

Contents

Editorial

The future JMG: www.jmedgenet.com eamonn r maher 729

Review article

Mutational analysis using oligonucleotide microarrays Joseph G Hacia, Francis S Collins 730

Commentary

Closing time for CATCH22 737

Original articles

A molecular investigation of true dominance in Huntington's disease Yolanda Narain, Andreas Wyttenbach, Julia Rankin, Robert A Furlong, David C Rubinsztein 739

A highly accurate, low cost test for BRCA1 mutations Nathalie J van Orsouw, Rahul K Dhandu, Youssef Elhaji, Steven A Narod, Frederick P Li, Charis Eng, Jan Vijg 747

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 Laurent Villard*, Sylvain Briault*, Anne-Marie Lossi, Christine Paringaux, Jérôme Belougne, Laurence Colleaux, D R Pincus, E Woollatt, James Lespinasse, Arnold Munnich, Claude Moraine, Michel Fontès, Jozef Gecz 754

X linked severe mental retardation, craniofacial dysmorphism, epilepsy, ophthalmoplegia, and cerebellar atrophy in a large South African kindred is localised to Xq24-q27 Arnold L Christianson, Roger E Stevenson, C H van der Meyden, Julie Pelsler, Francois W Theron, Petro L van Rensburg, Michael Chandler, Charles E Schwartz 759

Microdeletions in FMR2 may be a significant cause of premature ovarian failure Anna Murray, James Webb, Nick Dennis, Gerard Conway, Newton Morton 767

Specific polymorphisms in the RET proto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression Salud Borrego, María Eugenia Sáez, Agustín Ruiz, Oliver Gimm, Manuel López-Alonso, Guillermo Antiñolo, Charis Eng 771

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 Coëslie, Labarriere F, L Vallée, M F Croquette, A Hanauer 775

Defective PEX gene products correlate with the protein import, biochemical abnormalities, and phenotypic heterogeneity in peroxisome biogenesis disorders Nobuyuki Shimozawa, Atsushi Imamura, Zhongyi Zhang, Yasuyuki Suzuki, Tadao Oriti, Toshiro Tsukamoto, Takashi Osumi, Yukio Fujiki, Ronald J A Wanders, Guy Besley, Naomi Kondo 779

Maternal uniparental disomy for chromosome 14 in a boy with a normal karyotype Roel Hordijk, Henk Wierenga, Hans Scheffer, Beike Leegte, Robert M W Hofstra, Irene Stolte-Dijkstra 782

Schimke immuno-osseous dysplasia: case report and review of 25 patients Jorge M Saraiva, Alexandra Dinis, Cristina Resende, Emília Faria, Clara Gomes, A Jorge Correia, Júlia Gil, Nicolau da Fonseca 786

Letters to the editor

A missense mutation in both hMSH2 and APC in an Ashkenazi Jewish HNPCC kindred: implications for clinical screening zhi qiang yuan nora wong william d foulkes lesley alpert fortunato manganaro corinne andreutti-zaugg richard iggo kira anthony eugene hsieh mark redston leonard pinsky mark trifiro philip h gordon dana lasko 790

A PCR test for the detection of hypermethylated alleles at the retinoblastoma locus michael zeschnigk dietmar lohmann bernhard horsthemke 793

Frequency and predictive value of 22q11 deletion jesse li ling ian cross john burn paul c daniel janet e tawn louise parker bongani m mayosi saib s khogali baiping zhang hugh watkins 794

Cardiac and skeletal actin gene mutations are not a common cause of dilated cardiomyopathy Bongani M Mayosi, Saib S Khogali, Baiping Zhang, Hugh Watkins 796

Subclinical cognitive impairment in autosomal dominant "pure" hereditary spastic paraplegia e reid c grayson d c rubinsztein m t rogers j s rubinsztein hendrik van den berg raoul c m hennekam 797

Acute lymphoblastic leukaemia in a patient with cardiofaciocutaneous syndrome Hendrik van Den Berg, Raoul C M Hennekam 799