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

CONTENTS

MANUSCRIPT FORMAT ---------- **3-7**

Article Types ---------- 3

General Guidelines ---------- 3

Title Page ---------- 4

Abstract and Keywords ---------- 4

Main Document ---------- 4

Acknowledgments ---------- 4

Conflict of Interest ---------- 4

References ---------- 6

Figure Captions ---------- 7

Tables ---------- 7

DIGITAL ARTWORK PREPARATION ---------- **8**

General Guidelines ---------- 8

Black and White Art ---------- 8

Color Art ---------- 8

Art Labels ---------- 8

SUBMISSION PROCEDURE ---------- **9**

Submission Procedure ---------- 9

Revision Procedure ---------- 9

PRODUCTION PROCEDURE ---------- **9**

Page Proofs ---------- 9

Article Offprints ---------- 9

POLICY STATEMENTS ---------- **10**

Statement on Liability ---------- 10

Definition of Authorship ---------- 10

Copyright Statement ---------- 10

Statement of Ethics ---------- 10

Patient Permission Policy ---------- 10

EDITORIAL CONTACTS ---------- **12**

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

Thieme offers a language editing service for manuscripts, abstracts and theses in partnership with Enago, a world-leading provider of author services to researchers around the world. Authors can choose from a range of editing services and get their manuscripts edited by Enago's professional medical editors. Authors that wish to use this service will receive a 20% discount on all editing services. To find out more information or get a quote, please visit https://www.enago.com/thieme

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 acelluler 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

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

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.**
- Manuscripts must be submitted electronically at the following link: 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.

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.

PRODUCTION PROCEDURE

Page Proofs

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.

Article Offprints

You will be able to order offprints of your article in advance of its publication. Details and prices will be sent to you along with the page proofs. Upon publication, the corresponding author will receive a complimentary PDF of their article.

POLICY STATEMENTS

Statement on Liability

The legislation on product liability makes increased demands on the duty of care to be exercised by authors of scientific research and medical publications. This applies in particular to papers and publications containing therapeutic directions or instructions and doses or dosage schedules. We therefore request you to examine with particular care, also in your own interest, the factual correctness of the contents of your manuscript once it has been copyedited and returned to you in the form of galley proofs. The responsibility for the correctness of data and statements made in the manuscript rests entirely with the author.

Definition of Authorship

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

Copyright Statement

Submitted manuscripts must represent original research not previously published nor being considered for publication elsewhere. The editors and Thieme combat plagiarism, double publication, and scientific misconduct with the software CrossCheck powered by iThenticate. Your manuscript may be subject to an investigation and retraction if plagiarism is suspected.

If you plan to reproduce text, tables, or figures from a published source, you must first obtain written permission from the copyright holder (usually the publisher). This is required even if the material is from your own published work. For material never before published and given to you by another person, you must obtain permission from that person. Serious delays to publication can be incurred if permissions are not obtained.

As the author, it is your responsibility to obtain all permissions, pay any permission fees, furnish copies of permissions to Thieme with your manuscript, and include a credit line at the end of the figure caption, beneath the table, or in a text footnote.

Upon publication of an article, all rights are held by the publishers, including the rights to reproduce all or part of any publication. The reproduction of articles or illustrations without prior consent from the publisher is prohibited.

Statement of Ethics

This journal adheres to the ethical standards described by the Committee on Publication Ethics and the International Committee of Medical Journal Editors. Authors are expected to adhere to these standards.

For all manuscripts reporting data from studies involving human or animal participants, formal review and approval, or formal review and waiver (exemption), by an appropriate institutional review board (IRB) or ethics committee is required, as well as any necessary HIPAA consent, and should be described in the Methods section with the full name of the reviewing entity. All clinical trials must be registered in a public trials registry. Denote the registry and registry number.

Patient Permission Policy and Thieme GDPR Policy

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.

The personal rights of people who are recognizable on images must be protected. Please provide a written consent form for publication signed by every recognizable person. For persons under 18 years of age / persons supervised, please provide the signature of both parents / the legal guardian / supervisor. A suitable declaration of consent form can be obtained in our authors' lounge. Patient permission forms are available at www.thieme.com/journal-authors

We cannot accept or store illustrations in which personal data of third parties are included. Please submit images in completely anonymous form, free of personal data only! Such data may not only be directly visible in the image (e.g., a patient name or a date of birth in an X-ray image); they can also be included in the metadata of the image, which is accessible with the appropriate software. They may also be obscured by a cropping feature (such as PowerPoint or Word), but can be made visible underneath. If you have questions about data protection regulations, please contact us before submitting your manuscript.

EDITORIAL CONTACTS

Please contact the Editors or Thieme Publishers with any questions.

Editors in Chief
Pankaj Prasun, MD
Department of Genetics and Genomic Sciences,
Icahn School of Medicine at Mount Sinai,
One Gustave L. Levy Place - Box 1497
New York, NY 10029
Tel: 212 241 6947/Fax: 212-860-3316
pankaj.prasun@mssm.edu

Huseyin Caksen, MD, PhD
Necmettin Erbakan University, Meram Medical Faculty
Department of Pediatrics,
Divisions of Pediatric Neurology and Genetics
42080 Meram, Konya, Turkey
Tel: +90 332 223 75 46 / Fax: +90 332 223 61 81
huseyincaksen@hotmail.com

Thieme Publishers
journals@thieme.com

Thieme Publishers – Production Editor
Sudeshna Roy
Thieme Medical and Scientific Publishers Private Limited
A-12, Second Floor, Sector -2,
NOIDA -201301
Uttar Pradesh
India
Email: Sudeshna.Roy@thieme.in
Tel: +91 120 4556600/ Fax: +91 120 4556649