

Contents



Commentary

- 145** Apolipoprotein E polymorphisms and risk of malaria *M A Wozniak, E M Riley, R F Itzhaki*

Original articles

- 147** A genotype-phenotype correlation for *GJB2* (connexin 26) deafness *K Cryns, E Orzan, A Murgia, P L M Huygen, F Moreno, I del Castillo, G Parker Chamberlin, H Azaiez, S Prasad, R A Cucci, E Leonardi, R L Snoeckx, P J Govaerts, P H Van de Heyning, C M Van de Heyning, R J H Smith, G Van Camp*
- 155** Genetic testing in familial isolated hyperparathyroidism: unexpected results and their implications *J Warner, M Epstein, A Sweet, D Singh, J Burgess, S Stranks, P Hill, D Perry-Keene, D Learoyd, B Robinson, P Birdsey, E Mackenzie, B T Teh, J B Prins, J Cardinal*
- 161** Akt activation and localisation correlate with tumour invasion and oncogene expression in thyroid cancer *V Vasko, M Saji, E Hardy, M Kruhlak, A Larin, V Savchenko, M Miyakawa, OIsozaki, H Murakami, T Tsushima, K D Burman, C De Micco, M D Ringel*

Short reports

- 171** Mutations in *SCN9A*, encoding a sodium channel alpha subunit, in patients with primary erythralgia *Y Yang, Y Wang, S Li, Z Xu, H Li, L Ma, J Fan, D Bu, B Liu, Z Fan, G Wu, J Jin, B Ding, X Zhu, Y Shen*
- 175** BAC microarray analysis of 15q11-q13 rearrangements and the impact of segmental duplications *D P Locke, R Segraves, R D Nicholls, S Schwartz, D Pinkel, D G Albertson, E E Eichler*
- 183** Identification of a mutation in synapsin I, a synaptic vesicle protein, in a family with epilepsy *C C Garcia, H J Blair, M Seager, A Coulthard, S Tennant, M Buddless, A Curtis, J A Goodship*

Letters to JMG

- 188** A locus for autosomal dominant keratoconus maps to human chromosome 3p14-q13 *F Brancati, E M Valente, A Sarkozy, J Fehèr, M Castori, P Del Duca, R Mingarelli, A Pizzuti, B Dallapiccola*

- 193** Autosomal dominant axonal Charcot-Marie-Tooth disease type 2 (CMT2G) maps to chromosome 12q12-q13.3 *E Nelis, J Berciano, N Verpoorten, K Coen, I Dierick, V Van Gerwen, O Combarros, P De Jonghe, V Timmerman*
- 198** The performance of CGH array for the detection of cryptic constitutional chromosome imbalances *J Schoumans, B-M Anderlid, E Blennow, B T Teh, M Nordenskjöld*
- 203** Genotype and psychological phenotype in tuberous sclerosis *J C Lewis, H V Thomas, K C Murphy, J R Sampson*
- 208** Assignment of the locus for ichthyosis prematurity syndrome to chromosome 9q33.3-34.13 *J Klar, T Gedde-Dahl Jr, M Larsson, M Pigg, B Carlsson, D Tentler, A Vahlquist, N Dahl*
- 213** Acropectorovertebral dysgenesis (F syndrome) maps to chromosome 2q36 *H Thiele, C McCann, S van't Padje, G C Schwabe, H C Hennies, G Camera, J Opitz, R Laxova, S Mundlos, P Nürnberg*
- 219** Interleukin-1 cluster is associated with genetic risk for schizophrenia and bipolar disorder *S Papiol, A Rosa, B Gutiérrez, B Martín, P Salgado, R Catalán, B Arias, L Fañanás*
- 224** A new locus for recessive distal spinal muscular atrophy at Xq13.1-q21 *R I Takata, C E Speck Martins, M R Passosbueno, K T Abe, A L Nishimura, M Dorvalina Da Silva, A Monteiro Jr, M I Lima, F Kok, M Zatz*
- 230** Trinucleotide repeat expansion in SCA17/ TBP in white patients with Huntington's disease-like phenotype *P Bauer, F Laccone, A Rolfs, U Wüllner, S Bösch, H Peters, S Liebscher, M Scheible, J T Epplen, B H F Weber, E Holinski-Feder, H Weirich-Schwaiger, D J Morris-Rosendahl, J Andrich, O Riess*
- 233** Loci for primary ciliary dyskinesia map to chromosome 16p12.1-12.2 and 15q13.1-15.1 in Faroe Islands and Israeli Druze genetic isolates *D Jeganathan, R Chodhari, M Meeks, O Færoe, D Smyth, K Nielsen, I Amirav, A S Luder, H Bisgaard, R M Gardiner, E M K Chung, H M Mitchison*

contd...

