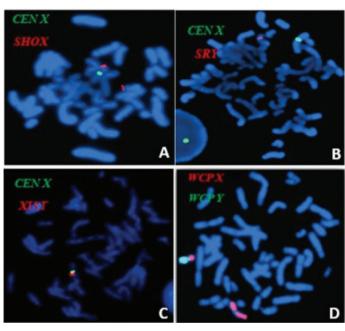
# JCRPE

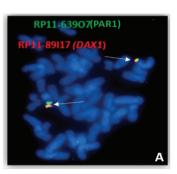
Journal of Clinical Research in Pediatric Endocrinology

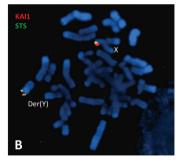
March 2023 volume 15 issue 1 www.jcrpe.org ISSN: 1308-5727

E-ISSN: 1308-5735



FISH analysis. A) FISH results using SHOX probe, two red spots were detected. B) FISH results using SRY probe, one red spot was detected. C) FISH results using XIST probe, one red spot was detected. D) FISH analysis using WCPX/WCPY showed the presence of a part from chromosome X on the Y chromosome





FISH analysis using specific probes:
A) NR0B1 probe showed its presence on both sex chromosomes (white arrows); B) KAL1 and STS probes showed their presence on both sex chromosomes

Anomalies in Human Sex Determination: Usefulness of a Combined Cytogenetic Approach to Characterize an Additional Case with Xp Functional Disomy Associated with 46,XY Gonadal Dysgenesis

Rjiba K et al. Page: 25-34







#### **Editor in Chief**

#### Feyza Darendeliler

#### Associate Editors Abdullah Bereket

Marmara University Faculty of Medicine, Department of Pediatric Endocrinology, İstanbul, Turkey abdullahbereket@gmail.com

orid.org/0000-0002-6584-9043

#### Damla Gökşen

Ege University Faculty of Medicine, Department of Pediatric Endocrinology, İzmir, Turkey damla.goksen@ege.edu.tr

orcid.org/0000-0001-6108-0591

#### Korcan Demir

Dokuz Eylül University Faculty of Medicine, Department of Pediatric Endocrinology, İzmir, Turkey korcandemir@qmail.com

orcid.org/0000-0002-8334-2422

#### Samim Özen

Ege University Faculty of Medicine, Department of Pediatric Endocrinology, İzmir, Turkey samim.ozen@ege.edu.tr

© orcid.org/0000-0001-7037-2713

#### **Serap Turan**

Marmara University Faculty of Medicine, Department of Pediatric Endocrinology, İstanbul, Turkey serap.turan@marmara.edu.tr

orcid.org/0000-0002-5172-5402

#### **English Language Editor**

Jeremy Jones, Kocaeli, Turkey

⊕ 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 pirinted 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.

#### **Editorial Board**

#### Ali Kemal Topaloğlu

Cukurova University Faculty of Medicine, Department of Pediatric Endocrinology, Adana. Turkey

#### Angel Ferrandez Longas

Children's Hospital Miguel Servet, Department of Pediatric Endocrinology, Zaragoza. Spain

#### Aysun Bideci

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

#### Banerjee Indi

Manchester University NHS Foundation Trust, Manchester, United Kingdom

#### Fima Lifshitz

Pediatric Sunshine Academics, Inc., Santa Barbara, USA

#### Hüseyin Onay

Ege University Faculty of Medicine, Department of Medical Genetics, İzmir, Turkev

#### Murat Bastepe

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

#### 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

Great Ormond Street Hospital for Children, Department of Pediatric Endocrinology, London, United Kingdom

#### Margaret C S Boguszewski

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

#### Merih Berberoğlu

Ankara University Faculty of Medicine, Department of Pediatric Endocrinology, Ankara, Turkey

#### Mitchell Geffner

Children's Hospital Los Angeles, Center for Endocrinology, Diabetes and Metabolism, Los Angeles, USA

#### Neslihan Güngör

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

#### Nurgün Kandemir

Hacettepe University Faculty of Medicine, Department of Pediatric Endocrinology, Ankara, Turkey

#### Güven Özkaya

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

#### Ömer Tarım

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

#### Pietro Galassetti

University of California, Pediatric Exercise and Genomics Research Center, Department of Pediatrics, California, USA

#### Robert Rapaport

Icahn School of Medicine at Mount Sinai, Kravis Children's Hospital at Mount Sinai, Department of Pediatric Endocrinology and Diabetes. New York. USA

#### Sandra L. Blethen

Emeritus Professor, Belmont, CA, USA

#### Thomas Allen Wilson

Stony Brook Children's Hospital, Department of Pediatric Endocrinology, New York, USA

#### Violeta lotova

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

#### Wayne Cutfield

University of Auckland, Liggins Institute, Department of Pediatric Endocrinology, Auckland, New Zealand



Publisher Contact

Address: Molla Gürani Mah. Kaçamak Sk. No: 21/1 34093 İstanbul, Turkey

Phone: +90 (212) 621 99 25 Fax: +90 (212) 621 99 27 E-mail: info@galenos.com.tr /yayin@galenos.com.tr

Web: www.galenos.com.tr Publisher Certificate Number: 14521 Printing at:

Son Sürat Daktilo Dijital Baskı San. Tic. Ltd. Şti.

Address: Gayrettepe Mah. Yıldızposta Cad. Evren Sitesi A Blok No: 32 D: 1-3

34349 Beşiktaş, İstanbul/Turkey Phone: +90 212 288 45 75 Date of printing: March 2023 ISSN: 1308-5727 E-ISSN: 1308-5735

www.jcrpe.org

#### AIMS AND SCOPE

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

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

JCRPE has an impact factor 2.016 in 2021. \*\*The 5-year impact factor 2.323 in 2021.

