

## Contents



### Review article

- 1** Fanconi anaemia *M D Tischkowitz, S V Hodgson*

### Original articles

- 11** A third MRX family (MRX68) is the result of mutation in the long chain fatty acid-CoA ligase 4 (FACL4) gene: proposal of a rapid enzymatic assay for screening mentally retarded patients *I Longo, S G M Frints, J-P Fryns, I Meloni, C Pescucci, F Ariani, M Borghgraef, M Raynaud, P Marynen, C Schwartz, A Renieri, G Froyen*
- 18** The human Y chromosome's azoospermia factor b (AZFb) region: sequence, structure, and deletion analysis in infertile men *A Ferlin, E Moro, A Rossi, B Dallapiccola, C Foresta*
- 25** Differential targets of CpG island hypermethylation in primary and metastatic head and neck squamous cell carcinoma (HNSCC) *D J Smiraglia, L T Smith, J C Lang, L J Rush, Z Dai, D E Schuller, C Plass*

### Short reports

- 34** In frame fibrillin-1 gene deletion in autosomal dominant Weill-Marchesani syndrome *L Faivre, R J Gorlin, M K Wirtz, M Godfrey, N Dagoneau, J R Samples, M Le Merrer, G Collod-Beroud, C Boileau, A Munnich, V Cormier-Daire*
- 37** Familial vestibulocerebellar disorder maps to chromosome 13q31-q33: a new nystagmus locus *N K Ragge, C Hartley, A M Dearlove, J Walker, I Russell-Eggitt, C M Harris*

### Letters to JMG

- 42** Identification of a mutation in the Indian Hedgehog (IHH) gene causing brachydactyly type A1 and evidence for a third locus *T J Kirkpatrick, K-S Au, J M Mastrobattista, M E McCready, D E Bulman, H Northrup*
- 45** Non-syndromic recessive auditory neuropathy is the result of mutations in the otoferlin (OTOF) gene *R Varga, P M Kelley, B J Keats, A Starr, S M Leal, E Cohn, W J Kimberling*

- 51** Familial Sotos syndrome is caused by a novel 1 bp deletion of the NSD1 gene *P Höglund, N Kurotaki, S Kytölä, N Miyake, M Somer, N Matsumoto*

- 55** P63 mutations are not a major cause of non-syndromic split hand/foot malformation *X J de Mollerat, D B Everman, C T Morgan, K B Clarkson, R C Rogers, R S Colby, A S Aylsworth, J M Graham Jr, R E Stevenson, C E Schwartz*

- 62** Beckwith-Wiedemann syndrome and assisted reproduction technology (ART) *E R Maher, L A Brueton, S C Bowdin, A Luharia, W Cooper, T R Cole, F Macdonald, J R Sampson, C L Barratt, W Reik, M M Hawkins*

- 65** Recessive multiple epiphyseal dysplasia (rMED): phenotype delineation in eighteen homozygotes for DTDST mutation R279W *D Ballhausen, L Bonafé, P Terhal, S L Unger, G Bellus, M Classen, B C Hamel, J Spranger, B Zabel, D H Cohn, W G Cole, J T Hecht, A Superfi-Furga*

- 72** Chromosome 15 maternal uniparental disomy and psychosis in Prader-Willi syndrome *A Vogels, G Matthijs, E Legius, K Devriendt, J-P Fryns*

- 74**  $\beta_1$ -adrenergic antagonists and melatonin reset the clock and restore sleep in a circadian disorder, Smith-Magenis syndrome *H De Leersnyder, J L Bresson, M-C de Blois, J-C Souberbielle, A Mogenet, B Delhotal-Landes, F Salefranque, A Munnich*

- 79** Instructions for authors

### Echo

- 24** COPD research is urged to get interactive

contd...

**NEW**  
**ONLINE**  
SUBMISSION  
**GO TO**  
**WEBSITE**  
TO SUBMIT YOUR  
MANUSCRIPT