



Cover credit: Schematic illustration of a typical work flow combining autozygosity mapping with exome sequencing, an approach that has led to the elucidation of hundreds of rare recessive diseases. From Kaya *et al.*, pg. 786.



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/>



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

Contents

Cancer genetics

- 721** An interstitial deletion within 9p21.3 and extending beyond *CDKN2A* predisposes to melanoma, neural system tumours and possible haematological malignancies *M J Baker, A M Goldstein, P L Gordon, K S Harbaugh, H B Mackley, M J Glantz, J J Drabick*
- 728** Germline *RRAS2* mutations are not associated with Noonan syndrome *J J Ceremask, A Yu, E Esquivel, C Lissewski, M Zenker, M L Loh, E Stieglitz*
- 729** New insights in the molecular signature of advanced medullary thyroid cancer: evidence of a bad outcome of cases with double *RET* mutations *C Romei, F Casella, A Tacito, V Bottici, L Valerio, D Viola, V Cappagli, A Matrone, R Ciampi, P Piaggi, C Ugolini, L Torregrossa, F Basolo, G Materazzi, P Vitti, R Elisei*

Cognitive and behavioural genetics

- 735** Mutations specific to the Rac-GEF domain of *TRIO* cause intellectual disability and microcephaly *R J Pengelly, S Greville-Heygate, S Schmidt, E G Seaby, M R Jabalameli, S G Mehta, M J Parker, D Goudie, C Fagotto-Kaufmann, C Mercer, the DDD Study, A Debant, S Ennis, D Baralle*

Developmental defects

- 743** *SETD2* and *DNMT3A* screen in the Sotos-like syndrome French cohort *C Tlemsani, A Luscan, N Leulliot, E Bieth, A Afenjar, G Baujat, M Doco-Fenzy, A Goldenberg, D Lacombe, L Lambert, S Odent, J Pasche, S Sigaudy, A Buffet, C Violle-Poirsier, A Briand-Suleau, I Laurendeau, M Chin, P Saugier-veber, D Vidaud, V Cormier-Daire, M Vidaud, E Pasmant, L Burglen*
- 752** Mutations in *MYT1*, encoding the myelin transcription factor 1, are a rare cause of OAVS *E Lopez, M Berenguer, A Tingaud-Sequeira, S Marlin, A Toutain, F Denoyelle, A Picard, S Charron, G Mathieu, H de Belvalet, B Arveiler, P J Babin, D Lacombe, C Rooryck*

November 2016 Volume 53 Issue 11

Genotype-phenotype correlations

- 761** Molecular findings from 537 individuals with inherited retinal disease *J M Ellingford, S Barton, S Bhaskar, J O'Sullivan, S G Williams, J A Lamb, B Panda, P I Sergouniotis, R L Gillespie, S P Daiger, G Hall, T Gale, I C Lloyd, P N Bishop, S C Ramsden, C C M Black*
- 768** The clinical, biochemical and genetic features associated with *RMND1*-related mitochondrial disease *Y S Ng, C L Alston, D Diodato, A A Morris, N Ulrick, S Kmoch, J Houšětk, D Martinelli, A Haghighi, M Atiq, M A Gamero, E Garcia-Martinez, H Kratochvílová, S Santra, R M Brown, G K Brown, N Ragge, A Monavari, K Pysden, K Ravn, J P Casey, A Khan, A Chakrapani, G Vassallo, C Simons, K McKeever, S O'Sullivan, A-M Childs, E Østergaard, A Vanderver, A Goldstein, J Vogt, R W Taylor, R McFarland*
- 776** Novel *LMNA* mutations cause an aggressive atypical neonatal progeria without progerin accumulation *C Soria-Valles, D Carrero, E Gabau, G Velasco, V Quesada, C Bárcena, M Moens, K Fieggen, S Möhrcken, M Owens, D A Puente, Ó Asensio, B Loeyes, A Pérez, V Benoit, W Wuyts, N Lévy, R C Hennekam, A De Sandre-Giovannoli, C López-Otín*

New loci

- 786** *KCNA4* deficiency leads to a syndrome of abnormal striatum, congenital cataract and intellectual disability *N Kaya, M Alsagob, M C D'Adamo, A Al-Bakheet, S Hasan, M Muccioli, F B Almutairi, R Almass, M Aldosary, D Monies, O M Mustafa, B Alyounes, R Kenana, J Al-Zahrani, E Naim, F S Binhumaid, A Qari, F Almutairi, B Meyer, T F Plageman, M Pessia, D Colak, M Al-Owain*