

Journal of Pediatric Genetics

Author Instructions

Thank you for contributing to *Journal of Pediatric Genetics*. Please read the instructions carefully and observe all the directions given. Failure to do so may result in unnecessary delays in publishing your article.

SUBMISSION CHECKLIST

All manuscripts must be submitted at the following link:

<http://www.editorialmanager.com/jpg>

- ☐ **AUTHOR INFORMATION**
 - All authors: full name, degrees, department, affiliation, e-mail address
 - Corresponding author: mailing address, telephone number
- ☐ **MANUSCRIPT FILE**
 - Must be digital - hard copy submissions are not accepted
- ☐ **ABSTRACT AND KEYWORDS**
 - See the section Article Types for word limits
- ☐ **REFERENCES**
 - Cited sequentially in AMA style
- ☐ **FIGURES AND TABLES**
 - Cited sequentially and included in the main document
- ☐ **ART FILES**
 - Must be saved separately from the main document
- ☐ **PERMISSIONS**
 - Required if you plan to reproduce content from a published source or include a photograph of a patient
 - Patient permission forms available at www.thieme.com/journal-authors

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MANUSCRIPT FORMAT

Article Types

The following graph shows what types of articles are accepted for publication, and what requirement they may have.

Article Type	Abstract Limit	Keywords Limit	Text Words (excluding refs) Limit	Tables/Figures Limit	References Limit
Original Article	Up to 250 words	3 to 6 keywords	Up to 4,000 words	Up to 5 tables/figures	Up to 40 references
Review Article	Up to 250 words	3 to 6 keywords	No limit	No limit	No limit
Case Based Review	Up to 250 words	3 to 6 keywords	Up to 2,500 words	Up to 5 tables/figures	Up to 50 references
Editorial	n/a	n/a	Up to 600 words	n/a	Up to 5 references
Book Review	n/a	n/a	Up to 600 words	n/a	n/a
Annual Meeting Abstract	n/a	n/a	No limit	No limit	No limit
Letters*	n/a	n/a	Up to 400 words	n/a	Up to 5 references

*Letters to the editor typically offer comments on material published in the Journal of Pediatric Genetics during the past few months. Letters should not exceed 400 words (inclusive of references). A maximum of 5 references are allowed, one of which should refer to the earlier work being discussed. Responses to published letters must meet similar requirements.

General Guidelines

- You must submit a digital copy of your manuscript. Hard copy submissions are not accepted.
- Keep the format of your manuscript simple and clear. We will set your manuscript according to our style—do not try to “design” the document.
- The manuscript, including the title page, abstract and keywords, text, references, figure captions, and tables should be typewritten, double-spaced in 12-point font with 1-inch margins all around and saved as one file.
- Each figure should be saved as its own separate file. Do not embed figures within the manuscript file. This requires special handling by Thieme’s Production Department.
- Keep abbreviations to a minimum and be sure to explain all of them the first time they are used in the text.
- The manuscripts should be written in American English.
- The authors should use Système International (SI) measurements. For clarity, nonmetric equivalents may be included in parentheses following the SI measurements.
- Use generic names for drugs. You may cite proprietary names in parentheses along with the name and location of the manufacturer.
- Credit suppliers and manufacturers of equipment, drugs, and other brand-name material mentioned in the manuscript within parentheses, giving the company name and primary location.

MANUSCRIPT FORMAT *continued*

Title Page

- This journal adheres to a single-blinded peer-review policy. The title page should be included in the main document.
- The title page should list the article title and the corresponding author's full name, degree, title, department, affiliation, mailing address, e-mail address, and telephone and fax numbers. It should also list the full name, degree, title, department, and affiliation of every co-author.

Abstract and Keywords

See the section Article Types for word limits.

The abstract should briefly outline the content of the article and any conclusions it may reach. The keywords should be words a reader would be likely to use in searching for the content of the article.

Main Document

- Please clearly distinguish the hierarchy of headings within the manuscript by using capital letters, underline, italic, and bold styles as necessary.
- As needed, use italic, superscripts, subscripts, and boldface, but otherwise do not use multiple fonts and font sizes.
- Do not insert page or section breaks except where noted in the Author Instructions.
- Use hard returns (the Enter key) only at the end of a paragraph, not at the end of a line. Allow lines of text to break automatically in your word-processing software. Do not justify your text.
- Use only one space, not two, after periods.
- Create tables using the Table function in Microsoft Word.

