

Contents



Review article

- 377** The p63 gene in EEC and other syndromes H G Brunner, B C J Hamel, H van Bokhoven

Original articles

- 382** Testing for osteogenesis imperfecta in cases of suspected non-accidental injury A Marlowe, M G Pepin, P H Byers
- 387** Genetic mapping of a susceptibility locus for disc herniation and spastic paraplegia on 6q23.3-q24.1 M Zortea, A Vettori, C P Trevisan, S Bellini, G Vazza, M Armani, A Simonati, M L Mostacciolo
- 391** A cryptic deletion of 2q35 including part of the *PAX3* gene detected by breakpoint mapping in a child with autism and a de novo 2;8 translocation I Borg, M Squire, C Menzel, K Stout, D Morgan, L Willatt, P C M O'Brien, M A Ferguson-Smith, H H Ropers, N Tommerup, V M Kalscheuer, D R Sargan

Letters to JMG

- 400** Association of an interleukin 1B gene polymorphism (-511) with Parkinson's disease in Finnish patients K M Mattila, J O Rinne, T Lehtimäki, M Röyttä, J-P Ahonen, M Hurme
- 403** An investigation of ACE as a risk factor for dementia and cognitive decline in the general population A G Yip, C Brayne, D Easton, D C Rubinstein, the Medical Research Council Cognitive Function and Ageing Study (MRC CFAS)
- 407** Change in the penetrance of founder *BRCA1/2* mutations? A retrospective cohort study W D Foulkes, J-S Brunet, N Wong, J Goffin, P O Chappuis
- 410** What do women really want to know? Motives for attending familial breast cancer clinics C J van Asperen, S van Dijk, M W Zoetewij, D R M Timmermans, G H de Bock, E J Meijers-Heijboer, M F Niermeijer, M H Breuning, J Kievit, W Otten
- 415** Molecular analysis of the *CBP* gene in 60 patients with Rubinstein-Taybi syndrome I Coupry, C Roudaut, M Stef, M-A Delrieu, M Marche, I Burgelin, L Taine, C Cruaud, D Lacombe, B Arveiler

- 422** Mutation screening of the *PKD1* transcript by RT-PCR S Burtey, A M Lossi, J Bayle, Y Berland, M Fontés

- 430** A novel 2 bp deletion in the *TM4SF2* gene is associated with MRX58 F E Abidi, E Holinski-Feder, O Rittinger, F Kooy, H A Lubs, R E Stevenson, C E Schwartz

- 434** Chromosome 2 aberrations in clinical cases characterised by high resolution multicolour banding and region specific FISH probes A Weise, H Starke, A Heller, H Tönnies, M Volleth, M Stumm, S Gabriele, A Nietzel, U Claussen, T Liehr

- 440** Towards a suggestive facial dysmorphism in adenylosuccinate lyase deficiency? M Holder-Espinasse, S Marie, G Bourrouillou, I Ceballos-Picot, M-C Nassogne, L Faivre, J Amiel, A Munich, M-F Vincent, V Cormier-Daire

- 443** Prenatal detection of cystic fibrosis by ultrasonography: a retrospective study of more than 346 000 pregnancies V Scotet, M De Braekeleer, M-P Audrézet, I Quére, B Mercier, I Duguépéroux, J Andrieux, M Blayau, C Férec

- 449** Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss S J Stern, K S Arnos, L Murrelle, K Oelrich Welch, W E Nance, A Pandya

Echoes

- 399** New mutations in hereditary pancreatitis
- 409** HLA-DR4 and risk of spondyloarthropathy
- 421** MICA-A5.1 weighs in with HLA-DR3/DQ2 in coeliac disease
- 454** Book reviews
- 456** Instructions for authors

contd...