



Cover credit: Regional association plots for loci associated with telomere length from Delgado DA, et al., page 68.



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



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://authors.bmj.com/open-access/>



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.

Contents

Cancer genetics

- 1** Paediatric ovarian tumours and their associated cancer susceptibility syndromes

C Goudie, L Witkowski, S Vairy, W G McCluggage, W D Foulkes

- 11** Extracolonic cancer risk in Dutch patients with APC (adenomatous polyposis coli)-associated polyposis

Z Ghorbanoghli, B A J Bastiaansen, A M J Langers, F M Nagengast, J-W Poley, J C H Hardwick, J J Koolstra, S Sanduleanu, W H de Vos tot Nederveen Cappel, B J M Witterman, H Morreau, E Dekker, H F A Vasen

- 15** The *BRCA1* c. 5096G>A p.Arg1699Gln (R1699Q) intermediate risk variant: breast and ovarian cancer risk estimation and recommendations for clinical management from the ENIGMA consortium

S Moghadas, H D Meeks, M P G Vreeswijk, L A M Janssen, Å Borg, H Ehrencrona, Y Paulsson-Karlsson, B Wappenschmidt, C Engel, A Gehrig, N Arnold, T Van Overeem Hansen, M Thomassen, U B Jensen, T A Kruse, B Ejlersen, A-M Gerdes, I S Pedersen, S M Caputo, F Couch, E J Hallberg, A M W van den Ouweland, M J Collée, E Teugels, M A Adank, R B van der Luit, A R Mensenkamp, J C Oostervijk, M J Blok, N Janin, K B M Claes, K Tucker, V Viassolo, A E Toland, D E Eccles, P Devilee, C J Van Asperen, A B Spurdle, D E Goldgar, E G García

Genotype-phenotype correlations

- 21** Phenotype-genotype correlations in Leigh syndrome: new insights from a multicentre study of 96 patients

K Sofou, I F M de Co, E Ostergaard, P Isohanni, K Naess, L De Meirleir, C Tzoulis, J Uusimaa, T Lönnqvist, L A Bindoff, M Tulinius, N Darin

New disease loci

- 28** Heterozygous mutations affecting the protein kinase domain of *CDK13* cause a syndromic form of developmental delay and intellectual disability

M J Hamilton, R C Caswell, N Canham, T Cole, H V Firth, N Foulds, K Heimdal, E Hobson, G Houge, S Joss, D Kumar, A K Lampe, I Maystadt, V McKay, K Metcalfe, R Newbury-Ecob, S-M Parke, L Robert, C F Rustad, E Wakeling, A O M Wilkie, The Deciphering Developmental Disorders Study, S R F Twigg, M Suri

January 2018 Volume 55 Issue 1

Neurogenetics

- 39** SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family

B Roeben, R Schüle, S Ruf, B Bender, B Alhaddad, T Benkert, T Meitinger, S Reich, J Böhringer, C-D Langhans, F M Vaz, S B Wortmann, T Marquardt, T B Haack, I Krägeloh-Mann, L Schöls, M Synofzik

- 48** A homozygous founder mutation in *TRAPPC6B* associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features

I Marin-Valencia, G Novarino, A Johansen, B Rosti, M Y Issa, D Musaev, G Bhat, E Scott, J L Silhavy, V Stanley, R O Rosti, J W Gleeson, F B Imam, M S Zaki, J G Gleeson

Complex traits

- 55** GWAS on prolonged gestation (post-term birth): analysis of successive Finnish birth cohorts

W Schierding, J Antony, V Karhunen, M Väärämäki, S Franks, P Elliott, E Kajantie, S Sebert, A Blakemore, J A Horsfield, M-R Järvelin, J M O'Sullivan, W S Cutfield

Telomere biology

- 64** Genome-wide association study of telomere length among South Asians identifies a second RTEL1 association signal

D A Delgado, C Zhang, L S Chen, J Gao, S Roy, J Shinkle, M Sabarinathan, M Argos, L Tong, A Ahmed, T Islam, M Rakibuz-Zaman, G Sarwar, H Shahriar, M Rahman, M Yunus, F Jasmine, M G Kibriya, H Ahsan, B L Pierce

Miscellaneous

- 72** Erratum: *FOXP1*-related intellectual disability syndrome: a recognisable entity