

## Review article

Spinal muscular atrophy: untangling the knot? *I Biros, S Forrest* 1

## Original articles

Sex related expressivity of the phenotype in coronal craniosynostosis caused by the recurrent P250R FGFR3 mutation *E Lajeunie, V El Ghouzzi, M Le Merrer, A Munnich, J Bonaventure, D Renier* 9

Molecular analysis of the APC gene in 205 families: extended genotype-phenotype correlations in FAP and evidence for the role of APC amino acid changes in colorectal cancer predisposition *Y L Wallis, D G Morton, C M McKeown, F Macdonald* 14

Molecular basis of iduronate-2-sulphatase gene mutations in patients with mucopolysaccharidosis type II (Hunter syndrome) *P Li, A B Bellows, J N Thompson* 21

Mucopolysaccharidosis type IIIB (Sanfilippo B): identification of 18 novel  $\alpha$ -N-acetylglucosaminidase gene mutations *S Bunge, A Knigge, C Steglich, W J Kleijer, O P van Diggelen, M Beck, A Gal* 28

Clinical and genetic studies on 12 preaxial polydactyly families and refinement of the localisation of the gene responsible to a 1.9 cM region on chromosome 7q36 *J Zguricas, H Heus, E Morales-Peralta, G Breedveld, B Kuyt, E F Mumcu, W Bakker, N Akarsu, S P J Kay, S E R Hovius, L Heredero-Baute, B A Oostra, P Heutink* 32

Cystic fibrosis carrier frequencies in populations of African origin *C Padoa, A Goldman, T Jenkins, M Ramsay* 41

Preimplantation genetic diagnosis for couples at high risk of Down syndrome pregnancy owing to parental translocation or mosaicism *C M Conn, J Cozzi, J C Harper, R M L Winston, J D A Delhanty* 45

Sotos syndrome and cutis laxa *S P Robertson, A Bankier* 51

## Short reports

A small interstitial deletion in the GPC3 gene causes Simpson-Golabi-Behmel syndrome in a Dutch-Canadian family *J Y Xuan, R M Hughes-Benzie, A E MacKenzie* 57

Myotonic dystrophy: the correlation of (CTG) repeat length in leucocytes with age at onset is significant only for patients with small expansions *M G Hamshere, H Harley, P Harper, J D Brook, J F Y Brookfield* 59

Unstable expansion of the CAG trinucleotide repeat in MAB21L1: report of a second pedigree and effect on protein expression *R L Margolis, O C Stine, C M Ward, M L Franz, A Rosenblatt, C Callahan, M Sherr, C A Ross, N T Potter* 62

Familial adenomatous polyposis associated with multiple adrenal adenomas in a patient with a rare 3' APC mutation *A Kartheuser, C Walon, S West, C Breukel, R Detry, A-C Gribomont, T Hamzehloei, P Hoang, D Maiter, J Pringot, J Rahier, P Meera Khan, A Curtis, J Burn, R Fodde, C Verellen-Dumoulin* 65

High prevalence of the C634Y mutation in the RET proto-oncogene in MEN 2A families in Spain *B Sánchez, M Robledo, J Biarnes, M-E Sáez, V Volpini, J Benítez, E Navarro, A Ruiz, G Antiñolo, S Borrego* 68

De novo 10q22 interstitial deletion *L Cook, D D Weaver, J K Hartsfield Jr, G H Vance* 71

Duplication of segment 1p21 following paternal insertional translocation,  $ins(6;1)(q25;p13.3p22.1)$  *A Utkus, I Sorokina, V Kucinskas, B Röthlisberger, D Balmer, L Brecevic, A Schinzel* 73

Molecular and cytogenetic characterisation of an unusual case of partial trisomy/partial monosomy 13 mosaicism:  $46,XX,r(13)(p11q14)/46,XX,der(13)t(13;13)(q10;q14)$  *M Gentile, A L Buonadonna, F Cariola, P Fiorente, M C Valenzano, Ginevra Guanti* 77

Anomalous right pulmonary artery origins in association with the fetal valproate syndrome *C N Mo, E J Ladusans* 83

## Letters to the Editor

Frequency of mutations for glycogen storage disease type II in different populations: the  $\Delta 525T$  and  $\Delta$ exon 18 mutations are not generally "common" in white populations *R Hirschhorn, M L Huie* 85

Infant methionine synthase variants and risk for spina bifida *G M Shaw, K Todoroff, R H Finnell, E J Lammer, D Leclerc, R A Gravel, R Rozen* 86

## Notice to contributors

88

# JMG

JOURNAL OF MEDICAL GENETICS

