

## Contents



### Review

- 529** The genetic and molecular bases of monogenic disorders affecting proteolytic systems *I Richard*
- 540** The location of constitutional neurofibromatosis 2 (NF2) splice site mutations is associated with the severity of NF2 *M E Baser, L Kuramoto, R Woods, H Joe, J M Friedman, A J Wallace, R T Ramsden, S Olschwang, E Bijlsma, M Kalamarides, L Papi, R Kato, J Carroll, C Lázaro, F Joncourt, D M Parry, G A Rouleau, D G R Evans*

### Hypothesis

- 547** Is the Ala12 variant of the PPARG gene an "unthrifty allele"? *E Ruiz-Narváez*

### Original articles

- 551** Homozygous mutations in *LPIN2* are responsible for the syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anaemia (Majeed syndrome) *P J Ferguson, S Chen, M K Tayeh, L Ochoa, S M Leal, A Pelet, A Munnich, S Lyonnet, H A Majeed, H El-Shanti*
- 558** Cloned fusion product from a rare t(15;19)(q13.2;p13.1) inhibit S phase in vitro *N Haruki, K S Kawaguchi, S Eichenberger, P P Massion, A Gonzalez, A F Gazdar, J D Minna, D P Carbone, T P Dang*
- 565** STRA13 expression and subcellular localisation in normal and tumour tissues: implications for use as a diagnostic and differentiation marker *A Ivanova, S-Y Liao, M I Lerman, S Ivanov, E J Stanbridge*

### Short reports

- 577** A new locus for hereditary haemorrhagic telangiectasia (*HHT3*) maps to chromosome 5 *S G Cole, M E Begbie, G M F Wallace, C L Shovlin*
- 583** Melanocortin-1 receptor gene variants affect pain and  $\mu$ -opioid analgesia in mice and humans *J S Mogil, J Ritchie, S B Smith, K Strasburg, L Kaplan, M R Wallace, R R Romberg, H Bijl, E Y Sarton, R B Fillingim, A Dahan*

### Letters to JMG

- 588** A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the *GJB2* gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment *F J del Castillo, M Rodríguez-Ballesteros, A Álvarez, T Hutchin, E Leonardi, C A de Oliveira, H Azaiez, Z Brownstein, M R Avenarius, S Marlin, A Pandya, H Shahin, K R Siemering, D Weil, W Wuyts, L A Aguirre, Y Martín, M A Moreno-Pelayo, M Villamar, K B Avraham, H-H M Dahl, M Kanaan, W E Nance, C Petit, R J H Smith, G Van Camp, E L Sartorato, A Murgia, F Moreno, I del Castillo*
- 595** A genome screen of families at high risk for Hodgkin lymphoma: evidence for a susceptibility gene on chromosome 4 *L R Goldin, M L McMaster, M Ter-Minassian, S Saddlemire, B Harmsen, G Lalonde, M A Tucker*
- 602** Breast and ovarian cancer risks to carriers of the BRCA1 5382insC and 185delAG and BRCA2 6174delT mutations: a combined analysis of 22 population based studies *A C Antoniou, P D P Pharoah, S Narod, H A Risch, J E Eyfjord, J L Hopper, N Loman, H Olsson, O Johannsson, Å Borg, B Pasini, P Radice, S Manoukian, D M Eccles, N Tang, E Olah, H Anton-Culver, E Warner, J Lubinski, J Gronwald, B Gorski, H Tulinius, S Thorlacius, H Eerola, H Nevanlinna, K Syrjäkoski, O-P Kallioniemi, D Thompson, C Evans, J Peto, F Lalloo, D G Evans, D F Easton*
- 604** Association of two tumour necrosis factor gene polymorphisms with the incidence of severe intraventricular haemorrhage in preterm infants *A Heep, A C Schueller, E Kattner, M Kroll, J Sander, M Wisbauer, P Bartmann, F Stueber*

### Correspondence

- e39** Update on the Manchester Scoring System for *BRCA1* and *BRCA2* testing *D G R Evans, F Lalloo, A Wallace, N Rahman*

### JMG Unlocked

Articles carrying the



logo are available free via the journal's website: [www.jmedgenet.com](http://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

- e40** Transmission of mutant alleles to female offspring of BRCA1 carriers in Poland  
*J Gronwald, T Byrski, T Huzarski, S A Narod, J Lubinski*
- e41** Re: Pitfalls of automated comparative sequence analysis as a single platform for routine clinical testing for NF1 (Messiaen and Wimmer) *J L Whittaker, C Mattocks, D Baralle, P Tarpey, C French-Constant, M Bobrow*
- e42** BRCA1 mutation and neuronal migration defect: implications for chemoprevention  
*D Eccles, D Bunyan, S Barker, B Castle*

---

### Electronic letters

- e43** Mutations in *FLNB* cause boomerang dysplasia *L S Bicknell, T Morgan, L Bonafé, M W Wessels, M G Bialer, P J Willems, D H Cohn, D Krakow, S P Robertson*
- e44** Association of a *COL1A1* polymorphism with lumbar disc disease in young military recruits *C Tilkeridis, T Bei, S Garantziotis, C A Stratakis*

- e45** A combination of genetic polymorphisms increases the risk of progressive disease in chronic hepatitis C *M M Richardson, E E Powell, H D Barrie, A D Clouston, D M Purdie, J R Jonsson*

---

### Online mutation reports

- e46** Significant association between *IRF6* 820G→A and non-syndromic cleft lip with or without cleft palate in the Thai population *C Srichomthong, P Siriwan, V Shotelersuk*
- e47** Molecular genetics of autosomal dominant retinitis pigmentosa (ADRP): a comprehensive study of 43 Italian families *C Ziviello, F Simonelli, F Testa, M Anastasi, S B Marzoli, B Falsini, D Ghiglione, C Macaluso, M P Manitto, C Garrè, A Ciccodicola, E Rinaldi, S Banfi*