

Contents



Hypothesis

- 401** *DFNA5*: hearing impairment exon instead of hearing impairment gene? L Van Laer, K Vrijens, S Thys, V F I Van Tendeloo, R J H Smith, D R Van Bockstaele, J-P Timmermans, G Van Camp

Original articles

- 407** Evidence for interaction between the *TCO* and *NMTC1* loci in familial non-medullary thyroid cancer J D McKay, D Thompson, F Lesueur, K Stankov, A Pastore, C Wattah, S Stolz, G Riccabona, R Moncayo, G Romeo, D E Goldgar
- 413** Non-random asynchronous replication at 22q11.2 favours unequal meiotic crossovers leading to the human 22q11.2 deletion A Baumer, M Riegel, A Schinzel

Short report

- 421** Prospective risk of cancer in *CDKN2A* germline mutation carriers A M Goldstein, J P Struewing, M C Fraser, M W Smith, M A Tucker

Letters to JMG

- 425** High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation J A Veltman, H G Yntema, D Lugtenberg, H Arts, S Briault, E H L P G Huys, K Osoegawa, P de Jong, H G Brunner, A Geurts van Kessel, H van Bokhoven, E F P M Schoenmakers
- 433** Molecular analysis of 20 patients with 2q37.3 monosomy: definition of minimum deletion intervals for key phenotypes M A Aldred, R O C Sanford, N S Thomas, M A Barrow, L C Wilson, L A Brueton, M C Bonaglia, R C M Hennekam, C Eng, N R Dennis, R C Trembath
- 440** Familial X/Y translocations associated with variable sexual phenotype A Sharp, K Kusz, J Jaruzelska, M Szarras-Czapnik, J Wolski, P Jacobs
- 445** Estimating the age of rare disease mutations: the example of Triple-A syndrome E Genin, A Tullio-Pelet, F Begeot, S Lyonnet, L Abel
- 450** A fifth locus for otosclerosis, *OTSC5*, maps to chromosome 3q22-24 K Van Den Bogaert, E M R De Leenheer, W Chen, Y Lee, P Nürnberg, R J E Pennings,

- 454** 5,10-methylenetetrahydrofolate reductase (*MTHFR*) 677C→T and 1298A→C mutations are associated with DNA hypomethylation R Castro, I Rivera, P Ravasco, M E Camilo, C Jakobs, H J Blom, I T de Almeida

- 459** Genetic analysis of PSORS2 markers in a UK dataset supports the association between RAPTOR SNPs and familial psoriasis F Capon, C Helms, C D Veal, D Tillman, A D Burden, J N Barker, A M Bowcock, R C Trembath

- 461** Systematic micro-array based identification of placental mRNA in maternal plasma: towards non-invasive prenatal gene expression profiling N B Y Tsui, S S C Chim, R W K Chiu, T K Lau, E K O Ng, T N Leung, Y K Tong, K C A Chan, Y M D Lo

- 468** An autosomal recessive cone-rod dystrophy associated with amelogenesis imperfecta M Michaelides, A Bloch-Zupan, G E Holder, D M Hunt, A T Moore

Medical genetics in practice

- 474** A new scoring system for the chances of identifying a *BRCA1/2* mutation outperforms existing models including BRCAPRO D G R Evans, D M Eccles, N Rahman, K Young, M Bulman, E Amir, A Shenton, A Howell, F Lalloo

Electronic letters

- e71** Selective disruption of muscle and brainspecific *BPAG1* isoforms in a girl with a $\delta;15$ translocation, cognitive and motor delay, and tracheo-oesophageal atresia R Giorda, A Cerritello, M C Bonaglia, S Bova, G Lanzi, E Repetti, S Giglio, C Baschiroto, T Pramparo, L Avolio, R Bragheri, P Maraschio, O Zuffardi
- e72** Site directed mutagenesis of hMLH1 exonic splicing enhancers does not correlate with splicing disruption P Lastella, N Resta, I Miccolis, A Quagliariella, G Guanti, A Stella

contd...

