



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 Feilletter, 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



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

Member since 2008
JM00012

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

equator
network

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.