



Cover credit: Missense and in frame pathogenic variants in CDAN1 and C15orf41 causing Congenital dyserythropoietic anaemia type I. See Olijnik *et al*, page 191.



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

## Contents

### Cancer genetics

- 145** Integrated case-control and somatic-germline interaction analyses of soft-tissue sarcoma  
*F Hu, Y Yu, J-S Chen, H Hu, P Scheet, C D Huff*

### Novel disease loci

- 155** Mutation in the MICOS subunit gene *APOO* (MIC26) associated with an X-linked recessive mitochondrial myopathy, lactic acidosis, cognitive impairment and autistic features  
*C Benincá, V Zanette, M Brischigliaro, M Johnson, A Reyes, D A do Valle, A J Robinson, A Degiorgi, A Yeates, B A Telles, J Prudent, E Baruffini, M L S F Santos, R L R de Souza, E Fernandez-Vizarra, A J Whitworth, M Zeviani*

- 168** Homozygous variants in *SYCP2L* cause premature ovarian insufficiency  
*W-B He, C Tan, Y-X Zhang, L-L Meng, F Gong, G-X Lu, G Lin, J Du, Y-Q Tan*

### Epigenetics

- 173** Paternal 132 bp deletion affecting *KCNQ1OT1* in 11p15.5 is associated with growth retardation but does not affect imprinting  
*T Eggermann, F Kraft, E Lausberg, K Ergezinger, E Kunstmann*

### Diagnostics

- 178** Improved molecular detection of mosaicism in Beckwith-Wiedemann Syndrome  
*S W Baker, K A Duffy, J Richards-Yutz, M A Deardorff, J M Kalish, A Ganguly*

## March 2021 Volume 58 Issue 3

### Genotype-phenotype correlations

- 185** Genetic and functional insights into CDA-I prevalence and pathogenesis  
*A-A Olijnik, N B A Roy, C Scott, J A Marsh, J Brown, K Lauschke, K Ask, N Roberts, D J Downes, S Brolih, E Johnson, B Xella, M Proven, R Hipkeiss, K Ryan, P Friske, J Mäkk, E-L M Stättin, N Sadasivam, L McIlwaine, Q A Hill, R Renella, J R Hughes, R J Gibbons, A Groth, P J McHugh, D R Higgs, V J Buckle, C Babbs*
- 196** *SETD1B*-associated neurodevelopmental disorder  
*A Roston, D Evans, H Gill, M McKinnon, B Isidor, B Cagné, J Mwenifumbo, C van Karnebeek, J An, S J M Jones, M Farrer, M Demos, M Connolly, W T Gibson, CAUSES Study, EPGEN Study*

### Neurogenetics

- 205** De novo variants in *SHAH1*, encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features  
*J Buratti, L Ji, B Keren, Y Lee, S Booke, S Erdin, S Y Kim, T B Palculic, V Meiner, J H Chae, C G Woods, A Tam, D Héron, F Cong, T Harel*

### Biochemical genetics

- 213** TRAP $\gamma$ -CDG shows asymmetric glycosylation and an effect on processing of proteins required in higher organisms  
*S Dittner-Moormann, C M Lourenco, J Reunert, R Nishinakamura, S S Tanaka, C Werner, V Debus, K-P Zimmer, G Wetzel, H Y Naim, Y Wada, S Rust, T Marquardt*

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

EDITOR'S CHOICE

This article has been made freely available online under the BMJ Journals open access scheme. See <http://authors.bmj.com/open-access/>

OPEN ACCESS

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

**recycle**  
When you have finished with this please recycle it

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