

Contents

Review

- 497** Apoptosis and cancer: mutations within caspase genes *S Ghavami, M Hashemi, S R Ande, B Yeganeh, W Xiao, M Eshraghi, C J Bus, K Kadkhoda, E Wiechec, A J Halayko, M Los*

Original articles

- 511** Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome *B W M van Bon, H C Mefford, B Menten, D A Koolen, A J Sharp, W M Nillesen, J W Innis, T J L de Ravel, C L Mercer, M Fichera, H Stewart, L E Connell, K Öunap, K Lachlan, B Castle, N Van der Aa, C van Ravenswaaij, M A Nobrega, C Serra-Juhé, I Simonic, N de Leeuw, R Pfundt, E M Bongers, C Baker, P Finnemore, S Huang, V K Maloney, J A Crolla, M van Kalmthout, M Elia, G Vandeweyer, J P Fryns, S Janssens, N Foulds, S Reitano, K Smith, S Parkel, B Loeys, C G Woods, A Oostra, F Speleman, A C Pereira, A Kurg, L Willatt, S J L Knight, J R Vermeesch, C Romano, J C Barber, G Mortier, L A Pérez-Jurado, F Kooy, H G Brunner, E E Eichler, T Kleefstra, B B A de Vries*

- 524** 17q21.31 microduplication patients are characterised by behavioural problems and poor social interaction *B Grisart, L Willatt, A Destrée, J-P Fryns, K Rack, T de Ravel, J Rosenfeld, J R Vermeesch, C Verellen-Dumoulin, R Sandford*

- 531** High resolution array analysis: diagnosing pregnancies with abnormal ultrasound findings *M Tyreman, K M Abbott, L R Willatt, R Nash, C Lees, J Whittaker, I Simonic*

- 542** In vitro splicing analysis showed that availability of a cryptic splice site is not a determinant for alternative splicing patterns caused by +1G→A mutations in introns of the dystrophin gene *Y Habara, Y Takeshima, H Awano, Y Okizuka, Z Zhang, K Saiki, M Yagi, M Matsuo*

August 2009 Volume 46 Issue 8

- 548** Natural course of Fabry disease: changing pattern of causes of death in FOS – Fabry Outcome Survey *A Mehta, J T R Clarke, R Giugliani, P Elliott, A Linhart, M Beck, G Sunder-Plassmann, on behalf of the FOS Investigators*

Letters to JMG


- 553** Association of *RASGRP1* with type 1 diabetes is revealed by combined follow-up of two genome-wide studies *H Q Qu, S F A Grant, J P Bradfield, C Kim, E Frackelton, H Hakonarson, C Polychronakos*
- 555** A functional haplotype variant in the *TBX22* promoter is associated with cleft palate and ankyloglossia *E Pauws, G E Moore, P Stanier*
- 562** Identification of loss-of-function mutations of *SLC35D1* in patients with Schneckbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases *T Furuichi, H Kayserili, S Hiraoka, G Nishimura, H Ohashi, Y Alanay, J C Lerena, A D Aslanger, H Koseki, D H Cohn, A Superti-Furga, S Unger, S Ikegawa*


Mutation report

- 569** Identification of 13 novel *NLRP7* mutations in 20 families with recurrent hydatidiform mole; missense mutations cluster in the leucine-rich region *C M Wang, P H Dixon, S Decordova, M D Hodges, N J Sebire, S Ozalp, M Fallahian, A Sensi, F Ashrafi, V Repiska, J Zhao, Y Xiang, P M Savage, M J Seckl, R A Fisher*

PostScript

- 576** Correspondence
- 576** Corrections

 This article has been chosen by the Editor to be of special interest or importance and is freely available online.

 Articles carrying the Unlocked Logo are freely available online under the BMJ Journals unlocked scheme. See <http://jmg.bmj.com/info/unlocked.dtl>

C O P E COMMITTEE ON PUBLICATION ETHICS

This journal is a member of and subscribes to the principles of the Committee on Publication Ethics
www.publicationethics.org.uk

 equator network

 recycle
When you have finished with this please recycle it