Journal of Medical Genetics

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.

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://jmj.bmj.com/site/about/guidelines.xhtml#open**

COPE 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.

Contents

March 2016 Volume 53 Issue 3

Diagnostics

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

Developmental defects

152 OPEN ACCESS 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 Sohal, DDD collaboration, S J Kühl, U Kini, A McNeill*

Complex traits

163 OPEN ACCESS 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*

190 EDITOR'S CHOICE Identification of novel genetic causes of Rett syndrome-*like* phenotypes *F Lopes, M Barbosa, A Ameur, 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 Gyllensten, 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 Dyment, C M Armour*

Methods

208 Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies *M Schueler, J Halbritter, I G Phelps, D A Braun, E A Otto, J D Porath, H Y Gee, J Shendure, B J O'Roak, 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*

J Med Genet: first published as on 1 March 2016. Downloaded from http://jmg.bmj.com/ on February 2, 2023 by guest. Protected by copyright.