

Contents

Genotype-phenotype correlations

- 419** Rhizomelic chondrodysplasia punctata and cardiac pathology *I C Huffnagel, S-A B Clur, A M Bams-Mengerink, N A Blom, R J A Wanders, H R Waterham, B T Poll-The*

New loci

- 425** Mutation in *ADAT3*, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus *A M Alazami, H Hijazi, M S Al-Dosari, R Shaheen, A Hashem, M A Aldahmesh, J Y Mohamed, A Kentab, M A Salih, A Awaji, T A Masoodi, F S Alkouraya*
- 431** A novel syndrome of hypohidrosis and intellectual disability is linked to *COG6* deficiency *R Shaheen, S Ansari, M J Alshammari, H Alkhalidi, H Alrukban, W Eyaid, F S Alkouraya*
- 437** A novel desmin mutation leading to autosomal recessive limb-girdle muscular dystrophy: distinct histopathological outcomes compared with desminopathies *N Cetin, B Balci-Hayta, H Gundesli, P Korkusuz, N Purali, B Talim, E Tan, D Selcen, S Erdem-Ozdamar, P Dincer*

Immunogenetics

- 444** Upregulation of *RCAN1* causes Down syndrome-like immune dysfunction *K R Martin, D Layton, N Seach, A Corlett, M J Barallobre, M L Arbonés, R L Boyd, B Scott, M A Pritchard*

Methods

- 455** Next generation diagnostics of cystic fibrosis and *CFTR*-related disorders by targeted multiplex high-coverage resequencing of *CFTR* *D Trujillano, M D Ramos, J González, C Tornador, F Sotillo, G Escaramis, S Ossowski, L Armengol, T Casals, X Estivill*

Developmental defects

- 463** Phenotype and genotype in 101 males with X-linked creatine transporter deficiency *J M van de Kamp, O T Betsalel, S Mercimek-Mahmutoglu, L Abulhoul, S Grünewald, I Anselm, H Azzouz, D Bratkovic, A de Brouwer, B Hamel, T Kleefstra, H Yntema, J Campistol, M A Vilaseca, D Cheillan, M D'Hooghe, L Diogo, P Garcia, C Valongo, M Fonseca, S Frints, B Wilcken, S von der Haar, H E Meijers-Heijboer, F Hofstede, D Johnson, S G Kant, L Lion-Francois, G Pitelet, N Longo, J A Maat-Kievit, J P Monteiro, A Munnich, A C Muntau, M C Nassogne, H Osaka, K Ounap, J M Pinard, S Quijano-Roy, I Poggenburg, N Poplawski, O Abdul-Rahman, A Ribes, A Arias, J Yapflio-Lee, A Schulze, C E Schwartz, S Schwenger, G Soares, Y Sznajer, V Valayannopoulos, H Van Esch, S Waltz, M M C Wamelink, P J W Pouwels, A Errami, M S van der Knaap, C Jakobs, G M Mancini, G S Salomons*

Genome-wide studies

- 473** Meta-analysis of genome-wide studies identifies *MEF2C* SNPs associated with bone mineral density at forearm *H-F Zheng, E L Duncan, L M Yerges-Armstrong, J Eriksson, U Bergström, P J Leo, W D Leslie, D Goltzman, J Blangero, D A Hanley, M A Carless, E A Streeten, M Lorentzon, M A Brown, T D Spector, U Pettersson-Kymmer, C Ohlsson, B D Mitchell, J B Richards*

Complex traits

- 479** An X chromosome-wide association analysis identifies variants in *GPR174* as a risk factor for Graves' disease *X Chu, M Shen, F Xie, X-J Miao, W-H Shou, L Liu, P-P Yang, Y-N Bai, K-Y Zhang, L Yang, Q Hua, W-D Liu, Y Dong, H-F Wang, J-X Shi, Y Wang, H-D Song, S-J Chen, Z Chen, W Huang*

Cancer genetics

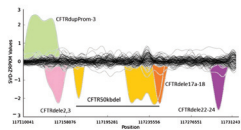
- 486** *CDH1* germline mutations and the hereditary diffuse gastric and lobular breast cancer syndrome: a multicentre study *P R Benusiglio, D Malka, E Rouleau, A De Pauw, B Buecher, C Noguès, E Fourme, C Colas, F Coulet, M Warcoin, S Grandjouan, A Sezeur, P Laurent-Puig, D Molière, C Tlemsani, M Di Maria, V Byrde, S Delalogue, M Blayau, O Caron*

PostScript

- 491** Letter

July 2013 Volume 50 Issue 7

JMG
Journal of Medical Genetics



BMJ

jmg.bmj.com

Cover credit: Depth of coverage and detection of structural variants in the *CFTR*, from D Trujillano *et al.*

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
www.publicationethics.org.uk

equator
network

recycle

When you have finished with this please recycle it