Thieme Editing Services

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Acknowledgments

The source of any financial support received and recognition of personal assistance for the work being published should be indicated at the end of the article, just before the Reference section, under the heading Acknowledgments.

Conflict of Interest

All authors (including corresponding and co-authors associated with the manuscript) must make a formal statement at the time of submission indicating any potential conflict of interest that might constitute an embarrassment to any of the authors if it were not to be declared and were to emerge after publication. Such conflicts might include, but are not limited to, shareholding in or receipt of a grant or consultancy fee from a company whose product features in the submitted manuscript or which manufactures a competing product. Types of conflicts include: Consulting, Royalties, Research Support, Institutional Support, Ownership, Stock/Options, Speakers Bureau, and Fellowship Support. Any commercial entity whose products are described, reviewed, evaluated, or compared in the manuscript, except for those disclosed in the Acknowledgments section, are potential conflicts.

This journal follows the guidelines of the [International Committee of Medical Journal Editors](#) and an [ICMJE disclosure of potential conflicts of interest \(COI\) form](#) must be submitted for each author at the time of manuscript submission. Forms must be submitted even if there is no conflict of interest. It is the responsibility of the corresponding author to ensure that all authors adhere to this policy prior to submission.

A conflict of interest statement must also be included in the manuscript after any "Acknowledgements" and "Funding" sections and should summarize all aspects of any conflicts of interest included on the ICMJE form. If there is no conflict of interest, authors must include 'Conflict of Interest: none declared'.

Please click <http://www.icmje.org/conflicts-of-interest> to download a Conflict of Interest form. The disclosure information is important in article processing. If the provided forms are incomplete or missing, it can cause delays in publishing of article.

Please note: The ICMJE Conflict of Interest forms can only be opened & edited with [Adobe Reader](#).

References

References should be the most recent and pertinent literature available. It is essential that they are complete and thoroughly checked. If the reference information is incomplete, good online sites to search for full details are the National Library of Medicine: www.nlm.nih.gov; Books in Print: www.booksinprint.com; PubMed: www.ncbi.nlm.nih.gov/PubMed/; or individual publisher Web sites.

- References must be listed in AMA style, using Index Medicus journal title abbreviations.
 - References follow the article text. Insert a page break between the end of text and the start of references.
 - References must be cited sequentially (NOT alphabetically) in the text using superscript numbers.
 - By way of exception to AMA style, do not italicize book titles or journal title abbreviations and do not put a period at the end of a reference.
 - List all author names, up to and including six names. For more than six authors, list the first three followed by et al.
 - References should be styled per the following examples:
1. Citing a journal article:
Newburger JW, Takahashi M, Burns JC, et al. The treatment of Kawasaki syndrome with intravenous gamma-globulin. *N Engl J Med* 1986;315:341–347
 2. Citing a chapter in a book:
Toma H. Takayasu's arteritis. In: Novick A, Scoble J, Hamilton G, eds. *Renal Vascular Disease*. Philadelphia: WB Saunders; 1995:47–62
 3. Citing a book:
Stryer L. *Biochemistry*. 2nd ed. San Francisco: WH Freeman; 1981:559–596
 4. Citing a thesis:
Stern I. Hemorrhagic Complications of Anticoagulant Therapy [Ph.D. dissertation]. Evanston, IL: Northwestern University; 1994
 5. Citing a government publication:
Food and Drug Administration. Jin Bu Huan Herbal Tablets. Rockville, MD: National Press Office; April 15, 1994. Talk Paper T94-22
 6. Citing an online article:
Rosenthal S, Chen R, Hadler S. The safety of acellular pertussis vaccine vs whole-cell pertussis vaccine [abstract]. *Arch Pediatr Adolesc Med* [serial online]. 1996;150:457–460. Available at: http://www.ama-assn.org/sci-pubs/journals/archive/ajdc/vol_150/no_5/abstract/htm. Accessed November 10, 1996
 7. Citing a symposium article:
Eisenberg J. Market forces and physician workforce reform: why they may not work. Paper presented at: Annual Meeting of the Association of American Medical Colleges; October 28, 1995; Washington, DC

