

Contents



Review

- 289** Genetics of dyslexia: the evolving landscape *J Schumacher, P Hoffmann, C Schmä, G Schulte-Körne, M M Nöthen*

Original articles

- 298** Abnormal sterol metabolism in holoprosencephaly: studies in cultured lymphoblasts *D Haas, J Morgenthaler, F Lacbawan, B Long, H Runz, S F Garbade, J Zschocke, R I Kelley, J G Okun, G F Hoffmann, M Muenke*
- 306** Genotypes and phenotypes in children with short stature: clinical indicators of *SHOX* haploinsufficiency *G Rappold, W F Blum, E P Shavrikova, B J Crowe, R Roeth, C A Quigley, J L Ross, B Niesler*
- 314** The human *GIMAP5* gene has a common polyadenylation polymorphism increasing risk to systemic lupus erythematosus *A Hellquist, M Zucchelli, K Kivinen, U Saarialho-Kere, S Koskenmies, E Widen, H Julkunen, A Wong, M-L Karjalainen-Lindsberg, T Skoog, J Vendelin, D S Cunningham-Graham, T J Vyse, J Kere, C M Lindgren*

Short report


- 322** Borate transporter *SLC4A11* mutations cause both Harboyan syndrome and non-syndromic corneal endothelial dystrophy *J Desir, G Moya, O Reish, N Van Regemorter, H Deconinck, K L David, F M Meire, M J Abramowicz*

Letters to JMG

- 327** Genetic heterogeneity in Rubinstein-Taybi syndrome: delineation of the phenotype of the first patients carrying mutations in *EP300* *D Bartholdi, J H Roelfsema, F Papadia, M H Breuning, D Niedrist, R C Hennekam, A Schinzel, D J M Peters*
- 334** L-2-hydroxyglutaric aciduria: characterisation of the molecular defect in a spontaneous canine model *J Penderis, J Calvin, C Abramson, C Jakobs, L Pettitt, M M Binns, N M Verhoeven, E O'Driscoll, S R Platt, C S Mellersh*
- 341** Exon skipping through the creation of a putative exonic splicing silencer as a consequence of the cystic fibrosis mutation R553X *I Aznarez, J Zielenski, J M Rommens, B J Blencowe, L-C Tsui*
- 347** FRA18C: a new aphidicolin-inducible fragile site on chromosome 18q22, possibly associated with in vivo chromosome breakage *K Debacker, B Winnepeninckx, N Ben-Porat, D FitzPatrick, R Van Luijk, S Scheers, B Kerem, R F Kooy*

Correspondence

- e78** Correction: Functional analysis of BRCA1 M1628V variant *M A Carvalho, A N A Monteiro*
- e79** No screening yet after a negative test for the family mutation *M M A Tilanus-Linthorst*

Articles carrying the  logo are freely available online under the BMJ Journals unlocked scheme, see <http://jmg.bmj.com/info/unlocked.dtl>