



Journal of
Medical Genetics
jmg.bmj.com



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

Contents

Clinical guidelines

503 Diagnosis and management in Rubinstein-Taybi syndrome: first international consensus statement

MCQS
OPEN ACCESS
D Lacombe, A Bloch-Zupan, C Bredrup, E B Cooper, S D Houge, S García-Miñaur, H Kayserili, L Larizza, V Lopez Gonzalez, L A Menke, D Milani, F Saettini, C A Stevens, L Tooke, J A Van der Zee, M M Van Genderen, J Van-Gils, J Waite, J-L Adrien, O Bartsch, P Bitoun, A H M Bouts, A M Cueto-González, E Domínguez-Garrido, F A Duijckers, P Fergelot, E Halstead, S A Huisman, C Meossi, J Mullins, S M Niekkel, C Oliver, E Prada, A Rei, I Riddle, C Rodríguez-Fonseca, R Rodríguez Pena, J Russell, A Saba, F Santos-Simarro, B N Simpson, D F Smith, M F Stevens, K Szakacson, E Taupiac, N Totaro, I Valenzuela Palafoll, D C M Van Der Kaay, M P Van Wijck, K Vyshka, S Wiley, R C Hennekam

Therapeutics

520 Head-to-head trial of pegunigalsidase alfa versus agalsidase beta in patients with Fabry disease and deteriorating renal function: results from the 2-year randomised phase III BALANCE study

OPEN ACCESS
E L Wallace, O Goker-Alpan, W R Wilcox, M Holidá, J Bernat, N Longo, A Linhart, D A Hughes, R J Hopkin, C Tøndel, M Langeveld, P Giraldo, A Pisani, D P Germain, A Mehta, P B Deegan, M J Mohar, D Ortiz, A Jovanovic, M Muriello, B A Barshop, V Kimonis, B Vujkovic, A Nowak, T Geberhiwot, I Kantola, J Knoll, S Walde, K Nedd, A Karaa, E Brill-Almon, S Alon, R Chertkoff, R Rocco, A Sakov, D G Warnock

531 Comment to: Head-to-head trial of pegunigalsidase alfa versus agalsidase beta in patients with Fabry disease and deteriorating renal function: results from the 2-year randomised phase III BALANCE study—determination of immunogenicity

M Lenders, E Brand

534 Response to commentary: Head-to-head trial of pegunigalsidase alfa versus agalsidase beta in patients with Fabry disease and deteriorating renal function: results from the 2-year randomised phase III BALANCE study – determination of immunogenicity

OPEN ACCESS
D G Warnock, E L Wallace

Genotype-phenotype correlations

536 Genotype and phenotype correlation of *PHACTR1*-related neurological disorders

Z Xu, L Saddleir, H Goel, X Jiao, Y Niu, Z Zhou, G de Valles-Ibáñez, G Poke, M Hildebrand, N Lieffering, J Qin, Z Yang

543 *GRN* mutation spectrum and genotype-phenotype correlation in Chinese dementia patients: data from PUMCH dementia cohort

C Liu, L Dong, J Wang, J Li, X Huang, D Lei, C Mao, S Chu, L Sha, Q Xu, B Peng, L Cui, J Gao

549 De novo heterozygous missense variants in *CELSR1* as cause of fetal pleural effusions and progressive fetal hydrops

M A de Koning, P A Pimienta Ramirez, M C Haake, X Han, M HA Ruiterkamp-Versteeg, N de Leeuw, U A Schatz, M Shoukier, E Rieger-Fackeldey, J U Ortiz, S G van Duinen, W M Klein, R S G M Witlox, R H Finnell, G W E Santen, Y Lei, M Suerink

553 Bi-allelic variants in chromatoid body protein TDRD6 cause spermiogenesis defects and severe oligoasthenoteratozoospermia in humans

R Guo, H Wu, X Zhu, G Wang, K Hu, K Li, H Geng, C Xu, C Zu, Y Gao, D Tang, Y Cao, X He

Neurogenetics

566 *ZNF142* mutation causes sex-dependent neurologic disorder

R Proskorovski-Ohayon, M Eskin-Schwartz, Z Shorer, R Kadir, D Halperin, M Drabkin, Y Yogev, S Aharoni, N Hadar, H Cohen, E Eremenko, Y Perez, O S Birk

578 Expanding the phenotype of Kleefstra syndrome: speech, language and cognition in 103 individuals

L D Morison, M G P Kennis, D Rots, A Bouman, J Kummeling, E Palmer, A P Vogel, F Liegeois, A Brignell, S Srivastava, Z Frazier, D Milnes, H Goel, D J Amor, I E Scheffer, T Kleefstra, A T Morgan

586 Exploring the molecular pathways linking sleep phenotypes and *POGZ*-associated neurodevelopmental disorder

B P Marquezini, M Moysés-Oliveira, A Kloster, L Cunha, T B Deconto, A C Mosini, P Guerreiro, M Paschalidis, L N G Adami, M L Andersen, S Tufik

Epigenetics

590 Whole-exome sequencing reveals causative genetic variants for several overgrowth syndromes in molecularly negative Beckwith-Wiedemann spectrum

K Higashimoto, F Sun, E Imagawa, K Saida, N Miyake, S Hara, H Yatsuki, M Kubiura-Ichimar, A Fujita, T Mizuguchi, N Matsumoto, H Soejima

595 Skewed X-chromosome inactivation drives the proportion of *DNAIF6*-defective airway motile cilia and variable expressivity in primary ciliary dyskinesia

L Thomas, L Cuisset, J-F Papon, A Tamalet, I Pin, R Abou Taam, C Faucon, G Montantini, S Tissier, P Duquesnoy, F Dastot - Le Moal, B Copin, N Carion, B Louis, S Chantot-Bastarud, J-P Siffroi, R Mitri, A Coste, E Escudier, G Thouvenin, S Amselem, M Legendre

Vision science

605 Cerebral visual impairment: genetic diagnoses and phenotypic associations

OPEN ACCESS
E Shaw, I Flitcroft, R Bowman, K Baker, Genomics England Research Consortium

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/>



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