

## Contents



### Review

- 1** Rett syndrome: clinical review and genetic update *L S Weaving, C J Ellaway, J Gécz, J Christodoulou*

### Original articles

- 8** The complex nature of constitutional de novo apparently balanced translocations in patients presenting with abnormal phenotypes *S M Gribble, E Prigmore, D C Burford, K M Porter, Bee Ling Ng, E J Douglas, H Fiegler, P Carr, D Kalaitzopoulos, S Clegg, R Sandstrom, I K Temple, S A Youings, N S Thomas, N R Dennis, P A Jacobs, J A Crolla, N P Carter*
- 17** Fine mapping of a region on chromosome 21q21.11–q22.3 showing linkage to type 1 diabetes *R Bergholdt, J Nerup, F Pociot*
- 26** Angiotensin converting enzyme gene polymorphism and cardiovascular morbidity and mortality: the Rotterdam Study *F A Seyed-Tabatabaei, A F C Schut, A Arias Vásquez, A M Bertoli-Avella, A Hofman, J C M Witteman, C M van Duijn*
- 31** A *PDGFRA* promoter polymorphism, which disrupts the binding of ZNF148, is associated with primitive neuroectodermal tumours and ependymomas *C De Bustos, A Smits, B Strömberg, V P Collins, M Nistér, G Afink*
- 38** Biochemical analysis of cultured chorionic villi for the prenatal diagnosis of peroxisomal disorders: biochemical thresholds and molecular sensitivity for maternal cell contamination detection *S Steinberg, S Katsanis, A Moser, G Cutting*
- 45** Multiple meningiomas: differential involvement of the *NF2* gene in children and adults *D G R Evans, C Watson, A King, A J Wallace, M E Baser*

### Short report

- 49** Array based CGH and FISH fail to confirm duplication of 8p22-p23.1 in association with Kabuki syndrome *J D Hoffman, Y Zhang, J Greshock, K L Ciprero, B S Emanuel, E H Zackai, B L Weber, J E Ming*

- 58** New insights into cystinuria: 40 new mutations, genotype–phenotype correlation, and digenic inheritance causing partial phenotype *M Font-Llitjós, M Jiménez-Vidal, L Bisceglia, M Di Perna, L de Sanctis, F Rousaud, L Zelante, M Palacín, V Nunes*
- 69** A report of a national mutation testing service for the *MEN1* gene: clinical presentations and implications for mutation testing *J W Cardinal, L Bergman, N Hayward, A Sweet, J Warner, L Marks, D Learoyd, T Dwight, B Robinson, M Epstein, M Smith, B T Teh, D P Cameron, J B Prins*

### Letters to JMG

- 75** Evidence for a gene influencing haematocrit on chromosome 6q23–24: genomewide scan in the Framingham Heart Study *J-P Lin, C J O'Donnell, D Levy, L A Cupples*
- 80** A new locus for autosomal recessive complicated hereditary spastic paraplegia (SPG26) maps to chromosome 12p11.1–12q14 *P A Wilkinson, M A Simpson, L Bastaki, H Patel, J A Reed, K Kalidas, E Samilchuk, R Khan, T T Warner, A H Crosby*
- 83** A full genome scan for gastric cancer *M Aoki, Y Yamamura, H Noshiro, K Sakai, J Yokota, T Kohno, T Tokino, S Ishida, S Ohyama, I Ninomiya, K Uesaka, M Kitajima, S Shimada, S Matsuno, M Yano, M Hiratsuka, H Sugimura, F Itoh, T Minamoto, Y Maehara, S Takenoshita, T Aikou, H Katai, K Yoshimura, T Takahashi, K Akagi, M Sairenji, K Yamamoto, T Sasazuki*
- 88** Identification of a new locus for isolated familial keratoconus at 2p24 *H Hutchings, H Ginisty, M Le Gallo, D Levy, F Stoësser, J F Rouland, J L Arné, M H Laloux, P Calvas, M P Roth, A Hovnanian, F Malecaze*
- 95** The potential for increased clinical sensitivity in genetic testing for polyposis colorectal cancer through the analysis of *MYH* mutations in North American patients *K Eliason, B C Hendrickson, T Judkins, M Norton, B Leclair, E Lyon, B Ward, W Noll, T Scholl*

### Medical genetics in practice

- 54** An aetiological classification of birth

contd...

**NEW  
ONLINE  
SUBMISSION**

**GO TO  
WEBSITE  
TO SUBMIT YOUR  
MANUSCRIPT**

## Contents ...contd

---

### Electronic letter

**e1** A third locus for dominant optic atrophy on chromosome 22q *F Barbet, S Hakiki, C Orssaud, S Gerber, I Perrault, S Hanein, D Ducroq, J-L Dufier, A Munnich, J Kaplan, J-M Rozet*

---

### Online mutation reports

**e2** Sensorineural hearing loss, striate palmoplantar hyperkeratosis, and knuckle pads in a patient with a novel connexin 26 (*GJB2*) mutation *N J Leonard, A L Krol, S Bleoo, M J Somerville*

**e3** A 17p11.2 germline deletion in a patient with Smith-Magenis syndrome and neuroblastoma *T Hienonen, H Sammalkorpi, P Isohanni, R Versteeg, R Karikoski, L A Aaltonen*

**e4** First occurrence of aprosencephaly/atelencephaly and holoprosencephaly in a family with a *SIX3* gene mutation and phenotype/genotype correlation in our series of *SIX3* mutations *L Pasquier, C Dubourg, M Gonzales, L Lazaro, V David, S Odent, F Encha-Razavi*

**e5** Prevalence of *BRCA2* mutations in a hospital based series of unselected breast cancer cases *S-W Kim, C S Lee, J V Fey, P I Borgen, J Boyd*