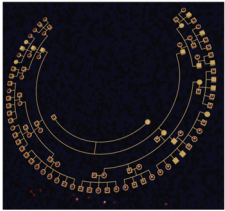


April 2013 Volume 50 Issue 4

**JMG**  
Journal of Medical Genetics



BMJ Journals

[jmg.bmj.com](http://jmg.bmj.com)

Cover credit: Dominant Jewelry.  
Stylised depiction of a large dominant pedigree. Ray-tracing rendition by Constantin Polychronakos.

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>

**C O P E** COMMITTEE ON PUBLICATION ETHICS

This journal is a member of and subscribes to the principles of the Committee on Publication Ethics  
[www.publicationethics.org.uk](http://www.publicationethics.org.uk)

**equator**  
network

**recycle**  
When you have finished with this please recycle it

## Contents

### Review

- 203** From the periphery to centre stage: de novo single nucleotide variants play a key role in human genetic disease *C-S Ku, E K Tan, D N Cooper*

### Complex traits

- 212** Meta-analysis identifies a *MECOM* gene as a novel predisposing factor of osteoporotic fracture *J-Y Hwang, S H Lee, M J Go, B-J Kim, I Kou, S Ikegawa, Y Guo, H-W Deng, S Raychaudhuri, Y J Kim, J H Oh, Y Kim, S Moon, D-J Kim, H Koo, M-J Cha, M H Lee, J Y Yun, H-S Yoo, Y-A Kang, E-H Cho, S-W Kim, K W Oh, M I Kang, H Y Son, S-Y Kim, G S Kim, B-G Han, Y S Cho, M-C Cho, J-Y Lee, J-M Koh*

### Genotype-phenotype correlations

- 220** CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders *C Thauvin-Robinet, A Munck, F Huet, A de Becdelièvre, C Jimenez, G Lalau, E Gautier, J Rollet, J Flori, R Nové-Josserand, J-C Soufir, A Haloun, D Hubert, E Houssin, G Bellis, G Rault, A David, L Janny, R Chiron, N Rives, D Hairion, P Collignon, A Valeri, G Karsenty, A Rossi, M-P Audrézet, C Férec, J Leclerc, M des Georges, M Claustres, T Bienvenu, B Gérard, P Boisseau, F Cabet-Bey, D Cheillan, D Feldmann, C Clavel, E Bieth, A Iron, B Simon-Bouy, V Izard, J Steffann, S Viville, C Costa, V Drouineaud, P Fauque, C Binquet, C Bonithon-Kopp, M A Morris, L Faivre, M Goossens, M Roussey, E Girodon, the collaborating working group on p.Arg117His*

- 228** Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing *L R Lopes, A Zekavati, P Syrris, M Hubank, C Giambartolomei, C Dalageorgou, S Jenkins, W McKenna, UK10k Consortium, V Plagnol, P M Elliott*

### New loci

- 240** Agenesis of corpus callosum and optic nerve hypoplasia due to mutations in *SLC25A1* encoding the mitochondrial citrate transporter *S Edvardson, V Porcelli, C Jalas, D Soiferman, Y Kellner, A Shaag, S H Korman, C L Pierri, P Scaria, N D Fraenkel, R Segel, A Schechter, A Frumkin, O Pines, A Saada, L Palmieri, O Elpeleg*

### Vision science

- 246** Whole exome sequencing identifies a mutation for a novel form of corneal intraepithelial dyskeratosis *V J Soler, K-N Tran-Viet, S D Galiacy, V Limviphuvadh, T P Klemm, E St Germain, P R Fournié, C Guillaud, S Maurer-Stroh, F Hawthorne, C Suarez, B Kantelip, N A Afshari, I Creveaux, X Luo, W Meng, P Calvas, M Cassagne, J-L Arné, S G Rozen, F Malecaze, T L Young*

### Cancer genetics

- 255** High cumulative risks of cancer in patients with *PTEN* hamartoma tumour syndrome *V Bubié, F Bonnet, V Brouste, S Hoppe, E Barouk-Simonet, A David, P Eder, A Botani, V Layet, O Caron, B Gilbert-Dussardier, C Delnatte, C Dugast, J-P Fricker, D Bonneau, N Sevenet, M Longy, F Caux, French Cowden Disease Network*

- 264** Melanoma prone families with *CDK4* germline mutation: phenotypic profile and associations with *MC1R* variants *H E Puntervoll, X R Yang, H H Vetti, I M Bachmann, M F Avril, M Benfodda, C Catricalà, S Dalle, A B Duval-Modeste, P Ghiorzo, P Grammatico, M Harland, N K Hayward, H-H Hu, T Jouary, T Martin-Denavit, A Ozola, J M Palmer, L Pastorino, D Pjanova, N Soufir, S J Steine, A J Straigos, L Thomas, J Tinat, H Tsao, R Vignalde, M A Tucker, B B-de Paillerets, J A Newton-Bishop, A M Goldstein, L A Akslen, A Molven*

### Correction

- 270** Correction