



## Contents

October 2008 Vol 45 No 10

### Reviews

- 609** Trichothiodystrophy: a systematic review of 112 published cases characterises a wide spectrum of clinical manifestations *S Faghri, D Tamura, K H Kraemer, J J DiGiovanna*
- 622** Mosaicism in sporadic neurofibromatosis type 1: variations on a theme common to other hereditary cancer syndromes?  
*H Kehrer-Sawatzki, D N Cooper*

### Commentary

- 632** Is genetics inhumane? *A Munnich*

### Original articles

- 635** Congenital heart disease is a feature of severe infantile spinal muscular atrophy  
*S Rudnik-Schöneborn, R Heller, C Berg, C Betzler, T Grimm, T Eggermann, K Eggermann, R Wirth, B Wirth, K Zerres*
- 639** The CTG repeat expansion size correlates with the splicing defects observed in muscles from myotonic dystrophy type 1 patients *A Botta, F Rinaldi, C Catalli, L Vergani, E Bonifazi, V Romeo, E Loro, A Viola, C Angelini, G Novelli*
- 647** Refinement of cortical dysgeneses spectrum associated with *TUBA1A* mutations  
*N Bahi-Buisson, K Poirier, N Boddaert, Y Saillour, L Castelnau, N Philip, G Buyse, L Villard, S Joriot, S Marret, M Bourgeois, H Van Esch, L Lagae, J Amiel, L Hertz-Pannier, A Roubertie, F Rivier, J M Pinard, C Beldjord, J Chelly*

### Short reports


- 654** A genome-wide association study identifies an association of a common variant in *TERT* with susceptibility to idiopathic pulmonary fibrosis  
*T Mushiroda, S Wattanapokayakit, A Takahashi, T Nukiwa, S Kudoh, T Ogura, H Taniguchi, M Kubo, N Kamatani, Y Nakamura, the Pirfenidone Clinical Study Group*
- 657** Contribution of *PTEN* large rearrangements in Cowden disease: a multiplex amplifiable probe hybridisation (MAPH) screening approach  
*F Chibon, C Primois, J-M Bressieux, D Lacombe, C Lok, L Mauriac, A Taieb, M Longy*


### Letter to JMG

- 666** Heart-hand syndrome of Slovenian type: a new kind of laminopathy *L Renou, S Stora, R Ben Yaou, M Volk, M Šinkovec, L Demay, P Richard, B Peterlin, G Bonne*
- 672** Mapping of 5q35 chromosomal rearrangements within a genomically unstable region *K Buysse, A Crepel, B Menten, F Pattyn, F Antonacci, J A Veltman, L A Larsen, Z Tümer, A de Klein, I van de Laar, K Devriendt, G Mortier, F Speleman*
- 679** Alteration of expression of muscle specific isoforms of the fragile X related protein 1 (FXR1P) in facioscapulohumeral muscular dystrophy patients *L Davidovic, S Sacconi, E G Bechara, S Delplace, M Allegra, C Desnuelle, B Bardoni*

### Mutation report

- 686** Early onset of hypokalaemic periodic paralysis caused by a novel mutation of the *CACNA1S* gene *S Chabrier, N Monnier, J Lunardi*

 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)

 equator network