

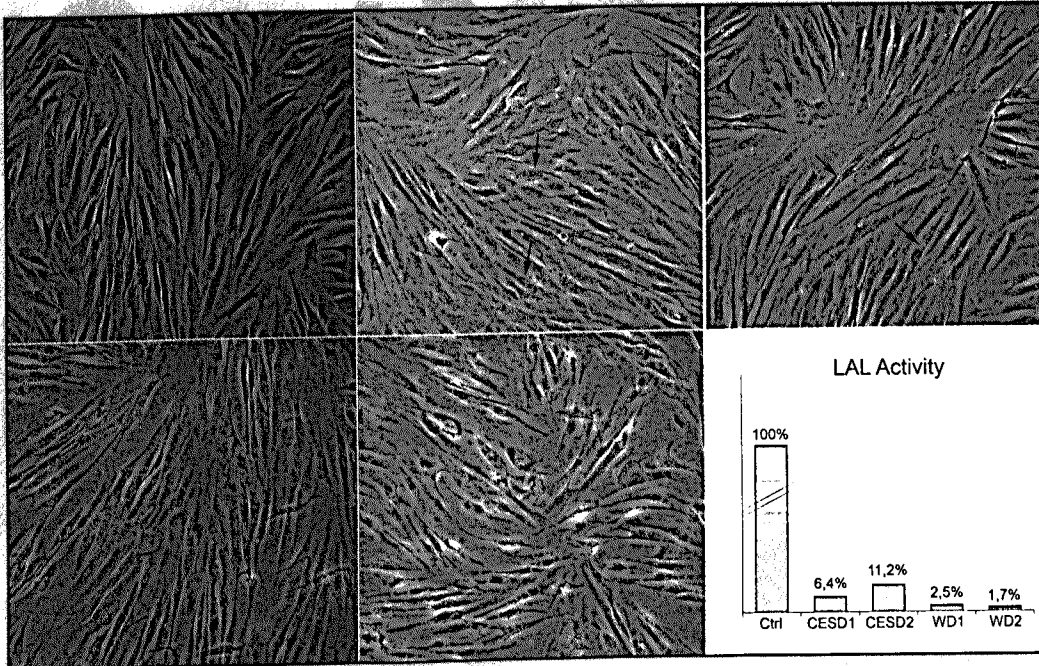
Molecular genetics and metabolism.
v. 105, no. 3 (Mar. 2012)
General Collection
W1 B1621R
2012-04-10 06:30:15

ISSN 1096-7192
Volume 105, Issue 3, March 2012

Molecular Genetics and Metabolism

Editor-in-Chief
Edward R.B. McCabe

PROPERTY OF THE
NATIONAL
LIBRARY OF
MEDICINE



020150 MOLECULAR GENETICS AND METABOLISM
2012 VOLUME 105 ISSUE 3
SISAC
030230
03269

Including the Program and Abstracts for
the Annual Meeting of the Society
for Inherited Metabolic Disorders

**DOCKET
ALARM**

Find authenticated court documents without watermarks at docketalarm.com.

Molecular Genetics and Metabolism

Volume 105 (3) 2012



ELSEVIER

Amsterdam • Boston • London • New York • Oxford • Paris • Philadelphia • San Diego • St. Louis

DOCKET
A L A R M

Find authenticated court documents without watermarks at docketalarm.com.

Molecular Genetics and Metabolism

Volume 105, Issue 3, March 2012

© 2012 Elsevier Inc. All rights reserved.

This journal and the individual contributions contained in it are protected under copyright by Elsevier Inc., and the following terms and conditions apply to their use:

Photocopying

Single photocopies of single articles may be made for personal use as allowed by national copyright laws. Permission of the Publisher and payment of a fee are required for all other photocopying, including multiple or systematic copying, copying for advertising or promotional purposes, resale, and all forms of document delivery. Special rates are available for educational institutions that wish to make photocopies for nonprofit educational classroom use. For information on how to seek permission visit www.elsevier.com/permissions or call: (+44) 1865 843830 (UK)/(+1) 215 239 3804 (USA).

Derivative Works

Subscribers may reproduce tables of contents or prepare lists of articles including abstracts for internal circulation within their institutions. Permission of the Publisher is required for resale or distribution outside the institution. Permission of the Publisher is required for all other derivative works, including compilations and translations (please consult www.elsevier.com/permissions).

Electronic Storage or Usage

Permission of the Publisher is required to store or use electronically any material contained in this journal, including any article or part of an article (please consult www.elsevier.com/permissions).

