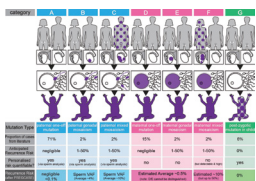


September 2023 Volume 60 Issue 9

**JMG**  
Journal of Medical Genetics



[jmg.bmj.com](http://jmg.bmj.com)



BMJ

Cover credit: Overview of the PREcision Genetic Counselling And REproduction (PREGCARE) strategy and stratification of de novo mutation (DNM) recurrence risk into seven different categories. See Kay *et al.*, page 926.



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

## Contents

### Cancer genetics

**835** *GOLM1*: expanding our understanding of melanoma susceptibility  
*E J Maas, C K Wallingford, E DeBortoli, D J Smit, B Betz-Stablein, L G Aoude, M S Stark, R A Sturm, H P Soyer, A M McInerney-Leo*

**838** Differential rates of germline heterozygote and mosaic variants in *NF2* may show varying propensity for meiotic or mitotic mutation  
*D G Evans, G J Burghel, M J Smith*

**842** Germline (epi)genetics reveals high predisposition in females: a 5-year, nationwide, prospective Wilms tumour cohort  
*U K Stoltze, M Hildonen, T V O Hansen, J Foss-Skeftjensvik, A Byrjalsen, M Lundsgaard, L Pignata, K Grønsvov, Z Tumer, K Schmiegelow, J S Brok, K A W Wadt*

### Therapeutics

**850** Effect of Migalastat on cArDiac InvOlvement in FabRry Disease: MAIORA study  
*A Camporeale, F Bandera, M Pieroni, F Pieruzzi, M Spada, A Bersano, L Econimo, C Lanzillo, M Rubino, R Mignani, I Motta, I Olivotto, I Tanini, R Valaperta, K Chow, I Baroni, S Boveri, F Graziani, S Pica, L Tondi, M Guazzi, M Lombardi*

### Genotype-phenotype correlations

**859** Pseudocoloboma-like maculopathy with biallelic *RDH12* missense mutations  
*C-Y Kuo, M-Y Chung, S-J Chen*

**866** The crucial role of titin in fetal development: recurrent miscarriages and bone, heart and muscle anomalies characterise the severe end of titinopathies spectrum  
*M F Di Feo, V Lillhack, M Jokela, M McEntagart, T Homfray, E Giorgio, G C Casalis Cavalchini, A Brusco, M Iascone, L Spaccini, P D'Oria, M Savarese, B Udd*

**874** Pathogenic variations in *MAML2* and *MAMLD1* contribute to congenital hypothyroidism due to dysmorphogenesis by regulating the Notch signalling pathway  
*F-Y Wu, R-M Yang, H-Y Zhang, M Zhan, P-H Tu, Y Fang, C-X Zhang, S-Y Song, M Dong, R-J Cui, X-Y Liu, L Yang, C-Y Yan, F Sun, R-J Zhang, Z Wang, J Liang, H-D Song, F Cheng, S-X Zhao*

## September 2023 Volume 60 Issue 9

### Neurogenetics

**885** Recurrent, founder and hypomorphic variants contribute to the genetic landscape of Joubert syndrome  
*V Serpieri, G Mortarini, H Loucks, T Biagini, A Micalizzi, I Palmieri, J C Dempsey, F D'Abrusco, C Mazzotta, R Battini, E S Bertini, E Boltshauser, R Borgatti, K Brockmann, S D'Arrigo, N Nardocci, R Fischetto, E Agolini, A Novelli, A Romano, R Romaniello, F Stanzial, S Signorini, P Strisciuglio, S Gana, T Mazza, D Doherty, E M Valente*

**894** Frontotemporal dementia presentation in patients with heterozygous p.H157Y variant of *TREM2*  
*N Ogonowski, H Santamaria-Garcia, S Baez, A Lopez, A Laserna, E Garcia-Cifuentes, P Ayala-Ramirez, I Zarante, F Suarez-Obando, P Reyes, M Kauffman, N Cochran, M Schulte, D W Sirkis, S Spina, J S Yokoyama, B L Miller, K S Kosik, D Matallana, A Ibañez*

### Phenotypes

**905** Seven cases of hereditary haemorrhagic telangiectasia-like hepatic vascular abnormalities associated with *EPHB4* pathogenic variants  
*A Guilhem, S Dupuis-Girod, O Espitia, S Rivière, J Seguiet, M Kerjoutan, C Lavigne, H Maillard, P Magro, L Alric, D Lipsker, A Parrot, V Leguy, C Vanlemmens, L Guibaud, M Viikula, M Eyries, P-J Valette, S Giraud*

### Diagnostics

**910** Parental mosaicism detection and preimplantation genetic testing in families with multiple transmissions of de novo mutations  
*N Xu, W Shi, X Cao, X Zhou, L Jin, H-F Huang, S Chen, C Xu*

### Vision science

**918** *PSMD3* gene mutations cause pathological myopia  
*J Chen, P Lian, X Zhao, J Li, X Yu, X Huang, S Chen, L Lu*

### Screening

**925** Providing recurrence risk counselling for parents after diagnosis of a serious genetic condition caused by an apparently de novo mutation in their child: a qualitative investigation of the PREGCARE strategy with UK clinical genetics practitioners  
*A C Kay, J Wells, N Hallowell, A Goriely*

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

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