



Cover credit: Overlapping Rett-syndrome like phenotypes and corresponding mutated genes from, Maciel *et al.*, pg 190.



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

Receive regular table of contents by email. Register using this QR code.



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



This article has been made freely available online under the BMJ Journals Open Access scheme. See <http://jmg.bmj.com/site/about/guidelines.xhtml#open>



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



When you have finished with this please recycle it

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

Contents

Diagnostics

- 145** The importance of genetic diagnosis for Duchenne muscular dystrophy *A Aantsma-Rus, I B Ginjaar, K Bushby*

OPEN ACCESS

Developmental defects

- 152** Deletions and de novo mutations of *SOX11* are associated with a neurodevelopmental disorder with features of Coffin–Siris syndrome *A Hempel, A T Pagnamenta, M Blyth, S Mansour, V McConnell, I Kou, S Ikegawa, Y Tsurusaki, N Matsumoto, A Lo-Castro, G Plessis, B Albrecht, A Battaglia, J C Taylor, M F Howard, D Keays, A S Sohail, DDD collaboration, S J Kühl, U Kini, A McNeill*

Complex traits

- 163** The regulatory element READ1 epistatically influences reading and language, with both deleterious and protective alleles *N R Powers, J D Eicher, L L Miller, Y Kong, S D Smith, B F Pennington, E G Willcutt, R K Olson, S M Ring, J R Gruen*

Cancer genetics

- 172** Low-level *APC* mutational mosaicism is the underlying cause in a substantial fraction of unexplained colorectal adenomatous polyposis cases *I Spier, D Drichel, M Kerick, J Kirfel, S Horpaopan, A Laner, S Holzapfel, S Peters, R Adam, B Zhao, T Becker, R P Lifton, S Perner, P Hoffmann, G Kristiansen, B Timmermann, M M Nöthen, E Holinski-Feder, M R Schweiger, S Aretz*

Genotype-phenotype correlations

- 180** Disruption of Golgi morphology and altered protein glycosylation in *PLA2G6*-associated neurodegeneration *M Davids, M S Kane, M He, L A Wolfe, X Li, M A Raihan, K R Chao, W P Bone, C F Boerkoel, W A Gahl, C Toro*

March 2016 Volume 53 Issue 3

- 190** Identification of novel genetic causes of Rett syndrome-like phenotypes *F Lopes, M Barbosa, A Aneur, G Soares, J de Sá, A I Dias, G Oliveira, P Cabral, T Temudo, E Calado, I F Cruz, J P Vieira, R Oliveira, S Esteves, S Sauer, I Jonasson, A-C Syvänen, U Gyllenstein, D Pinto, P Maciel*

New disease loci

- 200** Identification of a pathogenic *FTO* mutation by next-generation sequencing in a newborn with growth retardation and developmental delay *H Daoud, D Zhang, F McMurray, A Yu, S M Luco, J Vanstone, O Jarinova, N Carson, J Wickens, S Shishodia, H Choi, M A McDonough, C J Schofield, M-E Harper, D A Dymont, C M Armour*

Methods

- 208** Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies *M Schueler, J Halbrinter, I G Phelps, D A Braun, E A Otto, J D Porath, H Y Gee, J Shendure, B J O'Roake, J A Lawson, M M Nabhan, N A Soliman, D Doherty, F Hildebrandt*

Book review

- 216** Molecular insights into development in humans: *Studies in Normal Development and Birth Defects* *L A Jerome-Majewska*