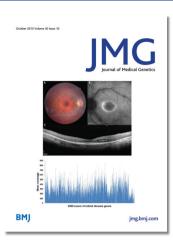
Journal of Medical Genetics

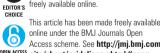


Cover credit: Exon coverage for NGS diagnosis of retinal disorders from Wang et al, p 674 in this issue.

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.



online under the BM.L. Journals Open Access scheme. See http://jmj.bmj.com/ OPEN ACCESS site/about/quidelines.xhtml#open



This journal is a member of and subscribes to the principles of the Committee on Publication

www.publicationethics.org.uk





Contents

Reviews

641 Digenic inheritance in medical genetics A A Schäffer

653 An overview on molecular biology of KIT/ PDGFRA wild type (WT) gastrointestinal stromal tumours (GIST) M Nannini, G Biasco, A Astolfi, M A Pantaleo

Developmental defects

662 Bicuspid aortic valve and aortic coarctation are linked to deletion of the X chromosome short arm in Turner syndrome C Bondy, V K Bakalov,

C Cheng, L Olivieri, D R Rosing, A E Arai

Complex traits

666 Large-scale genotyping identifies a new locus at 22g13.2 associated with female breast size J Li, J N Foo, N Schoof, J S Varghese, P Fernandez-Navarro, G L Gierach, S T Quek, M Hartman, S Nord, V N Kristensen, M Pollán, I D Figueroa, D I Thompson, Y Li, C C Khor, K Humphreys, J Liu, K Czene, P Hall

Genotype-phenotype correlations

674 Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next

generation sequencing X Wang, H Wang, V Sun, H-F Tuan, V Keser, K Wang, H Ren, I Lopez, J E Zaneveld, S Siddiqui, S Bowles, A Khan, J Salvo, S G Jacobson, A Jannaccone, F Wang, D Birch, I R Heckenlively, G A Fishman, E I Traboulsi, Y Li, D Wheaton, R K Koenekoop, R Chen

October 2013 Volume 50 Issue 10

Cancer genetics

689 Confirmation of papillary thyroid cancer susceptibility loci identified by genome-wide association studies of chromosomes 14g13, 9g22, 2g35 and 8p12 in a Chinese population Y-L Wang, S-H Feng, S-C Guo, W-J Wei, D-S Li, Y Wang, X Wang, Z-Y Wang, Y-Y Ma, L Jin, Q-H Ji, J-C Wang

Copy-number variation

696 CAG size-specific risk estimates for intermediate allele repeat instability in Huntington disease A Semaka, C Kay, C Doty, J A Collins, E K Bijlsma, F Richards, Y P Goldberg, M R Havden

Mutation report

704 Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders

S Bannwarth, V Procaccio, A S Lebre, C Iardel, A Chaussenot, C Hoarau, H Maoulida, N Charrier, X Gai, H M Xie, M Ferre, K Fragaki, G Hardy, B M de Camaret, S Marlin, C M Dhaenens, A Slama, C Rocher, J P Bonnefont, A Rötig, N Aoutil, M Gilleron, V Desquiret-Dumas, P Reynier, J Ceresuela, L Jonard, A Devos, C Espil-Taris, D Martinez, P Gaignard, K-H L Q Sang, P Amati-Bonneau, M J Falk, C Florentz, B Chabrol, I Durand-Zaleski, V Paquis-Flucklinger

