# Journal of Medical Genetics



## **Contents**

#### **Review**

Clinical and genetic delineation of neurodegeneration with brain iron accumulation

A Gregory, B J Polster, S J Hayflick

#### **Original articles**

- 81 Longitudinal study of neurofibromatosis 1 associated plexiform neurofibromas T Tucker, J M Friedman, R E Friedrich, R Wenzel, C Fünsterer, V-F Mautner
- **86** Chromosome 15q11–13 duplication syndrome brain reveals epigenetic alterations in gene expression not predicted from copy number *A Hogart, K N Leung, N J Wang, D J Wu, J Driscoll, R O Vallero, N C Schanen, J M LaSalle*
- 94 Minocycline promotes dendritic spine maturation and improves behavioural performance in the fragile X mouse model T V Bilousova, L Dansie, M Ngo, J Aye, J R Charles, D W Ethell, I M Ethell
- 103 Novel transglutaminase-1 mutations and genotype—phenotype investigations of 104 patients with autosomal recessive congenital ichthyosis in the USA S Farasat, M-H Wei, M Herman, D J Liewehr, S M Steinberg, S J Bale, P Fleckman, J R Toro

#### **Letters to JMG**

**112** Contribution of *TARDBP* mutations to sporadic amyotrophic lateral sclerosis *H Daoud*, *P N Valdmanis*, *E Kabashi*, *P Dion*, *N Dupré*, *W Camu*, *V Meininger*, *G A Rouleau* 

### February 2009 Vol 46 No 2

- Investigation of somatic *NKX2-5* mutations in
- 6 congenital heart disease J M Draus Jr, M A Hauck, M Goetsch, E H Austin III, A Tomita-Mitchell, M E Mitchell
- Detection of cryptic pathogenic copy number variations and constitutional loss of heterozygosity using high resolution SNP microarray analysis in 117 patients referred for cytogenetic analysis and impact on clinical practice D L Bruno, D Ganesamoorthy, J Schoumans, A Bankier, D Coman, M Delatycki, R J M Gardner, M Hunter, P A James, P Kannu, G McGillivray, N Pachter, H Peters, C Rieubland, R Savarirayan, I E Scheffer, L Sheffield, T Tan, S M White, A Yeung, Z Bowman, C Ngo, K W Choy, V Cacheux, L Wong, D J Amor, H R Slater
- 132 Effect of CHEK2 missense variant I157T on the risk of breast cancer in carriers of other CHEK2 or BRCA1 mutations C Cybulski, B Górski, T Huzarski, T Byrski, J Gronwald, T Debniak, D Wokołorczyk, A Jakubowska, P Serrano-Fernández, T Dork, S A Narod, J Lubiński

#### **Mutation report**

in patients with autosomal dominant optic atrophy N Fuhrmann, M V Alavi, P Bitoun, S Woernle, G Auburger, B Leo-Kottler, P Yu-Wai-Man, P Chinnery, B Wissinger



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

