



## Contents

### Review

- 289** The genetic basis of congenital hyperinsulinism  
*C James, R R Kapoor, D Ismail, K Hussain*



### Original articles

- 300** Polymorphisms in *C2*, *CFB* and *C3* are associated with progression to advanced age related macular degeneration associated with visual loss  
*P J Francis, S C Hamon, J Ott, R G Weleber, M L Klein*
- 308** Infantile cardiomyopathy caused by a mutation in the overlapping region of mitochondrial ATPase 6 and 8 genes  
*S M Ware, N El-Hassan, S G Kahler, Q Zhang, Y-WMa, E Miller, B Wong, R L Spicer, W J Craigen, B A Kozel, D K Grange, L-J Wong*
- 315** Replication of restless legs syndrome loci in three European populations  
*D Kemlink, O Polo, B Frauscher, V Gschliesser, B Högl, W Poewe, P Vodicka, J Vavrova, K Sonka, S Nevsimalova, B Schormair, P Lichtner, K Silander, L Peltonen, C Gieger, H E Wichmann, A Zimprich, D Roeske, B Müller-Myhsok, T Meitinger, J Winkelmann*
- 319** Evaluation of a surveillance programme for women with a family history of breast cancer  
*M M Reis, M Tavakoli, J Dewar, D Goudie, A Cook, L McLeish, D Young, J Kenyon, M Steel*
- 324** Premature death in adults with 22q11.2 deletion syndrome  
*A S Bassett, E W C Chow, J Husted, K A Hodgkinson, E Oechslin, L Harris, C Silversides*
- 331** A new nonsense mutation of *SMAD8* associated with pulmonary arterial hypertension  
*M Shintani, H Yagi, T Nakayama, T Saji, R Matsuoka*

### Short report

- 338** Germline mutation in *DOK7* associated with fetal akinesia deformation sequence  
*J Vogt, N V Morgan, T Marton, S Maxwell, B J Harrison, D Beeson, E R Maher*

### Letters to JMG

- 341** Predictive diagnosis of the cancer prone Li-Fraumeni syndrome by accident: new challenges through whole genome array testing  
*T Schwarzbraun, A C Obenaus, A Langmann, U Gruber-Sedlmayr, K Wagner, M R Speicher, P M Kroisel*
- 345** Point mutations and a large intragenic deletion in *SPG11* in complicated spastic paraplegia without thin corpus callosum  
*C Crimella, A Arnoldi, F Crippa, M L Mostacciolo, F Boaretto, M Sironi, M Grazia D'Angelo, S Manzoni, L Piccinini, A C Turconi, A Toscano, O Musumeci, S Benedetti, R Fazio, N Bresolin, A Daga, A Martinuzzi, M T Bassi*

### Mutation reports

- 352** Genetic diagnosis of familial hypercholesterolaemia: the importance of functional analysis of potential splice-site mutations  
*M Bourbon, M A Duarte, A C Alves, A M Medeiros, L Marques, A K Soutar*
- 358** Germline mutation of microRNA-125a is associated with breast cancer  
*W Li, R Duan, F Kooy, S L Sherman, W Zhou, P Jin*



EDITOR'S CHOICE

This article has been chosen by the Editor to be of special interest or importance and is freely available online.



Articles carrying the Unlocked Logo are freely available online under the BMJ Journals unlocked scheme.

See <http://jmg.bmj.com/info/unlocked.dtl>

C O P E COMMITTEE ON PUBLICATION ETHICS

This journal is a member of and subscribes to the principles of the Committee on Publication Ethics

[www.publicationethics.org.uk](http://www.publicationethics.org.uk)

equator network

recycle

When you have finished with this please recycle it