**NEW
ONLINE
SUBMISSION**
**GO TO
WEBSITE**
TO SUBMIT YOUR
MANUSCRIPT

Contents ...contd

- e73** A mitochondrial DNA duplication as a marker of skeletal muscle specific mutations in the mitochondrial genome
M Mancuso, C Vives-Bauza, M Filosto, R Marti, A Solano, J Montoya, J Gamez, S DiMauro, A L Andreu
- e74** Haplotype analysis of human *AMPD1* gene: origin of common mutant allele
K Toyama, H Morisaki, Y Kitamura, M Gross, T Tamura, Y Nakahori, J M Vance, M Speer, N Kamatani, T Morisaki
- e75** Spontaneous recovery of a childhood onset mitochondrial myopathy caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene
R Horváth, H Lochmüller, M Hoeltzenbein, J Müller-Höcker, B G Schoser, D Pongratz, M Jaksch
- e76** Genetic influences on the circulating cytokines involved in osteoclastogenesis
G Livshits, I Pantsulaia, S Trofimov, E Kobylansky
- e77** A novel form of syndromic cutis laxa with facial dysmorphism, cleft palate, and mental retardation
D Genevieve, C Baumann, C Huber, L Faivre, D Sanlaville, C Bodemer, S Hadj-Rabia, A Assoumou, A Verloes, F Raqbi, A Munnich, V Cormier-Daire
- e78** Angel shaped phalangeal dysplasia, hip dysplasia, and positional teeth abnormalities are part of the brachydactyly C spectrum associated with *CDMP-1* mutations
M Holder-Espinasse, F Escande, E Mayrargue, A Dieux-Coeslier, D Fron, A Douval-Bisser, O Boute-Benejean, Y Robert, N Porchet, S Manouvrier-Hanu
- e79** Evidence of differing genotypic effects of *PPAR α* in women and men
Q H Khan, D E Pontefract, S Iyengar, S Ye
- e80** Y-linked inheritance of non-syndromic hearing impairment in a large Chinese family
Q J Wang, C Y Lu, N Li, S Q Rao, Y B Shi, D Y Han, X Li, J Y Cao, L M Yu, Q Z Li, M X Guan, W Y Yang, Y Shen
- e82** Clusters of non-truncating mutations of P/Q type Ca^{2+} channel subunit *Ca $_v$ 2.1* causing episodic ataxia 2
E Mantuano, L Veneziano, M Spadaro, P Giunti, S Guida, M G Leggio, L Verriello, N Wood, C Jodice, M Frontali
- e83** A novel point mutation A170P in the *SHOX* gene defines impaired nuclear translocation as a molecular cause for Lér-Weill dyschondrosteosis and Langer dysplasia
N Sabherwal, R J Blaschke, A Marchini, D Heine-Suner, J Rosell, J Ferragut, W F Blum, G Rappold
- e84** Complement factor I: a susceptibility gene for atypical haemolytic uraemic syndrome
V Fremeaux-Bacchi, M-A Dragon-Durey, J Blouin, C Vigneau, D Kuypers, B Boudailliez, C Loirat, E Rondeau, W H Fridman
- e85** Screening of *MECP2* coding sequence in patients with phenotypes of decreasing likelihood for Rett syndrome: a cohort of 171 cases
F Kammoun, N de Roux, O Boespflug-Tanguy, L Vallée, R Seng, M Tardieu, P Landrieu
- e86** *P* gene mutations in patients with oculocutaneous albinism and findings suggestive of Hermansky-Pudlak syndrome
N A Garrison, Z Yi, O Cohen-Barak, M Huizing, L M Hartnell, W A Gahl, M H Brilliant
- e87** Broader geographical spectrum of Cohen syndrome due to *COH1* mutations
G H Mochida, A Rajab, W Eyaid, A Lu, D Al-Nouri, K Kosaki, M Noruzinia, P Sarda, J Ishihara, A Bodell, K Apse, C A Walsh
- e88** A germline mutation in *KIT* in familial diffuse cutaneous mastocytosis
X Tang, M Boxer, A Drummond, P Ogston, M Hodgins, A D Burden
- e89** Germline mutations of the *E-cadherin (CDH1)* and *TP53* genes, rather than of *RUNX3* and *HPP1*, contribute to genetic predisposition in German gastric cancer patients
G Keller, H Vogelsang, I Becker, S Plaschke, K Ott, G Suriano, A R Mateus, R Seruca, K Biedermann, D Huntsman, C Döring, E Holinski-Feder, A Neutzling, J R Siewert, H Höfler

Online mutation reports

- e81** Case report: a subject with a mutation in the ATG start codon of L-ferritin has no haematological or neurological symptoms
L Cremonesi, A Cozzi, D Girelli, F Ferrari, I Fermo, B Foglieni, S Levi, C Bozzini, M Campanini, M Ferrari, P Arosio