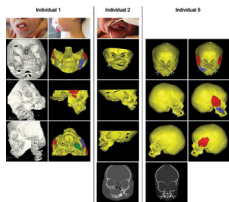


November 2023 Volume 60 Issue 11

JMG

Journal of Medical Genetics



jmg.bmj.com



BMJ

Cover credit: Maxillomandibular anomalies in bony syngnathia in individuals carrying variants in the *VGLL2* gene. See Agostini *et al.*, page 1086.



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

Contents

November 2023 Volume 60 Issue 11

Cancer genetics

1035 Position statement of the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) on *APC* I1307K and cancer risk

L Valle, L H Katz, A Latchford, P Mur, V Moreno, I M Frayling, B Heald, G Capellà, on behalf of the InSiGHT Council

1044 Ability of a polygenic risk score to refine colorectal cancer risk in Lynch syndrome

N Dueñas, H Klinkhammer, N Bonifaci, I Spier, A Mayr, E Hassanin, A Diez-Villanueva, V Moreno, M Pineda, C Maj, G Capellà, S Aretz, J Brunet

1052 Pancreatic cancer cluster region identified in *BRCA2*

J S Chian, W Xu, S M Wang

1057 *TP53* c.455C>T p.(Pro152Leu) pathogenic variant is a lower risk allele with attenuated risks of breast cancer and sarcoma

D G Evans, E F Harkness, E R Woodward

Genotype-phenotype correlations

1061 Analysis of Rotterdam Study cohorts confirms a previously identified *RIPOR2* in-frame deletion as a prevalent genetic factor in phenotypically variable adult-onset hearing loss (DFNA21) in the Netherlands

H M Velde, N C Homans, A Goedegebure, C P Laning, R J E Pennings, H Kremer

1067 Pulmonary function and structure abnormalities in children and young adults with osteogenesis imperfecta point to intrinsic and extrinsic lung abnormalities

B R Gochuico, M Hossain, S K Talvacchio, M X G Zuo, M Barton, A N Dang Do, J C Marini

1076 Genotypes and phenotypes of *DNM1* encephalopathy

J Kim, L-Y Teng, B Shaker, D Na, H Y Koh, S S Kwon, J S Lee, H D Kim, H-C Kang, S H Kim

Developmental defects

1084 Biallelic truncating variants in *VGLL2* cause syngnathia in humans

V Agostini, A Tessier, N Djaziri, R H Khonsari, E Galliani, Y Kurihara, M Honda, H Kurihara, K Hidaka, G Tunchbilek, A Picard, E Konas, J Amiel, C T Gordon

1092 Dominant negative variants in *IKZF2* cause ICHAD syndrome, a new disorder characterised by immunodysregulation, craniofacial anomalies, hearing impairment, athelia and developmental delay

A Mohajeri, M Vaseghi-Shanjani, J A Rosenfeld, G X Yang, H Lu, M Sharma, S Lin, A Salman, M Waqas, M Sababi Azamian, K C Worley, K L Del Bel, F K Kozak, R Rahmanian, C M Biggs, K J Hildebrand, S R Lalani, S K Nicholas, D A Scott, S Mostafavi, C van Karnebeek, E Henkelman, J Halparin, C L Yang, L Armstrong, Undiagnosed Diseases Network, Care4Rare Canada Consortium, S E Turvey, A Lehman

Neurogenetics

1105 HnRNPR strongly represses splicing of a critical exon associated with spinal muscular atrophy through binding to an exonic AU-rich element

T Jiang, R Qu, X Liu, Y Hou, L Wang, Y Hua

1116 Congenital mirror movements are associated with defective polymerisation of RAD51

O Trouillard, P Dupaigne, M Dunoyer, M Doulazmi, M K Herlin, S Frismand, A Riou, V Legros, G Chevreux, X Veaute, D Busso, C Fouquet, C Y Utagawa, C E Steiner, L Steinmetz, R S Honjo, C A Kim, L Wang, R Abourjaili-Bilodeau, P M Campeau, M Warman, M R Passos-Bueno, N C Hoch, D R Bertola

Novel disease loci

1127 Biallelic variants in *DNA2* cause poikiloderma with congenital cataracts and severe growth failure reminiscent of Rothmund-Thomson syndrome

R Di Lazzaro Filho, G L Yamamoto, T J Silva, L A Rocha, B D W Linnenkamp, M A A Castro, D Bartholdi, A Schaller, T Leeb, S Kelmann, C Y Utagawa, C E Steiner, L Steinmetz, R S Honjo, C A Kim, L Wang, R Abourjaili-Bilodeau, P M Campeau, M Warman, M R Passos-Bueno, N C Hoch, D R Bertola

1133 Exome sequencing links the SUMO protease SENP7 with fatal arthrogryposis multiplex congenita, early respiratory failure and neutropenia

N Samra, N S Jansen, I Morani, R R Kakun, R Zaid, T Paperna, M Garcia-Dominguez, Y Vimer, H Frankenthal, E S Shinwell, I Portnov, D Bakry, A Shalata, M Shapira Rootman, D Kidron, L A Claessens, R A Wevers, H Mandel, A C O Vertegaal, K Weiss

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/>

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.