of publication of the original item. Correspondence may be subjected to peer review and counter-replies are usually invited from the authors of the original publication.

The concise form of a letter may also be used to report exceptionally important clinical or research observations unrelated to a previous JIMD publication that merit communication but do not fulfil the requirements for scientific articles or short reports. These items will be peer reviewed and if accepted will be published under the heading “Observation”.

Letters should have no more than five authors. They have no abstract, are limited to a maximum of 500 words and should contain no more than two (if any) references. Additional

material can be added as supplementary material online. Letters will be fully printed; title and author(s) will be listed in bibliographical databases such as Medline.

### ***JIMD Reports***

Some manuscripts present scientifically sound research findings or clinical observations that are worth communicating but are of more limited interest to the readers of the JIMD and may be sufficiently summarised in an abstract of 250 words. In order to facilitate publication of these types of manuscripts, JIMD Reports has been introduced as a sister publication of the traditional JIMD. It is an independent periodical with its own ISSN number.

All manuscripts submitted as Reports to the JIMD website will be considered for JIMD Reports rather than for the traditional journal. They will undergo the same review process as Original Articles (and in exceptional cases may be reassigned for publication in the JIMD). In addition, the Editorial Team (based on the advice of reviewers and Communicating Editors) may reject Original Articles for publication in the JIMD but offer publication in JIMD Reports. After acceptance, articles in JIMD Reports are professionally typeset in the same manner as articles in the JIMD, and full documents are available online to SSIEM members and institutional subscribers via the Springer website. Articles from JIMD Reports are submitted to PubMedCentral and are listed in PubMed as well as other abstracting and indexing services. After an embargo period of 12 months, all manuscripts published in JIMD Reports (in contrast to JIMD articles) are available free of charge world-wide, thereby ensuring widest possible readership. In addition, titles and abstracts of Reports are printed in the print-only “Extras” section of the JIMD. It is recommended to make use of the full allowance of 250 words for the abstract of Reports to convey the message of the article to the readers of the print journal.

Reports follow the same rules as Full Articles; they should not be used as a form of preliminary communication. They may take the form of **Research Reports**, with content similar to that of original articles, or **Case Reports**. Case Reports will only be considered when they highlight some unusual or previously unrecorded feature relevant to the disorder or serve as an important reminder of clinical or biochemical features of a Mendelian disorder. Chance associations of two conditions or sporadic cases from new geographical locations (as opposed to systematic epidemiological studies) are not in themselves of sufficient scientific merit to justify publication.

### **Extras in the JIMD**

The Editors of the JIMD invite submission of short items that are interesting, stimulating, important or entertaining to professionals working in the field of inborn errors of metabolism. These items will not usually be reviewed outside the editorial board and usually will not be referenced in bibliographic databases. All items of this type should be submitted by email to the editorial office (editor@jimd.org); please provide full personal details for all authors of each contribution.

### ***Garrodian***

Small texts that are used to fill gaps (e.g. at the end of original articles) have been a long and cherished tradition in some journals. They usually have the added advantage of entertaining readers and stimulating thought. The Editors are happy to receive interesting stories or personal experiences of up to a few hundred words on topics such as:

- A patient / paper / experience that changed my practice

- A memorable patient / experience
- An error that proved educational or informative for lab operation or clinical care
- How I embarked on this career path and lessons learned along the way
- Any other story conveying instruction, pathos or humour

If the Garrodian refers to an identifiable person, written consent for publication from that person or an appropriate relative is required.

### ***Book Reviews***

Instructive reviews of up to 400 words are invited on new books published in the field of inborn errors of metabolism, or closely affiliated areas.

### ***Obituaries***

The Editors of the JIMD strongly encourage submission of obituary notices for all recently deceased SSIEM members or other persons in the field of inborn errors of metabolism. Obituary notices should be emailed to the editorial office. Please give your name and contact details, including a phone number and email address. Obituaries will be considered by the editorial board and may be shortened; they will be published (without proofs) with the name of the person(s) who submitted the notice.

Please provide:

1. The full **name** of the deceased
2. A photograph
3. A summary of **important data**:
  - a. *(Last) professional position and title, place of work*
  - b. *Date and place of birth*
  - c. *Primary degree with university and year when obtained*
  - d. *Additional professional qualifications with university and year when obtained*
  - e. *Date of death, cause of death*
4. The **main text** summarising important contributions and personal characteristics of the deceased. The last sentence should state the remaining relatives such as spouse and/or the number of children and grandchildren.

### **Formatting guidelines**

#### ***Language***

Please write your text in good English (American or British usage is accepted, but not a mixture of these).

#### ***Text formatting***

Standard text formatting is recommended in word, with the preferential use of Times New Roman, 12 font letters and double spaced text documents. The submission process automatically converts text files to pdf.

#### ***Units, Symbols, and Database References***

At the time of first mention, diseases, enzymes or genes should be referenced to the appropriate classification, nomenclature or database:

- Inherited diseases to the OMIM catalogue number (<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>)
- Enzymes to an Enzyme Commission (EC) number (<http://www.chem.qmul.ac.uk/iubmb/enzyme/>)

- Genes to the HUGO-approved gene symbol (<http://www.gene.ucl.ac.uk/nomenclature/>)

Authors should use SI units throughout the manuscript. Biochemical nomenclature should follow IUPAC-IUB recommendations (<http://www.chem.qmul.ac.uk/iupac/jcbtn/>). Nomenclature of mutations or genetic variants should follow HGVS recommendations (<http://www.hgvs.org/mutnomen/>). At the time of first mention, genetic variants should be described with both protein designation and DNA designation (based preferably on cDNA reference numbers).

