

Contents

Neurogenetics

- 73** Recurrent copy number variations as risk factors for neurodevelopmental disorders: critical overview and analysis of clinical implications *F Torres, M Barbosa, P Maciel*

Methods

- 91** CRISPR-Cas9 for medical genetic screens: applications and future perspectives *H-Y Xue, L-J Ji, A-M Gao, P Liu, J-D He, X-J Lu*
- 98** A targeted next-generation sequencing assay for the molecular diagnosis of genetic disorders with orodental involvement *M K Prasad, V Geoffroy, S Vicaire, B Jost, M Dumas, S Le Gras, M Switala, B Gasse, V Laugel-Haushalter, M Paschaki, B Leheup, D Droz, A Dalstein, A Loing, B Grollemund, M Muller-Bolla, S Lopez-Cazaux, M Minoux, S Jung, F Obry, V Vogt, J-L Davideau, T Davit-Beal, A-S Kaiser, U Moog, B Richard, J-J Morrier, J-P Duprez, S Odent, I Baillieux-Forestier, M M Rousset, L Meramedjian, A Toutain, C Joseph, F Giuliano, J-C Dahlet, A Courval, M El Alloussi, S Laouina, S Soskin, N Guffon, A Dieux, B Doray, S Feierabend, E Ginglinger, B Fournier, M de la Dure Molla, Y Alembik, C Tardieu, F Clauss, A Berdal, C Stoetzel, M C Manière, H Dollfus, A Bloch-Zupan*
- 111** WGSa: an annotation pipeline for human genome sequencing studies *X Liu, S White, B Peng, A D Johnson, J A Brody, A H Li, Z Huang, A Carroll, P Wei, R Gibbs, R J Klein, E Boerwinke*

- 123** Paraspinal neurofibromas and hypertrophic neuropathy in Noonan syndrome with multiple lentigines *E Conboy, R Dhamija, M Wang, J Xie, P J Dyck, A G Bridges, R J Spinner, A C Clayton, R E Watson, L Messiaen, D Babovic-Vuksanovic*
- 127** Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygous *OPA1* mutation *R Spiegel, A Saada, P J Flannery, F Burté, D Soiferman, M Khayat, V Eisner, E Vladovski, R W Taylor, L A Bindoff, A Shaag, H Mandel, O Schuler-Furman, S A Shalev, O Elpeleg, P Yu-Wai-Man*

New loci

- 132** Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the *Hikeshi* gene *S Edvardson, S Kose, C Jalas, A Fattal-Valevski, A Watanabe, Y Ogawa, H Mamada, A M Fedick, S Ben-Shachar, N R Treff, A Shaag, S Bale, J Gärtner, N Imamoto, O Elpeleg*
- 138** Homozygous missense mutation in the *LMAN2L* gene segregates with intellectual disability in a large consanguineous Pakistani family *R Rafiullah, M Aslamkhan, N Paramasivam, C Thiel, G Mustafa, S Wiemann, M Schlesner, R C Wade, G A Rappold, S Berkel*

Genotype-phenotype correlations

- 113** Mutation in cytochrome b gene of mitochondrial DNA in a family with fibromyalgia is associated with NLRP3-inflammasome activation *M D Cordero, E Alcocer-Cómez, F Marín-Aguilar, T Rybkina, D Cotán, A Pérez-Pulido, J M Alvarez-Suarez, M Battino, J A Sánchez-Alcazar, A M Carrión, O Culic, J M Navarro-Pando, P Bullón*



Cover credit: Paraspinal neurofibromas in Noonan syndrome from Babovic-Vuksanovic *et al.*, pg 123.



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

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



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

OPEN ACCESS 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 This journal is a member of and subscribes to the principles of the Committee on Publication Ethics <http://publicationethics.org/>
Member since 2008
JMG0012

equator network

recycle
When you have finished with this please recycle it

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