

## Contents



### Review article

- 537** Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome *R S Wildin, S Smyk-Pearson, A H Filipovich*

### Original articles

- 546** Prospective screening for subtelomeric rearrangements in children with mental retardation of unknown aetiology: the Amsterdam experience *C D M van Karnebeek, C Koevoets, S Sluijter, E K Bijlsma, D F M C Smeets, E J Redeker, R C M Hennekam, J M N Hoovers*
- 554** Cardiovascular manifestations in 75 patients with Williams syndrome *M Eronen, M Peippo, A Hiippala, M Raatikka, M Arvio, R Johansson, M Kähkönen*
- 559** Analysis of the p63 gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts *L L Barrow, H van Bokhoven, S Daack-Hirsch, T Andersen, S E C van Beersum, R Gorlin, J C Murray*

### Short reports

- 567** A novel locus for autosomal dominant non-syndromic deafness (DFNA41) maps to chromosome 12q24-qter *S H Blanton, C Y Liang, M W Cai, A Pandya, L L Du, B Landa, S Mummalani, K S Li, Z Y Chen, X N Qin, Y F Liu, T Balkany, W E Nance, X Z Liu*
- 571** *PTPN11* mutations in LEOPARD syndrome *E Legius, C Schrander-Stumpel, E Schollen, C Pulles-Heintzberger, M Gewillig, J-P Fryns*

### Letters to JMG

- 575** Characterisation of the growth regulating gene *IMP3*, a candidate for Silver-Russell syndrome *D Monk, L Bentley, C Beechey, M Hitchins, J Peters, M A Preece, P Stanier, G E Moore*
- 582** Silver-Russell phenotype in a patient with pure trisomy 1q32.1-q42.1: further delineation of the pure 1q trisomy syndrome *M M van Haelst, H J F M M Eussen, F Visscher, J L M de Ruijter, S L S Drop, D Lindhout, C H Wouters, L C P Govaerts*

- 586** *MECP2* gene nucleotide changes and their pathogenicity in males: proceed with caution *F Laccone, B Zoll, P Huppke, F Hanefeld, W Pepinski, R Trappe*

- 589** Twelve novel *FBN1* mutations in Marfan syndrome and Marfan related phenotypes test the feasibility of *FBN1* mutation testing in clinical practice *D J Halliday, S Hutchinson, L Lonie, J A Hurst, H Firth, P A Handford, P Wordsworth*

- 594** Deletion of the *SIM1* gene (6q16.2) in a patient with a Prader-Willi-like phenotype *L Faivre, V Cormier-Daire, J M Lapiere, L Colleaux, S Jacquemont, D Geneviève, P Saunier, A Munnich, C Turleau, S Romana, M Prieur, M C De Blois, M Vekemans*

- 597** The phenotype of survivors of campomelic dysplasia *S Mansour, A C Offiah, S McDowall, P Sim, J Tolmie, C Hall*

- 603** Dysosteosclerosis: a report of three new cases and evolution of the radiological findings *N H Elçioglu, A Vellodi, C M Hall*

- 608** A significant response to neoadjuvant chemotherapy in *BRCA1/2* related breast cancer *P O Chappuis, J Goffin, N Wong, C Perret, P Ghadirian, P N Tonin, W D Foulkes*

- 611** Incidence of non-founder *BRCA1* and *BRCA2* mutations in high risk Ashkenazi breast and ovarian cancer families *N D Kauff, P Perez-Segura, M E Robson, L Scheuer, B Siegel, A Schluger, B Rapaport, T S Frank, K Nafa, N A Ellis, G Parmigiani, K Offit*

### Book review

- 615** Counseling About Cancer: Strategies for Genetic Counseling *K Sweet, D G R Evans*
- 616** Instructions for authors

contd...

**NEW  
ONLINE  
SUBMISSION**

**GO TO  
WEBSITE**

**TO SUBMIT YOUR  
MANUSCRIPT**