

Contents



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

Reviews

- 705** Syndromes and constitutional chromosomal abnormalities associated with Wilms tumour *R H Scott, C A Stiller, L Walker, N Rahman*
- 716** Cerebral cavernous malformation: new molecular and clinical insights *N Revencu, M Vikkula*

Original articles

- 722** Hereditary haemorrhagic telangiectasia: mutation detection, test sensitivity and novel mutations *N L Prigoda, S Savas, S A Abdalla, B Piovesan, D Rushlow, K Vandezande, E Zhang, H Ozcelik, B L Gallie, M Letarte*
- 729** CDKL5 mutations cause infantile spasms, early onset seizures, and severe mental retardation in female patients *H L Archer, J Evans, S Edwards, J Colley, R Newbury-Ecob, F O'Callaghan, M Huyton, M O'Regan, J Tolmie, J Sampson, A Clarke, J Osborne*
- 735** High incidence of SHOX anomalies in individuals with short stature *C Huber, M Rosilio, A Munnich, V Cormier-Daire, the French SHOX GeNeSIS Module*
- 740** Polymorphisms in the VKORC1 gene are strongly associated with warfarin dosage requirements in patients receiving anticoagulation *T Li, L A Lange, X Li, L Susswein, B Bryant, R Malone, E M Lange, T-Y Huang, D W Stafford, J P Evans*
- 745** Polymorphisms in the xylosyltransferase genes cause higher serum XT-I activity in patients with pseudoxanthoma elasticum (PXE) and are involved in a severe disease course *S Schön, V Schulz, C Prante, D Hendig, C Szliska, J Kuhn, K Kleesiek, C Götting*

Short report

- 750** Fanconi anaemia complementation group B presenting as X linked VACTERL with hydrocephalus syndrome *S T Holden, J J Cox, I Kesterton, N S Thomas, C Carr, C G Woods*

Letters to JMG

- 755** Fumarate hydratase enzyme activity in lymphoblastoid cells and fibroblasts of individuals in families with hereditary leiomyomatosis and renal cell cancer *M Pithukpakorn, M-H Wei, O Toure, P J Steinbach, G M Glenn, B Zbar, W M Linehan, J R Toro*

- 763** Survey of the frequency of USH1 gene mutations in a cohort of Usher patients shows the importance of cadherin 23 and protocadherin 15 genes and establishes a detection rate of above 90% *A-F Roux, V Faugère, S Le Guédard, N Pallares-Ruiz, A Vielle, S Chambert, S Marlin, C Hamel, B Gilbert, S Malcolm, M Claustres, for the French Usher Syndrome Collaboration*

Electronic letters

- e46** A novel mutation in the mitochondrial tRNA^{Ser(AGY)} gene associated with mitochondrial myopathy, encephalopathy, and complex I deficiency *L-J C Wong, D Yim, R-K Bai, H Kwon, M M Vacek, J Zane, C L Hoppel, D S Kerr*
- e47** Measurement of mRNA of trophoblast-specific genes in cellular and plasma components of maternal blood *S Okazaki, A Sekizawa, Y Purwosunu, M Iwasaki, A Farina, T Okai*

Online mutation reports

- e48** Novel NHLRC1 mutations and genotype-phenotype correlations in patients with Lafora's progressive myoclonic epilepsy *S Singh, I Sethi, S Francheschetti, C Riggio, G Avanzini, K Yamakawa, A V Delgado-Escueta, S Ganesh*
- e49** The contribution of germline rearrangements to the spectrum of BRCA2 mutations *F Casilli, I Tourmier, O M Sinilnikova, F Coulet, F Soubrier, C Houdayer, A Hardouin, P Berthet, H Sobol, V Bourdon, D Muller, J P Fricker, C Capoulade-Metay, A Chompret, C Nogues, S Mazoyer, P Chappuis, P Mailet, C Philippe, A Lortholary, P Gesta, S Bézieau, C Toulas, L Gladiéff, C M Maugard, D M Provencher, C Dugast, C Delvincourt, T D Nguyen, L Faivre, V Bonadona, T Frébourg, R Lidereau, D Stoppa-Lyonnet, M Tosi*

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.