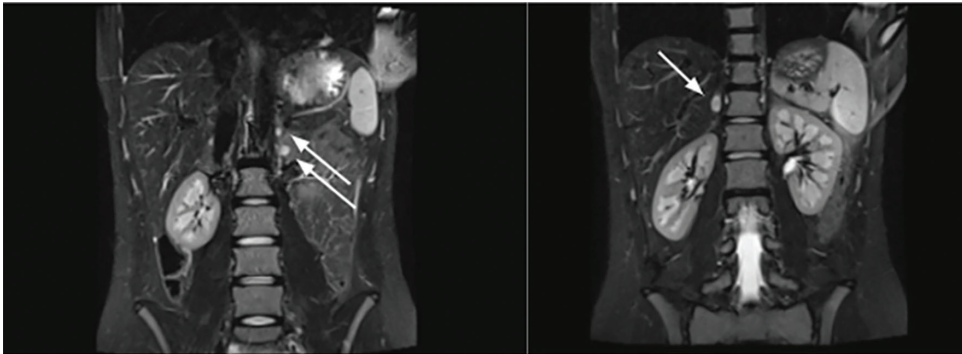


JCRPE

Journal of Clinical
Research in Pediatric
Endocrinology



Hereditary Pheochromocytoma as a Major Manifestation of von Hippel Lindau Disease (vHL) in Childhood: Long-term Follow-up of Five Patients with vHL from One Family

Pasternak-Pietrzak K et al.

Page: 12-19

June **2026**

Volume **18**

Issue **Suppl 1**



Official Journal of
Turkish Society for Pediatric
Endocrinology and Diabetes

Editor in Chief

Abdullah Bereket

Marmara University Faculty of Medicine, Department of Pediatric
Endocrinology, Istanbul, Türkiye
abdullahbereket@gmail.com

Associate Editors

Aneta Gawlik-Starzyk

Medical University of Silesia Faculty of Medical Sciences in Katowice,
Katowice, Poland
E-mail: agawlik@mp.pl

Damla Gökşen

Ege University Faculty of Medicine, Department of Pediatric Endocrinology,
Izmir, Türkiye
E-mail: damla.goksen@ege.edu.tr

Jan Idkowiak

Institute of Metabolism and Systems Research, College of Medical and Dental
Sciences, University of Birmingham, United Kingdom
E-mail: j.idkowiak@bham.ac.uk

Korcan Demir

Dokuz Eylül University Faculty of Medicine, Department of Pediatric
Endocrinology, Izmir, Türkiye
E-mail: korcandemir@gmail.com

Özlem Akgün-Doğan

Acıbadem University Faculty of Medicine, Departments of Pediatric Genetics
and Medical Genetics, Istanbul, Türkiye
E-mail: ozlemakgundogan@gmail.com

Samim Özen

Ege University Faculty of Medicine, Department of Pediatric Endocrinology,
Izmir, Türkiye
E-mail: samim.ozen@ege.edu.tr

Serap Demircioğlu Turan

Marmara University Faculty of Medicine, Department of Pediatric
Endocrinology, Istanbul, Türkiye
E-mail: serap.turan@marmara.edu.tr

Z. Alev Özön

Hacettepe University Faculty of Medicine, Department of Pediatrics, Division
of Pediatric Endocrinology, Ankara, Türkiye
E-mail: ozonalev@gmail.com

Honorary Editor

Feyza Darendeliler

Emeritus, Istanbul University Istanbul Faculty of Medicine, Department of
Pediatric Endocrinology, Istanbul, Türkiye
E-mail: feyzad@istanbul.edu.tr

Editorial Board

Ali Kemal Topaloğlu

University of Mississippi Medical Center, Department
of Pediatrics, Division of Pediatric Endocrinology,
Jackson, Mississippi, USA

Aysun Bideci

Gazi University Faculty of Medicine, Department of
Pediatric Endocrinology, Ankara, Türkiye

Ayça Aykut

Ege University Faculty of Medicine, Department of
Medical Genetics, Izmir, Türkiye

Banerjee Indi

Manchester University NHS Foundation Trust,
Manchester, United Kingdom

Belma Hailoğlu

Marmara University Faculty of Medicine, Department
of Pediatric Endocrinology, Istanbul, Türkiye

Gül Yeşiltepe Mutlu

Koç University Faculty of Medicine, Department of
Pediatric Endocrinology, Istanbul, Türkiye

Hakan Döneray

Atatürk University Faculty of Medicine, Department of
Pediatric Endocrinology, Erzurum, Türkiye

