



## Contents

### Reviews

- 769** Pseudomitochondrial genome haunts disease studies *Y-G Yao, Q-P Kong, A Salas, H-J Bandelt*
- 773** Genetics of infantile seizures with paroxysmal dyskinesia: the infantile convulsions and choreoathetosis (ICCA) and ICCA-related syndromes *J Rochette, P Roll, P Szepietowski*

### Original articles

- 780** A PCSK9 variant and familial combined hyperlipidaemia *M Abifadel, L Bernier, G Dubuc, G Nuel, J-P Rabès, J Bonneau, A Marques, M Marduel, M Devillers, A Munnich, D Erlich, M Varret, M Roy, J Davignon, C Boileau*
- 787** Mutations in *JARID1C* are associated with X-linked mental retardation, short stature and hyperreflexia *F E Abidi, L Holloway, C A Moore, D D Weaver, R J Simensen, R E Stevenson, R C Rogers, C E Schwartz*
- 794** Reversible phenotype in a mouse model of Hutchinson–Gilford progeria syndrome *H Sagelius, Y Rosengarten, E Schmidt, C Sonnabend, B Rozell, M Eriksson*
- 802** Neurodevelopmental abnormalities associated with severe congenital neutropenia due to the R86X mutation in the *HAX1* gene *N Ishikawa, S Okada, M Miki, K Shirao, H Kihara, M Tsumura, K Nakamura, H Kawaguchi, M Ohtsubo, S Yasunaga, K Matsubara, M Sako, J Hara, M Shiohara, S Kojima, T Sato, Y Takihara, M Kobayashi*

December 2008 Vol 45 No 12

- 808** DNA methylation in intron 1 of the frataxin gene is related to GAA repeat length and age of onset in Friedreich ataxia patients *I Castaldo, M Pinelli, A Monticelli, F Acquaviva, M Giacchetti, A Filla, S Sacchetti, S Keller, V E Avvedimento, L Chiariotti, S Cocozza*
- 813** Association of a null allele of *SPRN* with variant Creutzfeldt–Jakob disease *J A Beck, T A Campbell, G Adamson, M Poulter, J B Uphill, E Molou, J Collinge, S Mead*

### Letters to JMG

- 818** Association of haplotypes spanning *PDZ-GEF2*, *LOC728637* and *ACSL6* with schizophrenia in Han Chinese *X-j Luo, H-b Diao, J-k Wang, H Zhang, Z-m Zhao, B Su*
- 827** A novel mutation in the sulfate transporter gene *SLC26A2 (DTDST)* specific to the Finnish population causes de la Chapelle dysplasia *L Bonafé, J Hästbacka, A de la Chapelle, A B Campos-Xavier, C Chiesa, A Forlino, A Superti-Furga, A Rossi*

### PostScript

- 832** Correspondence
- 832** Corrections



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>

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)

