

Contents



This logo indicates an article that was published online first ahead of print

Original articles

- 625** Emerging patterns of cryptic chromosomal imbalance in patients with idiopathic mental retardation and multiple congenital anomalies: a new series of 140 patients and review of published reports *B Menten, N Maas, B Thienpont, K Buysse, J Vandesompele, C Melotte, T de Ravel, S Van Vooren, I Balikova, L Backx, S Janssens, A De Paepe, B De Moor, Y Moreau, P Marynen, J-P Fryns, G Mortier, K Devriendt, F Speleman, J R Vermeesch*
- 634** Mutations of human *TMHS* cause recessively inherited non-syndromic hearing loss *M I Shabbir, Z M Ahmed, S Y Khan, Saima Riazuddin, A M Waryah, S N Khan, R D Camps, M Ghosh, M Kabra, I A Belyantseva, T B Friedman, Sheikh Riazuddin*
- 641** Epidermolysis bullosa. I. Molecular genetics of the junctional and hemidesmosomal variants *R Varki, S Sadowski, E Pfendner, J Uitto*
- 653** Functional and clinical characterization of a mutation in *KCNJ2* associated with Andersen-Tawil syndrome *C-W Lu, J-H Lin, Y S Rajawat, H Jerng, T G Rami, X Sanchez, G DeFreitas, B Carabello, F DeMayo, D L Kearney, G Miller, H Li, P J Pfaffinger, N E Bowles, D S Khoury, J A Towbin*
- 660** Redefining the risks of prenatally ascertained supernumerary marker chromosomes: a collaborative study *M D Graf, L Christ, J T Mascarello, P Mowrey, M Pettenati, G Stetten, P Storto, U Surti, D L Van Dyke, G H Vance, D Wolff, S Schwartz*

Medical genetics in practice

- 665** Letting the family know: balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder *G K Suthers, J Armstrong, J McCormack, D Trott*

Letters to JMG

- 671** Tissue specific distribution of the 3243A→G mtDNA mutation *A L Frederiksen, P H Andersen, K O Kyvik, T D Jeppesen, J Vissing, M Schwartz*

- 678** Altered CD45 expression in C77G carriers influences immune function and outcome of hepatitis C infection *R Dawes, B Hennig, W Irving, S Petrova, S Boxall, V Ward, D Wallace, D C Macallan, M Thursz, A Hill, W Bodmer, P C L Beverley, E Z Tchilian*
- 685** Biochemical screening of type I collagen in osteogenesis imperfecta: detection of glycine substitutions in the amino end of the alpha chains requires supplementation by molecular analysis *W A Cabral, S Milgrom, A D Letocha, E Moriarty, J C Marini*
- 691** Predicting disease genes using protein-protein interactions *M Oti, B Snel, M A Huynen, H G Brunner*
- 699** X linked cone-rod dystrophy, *CORDX3*, is caused by a mutation in the *CACNA1F* gene *R Jalkanen, M Mäntyjärvi, R Tobias, J Isosomppi, E-M Sankila, T Alitalo, N T Bech-Hansen*

Electronic letters

- e39** Molecular and genomic characterisation of cryptic chromosomal alterations leading to paternal duplication of the 11p15.5 Beckwith-Wiedemann region *S Russo, P Finelli, M P Recalcati, S Ferraiuolo, F Cogliati, B Dalla Bernardina, M G Tibiletti, M Agosti, M Sala, M T Bonati, L Larizza*
- e40** Genetic predictors for acute experimental cold and heat pain sensitivity in humans *H Kim, D P Mittal, M J Iadarola, R A Dionne*
- e41** *STK11* status and intussusception risk in Peutz-Jeghers syndrome *N Hearle, V Schumacher, F H Menko, S Olschwang, L A Boardman, J J P Gille, J J Keller, A M Westerman, R J Scott, W Lim, J D Trimbath, F M Giardiello, S B Gruber, G J A Offerhaus, F W M D E Rooij, J H P Wilson, A Hansmann, G Möslein, B Royer-Pokora, T Vogel, R K S Phillips, A D Spigelman, R S Houlston*
- e42** *CYP19* haplotypes increase risk for Alzheimer's disease *R Huang, S E Poduslo*

JMG Unlocked

Articles carrying the



logo are available free via the journal's website: www.jmedgenet.com. Funding for this open access experiment is provided by the UK's Joint Information Systems Committee (JISC); see <http://jmg.bmjournals.com/cgi/content/full/42/2/97> for further information.

contd...

Contents ...contd

- e43** Association of lung function decline with the heme oxygenase-1 gene promoter microsatellite polymorphism in a general population sample. Results from the European Community Respiratory Health Survey (ECRHS), France A Guénéguou, B Leynaert, J Bénessiano, I Pin, P Demoly, F Neukirch, J Boczkowski, M Aubier

Correspondences

- e44** Reply to Dr Raux *et al.*: Molecular diagnosis of autosomal dominant early onset Alzheimer's disease: an update (*J Med Genet* 2005;42:793-5)
A J Larner, M Doran
- e45** Somatic mutations in cardiac malformations S M Reamon-Buettner, J Borlak