Hüseyin Onay

Ege University Faculty of Medicine, Department of
Medical Genetics, Izmir, Türkiye

Hussain Alsaffar

Sultan Qaboos University, Department of Child
Health, Oman

Justin Davies

University Hospital Southampton NHS Foundation
Trust, Southampton Children's Hospital, Paediatric
Endocrinology, Southampton; University of Southampton,
Faculty of Medicine, Hampshire, England

Khalid Hussain

Sidra Medicine, Department of Pediatric
Endocrinology, Doha, Qatar

Murat Bastepe

Massachusetts General Hospital, Harvard Medical
School Endocrine Unit, Boston, USA

Margaret C S Boguszewski, MD, PhD

Federal University of Paraná, Department of
Pediatrics, Curitiba, Brazil

Merih Berberoğlu

Emeritus Professor of Pediatrics, Ankara University
Faculty of Medicine, Department of Pediatric
Endocrinology, Ankara, Türkiye

Neslihan Güngör

Louisiana State University Health Sciences Center-
Shreveport, Department of Pediatric Endocrinology,
Louisiana, USA

Nurgün Kandemir

Emeritus Professor of Pediatrics, Hacettepe University
Faculty of Medicine, Department of Pediatric
Endocrinology, Ankara, Türkiye

Ömer Tarım

Bursa Uludağ University Faculty of Medicine,
Department of Pediatric Endocrinology, Bursa, Türkiye

Rasha Tarif

Ain Shams University Faculty of Medicine, Department
of Pediatrics, Cairo, Egypt

Violeta Iotova

Endo-ERN Work Package 'Education & Training'
Paediatric Chair, Department of Pediatrics, Medical
University of Varna, Varna, Bulgaria

Zehra Yavaş Abalı

Marmara University Faculty of Medicine, Department
of Pediatric Endocrinology, Istanbul, Türkiye

Zdeněk Šumník

Charles University and University Hospital Motol,
Department of Pediatrics, 2nd Faculty of Medicine,
Prag, Czech Republic

Güven Özkaya (Statistical Consultant)

Bursa Uludağ University Faculty of Medicine,
Department of Biostatistics, Bursa, Türkiye

English Language Editor and Assistant to the Editor in Chief

Jeremy Jones, *Kocaeli, Türkiye*

☞ The paper used to print this journal conforms to ISO 9706: 1994 standard (Requirements for Permanence).

The National Library of Medicine suggests that biomedical publications be printed on acid-free paper (alkaline paper).

Reviewing the articles' conformity to the publishing standards of the journal, typesetting, reviewing and editing the manuscripts and abstracts in English, creating links to source data, and publishing process are realized by Galenos.

ABOUT THE JOURNAL

The Journal of Clinical Research in Pediatric Endocrinology (JCRPE) is a peer-reviewed international journal that publishes high quality papers and reviews, which focus on clinical and translational research in Pediatric Endocrinology and Diabetes. JCRPE is among the leading journals in this field, committed to the publication of articles containing new clinical research that will have an immediate impact on clinical pediatric practice. In line with the journal's mission to disseminate knowledge and improve the care of children with endocrine disorders, articles in both current and previous issues are provided as free on-line open access to all readers globally, which also increases the visibility of the papers published in JCRPE.

JCRPE publishes original research articles, reviews, short communications, letters, case reports with novel insights, and other special features related to the field of pediatric endocrinology and diabetes. JCRPE is the official journal of Turkish Society for Pediatric Endocrinology and Diabetes and is published in English quarterly (March, June, September, December). The target audience includes physicians, researchers and other healthcare professionals in all areas of pediatric endocrinology and diabetes.

JCRPE is indexed in **PubMed/MEDLINE, Index Medicus/PubMed, PubMed Central (PMC), British Library, EBSCO, SCOPUS, EMBASE, Engineering Village, Reaxys, CINAHL, ProQuest, GALE, Turk Medline, Tübitak Ulakbim TR Index, Türkiye Citation Index, Science Citation Index-SCI-E, Hinari, GOALI, ARDI, OARE, AGORA, J-GATE, IdealOnline and DOAJ.**

JCRPE has an impact factor 1.5 in 2024.

****The 5-year impact factor 1.9 in 2024.**

The journal is printed on an acid-free paper.

Permissions

Requests for permission to reproduce published material should be sent to the publisher.

