

Review article

Complex and segmental uniparental disomy (UPD): review and lessons from rare chromosomal complements *D Kotzot* 497

Original articles

VHL c.505 T>C mutation confers a high age related penetrance but no increased overall mortality *B U Bender, C Eng, M Olschewski, D P Berger, J Laubenberger, C Altehöfer, G Kirste, M Orszagh, V van Velthoven, H Mioszcza, D Schmidt, H P H Neumann* 508

A common founder for the 35delG *GJB2* gene mutation in connexin 26 hearing impairment *L Van Laer, P Coucke, R F Mueller, G Caethoven, K Flothmann, S D Prasad, G P Chamberlin, M Houseman, G R Taylor, C M Van de Heyning, E Fransen, J Rowland, R A Cucci, R J H Smith, G Van Camp* 515

Predictive genetic testing in children and adults: a study of emotional impact *S Michie, M Bobrow, T M Marteau, FAP Collaborative Research Group* 519

Letters to the Editor

Dutch patients with glycogen storage disease type II show common ancestry for the 525delT and del exon 18 mutations *M G E M Ausems, K ten Berg, L A Sandkuijl, M A Kroos, A F J Bardoel, K N Roumelioti, A J J Reuser, R Sinke, C Wijmenga* 527

Recurrent mutations in the deafness gene *GJB2* (connexin 26) in British Asian families *S Rickard, D P Kelsell, T Sirimana, K Rajput, B MacArdle, M Bitner-Glindzicz* 530

A region of homozygosity within 22q11.2 associated with congenital heart disease: recessive DiGeorge/velocardiofacial syndrome? *J Henwood, C Pickard, J P Leek, C P Bennett, Y J Crow, J D R Thomson, M Ahmed, K G Watterson, J M Parsons, E Roberts, N J Lench* 533

High frequency of the ApoB-100 R3500Q mutation in Bulgarian hypercholesterolaemic subjects *A Horvath, A Savov, S Kirov, E Karshelova, I Paskaleva, A Goudev, V Ganev* 536

A community based study on intentions regarding predictive testing for hereditary breast cancer *M Welkenhuysen, G Evers-Kiebooms, M Decruyenaere, E Claes, L Denayer* 540

Ocular malformations, postaxial polydactyly, and delayed intramembranous ossification: a new autosomal dominant condition *D M Martin, J L Gorski* 547

Recessively inherited lower incisor hypodontia *S Pirinen, A Kentala, P Nieminen, T Varilo, I Thesleff, S Arte* 551

Congenital diaphragmatic hernia and interstitial deletion of chromosome 3 *P Brennan, G D Croaker, M Heath* 556

A supernumerary marker chromosome with a neocentromere derived from 5p14→pter *B Fritz, I Dietze, A Wandall, M Aslan, A Schmidt, E Kattner, R Schwerdtfeger, U Friedrich* 559

A case of Roberts syndrome described in 1737 *A W Bates* 565

Instructions for authors 568

Electronic letters

A novel mutation in a family with non-syndromic sensorineural hearing loss that disrupts the newly characterised OTOF long isoforms *M J Houseman, A P Jackson, L I Al-Gazali, R A Badin, E Roberts, R F Mueller* e25

MET mutation and familial gastric cancer *J D Chen, S Kearns, T Porter, F M Richards, E R Maher, B T Teh* e26

Agenesis of cruciate ligaments and menisci causing severe knee dysplasia in TAR syndrome *D Héron, C Bonnard, C Moraine, A Toutain* e27

JMG

JOURNAL OF MEDICAL GENETICS

www.jmedgenet.com

JOURNAL OF MEDICAL GENETICS

August 2001

