

## Contents

### Neurogenetics

**567** Advances in identification of genes involved in autosomal recessive intellectual disability: a brief review

*Y R Mir, R A H Kuchay*

**617** Novel *ASCC1* mutations causing prenatal-onset muscle weakness with arthrogryposis and congenital bone fractures

*J Böhm, E Malfatti, E Oates, K Jones, G Brochier, A Boland, J-F Deleuze, N B Romero, J Laporte*

### Cancer genetics

**574** Deleterious somatic variants in 473 consecutive individuals with ovarian cancer: results of the observational AGO-TR1 study (NCT02222883)

*J Hauke, E Hahnen, S Schneider, A Reuss, L Richters, S Kommos, A Heimbach, F Marmé, S Schmidt, K Prieske, H Gevensleben, A Burges, J Borde, N De Gregorio, P Nürnberg, A El-Balat, H Thiele, F Hilpert, J Altmüller, W Meier, D Dietrich, R Kimmig, B Schoemig-Markieska, K Kast, E Braicu, K Baumann, C Jackisch, T-W Park-Simon, C Ernst, L Hanker, J Pfisterer, A Schnelzer, A du Bois, R K Schmutzler, P Harter*

**581** Addition of a 161-SNP polygenic risk score to family history-based risk prediction: impact on clinical management in non-*BRCA1/2* breast cancer families

*I M M Lakeman, F S Hilbers, M Rodríguez-Girondo, A Lee, M P G Vreeswijk, A Hollestelle, C Seynaeve, H Meijers-Heijboer, J C Oosterwijk, N Hoogerbrugge, E Olah, H F A Vasen, C J van Asperen, P Devilee*

### Diagnostics

**590** Deciphering the complexity of the 4q and 10q subtelomeres by molecular combing in healthy individuals and patients with facioscapulohumeral dystrophy

*K Nguyen, N Brouqsault, C Chaix, S Roche, J D Robin, C Vovan, L Gerard, A Mégarbané, J A Urizberea, R Bellance, C Barnéas, A David, B Eymard, M Fradin, V Manel, S Sacconi, V Tiffreau, F Zagnoli, J-M Cuisset, E Salort-Campana, S Attarian, R Bernard, N Lévy, F Magdénier*

**602** Progression from islet autoimmunity to clinical type 1 diabetes is influenced by genetic factors: results from the prospective TEDDY study

*A Beyerlein, E Bonifacio, K Vehik, M Hippich, C Winkler, B I Frohnert, A K Steck, W A Hagopian, J P Krischer, Å Lernmark, M J Rewers, J-X She, J Toppari, B Akolkear, S S Rich, A-G Ziegler, the TEDDY Study Group*

### Complex traits

**607** Genome-wide association study identifies seven novel loci associating with circulating cytokines and cell adhesion molecules in Finns

*E Sliz, M Kalaoja, A Ahola-Olli, O Raitakari, M Perola, V Salomaa, T Lehtimäki, T Karhu, H Viinamäki, M Salmi, K Santalahi, S Jalkanen, J Jokelainen, S Keinänen-Kiukkaanniemi, M Männikkö, K-H Herzig, M-R Järvelin, S Sebert, J Kettunen*

### Genotype-phenotype correlations

**622** Bi-allelic loss of function variants of *TBX6* causes a spectrum of malformation of spine and rib including congenital scoliosis and spondylocostal dysostosis

*N Otomo, K Takeda, S Kawai, I Kou, L Guo, M Osawa, C Alev, N Kawakami, N Miyake, N Matsumoto, Y Yasuhiko, T Kotani, T Suzuki, K Uno, H Sudo, S Inami, H Taneichi, H Shigematsu, K Watanabe, I Yonezawa, R Sugawara, Y Taniguchi, S Minami, K Kaneko, M Nakamura, M Matsumoto, J Toguchida, K Watanabe, S Ikegawa*

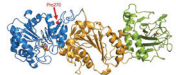
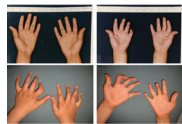
### Novel disease loci

**629** Pathogenic variants in *PLOD3* result in a Stickler syndrome-like connective tissue disorder with vascular complications

*L J Ewans, A Colley, C Gaston-Massuet, A Gualtieri, M J Cowley, M J McCabe, D Anand, S A Lachke, L Sciatti, F Forneris, Y Zhu, K Ying, C Walsh, E P Kirk, D Miller, C Giunta, D Sillence, M Dinger, M Buckley, T Roscioli*

September 2019 Volume 56 Issue 9

**JMG**  
Journal of Medical Genetics



jmg.bmj.com



BMJ

Cover credit: A pathogenic variant in *PLOD3* associated with a Stickler syndrome-like connective tissue disorder. See Ewans *et al*, pages 631, 632.



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



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.