

**Editor in Chief**  
Huw Dorkins (UK)

**Associate Editors**  
Constantin Polychronakos (Canada)  
HuiQi Qu (USA)  
Nada Jabado (Canada)  
Enza-Maria Valente (Italy)  
Mohnish Suri (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: 4.943

**Disclaimer:** The Editor of *Journal of Medical Genetics* has been granted editorial freedom and *Journal of Medical Genetics* is published in accordance with editorial guidelines issued by the World Association of Medical Editors and the Committee on Publication Ethics. *Journal of Medical Genetics* is primarily intended for healthcare professionals and its content is for information only. The Journal is published without any guarantee as to its accuracy or completeness and any representations or warranties are expressly excluded to the fullest extent permitted by law. Readers are advised to independently verify any information on which they choose to rely. Acceptance of advertising by *Journal of Medical Genetics* does not imply endorsement. BMJ Publishing Group Limited shall not be liable for any loss, injury or damage howsoever arising from *Journal of Medical Genetics* (except for liability which cannot be legally excluded).

**Copyright:** © 2020 BMJ Publishing Group Ltd. All rights reserved; no part of this publication may be reproduced in any form without permission.

*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 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 Board

<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 2020

**Print**  
£898

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

### Personal Rates 2020

**Print** (includes online access at no additional cost)  
£384

**Online only**  
£208

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  
E: [jmg@bmj.com](mailto:jmg@bmj.com)

### Production Editor

Tommaso Olivero  
E: [production.jmg@bmj.com](mailto:production.jmg@bmj.com)

### Customer support

For general queries and support with existing and new subscriptions:  
W: [support.bmj.com](mailto:support.bmj.com)  
T: +44 (0)20 7111 1105  
E: [support@bmj.com](mailto:support@bmj.com)

### Self-archiving and permissions

W: [bmj.com/company/products-services/rights-and-licensing/](http://bmj.com/company/products-services/rights-and-licensing/)  
E: [bmj.permissions@bmj.com](mailto:bmj.permissions@bmj.com)

### Advertising

W: [bmj.com/company/for-advertisers-and-sponsor/](http://bmj.com/company/for-advertisers-and-sponsor/)

### Display Advertising ROW

Sophie Fitzsimmons  
T: +44 (0)20 3655 5612  
E: [sfitzsimmons@bmj.com](mailto:sfitzsimmons@bmj.com)

### Online Advertising ROW

Marc Clifford  
T: +44 (0)20 3655 5610  
E: [mclifford@bmj.com](mailto:mclifford@bmj.com)

### Display & Online Advertising Americas

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

### Author Reprints

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

### Commercial Reprints ROW

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

### Commercial Reprints Americas

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/>