

Contents



Review

- 801** Clinical genetic counselling for familial cancers requires reliable data on familial cancer risks and general action plans
K Hemminki, C Eng

Original articles

- 808** TLR4 and TNF- α polymorphisms are associated with an increased risk for severe sepsis following burn injury
R C Barber, C C Aragaki, F A Rivera-Chavez, G F Purdue, J L Hunt, J W Horton
- 814** Sequence family variant loss from the AZFc interval of the human Y chromosome, but not gene copy loss, is strongly associated with male infertility
N Machev, N Saut, G Longepied, P Terriou, A Navarro, N Levy, M Guichaoua, C Metzler-Guillemain, P Collignon, A-M Frances, J Belougne, E Clemente, J Chiaroni, C Chevillard, C Durand, A Ducourneau, N Pech, K McElreavey, M-G Mattei, M J Mitchell
- 826** *FRG2*, an FSHD candidate gene, is transcriptionally upregulated in differentiating primary myoblast cultures of FSHD patients
T Rijkers, G Deidda, S van Koningsbruggen, M van Geel, R J L F Lemmers, J C T van Deutekom, D Figlewicz, J E Hewitt, G W Padberg, R R Frants, S M van der Maarel
- 853** Putative functional alleles of *DYX1C1* are not associated with dyslexia susceptibility in a large sample of sibling pairs from the UK
T S Scerri, S E Fisher, C Francks, I L MacPhie, S Paracchini, A J Richardson, J F Stein, A P Monaco
- 858** A new locus for a childhood onset, slowly progressive autosomal recessive spinocerebellar ataxia maps to chromosome 11p15
G J Breedveld, B van Wetten, G D te Raa, E Brusse, J C van Swieten, B A Oostra, J A Maat-Kievit
- 867** The ser9gly SNP in the dopamine D₃ receptor causes a shift from cAMP related to PGE₂ related signal transduction mechanisms in transfected CHO cells
M Hellstrand, E A Danielsen, V M Steen, A Ekman, E Eriksson, C L Nilsson
- 872** A gene responsible for autosomal dominant auditory neuropathy (AUNA1) maps to 13q14-21
T B Kim, B Isaacson, T A Sivakumaran, A Starr, B J B Keats, M M Lesperance
- 877** Increased recurrence risk in congenital disorders of glycosylation type Ia (CDG-Ia) due to a transmission ratio distortion
E Schallen, S Kjaergaard, T Martinsson, S Vuillaumier-Barrot, M Dunoe, L Keldermans, N Seta, G Matthijs

Miscellanea

- 813** Retraction statement

Short report

- 837** Neurofibromatous neuropathy in neurofibromatosis 1 (NF1)
R E Ferner, R A C Hughes, S M Hall, M Upadhyaya, M R Johnson

Letters to JMG

- 842** Missense mutations of *ACTA1* cause dominant congenital myopathy with cores
A M Kaindl, F Rüschenclorf, S Krause, H-H Goebel, K Koehler, C Becker, D Pongratz, J Müller-Höcker, P Nürnberg, G Stoltenburg-Didinger, H Lochmüller, A Huebner
- 849** A novel locus for autosomal recessive form of hypotrichosis maps to chromosome 3q26.33-q27.3
M Aslam, M H Chahrouh, A Razzaq, S Haque, K Yan, S M Leal, W Ahmad

NEW
ONLINE
SUBMISSION
GO TO
WEBSITE
TO SUBMIT YOUR
MANUSCRIPT

contd...

Contents ...contd

Electronic letter

- e116** Relation of type 2 diabetes to individual admixture and candidate gene polymorphisms in the Hispanic American population of San Luis Valley, Colorado
E J Parra, C J Hoggart, C Bonilla, S Dios, J M Norris, J A Marshall, R F Hamman, R E Ferrell, P M McKeigue, M D Shriver

Online mutation reports

- e117** *PTPN11* mutations in patients with LEOPARD syndrome: a French multicentric experience
B Keren, A Hadchouel, S Saba, Y Sznajder, D Bonneau, B Leheup, O Boute, D Gaillard, D Lacombe, V Layet, S Marlin, G Mortier, A Toutain, C Beylot, C Baumann, A Verloes, H Cavé, for the French Collaborative Noonan Study Group

- e118** Rapid detection of *CFTR* gene rearrangements impacts on genetic counselling in cystic fibrosis
F Niel, J Martin, F Dastot-Le Moal, B Costes, B Boissier, V Delattre, M Goossens, E Girodon
- e119** No live individual homozygous for a novel *endoglin* mutation was found in a consanguineous Arab family with hereditary haemorrhagic telangiectasia
A Karabegovic, M Shinawi, U Cymerman, M Letarte
- e120** Germline *CHEK2**1100delC mutations in breast cancer patients with multiple primary cancers
J Huang, S M Domchek, M S Brose, T R Rebbeck, K L Nathanson, B L Weber