Except as outlined above, no part of this publication may be reproduced, stored in a retrieval system, or transmitted in any form or by any means, electronic, mechanical, photocopying, recording, or otherwise, without prior written permission of the Publisher.

Notice

No responsibility is assumed by the Publisher for any injury and/or damage to persons or property as a matter of product liability, negligence, or otherwise, or from any use or operation of any methods, products, instructions, or ideas contained in the material herein. Because of rapid advances in the medical sciences, in particular, independent verification of diagnoses and drug dosages should be made.

Although all advertising material is expected to conform to ethical (medical) standards, inclusion in this publication does not constitute a guarantee or endorsement of the quality or value of such product or of the claims made of it by its manufacturer.

Funding Body Agreements and Policies

Elsevier has established agreements and developed policies to allow authors whose articles appear in journals published by Elsevier, to comply with potential manuscript archiving requirements as specified as conditions of their grant awards. To learn more about existing agreements and policies please visit <http://www.elsevier.com/fundingbodies>.

Publication information: *Molecular Genetics and Metabolism* (ISSN 1096-7192) is published monthly by Elsevier (Radarweg 29, 1043 NX Amsterdam, The Netherlands). Further information on this journal is available from the Publisher or from the Elsevier Customer Service Department nearest you or from this journal's website (<http://www.elsevier.com/locate/ymgme>). Information on other Elsevier products is available through Elsevier's website (<http://www.elsevier.com>). Periodicals Postage Paid at Rahway, NJ, and at additional mailing offices. **USA POSTMASTER:** Send change of address to *Molecular Genetics and Metabolism*, Elsevier Customer Service Department, 3251 Riverport Lane, Maryland Heights, MO 63043, USA.

Orders, claims, and journal inquiries: Please contact the Elsevier Customer Service Department nearest you: **St. Louis:** Elsevier Customer Service Department, 3251 Riverport Lane, Maryland Heights, MO 63043, USA; phone: (877) 8397126 [toll free within the USA]; (+1) (314) 4478878 [outside the USA]; fax: (+1) (314) 4478077; e-mail: JournalCustomerService-usa@elsevier.com. **Oxford:** Elsevier Customer Service Department, The Boulevard, Langford Lane, Kidlington, Oxford OX5 1GB, UK; phone: (+44) (1865) 843434; fax: (+44) (1865) 843970; e-mail: JournalsCustomerServiceEMEA@elsevier.com. **Tokyo:** Elsevier Customer Service Department, 4F Higashi-Azabu, 1-Chome Bldg.1-9-15 Higashi-Azabu, Minato-ku, Tokyo 106-0044, Japan; phone: (+81) (3) 5561 5037; fax: (+81) (3) 5561 5047; e-mail: JournalsCustomerServiceJapan@elsevier.com. **Singapore:** Elsevier Customer Service Department, 3 Killiney Road, #08-01 Winsland House I, Singapore 239519; phone: (+65) 63490222; fax: (+65) 67331510; e-mail: JournalsCustomerServiceAPAC@elsevier.com.

Author inquiries: For inquiries relating to the submission of articles (including electronic submission where available), please visit this journal's homepage at <http://www.elsevier.com/locate/ymgme>. You can track accepted articles at <http://www.elsevier.com/trackarticle> and set up e-mail alerts to inform you of when an article's status has changed. Also accessible from here is information on copyright, frequently asked questions, and more. For detailed instructions on the preparation of electronic artwork, please visit <http://www.elsevier.com/artworkinstructions>. Contact details for questions arising after acceptance of an article, especially those relating to proofs, will be provided by the publisher.

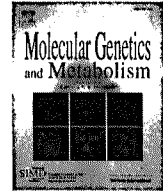
Advertising information: If you are interested in advertising or other commercial opportunities please e-mail Commercialsales@elsevier.com and your enquiry will be passed to the correct person who will respond to you within 48 hours.

Printed in the United States of America

Ⓢ The paper used in this publication meets the requirements of ANSI/NISO Z39.48-1992 (Permanence of Paper)

Sponsored supplements and/or commercial reprints: For more information, please contact Elsevier Life Sciences Commercial Sales, Radarweg 29, 1043 NX Amsterdam, The Netherlands; phone: (+31) (20) 4852939/2059; e-mail: LSCS@elsevier.com.