MANUSCRIPT FORMAT *continued*

Figure Captions

- Figures include photographs or radiographs, drawings, graphs, bar charts, flow charts, and pathways, but NOT lists or tables.
- Figures must be cited sequentially in the text. Number all figures (and corresponding figure captions) sequentially in the order they are cited in the text.
- Figure captions should be written after the reference list. Insert a page break between the end of references and the start of figure captions.
- Figure captions should include a description of the figure and/or each lettered part (A, B, etc.) and of any portions of the figure highlighted by arrows, arrowheads, asterisks, etc.
- For a figure borrowed or adapted from another publication (used with permission), add a credit line in parentheses at the end of each figure legend. This credit line should be a complete bibliographic listing of the source publication (as a reference), or other credit line as supplied by the copyright holder. For example (Reprinted with permission from Calfee DR, Wispelwey B. Brain abscess. *Semin Neurol* 2000;20:357.)

Tables

- Data given in tables should be commented on but not repeated in the text. Be sure that lists or columns of related data are composed in a word-processing program like the rest of the text.
- Do not intersperse tables in the text. Tables should appear after the figure captions. Insert a page break between the end of the figure captions and the start of the tables.
- Tables must be double-spaced and numbered in the same sequence they are cited in the text. A short descriptive title should be provided for each table.
- If a table contains artwork, supply the artwork separately as a digital file.
- For tables borrowed or adapted from another publication (used with permission), add a credit line as the first footnote beneath each table. This credit line should be a complete bibliographical listing of the source publication (as a reference), or other credit line as supplied by the copyright holder. For example, "Reprinted with permission from Calfee DR, Wispelwey B. Brain abscess. *Semin Neurol* 2000;20:357." ("Data from . . ." or "Adapted from . . ." may also be used, as appropriate.)
- Other footnotes for tables should be indicated in the table using superscript letters in alphabetical order.
- Any abbreviations used in the table should be explained at the end of the table in a footnote.

DIGITAL ARTWORK PREPARATION

General Guidelines

- It is best to use Adobe Photoshop to create and save images, and Adobe Illustrator for line art and labels.
- Do not submit art created in Microsoft Excel, Word, or PowerPoint. These files cannot be used by the typesetter.
- Save each figure in a separate file.
- Do not compress files.
- All black-and-white and color artwork should be at a resolution of 300 dpi (dots per inch) in TIFF format. Line art should be 1,200 dpi in EPS or TIFF format. Contact the Production Editor at Thieme if you are unsure of the final size.
- It is preferable for figures to be cropped to their final size (approximately 3½ inches for a single column and up to 7 inches for a double column), or larger, and in the correct orientation. If art is submitted smaller and then has to be enlarged, its resolution (dpi) and clarity will decrease.

Note: Lower resolutions (less than 300 dpi) and JPEG format (.jpg extension) for grayscale and color artwork are strongly discouraged due to the poor quality they yield in printing, which requires 300 dpi resolution for sharp, clear, detailed images. JPEG format, by definition, is a lower resolution (compressed) format designed for quick upload on computer screens.

Black-and-White Art

- Black-and-white artwork can be halftone (or grayscale) photographs, radiographs, drawings, line art, graphs, and flowcharts. Thieme will only accept digital artwork.
- If possible, do not send color art for conversion to black-and-white. Do the conversion yourself so that you can check the results and confirm in advance that no critical details are lost or obscured by the change to black-and-white.
- For best results, line art should be black on a white background. Lines and type should be clean and evenly dark. Avoid screens or cross-hatching, as they can darken or be uneven in printing and lead to unacceptable printing quality.

Color Art

- Color illustrations are expensive to produce and usually cannot be accepted unless the author is willing to cover the additional production costs incurred. Please check with the Editor in Chief or Thieme for details. We will convert color illustrations to black-and-white unless we receive a letter from the author assuming responsibility for the cost of printing color. Upon request, we will provide you with a cost estimate for the color printing.
- All color artwork should be saved in CMYK, not RGB.

Art Labels

- Arrows, asterisks, and arrowheads (or other markers) should be white in dark or black areas and black in light or white areas, and large in size. If not, these highlighting marks may become difficult to see when figures are reduced in size during the typesetting process.
- Use 1-point (or thicker) rules and leader lines.
- Capitalize the first word of each label and all proper nouns. Consider using all capitals if you need a higher level of labels.
- Where there are alternate terms or spellings for a named structure, use the most common one and make sure it is consistent with what is used in the text.
- Avoid using multiple fonts and font sizes for the labels; use only one or two sizes of a serif font.

