

Contents



Review

- 225** X-linked retinoschisis: an update
S K Sikkink, S Biswas, N R A Parry,
P E Stanga, D Trump

Original articles

- 233** Sensorineural deafness and male infertility: a contiguous gene deletion syndrome Y Z Zhang, M Malekpour,
N Al-Madani, K Kahrizi, M Zanganeh,
M Mohseni, F Mojahedi, A Daneshi,
H Najmabadi, R J H Smith
- 241** A comprehensive strategy for the subtyping of patients with Fanconi anaemia: conclusions from the Spanish Fanconi Anemia Research Network
J A Casado, E Callén, A Jacome, P Río,
M Castella, S Lobitz, T Ferro, A Muñoz,
J Sevilla, A Cantalejo, E Cela, J Cervera,
J Sánchez-Calero, I Badell, J Estella, A Dasí,
T Olivé, J J Ortega, A Rodriguez-Villa,
M Tapia, A Molinés, L Madero,
J C Segovia, K Neveling, R Kalb,
D Schindler, H Hanenberg, J Surrallés,
J A Bueren
- 250** Molecular characterisation of a mosaicism with a complex chromosome rearrangement: evidence for coincident chromosome healing by telomere capture and neo-telomere formation E Chabchoub,
L Rodríguez, E Galán, E Mansilla,
M L Martínez-Fernandez,
M L Martínez-Frías, J-P Fryns, J R Vermeesch
- 257** Chromosome 11 segmental paternal isodisomy in amniocytes from two fetuses with omphalocele: new highlights on phenotype–genotype correlations in Beckwith–Wiedemann syndrome
F R Grati, L Turolla, P D’Ajello, A Ruggeri,
M Miozzo, G Bracalente, D Baldo,
L Laurino, R Boldorini, E Frate, N Surico,
L Larizza, F Maggi, G Simoni

Short report

- 264** Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14
B Menten, K Buysse, F Zahir, J Hellermans,
S J Hamilton, T Costa, C Fagerstrom,
G Anadiotis, D Kingsbury, B C McGillivray,
M A Marra, J M Friedman, F Speleman,
G Mortier

Letters to JMG

- 269** Clinical and molecular cytogenetic characterisation of a newly recognized microdeletion syndrome involving 2p15-16.1 E Rajcan-Separovic, C Harvard,
X Liu, B McGillivray, J G Hall, Y Qiao,
J Hurlbert, J Hildebrand, E C R Mickelson,
J J A Holden, M E S Lewis
- 277** Congenital disorder of glycosylation type la presenting with hydrops fetalis
J M van de Kamp, D J Lefebre,
G J G Ruijter, S J Steggerda,
N S den Hollander, S M Willems,
G Matthijs, B J H M Poorthuis, R A Wevers
- 281** Exon deletions of *SPG4* are a frequent cause of hereditary spastic paraparesis
C Depienne, E Fedirko, S Forlani,
C Cazeneuve, P Ribai, I Feki, C Tallaksen,
K Nguyen, B Stankoff, M Ruberg,
G Stevanin, A Durr, A Brice
- 285** Phenotypic and population differences in the association between *CILP* and lumbar disc disease I M Virtanen, Y Q Song,
K M C Cheung, L Ala-Kokko, J Karppinen,
D W H Ho, K D K Luk, S P Yip, J C Y Leong,
K S E Cheah, P Sham, D Chan

Electronic letters

- e72** A survey of locus-specific database curation R G H Cotton, K Phillips,
O Horaitis
- e73** The Shwachman–Bodian–Diamond syndrome gene mutations cause a neonatal form of spondylometaphyseal dysplasia (SMD) resembling SMD Sedaghatian type G Nishimura,
E Nakashima, Y Hirose, T Cole, P Cox,
D H Cohn, D L Rimoin, R S Lachman,
Y Miyamoto, B Kerr, S Unger, H Ohashi,
A Superti-Furga, S Ikegawa

Online mutation report

- e74** Mutations in the ND5 subunit of complex I of the mitochondrial DNA are a frequent cause of oxidative phosphorylation disease M J Blok, L Spruijt, I F M de Coo,
K Schoonderwoerd, A Hendrickx,
H J Smeets

contd...

Contents ...contd

Correspondence

- e75** Array comparative genomic hybridisation analysis of boys with X-linked hypopituitarism identifies a 3.9 Mb duplicated critical region at Xq27 containing SOX3 N M Solomon, S A Ross, T Morgan, J L Belsky, F A Hol, P S Karnes, N J Hopwood, S E Myers, A S Tan, G L Wame, S M Forrest, P Q Thomas

- e76** Phenocopies in breast cancer 1 (BRCA1) families: implications for genetic counseling J Gronwald, C Cybulski, J Lubinski, S A Narod
- e77** Hypomethylation in the 11p15 telomeric imprinting domain in a patient with Silver–Russell syndrome with a CSH1 deletion (17q24) renders a functional role of this alteration unlikely T Eggermann, N Schönher, K Eggermann, H Wollmann