Galenos Publishing House

Address: Molla Gürani mah. Kaçamak Sok. 21/1 Fatih, İstanbul, Türkiye

Telephone: +90 212 621 99 25

Fax: +90 212 621 99 27

Web page: <http://www.galenos.com.tr/en>

E-mail: info@galenos.com.tr

Copyright Notice

The author(s) hereby affirms that the manuscript submitted is original, that all statement asserted as facts are based on author(s) careful investigation and research for accuracy, that the manuscript does not, in whole or part, infringe any copyright, that it has not been published in total or in part and is not being submitted or considered for publication in total or in part elsewhere.

Completed Copyright Assignment&Affirmation of Originality Form will be faxed to the JCRPE Editorial Office (Fax: +90 212 621 99 27).

By signing this form,

1. Each author acknowledge that he/she participated in the work in a substantive way and is prepared to take public responsibility for the work.
2. Each author further affirms that he or she has read and understands the "Ethical Guidelines for Publication of Research".
3. The author(s), in consideration of the acceptance of the manuscript for publication, does hereby assign and transfer to the Journal of Clinical Research in Pediatric Endocrinology all of the rights and interest in and the copyright of the work in its current form and in any form subsequently revised for publication and/or electronic dissemination.

Open Access Policy

This journal provides immediate open and free access to its content on the principle that making research freely available to the public supports a greater global exchange of knowledge.

Open Access Policy is based on the rules of the Budapest Open Access Initiative (BOAI) <http://www.budapestopenaccessinitiative.org/>. By "open access" to peer-reviewed research literature, we mean its free availability on the public internet, permitting any users to read, download, copy, distribute, print, search, or link to the full texts of these articles, crawl them for indexing, pass them as data to software, or use them for any other lawful purpose, without financial, legal, or technical barriers other than those inseparable from gaining access to the internet itself. The only constraint on reproduction and distribution, and the only role for copyright in this domain, should be to give authors control over the integrity of their work and the right to be properly acknowledged and cited.

This work is licensed under a Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 (CC BY-NC-ND) International License.

CC BY-NC-ND: This license allows reusers to copy and distribute the material in any medium or format in unadapted form only, for noncommercial purposes only, and only so long as attribution is given to the creator.

CC BY-NC-ND includes the following elements:

BY – Credit must be given to the creator

NC – Only noncommercial uses of the work are permitted

ND – No derivatives or adaptations of the work are permitted

INSTRUCTIONS TO AUTHORS

GENERAL INFORMATION

Manuscripts must be written in English and must meet the requirements of the journal. Papers that do not meet these requirements will be returned to the author for necessary revision before the review. Manuscripts submitted to JCRPE are evaluated by peer reviewers. Authors of manuscripts requiring modifications have two months to resubmit a revised paper. Manuscripts returned after this deadline will be treated as new submissions. The journal is in compliance with the uniform requirements for manuscripts submitted to biomedical journals published by the International Committee of Medical Journal Editors (NEJM 1997; 336:309-315, updated 2001). Upon submission of the manuscript, authors are to indicate the type of trial/research and provide the checklist of the following guidelines when appropriate: Consort statement for randomized controlled trials (Moher D, Schulz KF, Altman D, for the CONSORT Group. The CONSORT statement revised recommendations for improving the quality of reports of parallel group randomized trials. JAMA 2001 ; 285 : 1987 - 91), the QUOROM statement for meta-analysis and systemic reviews of randomized controlled trials (Moher D, Cook DJ, Eastwood S, Olkin I, Rennie D, Stroup DF. Improving the quality of reports of meta-analyses of randomized controlled trials: the QUOROM statement. Quality of Reporting of Meta-Analyses. Lancet 1999; 354 : 1896 – 900) and the MOOSE guidelines for meta-analysis and systemic reviews of observational studies (Stroup DF, Berlin JA, Morton SC, et al. Meta-analysis of observational studies in epidemiology: a proposal for reporting Meta-analysis of observational studies in Epidemiology (MOOSE) group. JAMA 2000; 283: 2008 – 12). Keywords are included according to MeSH (Medical Subject Headings) National Library of Medicine.

Once the manuscript is accepted to be published in The Journal of Clinical Research in Pediatric Endocrinology, it receives a Digital Object Identifier (DOI) number. Uncorrected full text files can be reached online via PubMed and Ahead of Print section of the journal's website (<http://www.jcrpe.org/ahead-of-print>).

All contents will be printed in black and white.

Article Publication Charges

As of January 1, 2025, "Journal of Clinical Research in Pediatric Endocrinology" applies an Article Processing Charge (APCs) for only accepted articles. No fees are requested from the authors during submission and evaluation process. All manuscripts must be submitted via <https://jcrpe.manuscriptmanager.net/>. An APC fee of and local taxes will be applied depending on the article type (see Table 1)

Table 1.

