



Contents

Review

- 793** Constitutional (germline) *MLH1* epimutation as an aetiological mechanism for hereditary nonpolyposis colorectal cancer *M P Hitchins, R L Ward*

Original articles

- 803** Clinical and cellular characterisation of Hermansky–Pudlak syndrome type 6 *M Huizing, B Pederson, R A Hess, A Griffin, A Helip-Wooley, W Westbroek, H Dorward, K J O'Brien, G Golas, E Tsilou, J G White, W A Gahl*
- 811** Addition of pathology and biomarker information significantly improves the performance of the Manchester scoring system for *BRCA1* and *BRCA2* testing *D G R Evans, F Lalloo, A Cramer, E A Jones, F Knox, E Amir, A Howell*
- 818** Penetrance of marked cognitive impairment in older male carriers of the *FMR1* gene premutation *M Sévin, Z Kutalik, S Bergman, M Vercelletto, P Renou, E Lamy, F J Vingerhoets, G Di Virgilio, P Boisseau, S Bezieau, L Pasquier, J-M Rival, J S Beckmann, P Damier, S Jacquemont*
- 825** Microdeletions including *YWHAE* in the Miller–Dieker syndrome region on chromosome 17p13.3 result in facial dysmorphisms, growth restriction, and cognitive impairment *S C Sreenath Nagamani, F Zhang, O A Shchelochkov, W Bi, Z Ou, F Scaglia, F J Probst, M Shinawi, C Eng, J V Hunter, S Sparagana, E Lagoë, C-T Fong, M Pearson, M Doco-Fenzy, E Landais, M Mozelle, A C Chinault, A Patel, C A Bacino, T Sahoo, S H Kang, S W Cheung, J R Lupski, P Stankiewicz*

December 2009 Volume 46 Issue 12

- 834** Enhancer deletions of the *SHOX* gene as a frequent cause of short stature: the essential role of a 250 kb downstream regulatory domain *J Chen, G Wildhardt, Z Zhong, R Röth, B Weiss, D Steinberger, J Decker, W F Blum, G Rappold*

Short report

- 840** Meta-analysis of vascular endothelial growth factor variations in amyotrophic lateral sclerosis: increased susceptibility in male carriers of the –2578AA genotype *D Lambrechts, K Poesen, R Fernández-Santiago, A Al-Chalabi, R Del Bo, P W J Van Vught, S Khan, S L Marklund, A Brockington, I van Marion, J Anneser, C Shaw, A C Ludolph, N P Leigh, G P Comi, T Gasser, P J Shaw, K E Morrison, P M Andersen, L H Van den Berg, V Thijs, T Siddique, W Robberecht, P Carmeliet*

Letters to JMG

- 847** 2q23.1 microdeletion identified by array comparative genomic hybridisation: an emerging phenotype with Angelman-like features? *S Jaillard, C Dubourg, M Gérard-Blanluet, A Delahaye, L Pasquier, C Dupont, C Henry, A-C Tabet, J Lucas, A Aboura, V David, B Benzacken, S Odent, E Pipiras*
- 856** Segregation of enlarged vestibular aqueducts in families with non-diagnostic *SLC26A4* genotypes *B Y Choi, A C Madeo, K A King, C K Zalewski, S P Pryor, J A Muskett, W E Nance, J A Butman, C C Brewer, A J Griffith*

Mutation report

- 862** Contribution of *RET*, *NTRK3* and *EDN3* to the expression of Hirschsprung disease in a multiplex family *A Sánchez-Mejías, R M Fernández, M López-Alonso, G Antiñolo, S Borrego*



EDITOR'S CHOICE

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



Articles carrying the Unlocked Logo are freely available online under the BMJ Journals unlocked scheme.

See <http://jmg.bmj.com/info/unlocked.dtl>



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

www.publicationethics.org.uk



When you have finished with this please recycle it