



Cover credit: Study of aggregation and cell area enlargement in cells transfected with a GFAP mutation. See Casasnovas *et al*, page 848.



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

## Contents

### Diagnostics

**783** Case for genome sequencing in infants and children with rare, undiagnosed or genetic diseases

OPEN ACCESS

*D Bick, M Jones, S L Taylor, R J Taft, J Belmont*

**792** CCMG practice guideline: laboratory guidelines for next-generation sequencing

OPEN ACCESS

*S Hume, T N Nelson, M Speevak, E McCready, R Agatep, H Feilottter, J Parboosingh, D J Stavropoulos, S Taylor, T L Stockley, On behalf of Canadian College of Medical Geneticists (CCMG)*

### Cognitive and behavioural genetics

**801** Genetic factors contributing to autism spectrum disorder in Williams-Beuren syndrome

OPEN ACCESS

*M Codina-Sola, M Costa-Roger, D Pérez-García, R Flores, M G Palacios-Verdú, I Cosco, L A Pérez-Jurado*

### Methods

**809** Impact of DNA source on genetic variant detection from human whole-genome sequencing data

OPEN ACCESS

*B Trost, S Walker, S A Haider, W W L Sung, S Pereira, C L Phillips, E J Higginbotham, L J Strug, C Nguyen, A Raajkumar, M J Szego, C R Marshall, S W Scherer*

### Genotype-phenotype correlations

**818** Differential disruption of autoinhibition and defect in assembly of cytoskeleton during cell division decide the fate of human *DIAPH1*-related cytoskeletopathy

*B J Kim, T Ueyama, T Miyoshi, S Lee, J H Han, H-R Park, A R Kim, J Oh, M Y Kim, Y S Kang, D Y Oh, J Yun, S M Hwang, N K D Kim, W-Y Park, S Kitajiri, B Y Choi*

## December 2019 Volume 56 Issue 12

### Biochemical genetics

**828** Intronic *SMCHD1* variants in FSHD: testing the potential for CRISPR-Cas9 genome editing

*R Goossens, M L van den Boogaard, R J L F Lemmers, J Balog, P J van der Vliet, I M Willemsen, J Schouten, I Maggio, N van der Stoep, R C Hoeben, S J Tapscott, N Geijsen, M A F V Gonçalves, S Sacconi, R Tawil, S M van der Maarel*

### Cancer genetics

**838** Clinical features and cancer risk in families with pathogenic *CDH1* variants irrespective of clinical criteria

*R M Xicola, S Li, N Rodriguez, P Reinecke, R Karam, V Speare, M H Black, H LaDuca, X Llor*

**844** Gastric cancer in Lynch syndrome is associated with underlying immune gastritis

*T Adar, M Friedman, L H Rodgers, K M Shannon, L R Zuberberg, D C Chung*

### Neurogenetics

**846** A novel mutation in the *GFAP* gene expands the phenotype of Alexander disease

*C Casasnovas, E Verdura, V Vélez, A Schlüter, A Pons-Escoda, C Homedes, M Ruiz, S Fourcade, N Launay, A Pujol*

### Novel disease loci

**850** Biallelic disruption of *PKDCC* is associated with a skeletal disorder characterised by rhizomelic shortening of extremities and dysmorphic features

*S A Sajan, J Ganesh, D N Shinde, Z Powis, M I Scarano, J Stone, S Winter, S Tang*

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  
JM00012

equator network

recycle  
When you have finished with this please recycle it

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