Previously published material should be acknowledged, and written permission from copyright holders must be obtained to reproduce figures, tables or substantial sections of text. Where a paper relies on material that is under consideration by or in press in another journal, a copy of this must be provided for the referees.

When writing the articles, please keep in mind the broad readership of the JIMD. For example, for methods that are widely reported or published it may be worthwhile to provide a brief two to three sentence description of the protocol to provide the reader with some insight into the methods used.

### ***References***

Consult a current issue of the journal. Citations in the text should use authors' names then the date, e.g. (Smith and Smith 1977); for 3 or more authors use et al, e.g. (Jones et al 1989).

The full references are listed in alphabetical order at the end of the paper. Authors are listed without 'and'. Give the first 3 authors plus et al when there are 7 or more authors. Both in the text and list use 'et al' without punctuation or italicization. Journal abbreviations follow Index Medicus or Chemical Abstracts. Examples are:

#### *Journals:*

Smith AL, Smith JD (1977) Hybridisation methods. *Nucl Acids Res* 8: 1095–1098.

#### *Chapter in an edited book:*

Weinstein L, Swartz MN (1974) Pathologic mechanisms of invading microorganisms. In Sodeman WA Jr, Sodeman WA, eds. *Pathogenic Physiology: Mechanisms of Disease*. Philadelphia: WB Saunders, 457–472.

To cite a web site in the text (but not a specific document), it is sufficient to give the address/URL (e.g., <http://www.ssiem.org>) without an entry in the reference list. However, when citing a specific web document or information, a standard citation in the text (e.g. Gaten 2000) and an entry in the reference list is required. Internet references should include the same information that would be provided for a printed source (or as much information as possible). The Web information is then placed at the end of the reference. It is important to use "Retrieved from" and the date because documents on the Web may change in content, move or be removed from a site altogether.

Reference to personal communications requires the explicit approval of the person quoted; written confirmation must be provided. Authors — not journal editors or copy editors — are responsible for the accuracy of all references, which includes verifying the source of email communications, before citing them as personal communications in manuscripts.

### ***Tables***

Please submit tables as editable text and not as images. Tables should be placed on separate page(s) at the end. Number tables consecutively in accordance with their appearance in the text and place any table notes below the table body. Be sparing in the use of tables and ensure that the data presented in them do not duplicate results described elsewhere in the article. Please avoid using vertical rules.

### ***Figures***

Please make sure that artwork files are in an acceptable format (TIFF, JPEG or PDF), and with the correct resolution (>300 dpi). If, together with your accepted article, you submit usable color figures, it will be ensured, at no additional charge, that these figures will appear in color online regardless of whether or not these illustrations are reproduced in color in the printed version. For color reproduction in print, you will receive information regarding the costs after receipt of your accepted article. Please indicate your preference for color: in print or online only. Figures should be submitted as separate files.

Ensure that each illustration has a caption. A caption should comprise a brief title (not on the figure itself) and a description of the illustration. Keep text in the illustrations themselves to a minimum but explain all symbols and abbreviations used. Figure legends should be included in the submitted manuscript at the end of the manuscript on a separate page.

### ***Online material***

Supplementary files will only appear as online material.

Supplementary material can support and enhance your scientific research. Supplementary files offer the author additional possibilities to publish supporting applications, high-resolution images, background datasets, sound clips and more. Please note that such items are published online exactly as they are submitted; there is no typesetting involved (supplementary data supplied as an Excel file or as a PowerPoint slide will appear as such online). Please submit the material together with the article and supply a concise and descriptive caption for each file.

### ***Research Materials***

It is assumed that authors whose research is published by the JIMD will make antibodies, cloned DNA sequences, and similar materials available to other investigators in non-commercial institutions, so as to permit replication of the reported work.

### **After Acceptance of a Manuscript**

#### ***Proofs***

Proofs will be sent to the corresponding author by email. Responses, with or without corrections, should be sent within 72 hours. Please do not correct or edit the PDF file. Extensive corrections must be clearly marked on a printout of the PDF file and should be sent by first-class mail (airmail overseas). Minor corrections (+/- 10) may be sent via email attachment to [proofscorrection@springer.com](mailto:proofscorrection@springer.com). Always quote the four-letter journal code (BOLI) and article number from your proof in the subject field of your email.

#### ***Page charges***

No page charges are levied on authors or their institutions except for colour pages. The corresponding author will be contacted regarding costs and invoicing if the printed manuscript includes colour figures. Colour page charges may be waived at the discretion of the editors.

### ***Copyright transfer***

Authors will be asked to transfer copyright of the article to the Publisher. This will ensure the widest possible dissemination of information under copyright laws.

### ***Open Access***

In addition to the normal publication process (whereby an article is submitted to the journal and access to that article is granted to customers who have purchased a subscription), Springer now provides an alternative publishing option: Springer Open Choice. A Springer Open Choice article receives all the benefits of a regular subscription-based article, but in addition is made available publicly through Springer's online platform SpringerLink. To publish via Springer Open Choice, upon acceptance please click on the link sent to you by email to complete the relevant order form and provide the required payment information. Payment must be received in full before publication or articles will be published as regular subscription-model articles. We regret that Springer Open Choice cannot be ordered for published articles. See also: [www.springeronline.com/openchoice](http://www.springeronline.com/openchoice).

### **Additional Information**

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