



Cover credit: Clinical and radiological phenotype of the two probands with a novel acromesomelic dysplasia. See Díaz-González *et al*, page 29.



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

## Contents

January 2022 Volume 59 Issue 1

### Neurogenetics

- 1** *NOTCH2NLC*-related disorders: the widening spectrum and genotype–phenotype correlation  
*Y Fan, Y Xu, C Shi*

### Cancer genetics

- 10** Genetic characterisation of sarcomatoid carcinomas reveals multiple novel actionable mutations and identifies *KRAS* mutation as a biomarker of poor prognosis  
*Y Ding, Y Shao, C Na, J C Yin, H Hua, R Tao, Y Jiang, R Hu, X He, C Miao, D Zhu, Z Zhang*

- 18** A germline 1;3 translocation disrupting the *VHL* gene: a novel genetic cause for von Hippel-Lindau  
**OPEN ACCESS**  
*C J Ricketts, C D Vocke, M Lang, X Chen, Y Zhao, B Tran, M Tandon, L S Schmidt, M W Ball, W M Linehan*

- 23** Stay at home: implementation and impact of virtualising cancer genetic services during COVID-19  
*M L Norman, J Malcolmson, S Randall Armel, B Gillies, B Ou, E Thain, J M McCuaig, R H Kim*

### Developmental defects

- 28** Biallelic cGMP-dependent type II protein kinase gene (*PRKG2*) variants cause a novel acromesomelic dysplasia  
*F Díaz-González, S Wadhwa, M Rodriguez-Zabala, S Kumar, M Aza-Carmona, L Senthordi-Montané, M Alonso, I Ahmad, S Zahra, D Kumar, N Kushwah, U Shamim, H Sait, S Kapoor, B Roldán, G Nishimura, A C Offiah, M Faruq, K E Heath*

### Phenotypes

- 39** Phenotypes in adult patients with Rett syndrome: results of a 13-year experience and insights into healthcare transition  
**OPEN ACCESS**  
*A Peron, M P Canevini, F Ghelma, R Arancio, M N Savini, A Vignoli*

### Genome-wide studies

- 46** Linkage analysis identifies an isolated strabismus locus at 14q12 overlapping with *FOXG1* syndrome region  
**OPEN ACCESS**  
*X (Cynthia) Ye, N M Roslin, A D Paterson, C J Lyons, V Pegado, P Richmond, C Shyr, O Fornes, X Han, M Higginson, C J Ross, D Giaschi, C Gregory-Evans, M S Patel, W W Wasserman*

### Genotype-phenotype correlations

- 56** Characteristics of germline mutations in Korean patients with pheochromocytoma/ paraganglioma  
*J H Kim, M J Kim, S H Kong, S J Kim, H Kang, C S Shin, S S Park, K E Lee, M-W Seong*
- 65** WFS1 protein expression correlates with clinical progression of optic atrophy in patients with Wolfram syndrome  
**OPEN ACCESS**  
*K Hu, M Zatyka, D Astuti, N Beer, R P Dias, A Kulkarni, J Ainsworth, B Wright, A Majander, P Yu-Wai-Man, D Williams, T Barrett*

### Diagnostics

- 75** Screening of CNVs using NGS data improves mutation detection yield and decreases costs in genetic testing for hereditary cancer  
*J M Moreno-Cabrera, J del Valle, L Feliubadaló, M Pineda, S González, O Campos, R Cuesta, J Brunet, E Serra, G Capellà, B Gel, C Lázaro*

### Biochemical genetics

- 79** MELAS-associated m.5541C>T mutation caused instability of mitochondrial tRNA<sup>Trp</sup> and remarkable mitochondrial dysfunction  
*K Ji, Y Lin, X Xu, W Wang, D Wang, C Zhang, W Li, Y Zhao, C Yan*
- 88** Dysfunction of *VIPR2* leads to myopia in humans and mice  
*F Zhao, Q Li, W Chen, H Zhu, D Zhou, P S Reinach, Z Yang, M He, A Xue, D Wu, T Liu, Q Fu, C Zeng, J Qu, X Zhou*

### Position statement

- 101** Canadian College of Medical Geneticists (CCMG) points to consider: resuming genetic services in a pandemic—a summary  
*L Chad, A J Dawson, E S-Y Goh*

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

**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

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

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