

Contents

Review

- 133** Role of *PRRT2* in common paroxysmal neurological disorders: a gene with remarkable pleiotropy *S E Heron, L M Dibbens*

Genotype-phenotype correlations

- 140** Genome-wide significant association of *ANKRD55* rs6859219 and multiple sclerosis risk *C M Lill, B-M M Schjerve, C Graetz, T Liu, V Damotte, D A Akkad, P Blaschke, L-A Gerdes, A Kroner, F Luessi, I Cournu-Rebeix, S Hoffjan, A Winkelmann, E Touze, F Pico, P Corcia, D Otaegui, A Antigüedad, A Alcina, M Comabella, X Montalban, J Olascoaga, F Matesanz, T Dörner, S-C Li, E Steinhagen-Thiessen, U Lindenberger, A Chan, P Rieckmann, H-P Hartung, O Aktas, P Lohse, M Buttmann, T Kümpfel, C Kubisch, U K Zettl, J T Epplen, B Fontaine, F Zipp, K Vandenbroeck, L Bertram*

Chromosomal rearrangements

- 144** Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations *C Schluth-Bolard, A Labalme, M-P Cordier, M Till, G Nadeau, H Tevissen, G Lesca, N Boutry-Kryza, S Rossignol, D Rocas, E Dubruc, P Edery, D Sanlaville*

New loci

- 151** Whole-exome sequencing identifies a mutation in the mitochondrial ribosome protein MRPL44 to underlie mitochondrial infantile cardiomyopathy *C J Carroll, P Isohanni, R Pöyhönen, L Euro, U Richter, V Brilhante, A Götz, T Lahtinen, A Paetau, H Pitko, B J Battersby, H Tyynismaa, A Suomalainen*
- 160** Mutations in *TMEM231* cause Meckel-Gruber syndrome *R Shaheen, S Ansari, E AL Mardawi, M J Alshammari, F S Alkurayia*

Copy-number variation

- 163** Deletions in 16q24.2 are associated with autism spectrum disorder, intellectual disability and congenital renal malformation *G R Handrigan, D Chitayat, A C Lionel, M Pinski, A K Vaags, C R Marshall, S Dyack, L F Escobar, B A Fernandez, J C Stegman, J A Rosenfeld, L G Shaffer, M Goodenberger, J C Hodge, J E Cain, R Babul-Hirji, D J Stavropoulos, V Yiu, S W Scherer, N D Rosenblum*

Developmental defects

- 174** Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome *C T Gordon, A Vuilloi, S Marlin, E Gerkes, A Henderson, A AlKindy, M Holder-Espinasse, S S Park, A Omarjee, M Sanchis-Borja, E B Bdira, M Oufadem, B Sikkema-Raddatz, A Stewart, R Palmer, R McGowan, F Petit, B Delobel, M R Speicher, P Aurora, D Kilner, P Pellerin, M Simon, J-P Bonnefont, E S Tobias, S Garcia-Miñaur, M Biner-Glindzicz, P Lindholm, B A Meijer, V Abadie, F Denoyelle, M-P Vazquez, C Rotley-Fast, V Couloigner, S Pierrot, Y Manach, S Breton, Yvonne M C Hendriks, A Munnich, L Jakobsen, P Kroisel, A Lin, L B Kaban, L Basel-Vanagaite, L Wilson, M L Cunningham, S Lyonnet, J Amiel*
- 187** Mutations of *NANOS1*, a human homologue of the *Drosophila* morphogen, are associated with a lack of germ cells in testes or severe oligo-astheno-teratozoospermia *K Kusz-Zamelczyk, M Sajek, A Spik, R Glazar, P Jędrzejczak, A Latos-Bieleńska, M Kotecki, L Pawelczyk, J Jaruzelska*

Screening

- 194** Mutations in *POLR3A* and *POLR3B* are a major cause of hypomyelinating leukodystrophies with or without dental abnormalities and/or hypogonadotropic hypogonadism *H Daoud, M Tétreault, W Gibson, K Guerrero, A Cohen, J Gburek-Augustat, M Synofzik, B Brais, C A Stevens, R Sanchez-Carpintero, C Goizet, S Naidu, A Vanderver, G Bernard*

Phenotypes

- 199** *TBC1D24* truncating mutation resulting in severe neurodegeneration *A Guven, A Tolun*

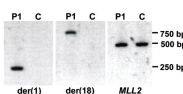
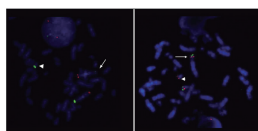
Correction

- 202** Correction

March 2013 Volume 50 Issue 3

JMG

Journal of Medical Genetics



BMJ Journals jmg.bmj.com

Cover credit: Validation of the use of next generation sequencing for the discovery of gene disruption by balanced translocations. From Schluth-Bolard *et al* in this issue (pg 144).

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