

Editor
Constantin Polychronakos
(Canada)

Associate Editors

HuiQi Qu (USA)
Diana Eccles (UK)
Nada Jabado (Canada)
Geoffrey Woods (UK)

Guidelines for Authors and Reviewers

Full instructions are available online at <http://jmg.bmj.com/fora>. Articles must be submitted electronically <http://jmg.bmj.com/submit>. Authors retain copyright but are required to grant Journal of Medical Genetics an exclusive licence to publish <http://jmg.bmj.com/licence-forms>

Twitter: @JMG_BMJ

Impact factor: 5.65

Disclaimer: Journal of Medical Genetics is owned and published by BMJ Publishing Group Ltd, a wholly owned subsidiary of the British Medical Association. The owner grants editorial freedom to the Editor of Journal of Medical Genetics.

Journal of Medical Genetics follows guidelines on editorial independence produced by the World Association of Medical Editors and the code on good publication practice of the Committee on Publication Ethics.

Journal of Medical Genetics is intended for medical professionals and is provided without warranty, express or implied. Statements in the journal are the responsibility of their authors and advertisers and not authors' institutions, the BMJ Publishing Group Ltd or the BMA unless otherwise specified or determined by law. Acceptance of advertising does not imply endorsement.

To the fullest extent permitted by law, the BMJ Publishing Group Ltd shall not be liable for any loss, injury or damage resulting from the use of Journal of Medical Genetics or any information in it whether based on contract, tort or otherwise. Readers are advised to verify any information they choose to rely on.

Copyright: © 2016 BMJ Publishing Group Ltd. All rights reserved; 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 the prior permission of Journal of Medical Genetics.

Journal of Medical Genetics is published by BMJ Publishing Group Ltd, typeset by Nova Techset Private Limited, Bengaluru & Chennai, India and printed in the UK on acid-free paper.

JMG Journal of Medical Genetics (ISSN No: 0022-2593) is published monthly by BMJ Publishing Group and is distributed in the USA by Air Business Ltd. Periodicals postage paid at Jamaica NY. 11431 POSTMASTER: Send address changes to Journal of Medical Genetics, Air Business Ltd, c/o Worldnet Shipping Inc., 156-15, 146th Avenue, 2nd Floor, Jamaica, NY 11434, USA.

Journal of Medical Genetics publishes original research and reviews relevant to medical genetics and particularly encourages submissions on the molecular basis of human disease, human cancer genetics, the clinical manifestations of genetics disorders, applications of molecular genetics to medical practice and the systematic evaluation of such applications

Editorial Committee

Stylianos Antonarakis (Switzerland) **Kari E North** (USA)
François Balloux (UK) **Peristera Paschou** (Greece)
Han Brunner (Netherlands) **Christopher E Pearson** (Canada)
Jean-Laurent Casanova (France) **Nicky Ragge** (UK)
David Collier (UK) **Brent Richards** (Canada)
David Cooper (UK) **Dan Rujescu** (Germany)
Robert C Elston (Australia) **Ravi Savarirayan** (Australia)
Asta Försti (Germany) **Chee-Sung Ku** (Sweden)
William Foulkes (Canada) **Helga Toriello** (USA)
Andrew Griffith (USA) **Massimo Zeviani** (Italy)
Hakon Hakonarson (Iceland) **Xue Zhang** (China)
Andrea Heinzmann (Germany)
Eric Legius (Belgium)

Stay a step ahead with Online First

We publish all our original articles online before they appear in a print issue. This means that the latest clinical research papers go straight from acceptance to your browser, keeping you at the cutting edge of Clinical Genetics. We update the site weekly so ensure it's as topical as possible. Find Online First articles at <http://jmg.bmj.com/onlinefirst.dtl>

Subscription Information

Journal of Medical Genetics is published monthly (subscribers receive all supplements)

Institutional Rates 2016

Print
£738; US\$1440; €997

Online
Site licences are priced on FTE basis and allow access by the whole institution.

Personal Rates 2016

Print (includes online access at no additional cost)
£314; US\$613; €424

Online only
£170; US\$332; €230

ISSN: 0022-2593 (print)
ISSN: 1468-6244 (online)

Personal print or online only and institutional print subscriptions may be purchased online at <http://group.bmj.com/group/subs-sales/subscriptions> (payment by Mastercard/Visa only).

Residents of some EC countries must pay VAT; for details call us or visit <http://group.bmj.com/group/subs-sales/subscriptions/subs-vat>

Contact Details

Editorial Office

BMJ Publishing Group Ltd, BMA House, Tavistock Square, London WC1H 9JR, UK
T: +44 (0)20 7383 6318
E: jmg@bmj.com

Production Editor

Laura Buchan
T: +44 (0)20 7874 0807
E: production.jmg@bmj.com

Permissions

<http://group.bmj.com/permissions>

Supplement Enquiries

T: +44 (0)20 7383 6057
E: journals@bmj.com

Subscriptions (except USA)

T: +44 (0)20 7111 1105
<http://support.bmj.com>

US Subscriptions

PP&F PO Box 361
Birmingham, AL 35201-0361
T: +1 800 348 6473 (toll free in the USA)
E: bmj-clinicalevidence@ebSCO.com

Display Advertising Sales

Sophie Fitzsimmons (Sales Manager)
T: +44 (0)20 7383 6783
E: sfitzsimmons@bmj.com
<http://group.bmj.com/group/advertising>

Online Advertising Sales

Marc Clifford (Sales Manager)
T: +44 (0)20 7383 6161
E: mclifford@bmj.com
<http://group.bmj.com/group/advertising>

Display & Online Advertising Sales (USA)

Jim Cunningham
T: +1 201 767 4170
E: jcunningham@cunnasso.com

Author Reprints

Reprints Administrator
E: admin.reprints@bmj.com

Commercial Reprints (except USA & Canada)

Nadia Gurney-Randall
T: +44 (0)20 8445 5825
M: +44 (0)7866 262344
E: ngurneyrandall@bmj.com

Commercial Reprints (USA & Canada)

Ray Thibodeau
T: +1 267 895 1758
M: +1 215 933 8484
E: ray.thibodeau@contentednet.com

For all other JMG journal contacts
<http://jmg.bmj.com/site/help/index.xhtml>