

Contents



This logo indicates an article that was published online first ahead of print

Review

- 97** Hereditary haemorrhagic telangiectasia: current views on genetics and mechanisms of disease *S A Abdalla, M Letarte*

Original articles

- 111** Breakpoints around the *HOXD* cluster result in various limb malformations *B Dlugaszewska, A Silahatoglu, C Menzel, S Kübart, M Cohen, S Mundlos, Z Tümer, K Kjaer, U Friedrich, H-H Ropers, N Tommerup, H Neitzel, V M Kalscheuer*
- 119** Survivin-directed RNA interference cocktail is a potent suppressor of tumour growth in vivo *H Caldas, M P Holloway, B M Hall, S J Qualman, R A Altura*
- 129** Type 1 diabetes and the *OAS* gene cluster: association with splicing polymorphism or haplotype? *M-C Tessier, H-Q Qu, R Fréchette, F Bacot, R Grabs, S P Taback, M L Lawson, S E Kirsch, T J Hudson, C Polychronakos*
- 133** Racial differences in the incidence of BRCA1 and BRCA2 mutations in a cohort of early onset breast cancer patients: African American compared to white women *B G Haffty, A Silber, E Matloff, J Chung, D Lannin*

Short reports

- 138** Cleft lip/palate and *CDH1/E-cadherin* mutations in families with hereditary diffuse gastric cancer *T Frebourg, C Oliveira, P Hochain, R Karam, S Manouvrier, C Graziadio, M Vekemans, A Hartmann, S Baert-Desurmont, C Alexandre, S Lejeune Dumoulin, C Marroni, C Martin, S Castedo, M Lovett, J Winston, J C Machado, T Attié, E W Jabs, J Cai, Ph Pellerin, J P Triboulet, M Scotte, F Le Pessot, A Hedouin, F Carneiro, M Blayau, R Seruca*
- 143** Disruption of *TCBA1* associated with a de novo t(1;6)(q32.2;q22.3) presenting in a child with developmental delay and recurrent infections *Y Yue, K Stout, B Grossmann, U Zechner, A Brinckmann, C White, D T Pilz, T Haaf*
- 148** Revisiting the craniosynostosis-radial ray hypoplasia association: Baller-Gerold syndrome caused by mutations in the *RECQL4* gene *L Van Maldergem, H A Siitonen, N Jalkh, E Chouery, M De Roy, V Delague, M Muenke, E W Jabs, J Cai, L L Wang, S E Plon, C Fourneau, M Kestilä, Y Gillerot, A Mégaarbané,*

Letters to JMG

- 153** Disruption of an exon splicing enhancer in exon 3 of *MLH1* is the cause of HNPCC in a Quebec family *S McVety, L Li, P H Gordon, G Chong, W D Foulkes*
- 157** Espin gene (*ESPN*) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation *F Donaudy, L Zheng, R Ficarella, E Ballana, M Carella, S Melchionda, X Estivill, J R Bartles, P Gasparini*
- 162** Congenital heart defects and genetic variants in the methylenetetrahydrofolate reductase gene *C A Hobbs, S J James, A Parsian, P A Krakowiak, S Jernigan, J J Greenhaw, Y Lu, M A Cleves*
- 167** The Pro279Leu variant in the transcription factor *MEF2A* is associated with myocardial infarction *P González, M Garcia-Castro, J R Reguero, A Batalla, Á G Ordóñez, R L Palop, I Lozano, M Montes, V Á Alvarez, E Coto*
- 170** A novel locus for autosomal dominant non-syndromic deafness, *DFNA53*, maps to chromosome 14q11.2-q12 *D Yan, X Ke, S H Blanton, X M Ouyang, A Pandya, L L Du, W E Nance, X Z Liu*
- 175** Sequence variation in mitochondrial complex I genes: mutation or polymorphism? *A L Mitchell, J L Elson, N Howell, R W Taylor, D M Turnbull*
- 180** Array-CGH detection of micro rearrangements in mentally retarded individuals: clinical significance of imbalances present both in affected children and normal parents *C Rosenberg, J Knijnenburg, E Bakker, A M Vianna-Morgante, W Sloos, P A Otto, M Kriek, K Hansson, A C V Krepisch-Santos, H Fiegler, N P Carter, E K Bijlsma, A van Haeringen, K Szuhai, H J Tanke*
- 187** Androgenetic/biparental mosaicism causes placental mesenchymal dysplasia *K A Kaiser-Rogers, D E McFadden, C A Livasy, J Dansereau, R Jiang, J F Knops, L Lefebvre, K W Rao, W P Robinson*

Miscellaneous

- 147** Echo: Carriers of mild *CFTR* mutations risk chronic pancreatitis

contd...

JMG Unlocked

Articles carrying the



logo are available free via the journal's website: www.jmedgenet.com. Funding for this open access experiment is provided by the UK's Joint Information Systems Committee (JISC); see <http://jmg.bmjournals.com/cgi/content/full/42/2/97> for further information.

Contents ...contd

Correspondence

- e3** In response to Cadet *et al* M B Delatycki,
K J Allen

Electronic letters

- e4** Association of TPH1 with suicidal behaviour and psychiatric disorders in the Chinese population X Liu, H Li, W Qin, G He, D Li, Y Shen, J Shen, N Gu, G Feng, L He
- e5** Loss of desmoplakin isoform I causes early onset cardiomyopathy and heart failure in a Naxos-like syndrome A Uzumcu, E E Norgett, A Dindar, O Uyguner, K Nisli, H Kayserili, S E Sahin, E Dupont, N J Severs, I M Leigh, M Yuksel-Apak, D P Kelsell, B Wollnik
- e6** Independent replication and initial fine mapping of 3p21-24 in Asperger syndrome K Rehnström, T Ylisaukko-oja, T Nieminen-von Wendt, S Sarenus, T Källman, E Kempas, L von Wendt, L Peltonen, I Järvelä
- e7** Linkage analysis in a large Swedish family supports the presence of a susceptibility locus for adenoma and colorectal cancer on chromosome 9q22.32-31.1 J Skoglund, T Djureinovic, X-L Zhou, J Vandrovцова, E Renkonen, L Iselius, M L Bisgaard, P Peltomäki, A Lindblom
- e8** Connective tissue dysplasia in five new patients with *NF1* microdeletions: further expansion of phenotype and review of the literature K A Mensink, R P Ketterling, H C Flynn, R A Knudson, N M Lindor, B A Heese, R J Spinner, D Babovic-Vuksanovic
- e9** Testing association between *LRRK2* and Parkinson's disease and investigating linkage disequilibrium C Paisán-Ruiz, E W Evans, S Jain, G Xiomerisiou, J R Gibbs, J Eerola, V Gourbali, O Hellström, J Duckworth, A Papadimitriou, P J Tienari, G M Hadjigeorgiou, A B Singleton