

## Contents

### Genotype-phenotype correlations

**503** Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families

*S Castro-Sánchez, M Álvarez-Satta, M Cortón, E Guillén, C Ayuso, D Valverde*

**514** Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity

*R Bachmann-Gagescu, J C Dempsey, I G Phelps, B J O'Roak, D M Knutzen, T C Rue, G E Ishak, C R Isabella, N Gordon, J Adkins, E A Boyle, N de Lacy, D O'Day, A Alswaid, R Ramadevi A, L Lingappa, C Lourenço, L Mantorell, Á Garcia-Cazorla, H Ozyürek, G Haliloğlu, B Tuysuz, M Topçu, University of Washington Center for Mendelian Genomics, P Chance, M A Parisi, I A Glass, J Shendure, D Doherty*

### New loci

**523** Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder

*L Zong, J Guan, M Ealy, Q Zhang, D Wang, H Wang, Y Zhao, Z Shen, C A Campbell, F Wang, J Yang, W Sun, L Lan, D Ding, L Xie, Y Qi, X Lou, X Huang, Q Shi, S Chang, W Xiong, Z Yin, N Yu, H Zhao, J Wang, J Wang, R J Salvi, C Petit, R J H Smith, Q Wang*

**532** Mutations in the mitochondrial cysteinyl-tRNA synthase gene, *CARS2*, lead to a severe epileptic encephalopathy and complex movement disorder

*C R CoughlinII, G H Scharer, M W Friederich, H-C Yu, E A Geiger, G Creadon-Swindell, A E Collins, A V Vanlander, R V Coster, C A Powell, M A Swanson, M Minczuk, J L K V Hove, T H Shaikh*

**541** Mutations in *SLC1A4*, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination

*N Damseh, A Simonin, C Jalas, J A Picoraro, A Shaag, M T Cho, B Yaacov, J Neidich, M Al-Ashhab, J Juusola, S Bale, A Telegrafi, K Retterer, J G Pappas, E Moran, J Cappell, K A Yeboa, B Abu-Libdeh, M A Hediger, W K Chung, O Elpeleg, S Edvardson*

**548** A mutation of *MET*, encoding hepatocyte growth factor receptor, is associated with human *DFNB97* hearing loss  
*G Mujtaba, J M Schultz, A Imtiaz, R J Morell, T B Friedman, S Naz*

### Cancer genetics

**553** Lynch syndrome caused by *MLH1* mutations is associated with an increased risk of breast cancer: a cohort study  
*E F Harkness, E Barrow, K Newton, K Green, T Clancy, F Laloo, J Hill, D G Evans*

**557** Multiple synchronous sites of origin of vestibular schwannomas in neurofibromatosis Type 2  
*S M Stivaros, A O Stemmer-Rachamimov, R Alston, S R Plotkin, J B Nadol, A Quesnel, J O'Malley, G A Whitfield, M G McCabe, S R Freeman, S K Lloyd, N B Wright, J-P Kilday, I D Kamaly-Asl, S J Mills, S A Rutherford, A T King, D G Evans*

**563** Hereditary diffuse gastric cancer syndrome: improved performances of the 2015 testing criteria for the identification of probands with a *CDH1* germline mutation  
*P R Benusiglio, C Colas, E Rouleau, N Uhrhammer, P Romero, A Remenieras, J Moretta, Q Wang, A De Pauw, B Buecher, D Stoppa-Lyonnet, E Mouret-Fourme, C Noguès, M D Maria, C Tlemsani, M Warcoin, S Grandjouan, D Malka, O Caron, M Blayau*

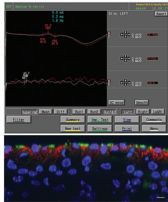
### Ethics and policy

**566** Streamlining review of research involving humans: Canadian models  
*Ma'n H Zawati, A Junker, B M Knoppers, V Rahimzadeh*

**571** Potential research participants support the return of raw sequence data  
*A Middleton, C F Wright, K I Morley, E Bragin, H V Firth, M E Hurles, M Parker, on behalf of the DDD study*

August 2015 Volume 52 Issue 8

**JMG**  
Journal of Medical Genetics



[jmg.bmj.com](http://jmg.bmj.com)



**BMJ**

Cover credit: Somatosensory evoked potentials in AIFM1 mutations and the localization of the gene in the inner ear, from Zong *et al*, pg 523.



Adopted as the official Journal of the Canadian College of Medical Geneticists

Receive regular table of contents by email. Register using this QR code.



This article has been chosen by the Editor to be of special interest or importance and is freely available online.



This article has been made freely available online under the BMJ Journals Open Access scheme. See <http://jmg.bmj.com/site/about/guidelines.xhtml#open>

**C O P E** COMMITTEE ON PUBLICATION ETHICS

This journal is a member of and subscribes to the principles of the Committee on Publication Ethics  
<http://publicationethics.org/>



**MCQs** The online version of this article contains multiple choice questions hosted on BMJ Learning.