

**Editorial**

The future JMG: [www.jmedgenet.com](http://www.jmedgenet.com) E R Maher

**Review article**

Mutational analysis using oligonucleotide microarrays J G Hacia, F S Collins

**Commentary**

Closing time for CATCH22 J Burn

**Original articles**

A molecular investigation of true dominance in Huntington's disease Y Narain, A Wyettbach, J Rankin, R A Furlong, D C Rubinstein

A highly accurate, low cost test for *BRCA1* mutations N J van Orsouw, R K Dhanda, Y Elhaji, S A Narod, F P Li, C Eng, J Vrij

Two unrelated patients with inversions of the X chromosome and non-specific mental retardation: physical and transcriptional mapping of their common breakpoint region in Xq13.1 L Villard, S Briault, A-M Lossi, C Paringaux, J Belouge, L Colleaux, D R Pincus, E Woollatt, J Espinasse, A Munnich, C Moraine, M Fontès, J Geczi

X linked severe mental retardation, craniofacial dysmorphology, epilepsy, ophthalmoplegia, and cerebellar atrophy in a large South African kindred is localised to Xq24-q27 A L Christianson, R E Stevenson, C H van der Meyden, J Pelser, F W Theron, P L van Rensburg, M Chandler, C E Schwartz

Microdeletions in *FMR2* may be a significant cause of premature ovarian failure A Murray, J Webb, N Dennis, G Conway, N Morton

Specific polymorphisms in the *RET* proto-oncogene are over-represented in patients with Hirschsprung disease

and may represent loci modifying phenotypic expression S Borrego, M E Sáez, A Ruiz, O Gimmi, M López-Alonso, G Antónolo, C Eng

**Short reports**

Unreported RSK2 missense mutation in two male sibs with an unusually mild form of Coffin-Lowry syndrome S Manouvrier-Hanu, J Amiel, S Jacquot, K Merienne, A Moerman, A Coëslier, F Labarrière, L Vallée, M F Croquette, A Hanauer

Defective PEX gene products correlate with the protein import, biochemical abnormalities, and phenotypic heterogeneity in peroxisome biogenesis disorders N Shimozawa, A Imamura, Z Zhang, Y Suzuki, T Orii, T Tsukamoto, T Osumi, Y Fujiki, R J A Wanders, F Besley, N Kondo

Maternal uniparental disomy for chromosome 14 in a boy with a normal karyotype R Hordijk, H Wierenga, H Scheffer, B Leegte, R M W Hofstra, I Stolte-Dijkstra

Schimke immuno-osseous dysplasia: case report and review of 25 patients J M Saraiva, A Dinis, C Resende, E Faria, C Gomes, A J Correia, J Gil, N da Fonseca

**Letters to the Editor**

A missense mutation in both *hMSH2* and *APC* in an Ashkenazi Jewish HNPCC kindred: implications for clinical screening Z Q Yuan, N Wong, W D Foulkes, L Albert, F Manganaro, C Andreutti-Zaugg, R Iggo, K Anthony, E Hsieh, M Redston, L Pinsky, M Trifiro, P H Gordon, D Lasko

A PCR test for the detection of hypermethylated alleles at the retinoblastoma locus M Zeschnigk, D Lohmann, B Horsthemke

Frequency and predictive value of 22q11 deletion J LiLing, I Cross, J Burn, C P Daniel, E J Tawn, L Parker

Cardiac and skeletal actin gene mutations are not a common cause of dilated cardiomyopathy B M Mayosi, S S Khogali, B Zhang, H Watkins

Subclinical cognitive impairment in autosomal dominant "pure" hereditary spastic paraparesis E Reid, C Grayson, D C Rubinstein, M T Rogers, J S Rubinstein

Acute lymphoblastic leukaemia in a patient with cardiofaciocutaneous syndrome H van Den Berg, R C M Hennekam

