

JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM

EDITOR-IN-CHIEF

Wieland Kiess, Leipzig

ASSOCIATE EDITORS

Abdullah Bereket, Istanbul

Feyza Darendeliler, Istanbul

Mehul Dattani, London

Jan Gustafsson, Uppsala

Feihong Luo, Shanghai

Veronica Mericq, Santiago

Christian Roth, Seattle, WA

Jorma Toppari, Turku

ASSISTANT MANAGING EDITOR

Melanie Penke, Leipzig

EDITORIAL BOARD

Tadej Battelino, Ljubljana

Atilla Buyukgebiz, Istanbul

Fernando Cassorla, Santiago de Chile

George Chrousos, Athens

Wayne Cutfield, Auckland

Hugo L. Fideleff, Buenos Aires

Eli HersHKovitz, Beersheba

Olaf Hiort, Lübeck

Stephen LaFranchi, Portland, OR

Roberto Lanes, Caracas

Angelika Mohn, Chieti

Ron Rosenfeld, Portland, OR

Allen W. Root, Saint Petersburg, FL

George Werther, Parkville

Zvi Zadik, Rehovot

DE GRUYTER

ABSTRACTED/INDEXED IN Baidu Scholar · Case · Celdes · Chemical Abstracts Service (CAS): CAPlus; SciFinder · CNKI Scholar (China National Knowledge Infrastructure) · CNPIEC · EBSCO (relevant databases) · EBSCO Discovery Service · Elsevier: EMBASE; Reaxys; SCOPUS · Gale/ Cengage · Genamics JournalSeek · Google Scholar · ICAP Alcohol Information Databases · Index Copernicus · J-Gate · JournalTOCs · Medline · Meta (formerly Scisearch) · Naviga (Softweco) · Primo Central (ExLibris) · ProQuest (relevant databases) · PubMed · PubsHub · ReadCube · ResearchGate · SCImago (SJR) · Sherpa/RoMEO · Summon (Serials Solutions/ProQuest) · TDOne (TDNet) · Thomson Reuters: Biological Abstracts; BIOSIS Previews: Current Contents/Life Sciences; Journal Citation Reports/Science Edition; Prou Science Integrity; Science Citation Index; Science Citation Index Expanded · Ulrich's Periodicals Directory/ulrichsweb · WorldCat (OCLC)

The publisher, together with the authors and editors, has taken great pains to ensure that all information presented in this work (programs, applications, amounts, dosages, etc.) reflects the standard of knowledge at the time of publication. Despite careful manuscript preparation and proof correction, errors can nevertheless occur. Authors, editors and publisher disclaim all responsibility for any errors or omissions or liability for the results obtained from use of the information, or parts thereof, contained in this work.

The citation of registered names, trade names, trademarks, etc. in this work does not imply, even in the absence of a specific statement, that such names are exempt from laws and regulations protecting trademarks etc. and therefore free for general use.

ISSN 0334-018X · e-ISSN 2191-0251

All information regarding notes for contributors, subscriptions, Open access, back volumes and orders is available online at www.degruyter.com/jpem.

RESPONSIBLE EDITOR(S)

Prof. Dr. Wieland Kiess, MD, Professor of Pediatrics, Director and Chair, Hospital for Children and Adolescents, Department of Women and Child Health, University Hospitals, University of Leipzig, Liebigstraße 20a, 04103 Leipzig, Germany, Tel.: +49 (0)341 972 60 00, Fax: +49 (0)341 972 60 09, e-mail: Wieland.Kiess@medizin.uni-leipzig.de

JOURNAL MANAGER Heike Jahnke, De Gruyter, Genthiner Straße 13, 10785 Berlin, Germany, Tel.: +49 (0)30 260 05 – 220, Fax: +49 (0)30 260 05 – 246, e-mail: jpem.editorial@degruyter.com

RESPONSIBLE FOR ADVERTISEMENTS Claudia Neumann, Genthiner Straße 13, 10785 Berlin, Germany, Tel.: +49 (0)30 260 05 – 226, Fax: +49 (0)30 260 05 – 264, e-mail: anzeigen@degruyter.com

© 2017 Walter de Gruyter GmbH, Berlin/Boston

TYPESETTING Compuscript Ltd., Shannon, Ireland

PRINTING Franz X. Stücker Druck und Verlag e.K., Ettenheim
Printed in Germany

Cover Illustration: medicalpicture/Friedhelm Achenbach



Offenlegung der Inhaber und Beteiligungsverhältnisse gem. § 7a Abs. 1 Ziff. 1, Abs. 2 Ziff. 3 des Berliner Pressegesetzes: Die Gesellschafter der Walter de Gruyter GmbH sind: Cram, Gisela, Rentnerin, Berlin; Cram, Elisabeth, Pensionärin, Rosengarten-Alvesen; Cram, Dr. Georg-Martin, Unternehmens-Systemberater, Stadtbergen; Cram, Maike, Wien (Österreich); Cram, Jens, Mannheim; Cram, Ingrid, Betriebsleiterin, Tuxpan/ Michoacan (Mexiko); Cram, Sabina, Mexico, DF (Mexiko); Cram, Silke, Wissenschaftlerin, Mexico DF (Mexiko); Cram, Björn, Aachen; Cram, Cram, Berit, Hamm; Cram-Gomez, Susana, Mexico DF (Mexiko); Cram-Heydrich, Walter, Mexico DF (Mexiko); Cram-Heydrich, Kurt, Angestellter, Mexico DF (Mexiko); Duvenbeck, Brigitta, Oberstudienrätin i.R., Bad Homburg; Gädeke, Gudula, M.A., Atemtherapeutin/Lehrerin, Tübingen; Gädeke, Martin, Einzelunternehmer, Ingolstadt; Lubasch, Dr. Annette, Ärztin, Berlin; Schütz, Dr. Christa, Ärztin, Mannheim; Schütz, Sonja, Berlin; Schütz, Juliane, Berlin; Schütz, Antje, Berlin; Schütz, Valentin, Mannheim; Seils, Dorothee, Apothekerin, Stuttgart; Seils, Gabriele, Journalistin, Berlin; Seils, Christoph, Journalist, Berlin; Siebert, John-Walter, Pfarrer, Oberstenfeld; Tran, Renate, Mediatorin, Zürich (Schweiz).

