

## Contents

### Congenital malformations

- 635** Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update  
*A Beza-Meireles, J Clayton-Smith, J M Saraiva, M Tassabehji*

### Complex traits

- 646** Moving beyond genetics: is *FAM13A* a major biological contributor in lung physiology and chronic lung diseases? *H Corvol, C A Hodges, M L Drumm, L Guillot*

### Genotype-phenotype correlations

- 650** Genotype phenotype associations across the voltage-gated sodium channel family  
*A Brunklaus, R Ellis, E Reavey, C Semsarian, S M Zuberi*
- 659** Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism  
*M Ansari, G Poke, Q Ferry, K Williamson, R Aldridge, A M Meynert, H Bengani, C Y Chan, H Kayserili, Ş Avci, R C M Hennekam, A K Lampe, E Redeker, T Homfray, A Ross, M F Smeland, S Mansour, M J Parker, J A Cook, M Split, R B Fisher, A Fryer, A C Magee, A Wilkie, A Barnicoat, A F Brady, N S Cooper, C Mercer, C Deshpande, C P Bennett, D T Pitz, D Ruddy, D Gilliers, D S Johnson, D Josifova, E Rosser, E M Thompson, E Wakeling, E Kinning, F Stewart, F Flinter, K M Girisha, H Cox, H V Firth, H Kingston, J S Wee, J A Hurst, J Clayton-Smith, J Tolmie, J Vogt, K Tatton-Brown, K Chandler, K Prescott, L Wilson, M Behnam, M McEntagart, R Davidson, S-A Lynch, S Sisodiya, S G Mehta, S A McKee, S Mohammed, S Holden, S-M Park, S E Holder, V Harrison, V McConnell, W K Lam, A J Green, D Donnai, M Bitner-Glindzicz, D E Donnelly, C Nelläker, M S Taylor, D R FitzPatrick*

- 669** Titin and desmosomal genes in the natural history of arrhythmogenic right ventricular cardiomyopathy  
*F Brun, C V Barnes, G Sinagra, D Slavov, G Barbati, X Zhu, S L Graw, A Spezzacatene, B Pinamonti, M Merlo, E E Salcedo, W H Sauer, M R G Taylor, L Mestroni, on behalf of the Familial Cardiomyopathy Registry*

### Genome-wide studies

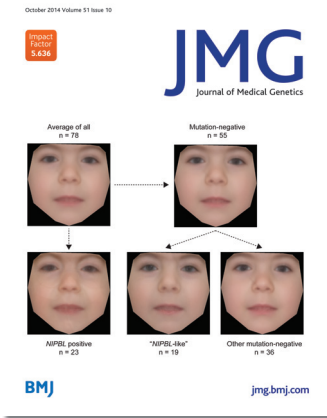
- 677** The clinical significance of small copy number variants in neurodevelopmental disorders  
*R Asadollahi, B Oneda, P Joset, S Azzarello-Burri, D Bartholdi, K Steindl, M Vincent, J Cobilanschi, H Sücht, R Baldinger, R Reissmann, I Sudholt, C T Thiel, A B Ekeci, A Reis, E K Bijlsma, J Andrieux, A Dieux, D FitzPatrick, S Rüter, A Baumer, B Latal, B Plecko, O G Jenni, A Rauch*

### Cancer genetics

- 689** Juvenile myelomonocytic leukaemia and Noonan syndrome  
*M Strullu, A Caye, J Lachenaud, B Cassinat, S Gazal, O Fenneteau, N Pouvreau, S Pereira, C Baumann, A Conet, N Sirvent, F Méchinand, I Guellec, D Adjaoud, C Paillard, C Alberti, M Zenker, C Chomienne, Y Bertrand, A Baruchel, A Verloes, H Cavé*

### New loci

- 699** Exome sequencing identifies *SLC17A9* pathogenic gene in two Chinese pedigrees with disseminated superficial actinic porokeratosis  
*H Cui, L Li, W Wang, J Shen, Z Yue, X Zheng, X Zuo, B Liang, M Gao, X Fan, X Yin, C Shen, C Yang, C Zhang, X Zhang, Y Sheng, J Gao, Z Zhu, D Lin, A Zhang, Z Wang, S Liu, L Sun, S Yang, Y Cui, X Zhang*



Cover credit: Computational reconstruction of average facial appearance, by genotype, from Ansari *et al.* pg 659.

Receive regular table of contents by email. Register using this QR code.



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://jmg.bmj.com/site/about/guidelines.xhtml#open>



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.