

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): CAlplus; SciFinder · Clarivate Analytics (formerly Thomson Reuters): Biological Abstracts; BIOSIS Previews; Current Contents/Life Sciences; Journal Citation Reports/Science Edition; Prous Science Integrity; Science Citation Index; Science Citation Index Expanded · 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 · KESLI-NDSL (Korean National Discovery for Science Leaders) · Medline · Meta (formerly Scisearch) · Microsoft Academic · Naviga (Softweco) · Primo Central (ExLibris) · ProQuest (relevant databases) · PubMed · PubsHub · ReadCube · ResearchGate · SCImago (SJR) · Sherpa/RoMEO · Summon (Serials Solutions/ProQuest) · TDNet · Ulrich's Periodicals Directory/ulrichsweb · WanFang Data · 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 top-ad Bernd Beutel, Schlossergäßchen 10, 69469 Weinheim, Germany, Tel.: +49 (0)62 01 290 92-0, Fax: +49 (0)62 01 290 92-20, e-mail: info@top-ad-online.de

© 2017 Walter de Gruyter GmbH, Berlin/Boston

TYPESETTING Compuscript Ltd., Shannon, Ireland

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

Cover Illustration: medicalpicture/Friedhelm Achenbach



Contents

Editorial

Wieland Kiess, Melanie Penke, Louise Kobelt, Tobias Lipek, Constance Henn, Ruth Gausche, Mandy Vogel and Freerk Prenzel

Endocrine aspects in cystic fibrosis — 805

Refers to articles on pp 807, 815, 885

Original Articles

Adela Chirita-Emandi, Sheila Shepherd, Andreas Kyriakou, Jane D. McNeilly, Carol Dryden, Donna Corrigan, Anne Devenny and Syed Faisal Ahmed

A retrospective analysis of longitudinal changes in bone mineral content in cystic fibrosis — 807

Nicolas Terliesner, Mandy Vogel, Anna Steighardt, Ruth Gausche, Constance Henn, Julia Hentschel, Thomas Kapellen, Sabine Klamt, Julia Gebhardt, Wieland Kiess and Freerk Prenzel

Cystic-fibrosis related-diabetes (CFRD) is preceded by and associated with growth failure and deteriorating lung function — 815

Rachel L. Lundberg, Katherine R. Marino, Aastha Jasrotia, Louise S. Maranda, Bruce A. Barton, Laura C. Alonso and Benjamin Udoka Nwosu

Partial clinical remission in type 1 diabetes: a comparison of the accuracy of total daily dose of insulin of <0.3 units/kg/day to the gold standard insulin-dose adjusted hemoglobin A_{1c} of ≤9 for the detection of partial clinical remission — 823

Nikhil Tenneti, Devi Dayal, Sheetal Sharda, Inusha Panigrahi, Mohammed Didi, Savita Verma Attri, Naresh Sachdeva and Anil Kumar Bhalla

Concentrations of leptin, adiponectin and other metabolic parameters in non-obese children with Down syndrome — 831

Navindra Persaud, Hedyeh Ziai, Gerald Lebovic, Jonathon L. Maguire, Marina Khovratovich, Janis A. Randall Simpson, Khosrow Adeli, Jill Hamilton, Brian W. McCrindle, Patricia C. Parkin and Catherine S. Birken, on behalf of the TARGet Kids! collaboration

Parent reported nutritional risk and laboratory indices of cardiometabolic risk and in preschool-aged children — 839

Emilio García-García, Mercedes López-González, Rosa Cabello-Laureano and Elena Navarro-González

Multinodular goiter in children: treatment controversies — 847

Marcella Pedullà, Giuseppina Rosaria Umano, Vincenzo Fierro, Francesco Capuano, Anna Di Sessa, Pierluigi Marzuillo, Laura Perrone and Emanuele Miraglia del Giudice

Atopy as a risk factor for subclinical hypothyroidism development in children — 851

Mehri Khatami, Mohammad Mehdi Heidari, Fatemeh Tabesh, Mahtab Ordooei and Zohreh Salehifar

Mutation analysis of the *NKX2.5* gene in Iranian pediatric patients with congenital hypothyroidism — 857

Pascal Amedro, Nabil Tahhan, Helena Bertet, Claire Jeandel, Sophie Guillaumont, Thibault Mura and Marie-Christine Picot

Health-related quality of life among children with Turner syndrome: controlled cross-sectional study — 863

Sarah Elitzur, Revital Houri-Shtrecher, Michal Yackobovitz-Gavan, Galia Avrahami, Shlomit Barzilai, Gil Gilad, Yael Lebenthal, Moshe Phillip, Batia Stark, Isaac Yaniv and Shlomit Shalitin

Growth and pubertal patterns in young survivors of childhood acute lymphoblastic leukemia — 869

Yun Sun, Yan-yun Wang and Tao Jiang

Clinical features and genotyping of patients with primary carnitine deficiency identified by newborn screening — 879

Letter to the Editor

Valérie Boudreau, Catherine Lehoux Dubois, Katherine Desjardins, Marjolaine Mailhot, François Tremblay and Rémi Rabasa-Lhoret

Sensitivity and specificity of cystic fibrosis-related diabetes screening methods: which test should be the reference method? — 885

Case Reports

Nursel Muratoğlu Şahin, Meliha Esra Bilici, Erdal Kurnaz, Melek Pala Akdoğan, Serdar Ceylaner and Zehra Aycan
Type 1 rhizomelic chondrodysplasia punctata with a homozygous *PEX7* mutation — 889

Tyler Fick, Julie Jack, Amy L. Pyle-Eilola and Rohan K. Henry
Severe hypertriglyceridemia at new onset type 1 diabetes mellitus — 893

Juliana Gabriel Ribeiro de Andrade, Liliana Aparecida Lucci De Angelo Andrade, Gil Guerra-Junior and Andréa Trevas Maciel-Guerra

45,X/46,XY ovotesticular disorder of sex development revisited: undifferentiated gonadal tissue may be mistaken as ovarian tissue — 899

Lorenzo Figà Talamanca, Luca Pasquini, Antonio Napolitano and Daniela Longo

MRI in medium-chain acyl-coenzyme A dehydrogenase deficiency: neuroimaging during the first month — 905