

Cover credit: Discovery of an *IFNLR1* mutation causing hearing loss and the zebrafish KD phenotype from Gao *X et al*, figures 1 and 3.



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

Contents

Cardiovascular genetics

- 287** New insights into *SERCA2a* gene therapy in heart failure: pay attention to the negative effects of B-type natriuretic peptides
Y Zhai, Y Luo, P Wu, D Li

Novel disease loci

- 298** Mutation of *IFNLR1*, an interferon lambda receptor 1, is associated with autosomal-dominant non-syndromic hearing loss
X Gao, Y-Y Yuan, Q-F Lin, J-C Xu, W-Q Wang, Y-H Qiao, D-Y Kang, D Bai, F Xin, S-S Huang, S-W Qiu, L-P Guan, Y Su, G-J Wang, M-Y Han, Y Jiang, H-K Liu, P Dai

Phenotypes

- 307** Hormonal, metabolic and skeletal phenotype of Schaaf-Yang syndrome: a comparison to Prader-Willi syndrome
J M McCarthy, B M McCann-Crosby, M E Rech, J Yin, C-A Chen, M A Ali, H N Nguyen, J L Miller, C P Schaaf
- 316** Chitayat-Hall and Schaaf-Yang syndromes: a common aetiology: expanding the phenotype of *MAGEL2*-related disorders
R Jobling, D J Stavropoulos, C R Marshall, C Cyttrynbaum, M M Axford, V Londero, S Moalem, J Orr, F Rossignol, F D Lopes, J Gauthier, N Alos, R Rupps, M McKinnon, S Adam, M J M Nowaczyk, S Walker, S W Scherer, C Nassif, F F Hamdan, C L Deal, J-F Soucy, R Weksberg, P Macleod, J L Michaud, D Chitayat

Cancer genetics

- 322** Risk factors for survival in patients with von Hippel-Lindau disease
J-Y Wang, S-H Peng, T Li, X-H Ning, S-J Liu, B-A Hong, J-Y Liu, P-J Wu, B-W Zhou, J-C Zhou, N-N Qi, X Peng, J-F Zhang, K-F Ma, L Cai, K Gong

Genotype-phenotype correlations


- 329** Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome
N Calmels, E Botta, N Jia, H Fawcett, T Nardo, Y Nakazawa, M Lanzafame, S Moriwaki, K Sugita, M Kubota, C Obringer, M-A Spitz, M Stefanini, V Laugel, D Orioli, T Ogi, A R Lehmann

Screening

- 344** Evaluation of the relative effectiveness of the 2017 updated Manchester scoring system for predicting *BRCA1/2* mutations in a Southeast Asian country
W Chew, R B Moorakonda, E Courtney, H Soh, S T Li, Y Chen, T Shaw, J C Allen, D Gareth R Evans, J Ngeow

Therapeutics

- 351** Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study
M Arends, M Biegstraaten, C Wanner, S Sirrs, A Mehta, P M Elliott, D Oder, O T Watkinson, D G Bichet, A Khan, M Iwanochko, F M Vaz, A B P van Kuilenburg, M L West, D A Hughes, C E M Hollak

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

EDITOR'S CHOICE


 This article has been made freely available online under the BMJ Journals Open Access scheme. See <http://authors.bmj.com/open-access/>

OPEN ACCESS

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

 equator network

 recycle
When you have finished with this please recycle it

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