



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 *FOXP1* 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 Kulkearni, 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^{Pro} 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.