SUBMISSION PROCEDURE

Article Processing Charge

Authors of articles for all Thieme subscription journals – including, of course, the *Journal of Pediatric Genetics* – have the option of paying an article processing charge (APC) so that their articles will be published on an Open Access basis. Learn more about Thieme's Open Access program by visiting [Open Access - Journal authors - Thieme Group](#). For the current pricing, please go to “APC” and select “Price List” (“Hybrid Open Access” price applies to this journal).

Submission Procedure

- Consult the checklist on the first page of this document to ensure that you are ready to submit your manuscript.
- Please note: **There are no submission charges to submit your manuscript to this journal.**
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<http://www.editorialmanager.com/jpg>
- Always review your manuscript before submitting it. You may stop a submission at any phase and save it to submit later. After submission, you will receive a confirmation email. You can also check the status of your manuscript by logging in to the submission system. The Editor in Chief will inform you via email once a decision has been made.

Preprint Server Statement

Journal of Pediatric Genetics encourages the submission of manuscripts that have been deposited in an initial draft version in preprint repositories such as Research Square, arXiv, and medRxiv. Drafts of short conference abstracts or degree theses posted on the website of the degree-granting institution, and draft manuscripts deposited on authors' or institutional websites are also welcome. All other prior publication is forbidden.

During submission, authors should (1) note use of the preprint repository in the cover letter, (2) state what adjustments and/or updates the draft has undergone between deposition and submission and (3) cite the preprint, including the DOI, as a reference in the manuscript.

After submission to the journal, and until a final decision has been made, authors are discouraged from depositing versions of their manuscript as preprints. Upon publication authors should add a link from the preprint to the published article. Twelve months after publication, authors can update the preprint with the accepted manuscript.

Revision Procedure

- Should the editors decide that your article requires a revision, you will need to make the changes via a word-processing program and resubmit it electronically.
- Log In to the submission system and find your article, which will be marked for revision.
- The best way to make revisions to your manuscript is by enabling the Track Changes mode in Microsoft Word, which will automatically highlight and mark up revised text. Please submit both a marked up copy and a clean copy of your revised manuscript to the submission system.
- Your original files will still be available after you upload your revised manuscript, so you should delete any redundant files before completing the submission.
- You will also be provided space in which to respond to the reviewers' and editors' comments. Please be as specific as possible in your response.

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Page proofs will be sent to you via email. The proofs will be in a PDF file format, which should be opened using Acrobat Reader software. You will receive further instructions with your proofs. Take this opportunity to check the typeset text for typographic and related errors. Elective alterations are difficult to accommodate owing to the associated time and expense of introducing them. Therefore, please be sure that when you submit your manuscript, it is accurate, complete, and final.

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Authorship credit should be based on criteria established by the [International Committee of Medical Journal Editors](#). Each author should have made the following contributions towards the completion of the manuscript:

1. Substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data
2. Drafting the article or revising it critically for important intellectual content
3. Final approval of the version to be published

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You must obtain a signed patient permission form for every patient whose recognizable photograph will be used. If you do not supply this, the identity of the patient must be obscured before the image is published; this could interfere with the instructive value of the photograph.

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Further Reading

Further general Thieme journal policies can be found [here](#). These include:

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 - Peer Review Model
 - Recommending Reviewers
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 - Integrity of the Scientific Record
 - Thieme's Archival Strategy
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 - Research Involving Human Subjects
 - Clinical Trial Registration
 - Reporting Guidelines
 - Research Involving Animal Studies
 - Research Involving Cell Lines
 - Research Involving Biological Samples and Specimen
 - Safety
 - Dual-Use Research of Concern
 - Competing Interests
 - Funding
- [Data and Supporting Evidence](#)
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 - Data Availability Policy Levels
 - Data Availability Statements
 - Data Repository
 - Data Mining
- [Misconduct](#)
 - Our Approach to Reported Misconduct
 - Our Malpractice Policies
 - Research Misconduct
 - Publication Misconduct: Data and Image Falsification and Fabrication

- Publication Misconduct: Paper Mills
 - Publication Misconduct: Plagiarism
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