

Journal of Medical Genetics

December 2017 Volume 54 Issue 12

JMG

Journal of Medical Genetics



Cover credit: Examples of vertebrates with disease-mimicking phenotypes from Emerling CA, *et al.*, page 792. Silhouettes and associated licenses from phylogic.org. Painting by Carl Buell (copyright John Gatesy).



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

Contents

Evolutionary genetics

- 787** Their loss is our gain: regressive evolution in vertebrates provides genomic models for uncovering human disease loci
C A Emerling, A D Widjaja, N N Nguyen, M S Springer

Diagnostics

- 795** Clinical genetic testing using a custom-designed steroid-resistant nephrotic syndrome gene panel: analysis and recommendations
E S Sen, P Dean, L Yarram-Smith, A Bierzynska, G Woodward, C Buxton, G Dennis, G I Welsh, M Williams, M A Saleem

Cancer genetics

- 805** Acute myeloid leukaemia in a case with Tatton-Brown-Rahman syndrome: the peculiar DNMT3A R882 mutation
I H I M Hollink, A M W van den Ouweland, H B Beverloo, S T C J M Arentsen-Peters, C M Zwaan, A Wagner

Neurogenetics

- 809** Confirmation of mutations in *PROSC* as a novel cause of vitamin B₆-dependent epilepsy
B Plecko, M Zwieler, A Begemann, D Mathis, B Schmitt, P Striano, M Baethmann, M S Vari, F Beccaria, F Zara, L M Crowther, P Joset, H Sticht, S M Papuc, A Rauch

- 815** A novel de novo dominant mutation in *ISCU* associated with mitochondrial myopathy

A Legati, A Reyes, C C Berti, O Stehling, S Marchet, C Lamperti, A Fenari, A J Robinson, U Mühlenhoff, R Lill, M Zeviani, P Goffrini, D Ghezzi

Developmental defects

- 825** Loss of function in *ROBO1* is associated with tetralogy of Fallot and septal defects
P Kruszka, P Tanpaiboon, K Neas, K Crosby, S I Berger, A F Martinez, Y A Addissie, Y Pongprot, R Sittivangkul, S Silvilairat, K Makonkawkeeyoon, L Yu, J Wynn, J T Bennett, H C Mefford, W T Reynolds, X Liu, M T M Mommersteeg, W K Chung, C W Lo, M Muenke

December 2017 Volume 54 Issue 12

- 830** Expanding the clinical spectrum of recessive truncating mutations of *KLHL7* to a Bohring-Opitz-like phenotype

A-L Bruel, S Bigoni, J Kennedy, M Whiteford, C Buxton, G Parmeggiani, M Wherlock, G Woodward, M Greenslade, M Williams, J St-Onge, A Ferlini, G Garani, E Ballardini, B W van Bon, R Acuna-Hidalgo, A Bohring, J-F Deleuze, A Boland, V Meyer, R Olaso, E Ginglinger, DDD Study, J-B Rivière, H G Brunner, A Hoischen, R Newbury-Ecob, L Faivre, C Thauvin-Robinet, J Thevenon

Epigenetics

- 836** *CTCF* deletion syndrome: clinical features and epigenetic delineation

I Hori, R Kawamura, K Nakabayashi, H Watanabe, K Higashimoto, J Tomikawa, D Ieda, K Ohashi, Y Negishi, A Hattori, Y Sugio, K Wakui, K Hata, H Soejima, K Kurosawa, S Saïtoh

Biochemical genetics

- 843** Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature

M Schiuff, C Roda, M-L Monin, A Arion, M Barth, N Bednarek, M Bidet, C Bloch, N Boddard, D Borgel, A Brassier, A Brice, A Bruneel, R Buissonnière, B Chabrol, M-C Chevalier, V Cormier-Daire, C De Barce, E De Maistre, A De Saint-Martin, N Dorison, V Drouin-Garraud, T Dupré, B Echenne, P Ederly, F Feillet, I Fontan, C Francannet, F Labarthe, C Gitiaux, D Héron, M Hully, S Lamoureux, D Martin-Coignard, C Mignot, G Morin, T Pascreau, O Pincemaille, M Polak, A Roubertie, C Thauvin-Robinet, A Toutain, G Viot, S Vuillaumier-Barrot, N Seta, P De Lonlay

Immunogenetics

- 853** Genetic landscape of interactive effects of *HLA-DRB1* alleles on susceptibility to ACPA(+) rheumatoid arthritis and ACPA levels in Japanese population

C Terao, Y Okada, K Ikari, Y Kochi, A Suzuki, K Ohmura, K Matsuo, A Taniguchi, M Kubo, S Raychaudhuri, K Yamamoto, H Yamanaka, Y Kamatani, T Mimori, F Matsuda

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/>

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.



54 (12)

J Med Genet 2017 54: 787-858

Updated information and services can be found at:
<http://jmg.bmj.com/content/54/12>

These include:

**Email alerting
service**

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
<http://group.bmj.com/group/rights-licensing/permissions>

To order reprints go to:
<http://journals.bmj.com/cgi/reprintform>

To subscribe to BMJ go to:
<http://group.bmj.com/subscribe/>