

## Contents

### Molecular medicine

- 359** Circular RNAs: a new frontier in the study of human diseases *Y Chen, C Li, C Tan, X Liu*



EDITOR'S CHOICE  
MCQs

### Cancer genetics

- 366** Multigene testing of moderate-risk genes: be mindful of the missense *E L Young, B J Feng, A W Stark, F Damiola, G Durand, N Forey, T C Franc, A Gammon, W K Kohlmann, K A Kaphingst, S McKay-Chopin, T Nguyen-Dumont, J Oliver, A M Paquette, M Patesi, N Robinot, J S Rosenthal, M Vallee, C Voegelé, J L Hopper, M C Southey, I L Andriulis, E M John, M Hashibe, J Gertz, Breast Cancer Family Registry, F Le Calvez-Kelm, F Lesueur, D E Goldgar, S V Tavtigian*



OPEN ACCESS

- 377** *HSP110* T17 simplifies and improves the microsatellite instability testing in patients with colorectal cancer *O Buhard, A Lagrange, A Guilloux, C Colas, M Chouchène, K Wanherdrick, F Coulet, E Guillermin, C Dorard, L Marisa, A Bokhari, M Greene, N El-Murr, S Bodo, M Muleris, I Sourouille, M Svrcek, P Cervera, H Blanché, J H Lefevre, Y Parc, C Lepage, C Chapusot, A-M Bouvier, M-P Gaub, J Selves, K Garrett, B Iacopetta, R Soong, R Hamelin, C Garrido, O Lascols, T André, J-F Fléjoux, A Collura, A Duval*

- 385** Identification of germline *DICER1* mutations and loss of heterozygosity in familial Wilms tumour *T B Palculict, E C Ruteshouser, Y Fan, W Wang, L Strong, V Huff*

### New loci

- 389** Deficiency of the alkaline ceramidase *ACER3* manifests in early childhood by progressive leukodystrophy *S Edvardson, J K Yi, C Jalas, R Xu, B D Webb, J Snider, A Fedick, E Kleinman, N R Treff, C Mao, O Elpeleg*

- 397** *UNC80* mutation causes a syndrome of hypotonia, severe intellectual disability, dyskinesia and dysmorphism, similar to that caused by mutations in its interacting cation channel *NALCN* *Y Perez, R Kadir, M Volodarsky, I Noyman, H Flusser, Z Shorer, L Gradstein, R Y Birnbaum, O S Birk*

### Genotype-phenotype correlations

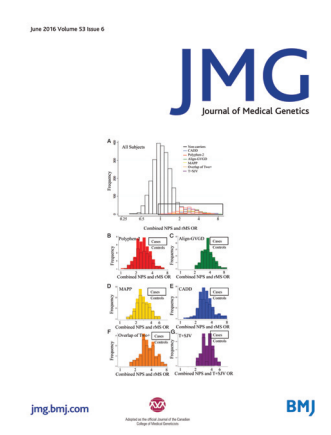
- 403** Clinical course of sly syndrome (mucopolysaccharidosis type VII) *A M Montaña, N Locke-Hocke, R D Steiner, B H Graham, M Szlago, R Greenstein, M Pineda, A Gonzalez-Meneses, M Çoker, D Bartholomew, M S Sands, R Wang, R Giugliani, A Macaya, G Pastores, A K Ketko, F Ezgü, A Tanaka, L Arash, M Beck, R E Falke, K Bhattacharya, J Franco, K K White, G A Mitchell, L Cimbalistiene, M Holtz, W S Sly*

### Phenotypes

- 419** Mutations in *EXOSC2* are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt *N Di Donato, T Neuhan, A-K Kahlert, B Klink, K Hackmann, I Neuhan, B Novotna, J Schallner, C Krause, I A Glass, S E Parnell, A Benet-Pages, A M Nissen, W Berger, J Altmüller, H Thiele, B H F Weber, E Schrock, W B Dobyns, A Bier, A Rump*
- 427** Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive *WNT1* mutations *K A Aldinger, N J Mendelsohn, B H Y Chung, W Zhang, D H Cohn, B Fernandez, F S Alkuraya, W B Dobyns, C J Curry*

### Correction

- 430** Erratum: A specific mutation in *TBL1XR1* causes Pierpont syndrome



Cover credit: Frequency distribution of breast-risk genotype scores from Young *et al.*, pg 366.



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

Receive regular table of contents by email. Register using this QR code.



This article has been chosen by the Editor to be of special interest or importance and is freely available online.



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>



This journal is a member of and subscribes to the principles of the Committee on Publication Ethics <http://publicationethics.org/>



When you have finished with this please recycle it

MCQs The online version of this article contains multiple choice questions hosted on BMJ Learning.