Article Type	Fee
Original article	\$ 350
Case Report	\$ 275
Noninvited Review	\$ 500

Please contact the editorial office for detailed information by the following link: info@jcrpe.org

*Please note that the Article Processing Charge (APC) will not affect neither the editorial and peer-review process nor the priority of the manuscripts by no means. All submissions will be evaluated by the Editorial Board and the external reviewers in terms of scientific quality and ethical standards.

MANUSCRIPT CATEGORIES

All manuscripts must adhere to the limitations, as described below, for text only; the word count does not include the abstract, references, or figure/table legends. The word count must be noted on the title page, along with the number of figures and tables. Original Articles should be no longer than 4000 words and include no more than six figures and tables and 50 references. The abstract of the article should not exceed 250 words.

Short Communications are short descriptions of focused studies with important, but very straightforward results. These manuscripts should be no longer than 2000 words, and include no more than two figures and tables and 20 references. The abstract of the article should not exceed 250 words.

Brief Reports are discrete, highly significant findings reported in a shorter format. The abstract of the article should not exceed 150 words and the text/article length should not exceed 1200 words. References should be limited to 12, a maximum of 2 figures or tables.

Clinical Reviews address important topics in the field of pediatric endocrinology. Authors considering the submission of uninvited reviews should contact the editors in advance to determine if the

topic that they propose is of current potential interest to the Journal. Reviews will be considered for publication only if they are written by authors who have at least three published manuscripts in the international peer reviewed journals and these studies should be cited in the review. Otherwise only invited reviews will be considered for peer review from qualified experts in the area. These manuscripts should be no longer than 5000 words and include no more than four figures and tables and 120 references. The abstract of the article should not exceed 250 words.

Case Reports are descriptions of a case or small number of cases revealing novel and important insights into a condition's pathogenesis, presentation, and/or management. These manuscripts should be 2500 words or less, with four or fewer figures and tables and 30 or fewer references. The abstract of the article should not exceed 250 words.

Consensus Statements may be submitted by professional societies. All such submission will be subjected to peer review, must be modifiable in response to criticisms, and will be published only if they meet the Journal's usual editorial standards. These manuscripts should typically be no longer than 4000 words and include no more than six figures and tables and 120 references.

Letters to the Editor may be submitted in response to work that has been published in the Journal. Letters should be short commentaries related to specific points of agreement or disagreement with the published work. Letters should be no longer than 500 words with no more than five complete references, and may not include any figures or tables.

Note on Prior Publication

The journal publishes original research and review material. Material previously published in whole or in part shall not be considered for publication. At the time of submission, authors must report that the manuscript has not been published elsewhere. Abstracts or posters displayed at scientific meetings need not be reported.

MANUSCRIPT SUBMISSION PROCEDURES

JCRPE only accepts electronic manuscript submission at the web site www.jcrpe.org

After logging on to the website www.jcrpe.org click 'Submit an Article' icon. All corresponding authors should be provided a password and a username after providing the information needed. If you already have an account from a previous submission, enter your username and password to submit a new or revised manuscript. If you have forgotten your username and/or password, e-mail the editorial office for assistance. After logging on the article submission system with your own password and username please read carefully the directions of the system to provide all needed information. Attach the manuscript, tables and figures and additional documents.

All Submissions Must Include:

1. A cover letter requesting that the manuscript be evaluated for publication in JCRPE and any information relevant to your manuscript. Cover letter should contain address, telephone, fax and e-mail address of the corresponding author.
2. Completed Copyright and Disclosure of Potential Conflicts of Interest Form. The corresponding author must acquire all of the authors' completed forms and mail to info@galenos.com.tr / yayin@galenos.com.tr or submit to the Manuscript Manager.
3. Completed Disclosure of Potential Conflict of Interest Form. The corresponding author must acquire all of the authors' completed disclosure forms and fax them to the editorial office at +90 212 621 99 27.

Authors must complete the online submission forms. If unable to successfully upload the files please contact the editorial office by e-mail.

MANUSCRIPT PREPARATION

General Format

The Journal requires that all submissions be submitted according to these guidelines:

- Text should be double spaced with 2.5 cm margins on both sides using 12-point type in Times Roman font.
- All tables and figures must be placed after the text and must be labeled.
- Each section (abstract, text, references, tables, figures) should start on a separate page.
- Manuscripts should be prepared as word document (*.doc) or rich text format (*.rtf).

