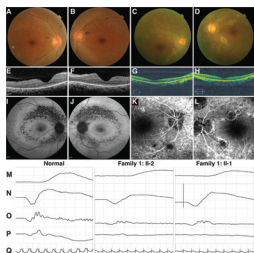


Journal of Medical Genetics

March 2017 Volume 54 Issue 3

JMG

Journal of Medical Genetics



jmg.bmj.com

BMJ

Cover credit: Retinal imaging and electroretinogram from cases of a new type of Usher syndrome due to *CEP78* mutations, from Fu *et al.*, pg. 192.



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

Contents

March 2017 Volume 54 Issue 3

Neurogenetics

- 145** Genetics insight into the amyotrophic lateral sclerosis/frontotemporal dementia spectrum



MCQs A-L Ji, X Zhang, W-W Chen, W-J Huang

Genotype-phenotype correlations

- 155** The importance of dynamic re-analysis in diagnostic whole exome sequencing
- A C Need, V Shashi, K Schoch, S Petrovski, D B Goldstein

Developmental defects

- 157** Chitayat syndrome: hyperphalangism, characteristic facies, hallux valgus and bronchomalacia results from a recurrent c.266A>G p.(Tyr89Cys) variant in the *ERF* gene
- M Balasubramanian, H Lord, S Levesque, H Guturu, F Thuriot, G Sillon, A M Wenger, D L Sureka, T Lester, D S Johnson, J Bowen, A R Calhoun, D H Viskochil, DDD Study, G Bejerano, J A Bernstein, D Chitayat

Functional genomics

- 166** Carriers of a *VEGFA* enhancer polymorphism selectively binding CHOP/DDIT3 are predisposed to increased circulating levels of thyroid-stimulating hormone
- T S Ahluwalia, J T Troelsen, M B-Harder, J B-Jensen, B H Thuesen, C Cerqueira, A Linneberg, N Grarup, O Pedersen, T Hansen, L T Dalgaard

Genome-wide studies

- 176** A novel *TRAPPC11* mutation in two Turkish families associated with cerebral atrophy, global retardation, scoliosis, achalasia and alacrima
- K Koehler, M P Milev, K Prematilake, F Reschke, S Kutzner, R Jühlen, D Landgraf, E Utine, F Hazan, G Diniz, M Schuelke, A Huebner, M Sacher

Genotype-phenotype correlations

- 186** Recessive progressive symmetric erythrokeratoderma results from a homozygous loss-of-function mutation of *KRT83* and is allelic with dominant monilethrix
- K Shah, M Ansar, Z Mughal, F S Khan, W Ahmad, T M Ferrara, R A Spritz

New loci

- 190** *CEP78* is mutated in a distinct type of Usher syndrome
- Q Fu, M Xu, X Chen, X Sheng, Z Yuan, Y Liu, H Li, Z Sun, H Li, L Yang, K Wang, F Zhang, Y Li, C Zhao, R Sui, R Chen
- 196** Mutations in the phosphatidylinositol glycan C (*PIGC*) gene are associated with epilepsy and intellectual disability
- S Edvardson, Y Murakami, T T M Nguyen, M Shahrouh, A St-Denis, A Shaag, N Damseh, F L Deist, Y Bryceson, B Abu-Libdeh, P M Campeau, T Kinoshita, Orly Elpeleg

Screening

- 202** *A de novo* missense mutation of *GABRB2* causes early myoclonic encephalopathy
- OPEN ACCESS** A Ishii, J-Q Kang, C C Schornak, C C Hernandez, W Shen, J C Watkins, R L Macdonald, S Hirose

Somatic mosaicism

- 212** A novel somatic mutation achieves partial rescue in a child with Hutchinson-Gilford progeria syndrome
- OPEN ACCESS** D Z Bar, M F Arlt, J F Brazier, W E Norris, S E Campbell, P Chines, D Larrieu, S P Jackson, F S Collins, T W Glover, L B Gordon

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 JM00012

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.



54 (3)

J Med Genet 2017 54: 145-216

Updated information and services can be found at:

<http://jmg.bmj.com/content/54/3>

These include:

**Email alerting
service**

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:

<http://group.bmj.com/group/rights-licensing/permissions>

To order reprints go to:

<http://journals.bmj.com/cgi/reprintform>

To subscribe to BMJ go to:

<http://group.bmj.com/subscribe/>