

# Journal of Medical Genetics

## AIMS AND SCOPE

The *Journal of Medical Genetics* publishes original research and reviews relevant to medical genetics and acts as a forum for discussion, debate, and information exchange through its correspondence column. The journal 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. The journal attempts to handle the review process and publication as expeditiously as possible. The review process is usually completed within 6 weeks, but can take longer in some instances. Accelerated publication is available where warranted by scientific urgency and importance.

## EDITOR

Eamonn R Maher (UK)

## ASSOCIATE EDITORS

Andrew Griffith (Sweden)  
Juha Kere (Sweden)  
Constantin Polychronakos (Canada)  
Guy Van Camp (Belgium)

## EDITORIAL COMMITTEE

L Aaltonen (Finland)  
SE Antonarakis (Switzerland)  
K Brøndum-Nielsen (Denmark)  
H Brunner (The Netherlands)  
J Burn (UK)  
PH Byers (USA)  
DF Callen (Australia)  
G Evers-Kiebooms (Belgium)  
RJM Gardner (Australia)  
RJ Gorlin (USA)  
AG Knudson Jr (USA)  
C Larsson (Sweden)  
E Legius (Belgium)  
Q Mehdi (Pakistan)  
A Munnich (France)  
S Narod (Canada)  
B Ponder (UK)  
R Savarirayan (Australia)  
KA Schneider (USA)  
T Strachan (UK)  
R Trembath (UK)  
H Torriello (USA)  
M Warman (USA)  
M Zeviani (Italy)  
Editor, *BMJ*

## CONTACT DETAILS

### Editorial Office

Editorial Assistant  
Journal of Medical Genetics  
BMJ Publishing Group Ltd  
BMA House  
Tavistock Square  
London WC1H 9JR, UK  
Tel: +44 (0)20 7383 6292  
Fax: +44 (0)20 7383 6668  
Email: [jmg@bmjgroup.com](mailto:jmg@bmjgroup.com)

### Permissions

See <http://journals.bmj.com/misc/permissions.dtl>

### Disclaimer

The *Journal of Medical Genetics* is published by BMJ Publishing Group Ltd, a wholly owned subsidiary of the British Medical Association.

The BMA grants editorial freedom to the Editor of the *Journal of Medical Genetics*. The *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.

The *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 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 shall not be liable for any loss, injury or damage resulting from the use of the *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.

## Supplement Enquiries

Gavin Stewart, Development Editor  
Tel: +44 (0)20 7383 6170  
Fax: +44 (0)20 7383 6668  
Email: [gstewart@bmjgroup.com](mailto:gstewart@bmjgroup.com)

## Subscriptions (except USA)

Subscription Manager, BMJ Journals  
BMJ Publishing Group Ltd  
PO BOX 299  
London WC1H 9TD, UK  
Tel: +44 (0)20 7383 6270  
Fax: +44 (0)20 7383 6402  
Email: [subscriptions@bmjgroup.com](mailto:subscriptions@bmjgroup.com)  
<http://jmg.bmj.com/subscriptions>

## US Subscriptions

BMJ Publishing Group  
PO Box 281  
Annapolis Junction, MD 20701-0281, USA  
Tel: +1 800 348 6473 toll free  
Fax: +1 801 206 9789  
Email: [bmjpg@pmds.com](mailto:bmjpg@pmds.com)

## Advertising

Advertising Manager, BMJ Journals  
Tel: +44 (0)20 7383 6181  
Fax: +44 (0)20 7383 6556  
Email: [ecurrer@bmjgroup.com](mailto:ecurrer@bmjgroup.com)  
[www.bmjpg.com/advertising](http://www.bmjpg.com/advertising)

## Author reprints

Sheila Williams  
Tel: +44 (0)20 7383 6305  
Fax: +44 (0)20 7383 6699  
Email: [swilliams@bmjgroup.com](mailto:swilliams@bmjgroup.com)

## Commercial Reprints (except USA & Canada)

Nadia Gurney-Randall  
Tel: +44 (0)20 8445 5825  
Fax: +44 (0)20 8445 5870  
Email: [ngurneyrandall@bmjgroup.com](mailto:ngurneyrandall@bmjgroup.com)

## Commercial Reprints (USA & Canada)

Marsha Fogler  
PO Box 3227  
Cherry Hill, NJ 08034, USA  
Tel: +1 800 482 1450  
Fax: +1 609 482 7414  
Email: [fogler@erols.com](mailto:fogler@erols.com)

## SUBSCRIPTION INFORMATION

*Journal of Medical Genetics* is published monthly (subscribers receive all supplements)  
ISSN: 0022 2593 (print); 1468 6244 (online)

## INSTITUTIONAL RATES

Print – 2007 rates: £414; US\$766; €613  
Online – Site licences are priced on FTE basis and allow access by the whole institution. Print is available at deeply discounted rates for online subscribers; details available online at <http://journals.bmj.com/subscriptions> or contact the Subscription Manager in the UK (see earlier)

## PERSONAL RATES

Print (includes online access at no additional cost) – 2007 rates: £177; US\$327; €262  
Online only – 2007 rates: £95; US\$176; €141

## HOW TO SUBSCRIBE

- Subscribers may pay by cheque\*, Switch, or credit card (Mastercard, Visa, American Express)
- Orders may be placed with any leading subscription agent or bookseller
- Call our subscription hotline on +44 (0)20 7383 6270; fax hotline +44 (0)20 7383 6402
- All enquiries and single copy sales should be addressed to the UK office
- Personal print or online only and institutional print subscriptions may be purchased online at <http://jmg.bmj.com/subscriptions> (payment by Visa/MasterCard only)

\*UK cheques must be drawn on a UK bank account; US cheques must be drawn on a US bank

Residents of some EC countries and Canada must pay VAT; for details, call us or visit <http://journals.bmj.com/subscriptions/cost2007.dtl>

## COPYRIGHT

© 2007 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 the publisher.

Authors are required to grant the *Journal of Medical Genetics* an exclusive licence to publish; further details available online at <http://jmg.bmj.com/ifora>

- *Journal of Medical Genetics* is published by the BMJ Publishing Group Ltd, copyedited by Macmillan India Ltd, typeset by The Charlesworth Group, and printed in UK on acid-free paper by Latimer Trend & Co. Limited, Plymouth.
- Periodicals postage paid, Rahway, NJ. Postmaster: send address changes to: *Journal of Medical Genetics*, c/o Mercury Airfreight International Ltd, 365 Blair Road, Avenel, NJ 07001, USA



[www.publicationethics.org.uk](http://www.publicationethics.org.uk)

This journal is a member of and subscribes to the principles of the Committee on Publication Ethics