INSTRUCTIONS TO AUTHORS

Title Page

The title page should include the following:

- Full title
- Short title of not more than 40 characters for page headings
- Authors' names, and institutions, and e-mail addresses
- Corresponding author's e-mail and post address, telephone and fax numbers
- At least five and maximum eight keywords. Do not use abbreviations in the keywords
- Word count (excluding abstract, figure legends and references)
- Name and address of person to whom reprint requests should be addressed
- Any grants or fellowships supporting the writing of the paper
- The acknowledgements, if there are any
- If the content of the manuscript has been presented before, the time and place of the presentation
- The ORCID (Open Researcher and Contributor ID) number of the all authors should be provided while sending the manuscript. A free registration can be done at <http://orcid.org>.

Structured Abstracts (According to the The Journal of the American Medical Association)

Original Articles should be submitted with structured abstracts of no more than 250 words. All information reported in the abstract must appear in the manuscript. The abstract should not include references. Please use complete sentences for all sections of the abstract. Structured abstract should include background, objective, methods, results and conclusion.

What is already known on this topic?

What this study adds?

These two items must be completed before submission. Each item should include at most 2-3 sentences and at most 50 words focusing on what is known and what this study adds. Review papers do not need to include these boxes.

Introduction

The article should begin with a brief introduction stating why the study was undertaken within the context of previous reports.

Experimental Subjects

All clinical investigations described in submitted manuscripts must have been conducted in accordance with the guidelines in the Declaration of Helsinki and has been formally approved by the appropriate institutional review committees. All manuscripts must indicate that such approval was obtained and that informed consent was obtained from subjects in all experiments involving humans. The study populations should be described in detail. Subjects must be identified only by number or letter, not by initials or names. Photographs of patients' faces should be included only if scientifically relevant. Authors must obtain written consent from the patient for use of such photographs.

Clinical Trials Registration

All clinical trials must be registered in a public trials registry acceptable to the International Committee of Medical Journals Editors (ICMJE). Authors of randomized controlled trials must adhere to the CONSORT guidelines, and provide both a CONSORT checklist (for protocols, see the SPIRIT guidance) and flow diagram. We require that you choose the MS Word template at www.consort-statement.org for the flow chart and cite/upload it in the manuscript as a figure. In addition, submitted manuscripts must include the unique registration number in the Abstract as evidence of registration.

You can register for clinical trials by visiting the following link:

<https://clinicaltrials.gov/>

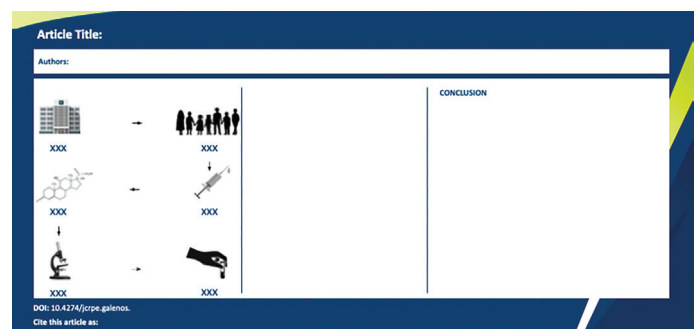
To register the relevant record in the system and learn more about the protocol to be followed, please review the link below:

<https://classic.clinicaltrials.gov/ct2/manage-recs/how-register>

Graphical Abstract

The Journal of Clinical Research in Pediatric Endocrinology aims to enhance the visibility of authors' publications and contribute to the scientific community as much as possible. In this regard, our journal encourages authors to publish graphical abstracts. The provided example abstract below reflects the standards accepted by our journal. Therefore, our journal expects authors to adhere to the following instructions:

- 1) Unless authors specify otherwise, the vectors used in the example will be used as the standard. Information to be placed beneath the vectors will be provided by the authors.
- 2) The article graphics used in the example will be revised with graphics of the authors' preference. Once these two stages are completed successfully, the prepared graphical abstract will be integrated into the article. Our journal aims to support authors in this process and enhance the impact of their scientific work.



Experimental Animals

A statement confirming that all animal experimentation described in the submitted manuscript was conducted in accord with accepted standards of humane animal care, according to the Declaration of Helsinki and Genova Convention, should be included in the manuscript.

Materials and Methods

These should be described and referenced in sufficient detail for other investigators to repeat the work. Ethical consent should be included as stated above.

The name of the ethical committee, approval number should be stated. At the same time, the Ethics Committee Approval Form should be uploaded with the article.

Results

The Results section should briefly present the experimental data in text, tables, and/or figures. Do not compare your observations with that of others in the results section.

The raw results of weight, length/height, body mass index, and blood pressure measurements can not be compared among groups since they normally change with age and according to gender. Instead, standard deviation scores of those values should be reported and compared.

