

Contents

Clinical guidelines

- 431** The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists *K Boycott, T Hartley, S Adam, F Bernier, K Chong, B A Fernandez, J M Friedman, M T Geraghty, S Hume, B M Knoppers, A-M Laberge, J Majewski, R Mendoza-Londono, M S Meyn, J L Michaud, T N Nelson, J Richer, B Sadikovic, D L Skidmore, T Stockley, S Taylor, C van Karnebeek, Ma'n H Zawati, J Lauzon, C M Armour, on behalf of the Canadian College of Medical Geneticists*

Screening

- 438** Canadian Open Genetics Repository (COGR): a unified clinical genomics database as a community resource for standardising and sharing genetic interpretations *J Lerner-Ellis, M Wang, S White, M S Lebo, and the Canadian Open Genetics Repository Group*

Genotype-phenotype correlations

- 446** A prospective study validating a clinical scoring system and demonstrating phenotypical-genotypical correlations in Silver-Russell syndrome *S Azzi, J Salem, N Thibaud, S Chantot-Bastaraud, E Lieber, I Netchine, M D Harbison*

Phenotypes

- 454** Pallister-Killian syndrome: a study of 22 British patients *M Blyth, V Maloney, S Beal, M Collinson, S Huang, J Crolla, I K Temple, D Baralle*

Cancer genetics

- 465** A risk prediction algorithm for ovarian cancer incorporating *BRCA1*, *BRCA2*, common alleles and other familial effects *S Jervis, H Song, A Lee, E Dicks, P Harrington, C Baynes, R Manchanda, D F Easton, I Jacobs, P P D Pharoah, A C Antoniou*

Copy-number variation

- 476** Microdeletions on 6p22.3 are associated with mesomelic dysplasia Savarirayan type *R Flöttmann, J Wagner, K Kobus, C J Curry, R Savarirayan, G Nishimura, N Yasui, J Spranger, H Van Esch, M J Lyons, B R DuPont, A Dwivedi, E Klopocki, D Horn, S Mundlos, M Spielmann*

Genome-wide studies

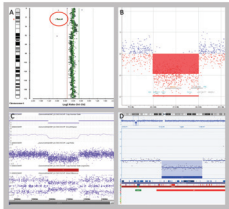
- 484** Rare genetic variants in Tunisian Jewish patients suffering from age-related macular degeneration *E Pras, D Kristal, N Shoshany, D Volodarsky, I Vulih, G Celniker, O Isakov, N Shomron, E Pras*

Neurogenetics

- 493** Loss-of-function de novo mutations play an important role in severe human neural tube defects *P Lemay, M-C Guyot, É Tremblay, A Dionne-Laporte, D Spiegelman, É Henrion, O Diallo, P De Marco, E Merello, C Massicotte, V Désilets, J L Michaud, G A Rouleau, V Capra, Z Kibar*
- 498** Prevalence of *MLH1* constitutional epimutations as a cause of Lynch syndrome in unselected versus selected consecutive series of patients with colorectal cancer *A Castillejo, E Hernández-Illán, M Rodríguez-Soler, L Pérez-Carbonell, C Egoavil, V M Barberá, M-I Castillejo, C Guarinos, E Martínez-de-Dueñas, M-J Juan, A-B Sánchez-Heras, Z García-Casado, C Ruiz-Ponte, A Brea-Fernández, M Juárez, L Bujanda, J Clofent, X Llor, M Andreu, A Castells, A Carracedo, C Alenda, A Payá, R Jover, J-L Soto*

July 2015 Volume 52 Issue 7

JMG
Journal of Medical Genetics



jmg.bmj.com

BMJ

Cover credit: 6p22 microdeletions from Flöttmann *et al*, pg 476.



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>

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/>



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