



Cover credit: Structural abnormalities detected on MRI of patients with bi-allelic *TTC5* variants. See Rasheed *et al*, page 242.



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

## Contents

April 2021 Volume 58 Issue 4

### Screening

- 217** Targeting lung cancer screening to individuals at greatest risk: the role of genetic factors



*M B Lebrecht, E J Crosbie, M J Smith, E R Woodward, D G Evans, P A J Crosbie*

### Cancer genetics

- 227** Sporadic vestibular schwannoma: a molecular testing summary

*K V Sadler, N L Bowers, C Hartley, P T Smith, S Tobi, A J Wallace, A King, S K W Lloyd, S Rutherford, O N Pathmanaban, C Hammerbeck-Ward, S Freeman, E Stapleton, A Taylor, A Shaw, D Halliday, M J Smith, D G Evans*

- 234** Loss-of-function variants in *POT1* predispose to uveal melanoma

*V Nathan, J M Palmer, P A Johansson, H R Hamilton, S K Warriar, W Glasson, L A McGrath, V F S Kahl, R S Vasireddy, H A Pickett, K M Brooks, A L Pritchard, N K Hayward*

### Neurogenetics

- 237** Bi-allelic *TTC5* variants cause delayed developmental milestones and intellectual disability

*A Rasheed, E Gumus, M Zaki, K Johnson, H Manzoor, G LaForce, D Ross, J McEvoy-Venneri, V Stanley, S Lee, A Virani, T Ben-Omran, J G Gleeson, S Naz, A Schaffer*

### Novel disease loci

- 247** *TMEM16A* deficiency: a potentially fatal neonatal disease resulting from impaired chloride currents

*J H Park, J Ousingsawat, I Cabrera, R E Bettels, J Große-Onnebrink, C Schmalstieg, S Biskup, J Reunert, S Rust, R Schreiber, K Kunzelmann, T Marquardt*

### Developmental defects

- 254** Absence of SCAPER causes male infertility in humans and *Drosophila* by modulating microtubule dynamics during meiosis

*O Wormsler, Y Levy, A Bakhrat, S Bonaccorsi, L Crazziadio, M Gatti, A AbuMadighem, R J McKenney, K Okada, S El Riati, I Har-Vardi, M Huleihel, E Levitas, O S Birk, U Abdu*

### Genotype-phenotype correlations

- 264** Genotype-phenotype correlations for pancreatic cancer risk in Dutch melanoma families with pathogenic *CDKN2A* variants



*K A Overbeek, M DM Rodríguez-Girondo, A Wagner, N van der Stoep, P C van den Akker, J C Oosterwijk, T A van Os, L E van der Kolle, H F A Vasen, F J Hes, D L Cahen, M J Bruno, T P Potjer*

- 270** Analysis of genotype-phenotype correlations in *PAX6*-associated aniridia

*T A Vasilyeva, A V Marakhonov, A A Voskresenskaya, V V Kadyshev, B Käsmann-Kellner, N V Sukhanova, L A Katargina, S I Kutsev, R A Zinchenko*

### Diagnostics

- 275** Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study

*S Shickh, M Gutierrez Salazar, K-R Zakeer, C Lázaro, J Gu, J Goltz, D Kleinman, A Noor, S Khalouei, C Mighton, E Reble, R Kodida, Y Bombard, S DiTroia, S Baxter, N Watkins, M Care, A Adler, S Horsburgh, O Morar, J Murphy, D-L Nevay, M Szybowska, M Aronson, S Panchal, R Godoy, S Holter, S Randall Armel, K Semotiuk, C Elser, R H Kim, D Chitayat, J So, H Faghfoury, J Silver, C F Morel, J Lerner-Ellis*

- 284** Comprehensive genetic sequence and copy number analysis for Charcot-Marie-Tooth disease in a Canadian cohort of 2517 patients

*M Volodarsky, J Kerkhof, A Stuart, M Levy, L I Brady, M Tarnopolsky, H Lin, P Ainsworth, B Sadikovic*



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.