Discussion

The Discussion should focus on the interpretation and significance of the findings with concise objective comments that describe their relation to other work in that area and contain study limitations.

Study Limitations

Limitations of the study should be detailed. In addition, an evaluation of the implications of the obtained findings/results for future research should be outlined.

Conclusion

The conclusion of the study should be highlighted.

Acknowledgments (Not Required for Submission)

An acknowledgment is given for contributors who may not be listed as authors, or for grant support of the research.

INSTRUCTIONS TO AUTHORS

Authorship Contribution

The kind of contribution of each author should be stated.

References

Authors are required to provide an active access link and, where available, the DOI (Digital Object Identifier) for each reference listed in the bibliography at the time of manuscript submission. References lacking DOI information should include a direct and verifiable URL to the source. Submissions with incomplete reference metadata may be returned to authors for correction prior to peer review.

References to the literature should be cited in numerical order (in parentheses) in the text and listed in the same numerical order at the end of the manuscript on a separate page or pages. The author is responsible for the accuracy of references.

Number of References: Case Report max 30 / Original Articles max 50

Examples of the reference style are given below. Further examples will be found in the articles describing the Uniform Requirements for Manuscripts Submitted to Biomedical Journals (Ann Intern Med. 1988; 208:258-265, Br Med J. 1988; 296:401-405). The titles of journals should be abbreviated according to the style used in the Index Medicus.

Journal Articles and Abstracts: List all authors. The citation of unpublished observations, of personal communications is not permitted in the bibliography. The citation of manuscripts in press (i.e., accepted for publication) is permitted in the bibliography; the name of the journal in which they appear must be supplied. Citing an abstract is not recommended.

Books: List all authors or editors.

Sample References

Papers Published in Periodical Journals: Gungor N, Saad R, Janosky J, Arslanian S. Validation of surrogate estimates of insulin sensitivity and insulin secretion in children and adolescents. J Pediatr 2004;144:47-55.

Papers Only Published with DOI Numbers: Knops NB, Sneeuw KC, Brand R, Hile ET, de Ouden AL, Wit JM, Verloove-Vanhorick SP. Catch-up growth up to ten years of age in children born very preterm or with very low birth weight. BMC Pediatrics 2005 doi: 10.1186/1471-2431-5-26.

Book Chapters: Darendeliler F. Growth Hormone Treatment in Rare Disorders: The KIGS Experience. In: Ranke MB, Price DA, Reiter EO (eds). Growth Hormone Therapy in Pediatrics: 20 Years of KIGS. Basel, Karger, 2007;213-239.

Books: Practical Endocrinology and Diabetes in Children. Raine JE, Donaldson MDC, Gregory JW, Savage MO. London, Blackwell Science, 2001;37-60.

Tables

Tables must be constructed as simply as possible. Each table must have a concise heading and should be submitted on a separate page. Tables must not simply duplicate the text or figures. Number all tables in the order of their citation in the text. Include a title for each table (a brief phrase, preferably no longer than 10 to 15 words). Include all tables in a single file following the manuscript.

Figures Legends

Figure legends and titles should be submitted on a separate page. Figure legends and titles should be clear and informative. Tables and figures should work under "windows". Number all figures (graphs, charts, photographs, and illustrations) in the order of their citation in the text. Include a title for each figure (a brief phrase, preferably no longer than 10 to 15 words).

Figures & Images

At submission, the following file formats are acceptable: AI, EMF, EPS, JPG, PDF, PPT, PSD, TIF. Figures may be embedded at the end of the manuscript text file or loaded as separate files for submission purposes.

All images MUST be at or above intended display size, with the following image resolutions: Line Art 800 dpi, Combination (Line Art + Halftone) 600 dpi, Halftone 300 dpi. See the Image quality specifications chart for details. Image files also must be cropped as close to the actual image as possible.

Units of Measure

Results/Tables should be expressed in metric units. If needed please apply this usage in your manuscript.

If p values are significant in the tables you have prepared, the relevant p values should be indicated in bold font.

Validation of Data and Statistical Analysis

Assay validation: Bioassay and radioimmunoassay potency estimates should be accompanied by an appropriate measure of the precision of these estimates. For bioassays, these usually will be the standard deviation, standard error of the mean, confidence limits. For both bioassays and radioimmunoassays, it is necessary to include data relating to within-assay and between-assay variability. If all relevant comparisons are made within the same assay, the latter may be omitted. Statistical analysis should be done accurately and with precision. Please consult a statistician if necessary.

