# Journal of Medical Genetics



Cover credit: Indigenous Filipino pedigree with otitis media. See Frank *et al*, page 443.

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

## Contents


July 2021 Volume 58 Issue 7



### Cancer genetics

**433** Comprehensive characteristics of somatic mutations in the normal tissues of patients with cancer and existence of somatic mutant clones linked to cancer development
*J-H Oh, C O Sung*

### Complex traits

**442** Otitis media susceptibility and shifts in the head and neck microbiome due to *SPINK5* variants
*D N Frank, A P J Giese, L Hafren, T C Bootpetch, T K L Yarza, M J Steritz, M Pedro, P J Labra, K A Daly, M L C Tantoco, W Szeremeta, M R T Reyes-Quintos, N Ahankoob, E G d.V Llanes, H S Pine, S Yousaf, D Ir, E Einarsdottir, R A R de la Cruz, N R Lee, R M A Nonato, C E Robertson, K M C Ong, J P M Magno, A N E Chiong, M C Espiritu-Chiong, M L San Agustin, T L G Cruz, G T Abes, M J Bamshad, E M Cutiongco-de la Paz, J Kere, D A Nickerson, K L Mohlke, S Riazuddin, A Chan, P S Mattila, S M Leal, A F Ryan, Z M Ahmed, T Chonmaitree, M M Sale, C M Chiong, R L P Santos-Cortez*

### Developmental defects

**453** *DLG5* variants are associated with multiple congenital anomalies including ciliopathy phenotypes
*J Marquez, N Mann, K Arana, E Deniz, W Ji, M Konstantino, E K Mis, C Deshpande, L Jeffries, J McGlynn, H Hugo, E Widmeier, M Konrad, V Tasic, R Morotti, J Baptista, S Ellard, S A Lakhani, F Hildebrandt, M K Khokha*

**465** Congenital sensorineural hearing loss as the initial presentation of *PTPN11*-associated Noonan syndrome with multiple lentigines or Noonan syndrome: clinical features and underlying mechanisms
*X Gao, S-S Huang, S-W Qiu, Y Su, W-Q Wang, H-Y Xu, J-C Xu, D-Y Kang, P Dai, Y-Y Yuan*

### Genotype-phenotype correlations

**475** Heterozygous *KIF1A* variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders
*F Nicita, M Ginevrino, L Travaglini, S D'Arrigo, G Zorzi, R Borgatti, G Terrone, M Catteruccia, G Vasco, V Brankovic, S Siliquini, S Romano, C Veredice, M Pedemonte, M Armando, D Lettori, F Stregapede, L Bosco, A Sferra, V Tessarollo, R Romaniello, G Ristori, E Bertini, E M Valente, G Zanni*

### Neurogenetics

**484** *CIC de novo* loss of function variants contribute to cerebral folate deficiency by downregulating *FOLR1* expression
*X Cao, A Wolf, S-E Kim, R M Cabrera, B J Wlodarczyk, H Zhu, M Parker, Y Lin, J W Steele, X Han, V T Ramaekers, R Steinfeld, R H Finnell, Y Lei*

**495** Biallelic variants in *ADARB1*, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy
*R Maroofian, J Sedmík, N Mazaheri, M Scala, M S Zaki, L P Keegan, R Azizimalamiri, M Issa, G Shariati, A Sedaghat, C Beetz, P Bauer, H Galehdari, M A O'Connell, H Houlden*


EDITOR'S CHOICE This article has been chosen by the Editor to be of special interest or importance and is freely available online.

OPEN ACCESS This article has been made freely available online under the BMJ Journals open access scheme. See **http://authors.bmj.com/open-access/**

COPE Member since 2008 JM00012 This journal is a member of and subscribes to the principles of the Committee on Publication Ethics **http://publicationethics.org/**

recycle When you have finished with this please recycle it

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