



Cover credit: Retinal and visual field findings in four unrelated patients with a missense variant, and a branchpoint variant, in *BBS1*. See Fadaie *et al*, page 441.



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

Contents

Developmental defects

- 417** Oculo-auriculo-vertebral spectrum: new genes and literature review on a complex disease
A Tingaud-Sequeira, A Trimouille, T Sagardoy, D Lacombe, C Rooryck

Vision science

- 428** Activation of cryptic donor splice sites by non-coding and coding *PAX6* variants contributes to congenital aniridia
M Tarilonte, P Ramos, J Moya, G Fernandez-Sanz, F Blanco-Kelly, S T Swafiri, C Villaverde, R Romero, A Tamayo, B Gener, P Calvas, C Ayuso, M Corton

- 438** *BBS1* branchpoint variant is associated with non-syndromic retinitis pigmentosa
Z Fadaie, L Whelan, A Dockery, C H Z Li, L I van den Born, C B Hoyng, C Gilissen, J Corominas, C Rowlands, R Megaw, A K Lampe, F P M Cremers, G J Farrar, J M Ellingford, P F Kenna, S Roosing

Neurogenetics

- 445** High efficiency and clinical relevance of exome sequencing in the daily practice of neurogenetics
Q Thomas, A Viobello, F Tran Mau-Them, Y Duffourd, A Fromont, M Giroud, B Daubail, A Jacquin-Piques, M Hervieu-Begue, T Moreau, G-V Osseby, P Garret, S Nambot, J Delanne, A-L Bruel, A Sorlin, P Callier, A-S Denomme-Pichon, L Faivre, Y Béjot, C Philippe, C Thauvin-Robinet, S Moutton

- 453** Homozygous mutation in *MCM7* causes autosomal recessive primary microcephaly and intellectual disability
E Ravindran, C Gutierrez de Velazco, A Ghazanfar, N Kraemer, S Zaout, A Waheed, M Hanif, S Mughal, A Prigione, N Li, X Fang, H Hu, A M Kaindl

- 462** Genetic origin of sporadic cases and RNA toxicity in neuronal intranuclear inclusion disease
J Deng, B Zhou, J Yu, X Han, J Fu, X Li, X Xie, M Zhu, Y Zheng, X Guo, P Li, Q Wang, J Liu, W Zhang, Y Yuan, S Yao, Z Wang, D Hong

May 2022 Volume 59 Issue 5

Genotype-phenotype correlations

- 470** Rising of *LOXHD1* as a signature causative gene of down-sloping hearing loss in people in their teens and 20s
B J Kim, H W Jeon, W Jeon, J H Han, J Oh, N Yi, M Y Kim, M Kim, J N Kim, B H Kim, J Y Hyon, D Kim, J-W Koo, D-Y Oh, B Y Choi

Cancer genetics

- 481** Characterisation of protein-truncating and missense variants in *PALB2* in 15 768 women from Malaysia and Singapore
P S Ng, R ACM Boonen, E Wijaya, C E Chong, M Sharma, S Knaup, S Mariapun, W K Ho, J Lim, S-Y Yoon, N A Mohd Taib, M H See, J Li, S H Lim, E Y Tan, B K-T Tan, S-M Tan, V K-M Tan, R M van Dam, K Rahmat, C H Yip, S Carvalho, C Lucarini, C Baynes, A M Dunning, A Antoniou, H van Attikum, D F Easton, M Hartman, S H Teo
- 492** High cumulative risk of colorectal cancers and desmoid tumours and fibromatosis in South Asian APC mutation carriers
S Ashar, A Lipsa, N Khan, R Sarin

Phenotypes

- 496** Clinical and subclinical findings in heterozygous *ABCC6* carriers: results from a Belgian cohort and clinical practice guidelines
L Nollet, L Campens, J De Zaeytijd, B Leroy, D Hemelsoet, P J Coucke, O M Vanakker
- 505** Patients with *KCNH1*-related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome
M Aubert Mucca, O Patat, S Whalen, L Arnaud, G Barcia, J Buratti, B Cogné, D Doummar, C Karsenty, S Kenis, E Leguern, G Lesca, C Nava, M Nizon, A Piton, S Valence, L Villard, S Weckhuysen, B Keren, C Mignot
- 511** Recurrent *de novo* missense variants in *GNB2* can cause syndromic intellectual disability
N B Tan, A T Pagnamenta, M P Ferla, J Gadian, B H Y Chung, M C Y Chan, J L F Fung, E Cook, S Guter, F Boschann, A Heinen, J Schallner, C Mignot, B Keren, S Whalen, C Sarret, D Mittag, L Demmer, R Stapleton, K Saida, N Matsumoto, N Miyake, R Sheffer, H Mor-Shaked, C P Barnett, A B Byrne, H S Scott, A Kraus, G Cappuccio, N Brunetti-Pierri, R Iorio, F Di Dato, L S Pais, A Yeung, T Y Tan, J C Taylor, J Christodoulou, S M White

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/>
Member since 2008
JMG0012

recycle
When you have finished with this please recycle it

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