Proofs and Reprints

Proofs and a reprint order are sent to the corresponding author. The author should designate by footnote on the title page of the manuscript the name and address of the person to whom reprint requests should be directed. The manuscript when published will become the property of the journal.

Page and Other Charges

Archiving

The editorial office will retain all manuscripts and related documentation (correspondence, reviews, etc.) for 12 months following the date of publication or rejection.

Submission Preparation Checklist

As part of the submission process, authors are required to check off their submission's compliance with all of the following items, and submissions may be returned to authors that do not adhere to these guidelines.

1. The submission has not been previously published, nor is it before another journal for consideration (or an explanation has been provided in Comments to the Editor).
2. The submission file is in Microsoft Word, RTF, or WordPerfect document file format. The text is double-spaced; uses a 12-point font; employs italics, rather than underlining (except with URL addresses); and all illustrations, figures, and tables are placed within the text at the appropriate points, rather than at the end. Please do not send the manuscript in docx.
3. Where available, URLs for the references have been provided.
4. A completed Copyright ve Disclosure of Potential Conflicts of Interest Form must be uploaded with other files during the submission. The corresponding author must acquire all of the authors' completed forms and mail to info@galenos.com.tr / yayin@galenos.com.tr or submit to the Manuscript Manager.
5. The text adheres to the stylistic and bibliographic requirements outlined in the Author Guidelines, which is found in About the Journal.
6. Completed Disclosure of Potential Conflict of Interest Form. The corresponding author must acquire all of the authors' completed disclosure forms and fax them, together, to the editorial office along with the Author Disclosure Summary.

Privacy Statement

The names and email addresses entered in this journal site will be used exclusively for the stated purposes of this journal and will not be made available for any other purpose or to any other party.

Peer Review Process

1. The manuscript is assigned to an editor, who reviews the manuscript and makes an initial decision based on manuscript quality and editorial priorities.
2. For those manuscripts sent for external peer review, the editor assigns reviewers to the manuscript.
3. The reviewers review the manuscript.
4. The editor makes a final decision based on editorial priorities, manuscript quality, and reviewer recommendations.
5. The decision letter is sent to the author.

INSTRUCTIONS TO AUTHORS

The Reviewer is Asked to Focus on the Following Issues:

1. General recommendation about the manuscript

How original is the manuscript?
Is it well presented?
How is the length of the manuscript?

2. Publication timing, quality, and priority

How important is the manuscript in this field?
Does it present original data?
Does it carry priority in publishing?

3. Specific questions regarding the quality of the manuscript

Does the title describe the study accurately?
Is the abstract informative and clear?
Do the authors state the study question in the introduction?
Are the methods clear?
Are ethical guidelines met?
Are statistical analyses appropriate?
Are the results presented clearly?
Does the discussion cover all of the findings?
Are the references appropriate for the manuscript?

4. Remarks to the editor

Accepted in its present form
Accepted after modest revisions
Reconsidered for acceptance after major changes
Rejected

5. Remarks to the author

What would be your recommendations to the author?
Conflict of interest statement for the reviewer (Please state if a conflict of interest is present)
For further instructions about how to review, see Reviewing Manuscripts for Archives of Pediatrics & Adolescent Medicine by Peter Cummings, MD, MPH; Frederick P. Rivara, MD, MPH in Arch Pediatr Adolesc Med. 2002;156:11-13.

GUIDELINES FOR MANUSCRIPT PREPARATION

The authors can also benefit from the following guidelines in the process of preparing the article:
Clinical Trials
Observational Studies
Systematic Review
Diagnostic and Prognostic Studies

