

Contents



Original articles

- 457** The effect of a single *BRCA2* mutation on cancer in Iceland *H Tulinius, G H Olafsdottir, H Sigvaldason, A Arason, R B Barkardottir, V Egilsson, H M Ogmundsdottir, L Tryggvadottir, S Gudlaugsdottir, J E Eyfjord*
- 463** Genetic and functional analysis of the von Hippel-Lindau (*VHL*) tumour suppressor gene promoter *M Zatyka, C Morrissey, I Kuzmin, M I Lerman, F Latif, F M Richards, E R Maher*
- 473** Linkage of otosclerosis to a third locus (*OTSC3*) on human chromosome 6p21.3-22.3 *W Chen, C A Campbell, G E Green, K Van Den Bogaert, C Komodakis, L S Manolidis, E Aconomou, Y Kyamides, K Christodoulou, C Faghel, C M Giguère, R L Alford, S Manolidis, G Van Camp, R J H Smith*
- 478** Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with *FOXC2* mutations or linkage to 16q24 *G Brice, S Mansour, R Bell, J R O Collin, A H Child, A F Brady, M Sarfarazi, K G Burnand, S Jeffery, P Mortimer, V A Murday*

Short report

- 484** Novel autosomal dominant mandibulofacial dysostosis with ptosis: clinical description and exclusion of *TCOF1* *P Hedera, H V Toriello, E M Petty*

Letters to JMG

- 489** Linkage stratification and mutation analysis at the *parkin* locus identifies mutation positive Parkinson's disease families *W C Nichols, N Pankratz, S K Uniacke, M W Pauciulo, C Halter, A Rudolph, P M Conneally, T Foroud, and the Parkinson Study Group*
- 493** Screening of *TCOF1* in patients from different populations: confirmation of mutational hot spots and identification of a novel missense mutation that suggests an important functional domain in the protein treacle *A Splendore, E W Jabz, M R Passos-Bueno*

- 496** Molecular studies in 10 cases of Rubinstein-Taybi syndrome, including a mild variant showing a missense mutation in codon 1175 of *CREBBP* *O Bartsch, K Locher, P Meinecke, W Kress, E Seemanová, A Wagner, K Ostermann, G Rödel*
- 502** Q829X, a novel mutation in the gene encoding otoferlin (*OTOF*), is frequently found in Spanish patients with prelingual non-syndromic hearing loss *V Migliosi, S Modamio-Høybjør, M A Moreno-Pelayo, M Rodríguez-Ballesteros, M Villamar, D Tellería, I Menéndez, F Moreno, I del Castillo*
- 507** Localisation of the Y chromosome stature gene to a 700 kb interval in close proximity to the centromere *S Kirsch, B Weiss, S Kleiman, K Roberts, J Pryor, A Milunsky, A Ferlin, C Foresta, G Matthijs, G A Rappold*
- 514** Deletion of 9p associated with gonadal dysfunction in 46,XY but not in 46,XX human fetuses *F Vialard, C Ottolenghi, M Gonzales, A Choiset, S Girard, J P Siffroi, K McElreavey, C Vibert-Guigue, M Sebaoun, N Joyé, M F Portnoi, F Jaubert, M Fellous*
- 519** Maternal uniparental disomy 12 in a healthy girl with a 47,XX,+der(12)(:p11→q11:)/46,XX karyotype *F von Eggeling, C Hoppe, U Bartz, H Starke, G Houge, U Claussen, G Ernst, D Kotzot, T Liehr*
- 522** Predictive testing in the context of pregnancy: experience in Huntington's disease and autosomal dominant cerebellar ataxia *G Lesca, C Goizet, A Dürr on behalf of the French Group for Presymptomatic Testing in Neurogenetic Disorders*
- 526** Concerns of women presenting to a comprehensive cancer centre for genetic cancer risk assessment *D J MacDonald, J Choi, B Ferrell, S Sand, S McCaffrey, K R Blazer, M Grant, J N Weitzel*
- 531** Management of women with a family history of breast cancer in the North West Region of England: training for implementing a vision of the future *M McAllister, K O'Malley, P Hopwood, B Kerr, A Howell, D G R Evans*

contd...

NEW

**ONLINE
SUBMISSION**

**GO TO
WEBSITE**

**TO SUBMIT YOUR
MANUSCRIPT**