



Contents

Position Statement

- 417** UK consensus recommendations for clinical management of cancer risk for women with germline pathogenic variants in cancer predisposition genes: *RAD51C, RAD51D, BRIP1* and *PALB2*

H Hanson, A Kulkarni, L Loong, G Kavanaugh, B Torr, S Allen, M Ahmed, A C Antoniou, R Cleaver, T Dabir, D G Evans, E Golightly, R Jewell, K Kohut, R Manchanda, A Murray, J Murray, K-R Ong, A N Rosenthal, E R Woodward, D M Eccles, C Turnbull, M Tischkowitz, On behalf of Consensus meeting attendees, F Lalloo

Biochemical genetics

- 430** Long-term outcomes of very early treated infantile-onset Pompe disease with short-term steroid premedication: experiences from a nationwide newborn screening programme

C-F Yang, T-W E Liao, Y-L Chu, L-Z Chen, L-Y Huang, T-F Yang, H-C Ho, S-M Kao, D-M Niu

Cancer genetics

- 440** The avoiding late diagnosis of ovarian cancer (ALDO) project: a pilot national surveillance programme for women with pathogenic germline variants in *BRCA1* and *BRCA2*

S Philpott, M Raikou, R Manchanda, M Lockley, N Singh, M Scott, D G Evans, J Adlard, M Ahmed, R Edmondson, E R Woodward, A Lammisio, J Balega, A F Brady, A Sharma, L Izatt, A Kulkarni, V Tripathi, J S Solomons, K Hayes, H Hanson, K Snape, L Side, S Skates, A McGuire, A N Rosenthal

- 450** Comprehensive RNA and protein functional assessments contribute to the clinical interpretation of *MSH2* variants causing in-frame splicing alterations

L Meulemans, S Baert Desurmont, M-C Wail, G Castelain, A Killian, J Hauchard, T Frebourg, F Coulet, A Martins, M Muleris, P Gaildrat

- 460** *APC* germline pathogenic variants and epithelial ovarian cancer: causal or coincidental findings?

R Vibert, J Le Gall, B Buecher, E Mouret-Fourme, G Bataillon, V Becette, O Trabelsi-Grati, V Moncoutier, C Dehainault, J Carriere, M Schwartz, V Suyheng, I Bieche, C Colas, A Vincent-Salomon, D Stoppa-Lyonnet, L Golmard

Phenotypes

- 464** Hereditary haemorrhagic telangiectasia in Danish patients with pathogenic variants in *SMAD4*: a nationwide study

A M Jelsing, A Kjeldsen, L L Christensen, B Bertelsen, J G Karstensen, K Brusgaard, P M Torrington

Chromosomal rearrangements

- 469** Disruption of the topologically associated domain at Xp21.2 is related to 46,XY gonadal dysgenesis

J A Meinel, V Yumiceba, A Kunstner, K Schultz, N Kruse, F J Kaiser, P-M Holterhus, A Claviez, O Hiort, H Busch, M Spielmann, R Werner

Genotype-phenotype correlations

- 477** Genotype-phenotype correlations and clinical outcomes of patients with von Hippel-Lindau disease with large deletions

K Zhang, W Yang, K Ma, J Qiu, L Li, Y Xu, Z Zhang, C Yu, J Zhou, Y Gong, L Cai, K Gong

- 484** Identifying the molecular drivers of ALS-implicated missense mutations

S Portelli, A Albanaz, D E V Pires, D B Ascher

Copy-number variation

- 491** Recurrent 17q12 microduplications contribute to renal disease but not diabetes

S Cannon, R Clissold, K Sulecharoen, M Tuke, G Hawkes, R N Beaumont, A R Wood, M Gilchrist, A T Hattersley, R A Oram, K Patel, C Wright, M N Weedon

Novel disease loci

- 498** Heterozygous pathogenic variants involving *CBFB* cause a new skeletal disorder resembling cleidocranial dysplasia

T Beylajens, E Boudin, N Revencu, N Boeckx, M Bertrand, L Schütz, T B Haacke, A Weber, E Biliouri, M Vinkšelj, A Zagožen, B Peterlin, S Pai, A Telegrafi, L B Henderson, C Ellis, L Turner, W Wuyts, W Van Hul, G Hendrickx, G R Mortier

Structural variation

- 505** Conclusion of diagnostic odysseys due to inversions disrupting *GLI3* and *FBN1*

A T Pagnamenta, J Yu, J Evans, P Twiss, Genomics England Research Consortium, Musculoskeletal GeCIP MDT, A C Offiah, M Wafik, S G Mehta, M K Javaid, S F Smithson, J C Taylor

Neurogenetics

- 511** Clinical, neuroimaging and molecular characteristics of *PPP2R5D*-related neurodevelopmental disorders: an expanded series with functional characterisation and genotype-phenotype analysis

N Oyama, P Vaneynde, S Reynhout, E M Pao, A Timms, X Fan, K Foss, R Derua, V Janssens, W Chung, G M Mirzaa

Cover credit: Radiographic features in four subjects carrying pathogenic variants in *CBFB*. See Beylajens *et al*, page 501.



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.