L I P I N E X 1016



Volume 105, Issues 3, March 2012

Special Issue: SIMD Abstracts for 2012

CONTENTS

Abstract/indexed in: BIOSIS, Chemical Abstracts, Current Contents/Life Sciences, EMBASE, EMBiology, MEDLINE®, and Science Citation Index. Also covered in the abstract and citation database SciVerse Scopus®. Full text available on SciVerse ScienceDirect®

SPECIAL SECTION: PROGRAM AND ABSTRACTS FOR THE ANNUAL MEETING OF THE SOCIETY FOR INHERITED METABOLIC DISORDERS

273 Program for SIMD annual meeting: March 31–April 3, 2012, The Westin Charlotte, Charlotte, NC

EDITORIAL FROM THE SIMD BOARD OF DIRECTORS

367 SIMD statement on investigational new drugs for rare disease therapies
Nicola Longo

MINIREVIEW

368 Treatable inborn errors of metabolism causing intellectual disability: A systematic literature review
Clara D.M. van Karnebeek, Sylvia Stockler

REGULAR ARTICLES

- 382 **Recombination mapping using Boolean logic and high-density SNP genotyping for exome sequence filtering**
Thomas C. Markello, Ted Han, Hannah Carlson-Donohoe, Chidi Ahaghotu, Ursula Harper, MaryPat Jones, Settara Chandrasekharappa, Yair Anikster, David R. Adams, NISC Comparative Sequencing Program, William A. Gahl, Cornelius F. Boerkoel
- 390 **Diet in phenylketonuria: A snapshot of special dietary costs and reimbursement systems in 10 international centers**
A. Belanger-Quintana, K. Dokoupil, H. Gokmen-Ozel, A.M. Lammardo, A. MacDonald, K. Motzfeldt, M. Nowacka, M. Robert, M. van Rijn, K. Ahring
- 396 **Long-term betaine therapy in a murine model of cystathionine beta-synthase deficient homocystinuria: Decreased efficacy over time reveals a significant threshold effect between elevated homocysteine and thrombotic risk**
Kenneth N. Maclean, Hua Jiang, Lori S. Greiner, Robert H. Allen, Sally P. Stabler