Contents

Editorial

Wieland Kiess, Melanie Penke, Jürgen Kratzsch and Roland Pfaeffle

Disorders of sex development — 1

Refers to articles on pp 3, 11, 19

Original Articles

Leena Nahata, Sonja I. Ziniel, Katharine C. Garvey, Richard N. Yu and Laurie E. Cohen

Fertility and sexual function: a gap in training in pediatric endocrinology — 3

Yasmeen Ganie, Colleen Aldous, Yusenhta Balakrishna and Rinus Wiersma

Disorders of sex development in children in KwaZulu-Natal Durban South Africa: 20-year experience in a tertiary centre — 11

Chupong Ittiwut, Jaturong Pratuangdejkul, Vichit Supornsilchai, Sasipa Muensri, Yodporn Hiranras, Taninee Sahakitrungruang, Suttipong Watcharasindhu, Kanya Suphapeetiporn and Vorasuk Shotelersuk

Novel mutations of the *SRD5A2* and *AR* genes in Thai patients with 46, XY disorders of sex development — 19

Catherine Mainguy, Gabriel Bellon, Véronique Delaup, Tiphane Ginoux, Behrouz Kassai-Koupai, Stéphane Mazur, Muriel Rabilloud, Laurent Remontet and Philippe Reix

Sensitivity and specificity of different methods for cystic fibrosis-related diabetes screening: is the oral glucose tolerance test still the standard? — 27

Prawej Ansari, Nadia Afroz, Shahnaz Jalil, Sohel Bin Azad, Md. Gazi Mustakim, Sabbir Anwar, S.M. Nazmul Haque, Saad Mosharraf Hossain, Ramisha Rifat Tony and J.M.A. Hannan

Anti-hyperglycemic activity of *Aegle marmelos* (L.) corr. is partly mediated by increased insulin secretion, α -amylase inhibition, and retardation of glucose absorption — 37

Selin Elmaogullari, Fatma Demirel and Nihal Hatipoglu
Risk factors that affect metabolic health status in obese children — 49

Johann Daxer, Theresa Hertrich, Ying Y. Zhao, Mandy Vogel, Andreas Hiemisch, Kathrin Scheuermann, Antje Körner, Jürgen Kratzsch, Wieland Kiess and Mirja Quante
Nocturnal levels of chemerin and progranulin in adolescents: influence of sex, body mass index, glucose metabolism and sleep — 57

Reiko Saito, Shunsuke Araki, Yukiyo Yamamoto and Koichi Kusuhara

Elevated endogenous secretory receptor for advanced glycation end products (esRAGE) levels are associated with circulating soluble RAGE levels in diabetic children — 63

Wojciech Szypowski, Karolina Kunecka, Beata Zduńczyk, Katarzyna Piechowiak, Maria Dyczek, Karolina Dąbrowa, Anna Wojtyra, Zuzanna Kaczmarek and Agnieszka Szypowska

Food exchange estimation by children with type 1 diabetes at summer camp — 71

Toshihide Kubo, Kyohei Takahashi, Mahoko Furujo, Yuki Hyodo, Hiroki Tsuchiya, Mariko Hattori, Shoko Fujinaga and Kenji Urayama

Usefulness of non-fasting lipid parameters in children — 77

Niels H. Birkebaek, David M. Hougaard and Arieh S. Cohen

Monitoring steroid replacement therapy in children with congenital adrenal hyperplasia — 85

Ho Ming Luk

Clinical and molecular characterization of Beckwith-Wiedemann syndrome in a Chinese population — 89

Karolina Antosik, Piotr Gnyś, Przemysława Jarosz-Chobot, Małgorzata Myśliwiec, Agnieszka Szadkowska, Maciej Matecki, Wojciech Młynarski and Maciej Borowiec
An analysis of the sequence of the *BAD* gene among patients with maturity-onset diabetes of the young (MODY) — 97

Case Reports

Janel Darcy Hunter, Melissa Ann Davis and Jennifer Rachel Law

Hypogonadotropic hypogonadism in a female patient with congenital arhinia — 101

Maria F. Contreras, Manish Raisingani, Kris Prasad, Bonita Franklin and Bina Shah

Transdermal testosterone gel for induction and continuation of puberty in adolescent boys with hepatic dysfunction — 105

Gabriela A. Vasques, Alfonso Hisado-Oliva, Mariana F.A. Funari, Antonio M. Lerario, Elisangela P.S. Quedas, Paulo Solberg, Karen E. Heath and Alexander A.L. Jorge
Long-term response to growth hormone therapy in a patient with short stature caused by a novel heterozygous mutation in *NPR2* — 111

Kosei Hasegawa, Chikahiko Numakura, Hiroyuki Tanaka, Mahoko Furujo, Toshihide Kubo, Yousuke Higuchi, Miho Yamashita and Hirokazu Tsukahara

Three cases of Japanese acromicric/geleophysic dysplasia with *FBN1* mutations: a comparison of clinical and radiological features — 117