

Contents



Reviews

- 369** Genetics of the polymicrogyria syndromes
A Jansen, E Andermann
- 379** Genetics of congenital hypothyroidism
S M Park, V K K Chatterjee

Original articles

- 390** Reverse cascade screening of newborns for hereditary haemochromatosis: a model for other late onset diseases?
E Cadet, D Capron, M Gallet, M-L Omanga-Léké, H Boutignon, C Julier, K J H Robson, J Rochette
- 396** Angiotensin II type I receptor gene polymorphism: anthropometric and metabolic syndrome traits
M R Abdollahi, T R Gaunt, H E Syddall, C Cooper, D I W Phillips, S Ye, I N M Day
- 402** Evidence of an association between genetic variation of the coactivator PGC-1 β and obesity
G Andersen, L Wegner, K Yanagisawa, C S Rose, J Lin, C Glümer, T Drivsholm, K Borch-Johnsen, T Jørgensen, T Hansen, B M Spiegelman, O Pedersen

Short reports

- 408** A new syndrome, congenital extraocular muscle fibrosis with ulnar hand anomalies, maps to chromosome 21qter
T Tükel, A Uzumcu, A Gezer, H Kayserili, M Yüksel Apak, O Uyguner, S H Gultekin, H-C Hennies, P Nurnberg, R J Desnick, B Wollnik
- 416** Identification of discrete chromosomal deletion by binary recursive partitioning of microarray differential expression data
X Zhou, S W Cole, N P Rao, Z Cheng, Y Li, J McBride, D T W Wong

Letters to JMG

- 420** Variability of sexual phenotype in 46,XX(SRY+) patients: the influence of spreading X inactivation versus position effects
A Sharp, K Kusz, J Jaruzelska, W Tapper, M Szarras-Czapnik, J Wolski, P Jacobs
- 428** *STK11* genotyping and cancer risk in Peutz-Jeghers syndrome
V Schumacher, T Vogel, B Leube, C Driemel, T Goecke, G Möslein, B Royer-Pokora

- 436** Novel association of *RP1* gene mutations with autosomal recessive retinitis pigmentosa
S Khaliq, A Abid, M Ismail, A Hameed, A Mohyuddin, P Lall, A Aziz, K Anwar, S Q Mehdi
- 439** Protective and susceptibility effects of *hSKCa3* allelic variants on juvenile myoclonic epilepsy
J Vijai, A Kapoor, H M Ravishankar, P J Cherian, G Kuruttukulam, B Rajendran, R Sridharan, G Rangan, A S Girija, S Jayalakshmi, S Mohandas, K S Mani, K Radhakrishnan, A Anand
- 443** Identification of a novel mutation disrupting the DNA binding activity of GCM2 in autosomal recessive familial isolated hypoparathyroidism
L Baumber, C Tufarelli, S Patel, P King, C A Johnson, E R Maher, R C Trembath

Correspondence

- e24** BRCA1 mutation and neuronal migration defect: implications for chemoprevention
D Eccles, D Bunyan, S Barker, B Castle
- e25** Pitfalls of automated comparative sequence analysis as a single platform for routine clinical testing for NF1
L M Messiaen, K Wimmer

Electronic letter

- e26** Is maternal duplication of 11p15 associated with Silver-Russell syndrome?
T Eggermann, E Meyer, C Obermann, I Heil, H Schüller, M B Ranke, K Eggermann, H A Wollmann

Online mutation reports

- e27** BRCA1:185delAG found in the San Luis Valley probably originated in a Jewish founder
I Makriyanni, N Hamel, S Ward, W D Foulkes, S Graw
- e28** Novel mutations in *COX15* in a long surviving Leigh syndrome patient with cytochrome c oxidase deficiency
M Bugiani, V Tiranti, L Farina, G Uziel, M Zeviani
- e29** Unconventional intronic splice site mutation in *SCN5A* associates with cardiac sodium channelopathy
T Rossenbacher, E Schollen, C Kuipéri, T J L de Ravel, K Devriendt, G Matthijs, D Collen, H Heidbüchel, P Carmeliet

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.

contd...

Contents ...contd

Online mutation reports

- e30** Search for genetic variants associated with cutaneous malignant melanoma in the Ashkenazi Jewish population
J C Y Loo, A D Paterson, A Hao, M Shennan, H Peretz, Y Sidi, D Hogg, E Yakobson
- e31** Large genomic rearrangements of both *BRCA2* and *BRCA1* are a feature of the inherited breast/ovarian cancer phenotype in selected families
A M Woodward, T A Davis, A G S Silva, kConFab Investigators, J A Kirk, J A Leary
- e32** GATA4 zinc finger mutations as a molecular rationale for septation defects of the human heart
S M Reamon-Buettnner, J Borlak