



## Contents

March 2020 Volume 57 Issue 3

### Clinical guidelines

- 145** Predictability and inconsistencies of cognitive outcome in patients with phenylketonuria and personalised therapy: the challenge for the future guidelines  
*V Leuzzi, F Chiarotti, F Nardecchia, D van Vliet, F J van Spronsen*

### Genotype-phenotype correlations

- 151** Myoclonic epilepsy, parkinsonism, schizophrenia and left-handedness as common neuropsychiatric features in 22q11.2 deletion syndrome  
*M Fanella, M Frascarelli, C Lambiase, A Morano, M Unolt, N Liberati, J Fattouch, A Buzzanca, T Accinni, M Ceccanti, A Viganò, M Biondi, C Colonnese, A T Giallonardo, F Di Fabio, A Pizzuti, C Di Bonaventura, A Berardelli*

### Diagnostics

- 160** Increasing knowledge in *IGF1R* defects: lessons from 35 new patients  
*E Giabicani, M Willems, V Steumou, S Chantot-Bastaraud, N Thibaud, W Abi Habib, S Azzi, B Lam, L Bérard, H Bony-Trifunovic, C Brachet, E Brischoux-Boucher, E Caldagues, R Coutant, M-L Cuvelier, G Gelwane, I Guemas, M Houang, B Isidor, C Jeandel, J Lespinasse, C Naud-Saudreau, M Jesuran-Perelroizen, L Perrin, J Piard, C Sechter, P-F Souchon, C Storey, D Thomas, Y Le Bouc, S Rossignol, I Netchine, F Brioude*

- 169** Genetic aetiology of early infant deaths in a neonatal intensive care unit  
*L Yang, X Liu, Z Li, P Zhang, B Wu, H Wang, L Hu, G Cheng, L Wang, W Zhou*

### Neurogenetics

- 178** Distal hereditary motor neuronopathy of the Jerash type is caused by a novel *SIGMAR1* c.500A>T missense mutation  
*A Ververis, R Dajani, P Koutsou, A Aloqaily, C Nelson-Williams, E Loring, A Arafat, A F Mubaidin, K Horany, M B Bader, Y Al-Baho, B Ali, A Muhtaseb, T DeSprenza Jr, A A Al-Qudah, L T Middleton, E Zamba-Papanicolaou, R Lifton, K Christodoulou*

### Gametes

- 187** Homozygous mutations in *REC114* cause female infertility characterised by multiple pronuclei formation and early embryonic arrest  
*W Wang, J Dong, B Chen, J Du, Y Kuang, X Sun, J Fu, B Li, J Mu, Z Zhang, Z Zhou, Z Lin, L Wu, Z Yan, X Mao, Q Li, L He, L Wang, Q Sang*

### Novel disease loci

- 195** Linked-read genome sequencing identifies biallelic pathogenic variants in *DONSON* as a novel cause of Meier-Gorlin syndrome  
*K M Knapp, R Sullivan, J Murray, G Gimenez, P Arn, P D'Souza, A Gezirici, W G Wilson, A P Jackson, C Ferreira, L S Bicknell*

### Cancer genetics

- 203** *NEK11* as a candidate high-penetrance melanoma susceptibility gene  
*E Christodoulou, R van Doorn, M Visser, A Teunisse, M Versluis, P van der Velden, N K Hayward, A Jochemsen, N Gruis*

### Somatic mosaicism

- 212** Postzygotic mosaicism in cerebral cavernous malformation  
*M Rath, A Pagenstecher, A Hoischen, U Felber*

Cover credit: Clinical characteristics of DONSON-MGORS individuals. See Knapp *et al*, page 199.



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



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://authors.bmj.com/open-access/>



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



When you have finished with this please recycle it

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