CONTENTS

CASE REPORTS

- 1 **A Rare Presentation of 17 α -Hydroxylase/17,20-Lyase Deficiency in a Patient with Non-Hodgkin's Lymphoma: A Case Report**
Niran Tekkeli, İlknur Kurt, Nevin Yalman, Çetin Timur, Şenol Demir, Elif Sağsak; İstanbul, Türkiye
- 6 **Autosomal Recessive Hypophosphatemic Rickets Type 2 Associated with a Novel *ENPP1* Variant in a Taiwanese Girl**
Han-Yi Lin, Ni-Chung Lee, Meng-Ju Melody Tsai, Ting-Ming Wang, Yi-Ching Tung; Hsinchu, Taipei, Yunlin, Taiwan
- 12 **Hereditary Pheochromocytoma as a Major Manifestation of von Hippel Lindau Disease (vHL) in Childhood: Long-term Follow-up of Five Patients with vHL from One Family**
Katarzyna Pasternak-Pietrzak, Agata Kozłowska, Elzbieta Moszczyńska; Warsaw, Poland
- 20 **A Rare Cause of Proportional Short Stature and Puberty Precocity: Floating-Harbor Syndrome**
Duygu Çetinkaya, Gönül Büyükyılmaz, Esra Kılıç; Ankara, Türkiye
- 25 **A Rare Coexistence of Turner Syndrome and Mycosis Fungoides: A Case Report**
Ozge Bayrak Demirel, Esin Karakılıç-Ozturan, Tugba Atci, Sule Ozturk Sari, Can Baykal, Asli Derya Kardelen, Melek Yıldız, Sukran Poyrazoglu, Firdevs Bas, Feyza Darendeliler; İstanbul, Türkiye
- 30 **Unraveling a Genetic Puzzle: Could *MAP3K7* Be a Candidate Gene for RASopathies?**
Sirmen Kızılcan Çetin, Zeynep Şıklar, Zehra Aycan, Elif Özsu, Serdar Ceylaner, Merih Berberoğlu; Ankara, Türkiye
- 37 **Gonadoblastoma with Dysgerminoma in a Virilized Adolescent with Karyotype 46,XX: A Case Report and Review of the Literature**
Tugce Kandemir, Esin Karakılıç Ozturan, Ozlem Dural, Ayca Dilruba Aslanger, Elif Inan Balci, Aysel Bayram, Semen Onder, Asli Derya Kardelen, Melek Yıldız, Sukran Poyrazoglu, Firdevs Bas, Feyza Darendeliler; İstanbul, Türkiye
- 45 **Permanent Neonatal Diabetes with High Insulin Requirements due to a New Variant in the *INS* Gene**
Johana Andrea Botero Hernández, Gina González-Valencia, Vanessa Suarez, Gabriel del Castillo; Medellín, Nariño, Colombia
- 50 **A Novel *SRD5A2* Loss-of-Function Variant in a Chinese Child with 5 α -reductase Type 2 Deficiency**
Peng Zhou, Juanjuan Lyu, Xiaomei Sun, Ying Liu, Chuanjie Yuan, Jin Wu; Chengdu, China
- 54 **A Rare Cause of Neonatal Salt Wasting Syndrome: Clinical Management of a Case Diagnosed with Pseudohypoaldosteronism due to a Novel Homozygous Variant in the *SCNN1B* Gene**
Berna Singin, Zeynep Donbaloğlu, Ebru Barsal Çetiner, Kürşat Çetin, Nurten Özkan Zarif, Kıymet Çelik, Ercan Mıhçı, Özden Altıok Clark, Hale Tuhan, Mesut Parlak; Antalya, Türkiye
- 59 **Clinical and Molecular Landscape of Weiss-Kruszka Syndrome: A Case Report and Literature Review**
Lele Li, Chunxiu Gong; Beijing, China
- 64 **Cabergoline Induced Pathological Gambling in an Adolescent with Prolactinoma**
Ummahan Tercan, Ezgi Sarban, Melek Yıldız, Özlem Nida Erbaşı, Mine Özkan, Asli Derya Kardelen, Şükran Poyrazoğlu, Firdevs Baş, Feyza Darendeliler; İstanbul, Türkiye
- 70 **Atypical Presentation of New Onset Diabetes with Hyperglycemic Hyperosmolar State in Two Toddlers**
Alina Haque, Esther E. Bell-Sambataro, Foram Patel, Leena Mamilly, Kathryn Obrynba, Jennifer M. Ladd; Ohio, Florida, Texas, United States
- 75 **Thauvin-Robinet-Faivre Syndrome: A *FIBP* Variant in an Adolescent with Segmental Overgrowth and Thyroid Carcinoma**
Ülkü Gül Şiraz, Deniz Koçak Göl, Meino Rohlf, Christoph Klein, Ekrem Ünal, Nihal Hatipoğlu; Kayseri, Gaziantep, Türkiye; Munich, Germany
- 83 **Xp21 Contiguous Gene Deletion Syndrome: Diagnosis, Treatment, and a Review of the Literature on a Rare Genetic Disorder**
Berna Singin, Zeynep Donbaloğlu, Ebru Barsal Çetiner, Aynur Bedel, Kürşat Çetin, Belgin Akcan Paksoy, Tarkan Kalkan, Halide Akbaş, Hale Ünver Tuhan, Mesut Parlak; Antalya, Türkiye
- 92 **Four Consecutive False Negative Newborn Screens in a Patient with Classical Congenital Adrenal Hyperplasia: A Case Report**
Patrick Rizzuto, Mariam Gangat, Ahmed Khattab, Ian Marshall; New Jersey, United States