

**Editor**  
Constantin Polychronakos  
(Canada)

**Associate Editors**  
HuiQi Qu (USA)  
Diana Eccles (UK)  
Nada Jabado (Canada)  
Enza-Maria Valente (Italy)  
Suri Mohnish (UK)

**Guidelines for Authors and Reviewers**

Full instructions are available online at <http://jmg.bmj.com/pages/authors>. 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://authors.bmj.com/submitting-your-paper/copyright-and-authors-rights>).

Twitter: @JMG\_BMJ

**Impact factor:** 5.451

**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:** © 2018 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 Exeter Premedia Services Private Ltd, 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

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

## 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/content/early/recent>

## Subscription Information

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

### Institutional Rates 2018

**Print**  
£814

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

### Personal Rates 2018

**Print** (includes online access at no additional cost)  
£347

**Online only**  
£188

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

Personal print or online only and institutional print subscriptions may be purchased online at <http://journals.bmj.com/content/subscribers> (payment by Mastercard/Visa only).

Residents of some EC countries must pay VAT; for details call us or visit <http://journals.bmj.com/content/subscribers>

## 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](mailto:jmg@bmj.com)

### Production Editor

Tommaso Olivero  
T: +44 (0)20 7874 0737  
E: [production.jmg@bmj.com](mailto:production.jmg@bmj.com)

### Permissions

<http://www.bmj.com/company/products-services/rights-and-licensing/permissions>

### Supplement Enquiries

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

### Subscriptions

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

### Display Advertising Sales

Sophie Fitzsimmons (Sales Manager)  
T: +44 (0)20 7383 6783  
E: [sfitzsimmons@bmj.com](mailto:sfitzsimmons@bmj.com)  
<http://www.bmj.com/company/raise-visibility-and-reach>

### Online Advertising Sales

Marc Clifford (Sales Manager)  
T: +44 (0)20 7383 6161  
E: [mclifford@bmj.com](mailto:mclifford@bmj.com)  
<http://www.bmj.com/company/raise-visibility-and-reach>

### Display & Online Advertising Sales (USA)

American Medical Communications (AMC)  
Ron Gordon (National Accounts Manager)  
T: +1 973 214 4374  
E: [rgordon@americanmedicalcomm.com](mailto:rgordon@americanmedicalcomm.com)

### Author Reprints

Reprints Administrator  
E: [admin.reprints@bmj.com](mailto:admin.reprints@bmj.com)

### Commercial Reprints (except USA & Canada)

Nadia Gurney-Randall  
T: +44 (0)20 8445 5825  
M: +44 (0)7866 262 344  
E: [ngurneyrandall@bmj.com](mailto:ngurneyrandall@bmj.com)

### Commercial Reprints (USA & Canada)

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

**For all other JMG journal contacts**  
<http://jmg.bmj.com/pages/contact-us/>