NEW
ONLINE
SUBMISSION
GO TO
WEBSITE
TO SUBMIT YOUR
MANUSCRIPT

Contents ...contd

Miscellanea

- 146** Correction
186 Book review
187 Echo
192 Echo
202 Echo
239 Correction
240 Correction

Electronic letters

- e22** *DJ-1* mutations are a rare cause of recessively inherited early onset parkinsonism mediated by loss of protein function P J Lockhart, S Lincoln, M Hulihan, J Kachergus, K Wilkes, G Bisceglia, D C Mash, M J Farrer
- e23** The impact of proband mediated information dissemination in families with a *BRCA1/2* gene mutation E Sermijn, G Goelen, E Teugels, L Kaufman, M Bonduelle, B Neyns, B Poppe, A De Paepe, J De Grève
- e24** Subcellular localisation of marenstrin/pyrin isoforms carrying the most common mutations involved in familial Mediterranean fever in the presence or absence of its binding partner ASC C Cazeneuve, S Papin, I Jéru, P Duquesnoy, S Anselem
- e25** Early onset, non-progressive, mild cerebellar ataxia co-segregating with a familial balanced translocation t(8;20)(p22;q13) J M Hertz, B Sivertsen, A Silahatoglu, M Bugge, V Kalscheuer, A Weber, J Wirth, H-H Ropers, N Tommerup, Z Tümer
- e26** Tetrasomy 21pterRq21.2 in a male infant without typical Down's syndrome dysmorphic features but moderate mental retardation I Rost, H Fiegler, C Fauth, P Carr, T Bettecken, J Kraus, C Meyer, A Enders, A Wirtz, T Meitinger, N P Carter, M R Speicher
- e27** *CLCA1* gene polymorphisms in chronic obstructive pulmonary disease A E Hegab, T Sakamoto, Y Uchida, A Nomura, Y Ishii, Y Morishima, M Mochizuki, T Kimura, W Saitoh, H H Massoud, H M Massoud, K M Hassanein, K Sekizawa
- e28** Hepatic lipase C-480T polymorphism modifies the effect of HDL cholesterol on the risk of acute myocardial infarction in men: a prospective population based study Y-M Fan, J T Salonen, T A Koivu, T-P Tuomainen, K Nyssönen, T A Lakka, R Salonen, K Seppänen, S T Nikkari, E Tahvanainen, T Lehtimäki

- e29** A new mutation of the lamin A/C gene leading to autosomal dominant axonal neuropathy, muscular dystrophy, cardiac disease, and leconychia C Goizet, R Ben Yaou, L Demay, P Richard, S Bouillot, M Rouanet, E Hermosilla, G Le Masson, A Lagueny, G Bonne, X Ferrer

Online mutation reports

- e30** Genetic and epigenetic profile of sporadic pheochromocytomas A Cascon, S Ruiz-Llorente, M F Fraga, R Leton, D Telleria, J Sastre, J Jose Diez, G Martinez Diaz-Guerra, J A Diaz Perez, J Benitez, M Esteller, M Robledo
- e31** Extending the p16-Leiden tumour spectrum by respiratory tract tumours R A Oldenburg, W H de Vos tot Nederveen Cappel, M van Puijbroek, A van den Ouweland, E Bakker, G Griffioen, P Devilee, C J Cornelisse, H Meijers-Heijboer, H F A Vasen, H Morreau
- e32** Germline and de novo mutations in the *HRPT2* tumour suppressor gene in familial isolated hyperparathyroidism (FIHP) A Villablanca, A Calender, L Forsberg, A Höög, J-D Cheng, D Petillo, C Bauters, K Kahnoski, T Ebeling, P Salmela, A-L Richardson, L Delbridge, A Meyrier, C Proye, J D Carpten, B T Teh, B G Robinson, C Larsson
- e33** RYR1 mutations in UK central core disease patients: more than just the C-terminal transmembrane region of the RYR1 gene S Shepherd, F Ellis, J Halsall, P Hopkins, R Robinson
- e34** A founder *MLH1* mutation in families from the districts of Modena and Reggio-Emilia in northern Italy with hereditary nonpolyposis colorectal cancer associated with protein elongation and instability O Caluseriu, C Di Gregorio, E Lucci-Cordisco, M Santarosa, J Trojan, A Brieger, P Benatti, M Pedroni, T Colibazzi, A Bellacosa, G Neri, M Ponz de Leon, A Viel, M Genuardi
- e35** Novel mutations in the *KCNQ2* gene link epilepsy to a dysfunction of the *KCNQ2*-calmodulin M C Richards, S E Heron, H E Spendlove, I E Scheffer, B Grinton, S F Berkovic, J C Mulley, A Davy

Electronic pages

- e36** Author index
e37 Subject index