- 404 **Impact of enzyme activity assay on indication in liver transplantation for ornithine transcarbamylase deficiency**
Taiichi Wakiya, Yukihiko Sanada, Taizen Urahashi, Yoshiyuki Ihara, Naoya Yamada, Noriki Okada, Kentaro Ushijima, Shinya Otomo, Koichi Sakamoto, Kei Murayama, Masaki Takayanagi, Kenichi Hakamada, Yoshikazu Yasuda, Koichi Mizuta
- 408 **Exploring the transcriptomic variation caused by the Finnish founder mutation of lysinuric protein intolerance (LPI)**
Maaria Tringham, Johanna Kurko, Laura Tanner, Johannes Tuikkala, Olli S. Nevalainen, Harri Niinikoski, Kirsti Nântö-Salonen, Marja Hietala, Olli Simell, Juha Mykkänen
- 416 **Combined extraction of acyl carnitines and 26:0 lysophosphatidylcholine from dried blood spots: Prospective newborn screening for X-linked adrenoleukodystrophy**
Yana Slanders, Ann B. Moser, Walter C. Hubbard, Lisa E. Kratz, Richard O. Jones, Gerald V. Raymond
- 421 **Enzymatic activity and genetic variation in SCD1 modulate the relationship between fatty acids and inflammation**
Carolina Stryjecki, Kaitlin Roke, Shannon Clarke, Daiva Nielsen, Alaa Badawi, Ahmed El-Sohemy, David W.L. Ma, David M. Mutch
- 428 **Early cognitive development in children with infantile Pompe disease**
Gail A. Spiridigliozzi, James H. Heller, Laura E. Case, Harrison N. Jones, Priya S. Kishnani
- 433 **Fanconi-Bickel syndrome: GLUT2 mutations associated with a mild phenotype**
Sarah Catharina Grünert, Karl Otfried Schwab, Martin Pohl, Jörn Oliver Sass, René Santer
- 438 **Morquio A syndrome due to Maternal Uniparental Isodisomy of the telomeric end of chromosome 16**
S. Catarzi, L. Giunti, F. Papadia, O. Gabrielli, R. Guerrini, M.A. Donati, M. Genuardi, A. Morrone
- 443 **Anti- α -galactosidase A antibody response to agalsidase beta treatment: Data from the Fabry Registry**
William R. Wilcox, Gabor E. Linthorst, Dominique P. Germain, Ulla Feldt-Rasmussen, Stephen Waldek, Susan M. Richards, Dana Beitner-Johnson, Marta Cizmarik, J. Alexander Cole, Wytke Kingma, David G. Warnock
- 450 **Lysosomal lipase deficiency: Molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease**
Tommaso Fasano, Livia Pisciotta, Letizia Bocchi, Ornella Guardamagna, Paola Assandro, Claudio Rabacchi, Paolo Zanoni, Mirella Filocamo, Stefano Bertolini, Sebastiano Calandra
- 457 **Non-invasive evaluation of buccal respiratory chain enzyme dysfunction in mitochondrial disease: Comparison with studies in muscle biopsy**
Michael J. Goldenthal, Teddy Kuruvilla, Shirish Damle, Leon Salganicoff, Sudip Sheth, Nidhi Shah, Harold Marks, Divya Khurana, Ignacio Valencia, Agustin Legido
- 463 **Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations**
Valerio Leoni, Laura Strittmatter, Giovanna Zorzi, Federica Zibordi, Sabrina Dusi, Barbara Garavaglia, Paola Venco, Claudio Caccia, Amanda L. Souza, Amy Deik, Clary B. Clish, Marco Rimoldi, Emilio Ciusani, Enrico Bertini, Nardo Nardocci, Vamsi K. Mootha, Valeria Tiranti
- 472 **ALS patients with mutations in the SOD1 gene have an unique metabolomic profile in the cerebrospinal fluid compared with ALS patients without mutations**
Anna Wuolikainen, Peter M. Andersen, Thomas Moritz, Stefan L. Marklund, Henrik Antti
- 479 **A novel splicing mutation causes analbuminemia in a Portuguese boy**
Gianluca Caridi, Monica Dagnino, Marco Di Duca, Helena Pinto, Maria do Céu Espinheira, António Guerra, Susana Fernandes, Monica Campagnoli, Monica Galliano, Lorenzo Minchiotti
- 484 **Dyrk1a activates antioxidant NQO1 expression through an ERK1/2-Nrf2 dependent mechanism**
Christophe Noll, Asma Tlili, Clémentine Ripoll, Ludovic Mallet, Jean-Louis Paul, Jean-Maurice Delabar, Nathalie Janel
- 489 **Genetic association between WNT10B polymorphisms and obesity in a Belgian case-control population is restricted to males**
J.K. Van Camp, S. Beckers, D. Zegers, A. Verrijken, L.F. Van Gaal, W. Van Hul
- 494 **Genetic contribution to C-reactive protein levels in severe obesity**
Geneviève Faucher, Frédéric Guénard, Luigi Bouchard, Véronique Garneau, Valérie Turcot, Alain Houde, André Tchernof, Jean Bergeron, Yves Deshaies, Frédéric-Simon Hould, Stéfane Lebel, Picard Marceau, Marie-Claude Vohl
- 502 **Population variability in CD38 activity: Correlation with age and significant effect of TNF- α -308G>A and CD38 184C>G SNPs**
Valeria Polzonetti, Francesco M. Carpi, Daniela Micozzi, Stefania Pucciarelli, Silvia Vincenzetti, Valerio Napolioni
- 508 **Association study of common variants in the sFRP1 gene region and parameters of bone strength and body composition in two independent healthy Caucasian male cohorts**
Eveline Boudin, Elke Pitters, Erik Franssen, Torben Leo Nielsen, Marianne Andersen, Greet Roef, Youri Taes, Kim Brixen, Wim Van Hul

Explore Litigation Insights

Docket Alarm provides insights to develop a more informed litigation strategy and the peace of mind of knowing you're on top of things.

Real-Time Litigation Alerts



Keep your litigation team up-to-date with **real-time alerts** and advanced team management tools built for the enterprise, all while greatly reducing PACER spend.

Our comprehensive service means we can handle Federal, State, and Administrative courts across the country.

Advanced Docket Research



With over 230 million records, Docket Alarm's cloud-native docket research platform finds what other services can't. Coverage includes Federal, State, plus PTAB, TTAB, ITC and NLRB decisions, all in one place.

Identify arguments that have been successful in the past with full text, pinpoint searching. Link to case law cited within any court document via Fastcase.

Analytics At Your Fingertips



Learn what happened the last time a particular judge, opposing counsel or company faced cases similar to yours.

Advanced out-of-the-box PTAB and TTAB analytics are always at your fingertips.

API

Docket Alarm offers a powerful API (application programming interface) to developers that want to integrate case filings into their apps.

LAW FIRMS

Build custom dashboards for your attorneys and clients with live data direct from the court.

Automate many repetitive legal tasks like conflict checks, document management, and marketing.

FINANCIAL INSTITUTIONS

Litigation and bankruptcy checks for companies and debtors.

E-DISCOVERY AND LEGAL VENDORS

Sync your system to PACER to automate legal marketing.