

Contents

Review

- 215** Normal and aberrant splicing of *LMNA*
Y-B Luo, Frank L Mastaglia, S D Wilton

New loci

- 224** *NECAP1* loss of function leads to a severe infantile epileptic encephalopathy
A M Alazami, H Hijazi, A Y Kentab, F S Alkuraya

Epigenetics

- 229** Genome-wide DNA methylation analysis of patients with imprinting disorders identifies differentially methylated regions associated with novel candidate imprinted genes
L E Docherty, F I Rezwani, R L Poole, H Jagoe, H Lake, G A Lockett, H Arshad, D I Wilson, J W Holloway, I K Temple, D J G Mackay

Genotype-phenotype correlations

- 239** Truncating mutations in *TAF4B* and *ZMYND15* causing recessive azoospermia
Ö Ayhan, M Balkan, A Guven, R Hazan, M Atar, A Tok, A Tolun

Cancer genetics

- 245** Functional analysis of *MSH2* unclassified variants found in suspected Lynch syndrome patients reveals pathogenicity due to attenuated mismatch repair
E AL Wielders, J Hettinger, R Dekker, C M Kets, M J Ligtenberg, A R Mensenkamp, A MW van den Ouweland, J Prins, A Wagner, W NM Dinjens, H J Dubbink, L P van Hest, F Menko, F Hogervorst, S Verhoef, H te Riele

Chromosomal rearrangements

- 254** A novel immunodeficiency syndrome associated with partial trisomy 19p13
M G Seidel, C Duerr, S Woutsas, A Schwerin-Nagel, K Sadeghi, J Neesen, S Uhrig, E Santos-Valente, W F Pickel, W Schwinger, C Urban, K Boztug, E Förster-Waldl

- 264** Next generation sequencing of chromosomal rearrangements in patients with split-hand/split-foot malformation provides evidence for *DYNC111* exonic enhancers of *DLX5/6* expression in humans
H L Allen, R Caswell, W Xie, X Xu, C Wragg, P D Turnpenny, C L S Turner, M N Weedon, S Ellard

Phenotypes

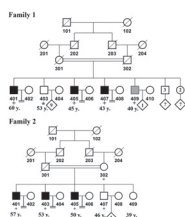
- 268** Conotruncal malformations and absent thymus due to a deleterious *NKX2-6* mutation
A Ta-Shma, N El-lahham, S Edvardson, P Stepensky, A Nir, Z Perles, S Gavri, J Golender, N Yaakobi-Simhayoff, A Shaag, A J J T Rein, O Elpeleg
- 271** A syndrome of congenital hyperinsulinism and rhabdomyolysis is caused by *KCNJ11* mutation
M Albaqumi, F A Alhabib, H E Shamseldin, F Mohammed, F S Alkuraya

Genome-wide studies

- 275** *POMK* mutation in a family with congenital muscular dystrophy with merosin deficiency, hypomyelination, mild hearing deficit and intellectual disability
A von Renesse, M V Petkova, S Lützkendorf, J Heinemeyer, E Gill, C Hübner, A von Moers, W Stenzel, M Schuelke

April 2014 Volume 51 Issue 4

JMG
Journal of Medical Genetics



BMJ

jmg.bmj.com

Cover credit: Azoospermia kindreds from Ayhan *et al.* pg 239.

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://jmg.bmj.com/site/about/guidelines.xhtml#open>

C O P E COMMITTEE ON PUBLICATION ETHICS

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