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. Kacamak Sok. 21/1 Fatih, Istanbul, Turkey

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 access to its content on the principle that making research freely available to the public supports a greater global exchange of knowledge.

This work is licensed under a Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International License.

#### **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, Schultz 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.

#### NEW

Article Publication Charges for accepted case reports is \$100. Please contact the editorial office for detailed information by the following link:

#### info@icrpe.org

In case of exceeding 4000 word limit, the author is charged with \$50 for each

All other forms of articles are free of publication charge.

#### **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.

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.

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

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.

Consensus Statements may be submitted by professional societies. All such submission will be subjected to peer review, must be modifiable in

www.jcrpe.org

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

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

#### MANUSCRIPT SUBMISSION PROCEDURES

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

After logging on to the website www.jcrpe.org click 'online manuscript submission' 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
- 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

- 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).

#### 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
- 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

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**

For clinical trial reports to be considered for publication in the Journal, prospective registration, as endorsed by the International Conference of Medical Journal Editors, is required. We recommend use of http://www. clinicaltrials.gov.

#### **Experimental Animals**

A statement confirming that all animal experimentation described in the submitted manuscript was conducted in accord with accepted standards of

## JCRP E Journal of Clinical Research in Pediatric Endocrinology www.jcrpe.org

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

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.

#### 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.

#### **Authorship Contribution**

The kind of contribution of each author should be stated.

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

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.

#### www.jcrpe.org

#### **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
- 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 aquire 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.

#### 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.

#### **Review**

1 Specific Functions of Melanocortin 3 Receptor (MC3R)

Tulin Yanik, Seyda Tugce Durhan

#### **Original Articles**

- 7 Factors Affecting Thyroid Elastography in Healthy Children and Patients with Hashimoto's Thyroiditis Serkan Bilge Koca, Turgut Seber
- 16 Endocrine Abnormalities and Growth Characterization in Colombian Pediatric Patients with 22q11 Deletion Syndrome Juan Lasprilla-Tovar, Nora Alejandra Zuluaga, Carolina Forero, Oscar Correa-Jiménez, Javier Mauricio Sierra
- Anomalies in Human Sex Determination: Usefulness of a Combined Cytogenetic Approach to Characterize an Additional Case with Xp Functional Disomy Associated with 46,XY Gonadal Dysgenesis

  Khouloud Rjiba, Wafa Slimani, Meriem Gaddas, Ikbel Hadj Hassine, Afef Jelloul, Hela Ben Khelifa, Fethi El Amri, Monia Zaouali,

  Kenneth Mcelreavey, Ali Saad, Soumaya Mougou-Zerelli
- 35 Clinical Utility and Outcome Prediction of Early ZnT8-IgG Testing and Titer in Type 1 Diabetes Amanda R. Dahl, Sarah Jenkins M., Sean J. Pittock, Siobhan T. Pittock
- 42 Comparison of Commonly Used Methods to Predict the Final Height in Constitutional Tall Stature Alma Kamar Matias, Evgenia Muginshtein-Simkovitch, Gilad Twig, Lilos Pearl, Zvi Laron
- Frequency, Clinical Characteristics and Predictors of Ketoacidosis at Diagnosis of Type One Diabetes Mellitus in Children and Adolescents from Jordan

  Rasha Odeh, Lobna Gharaibeh, Amirah Daher, Jumana Albaramki, Bahaa Ashour, Fatima Al Barakat, Dina Dahabreh, Hiba Hadadin,
- The Relationship Between Premature Adrenarche and Platelet Aggregation Ahmet Bolat, Cengiz Zeybek, Orhan Gürsel, Onur Akın, Mehmet Emre Taşçılar
- Factors Associated with Low Bone Mineral Density at the Time of Diagnosis in Children with Celiac Disease Emine Çamtosun, Fatma İlknur Varol, Şükrü Güngör, Mukadder Ayşe Selimoğlu
- Increased Carotid Intima-media Thickness and Its Association with Carbohydrate Metabolism and Adipocytokines in Children Treated with Recombinant Growth Hormone

  Seha Saygılı, Mehmet Kocaağa, Gamze Kaya, Mine Şükür, Firdevs Baş, Şükran Poyrazoğlu, Rüveyde Bundak, Feyza Darendeliler

#### **Case Reports**

Tala Melhem, Abeer Alassaf

- Weight Loss During Topiramate Treatment in a Severely Obese Adolescent with Congenital Adrenal Hyperplasia and Migraine Amy Seagroves, Heather M. Ross, Alaina P. Vidmar, Mitchell E. Geffner, William S. Kim, Darryl Hwang, Claudia Borzutzky, Nicole R. Fraga, Mimi S. Kim
- Rare Coexistence of Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency and Turner Syndrome: A Case Report and Brief Literature Review Isabel Inácio, Joana Serra-Caetano, Rita Cardoso, Isabel Dinis, Alice Mirante



- **90** Expanding the Phenotype of *TRMT10A* Mutations: Case Report and a Review of the Existing Cases Zeynep Şıklar, Tuğba Kontbay, Kevin Colclough, Kashyap A. Patel, Merih Berberoğlu
- 97 Compound Heterozygous Variants in FAM111A Cause Autosomal Recessive Kenny-Caffey Syndrome Type 2 Erdal Eren, Havva Tezcan Ünlü, Serdar Ceylaner, Ömer Tarım
- Maltodextrin May Be a Promising Treatment Modality After Near-total Pancreatectomy in Infants Younger Than Six Months with Persistent Hyperinsulinism: A Case Report Yasemin Denkboy Öngen, Erdal Eren, Halil Sağlam