

Contents

Original articles

Clustering of Y chromosome deletions in subinterval E of interval 6 supports the existence of an oligozoospermia critical region outside the DAZ gene L Stuppia, V Gatta, G Mastroprimiano, F Pompetti, G Calabrese, P G Franchi, E Morizio, R Mingarelli, M Nicolai, R Tenaglia, L Impronta, V Sforza, S Bisceglia, G Palka 881

Low frequency of BRCA1 germline mutations in 45 German breast/ovarian cancer families U Hamann, M Häner, U Stosiek, G Bastert, R J Scott 884

Atypical hereditary neuropathy with liability to pressure palsies (HNPP): the value of direct DNA diagnosis M Sessa, R Nemni, A Quattrini, U Del Carro, L Wrabetz, N Canal 889

Mutation and haplotype analysis of phenylalanine hydroxylase alleles in classical PKU patients from the Czech Republic: identification of four novel mutations L Kozák, M Blažková, V Kuhrová, A Pijáčková, Š Ržičková, S Šrástná 893

Different proximal and distal rearrangements of chromosome 7q associated with holoprosencephaly B Benzacken, J P Siffroi, C Le Bourhis, K Krabchi, N Joyé, F Maschino, F Viguié, J Soulié, M Gonzales, G Migné, M Bucourt, F Encha-Razavi, Lionel Carbillon, J L Taillemite 899

Familial streptomycin ototoxicity in a South African family: a mitochondrial disorder J C Gardner, R Goliath, D Viljoen, S Sellars, G Cortopassi, T Hutchin, J Greenberg, P Beighton 904

DNA testing for fragile X syndrome: implications for parents and family M A van Rijn, B B A de Vries, A Tibben, A M W van den Ouweland, D J J Halley, M F Niermeijer 907

Rapid identification of multiple supernumerary ring chromosomes with a new FISH technique C Mackie-Ogilvie, K Waddle, J Mandeville, M J Seller, Z Docherty 912

Syndrome of the month

Prader-Willi syndrome S B Cassidy 917

Short reports

Prenatal diagnosis of the fragile X syndrome: loss of mutation owing to a double recombinant or gene conversion event at the FMR1 locus M Losekoot, E Hoogendoorn, R Olmer, C C A M Jansen, J C Oosterwijk, A M W van den Ouweland, D J J Halley, S T Warren, R Willemsen, B A Oostra, E Bakker 924

Conotruncal heart defect/microphthalmia syndrome: delineation of an autosomal recessive syndrome M C Digilio, B Marino, A Giannotti, B Dallapiccola 927

Paternal transmission of congenital myotonic dystrophy C E M de Die-Smulders, H J M Smeets, W Loots, H B M Anten, J F Mirandolle, J P M Geraedts, C J Höweler 930

Misleading linkage results in an NF2 presymptomatic test owing to mosaicism E K Bijlsma, A J Wallace, D G R Evans 934

Associated malformations in the family of a patient with Meckel syndrome: heterozygous expression? R Gulati, S R Phadke, S S Agarwal 937

Mucopolysaccharidosis type I: identification of novel mutations that cause Hurler/Scheie syndrome in Chinese families G-J Lee-Chen, T-R Wang 939

Radial aplasia and chromosome 22q11 deletion M C Digilio, A Giannotti, B Marino, A M Guadagni, M Orzalesi, B Dallapiccola 942

Double partial trisomy 9q34.1→qter and 21pter→q22.11: FISH and clinical findings T Mattina, M Pierluigi, D Mazzone, S Scardilli, C Perfumo, F Mollica 945

Germline duplication of chromosome 2p and neuroblastoma J S Patel, J Pearson, L Willatt, T Andrews, R Beach, A Green 949

Median cleft of upper lip and pedunculated skin masses associated with de novo reciprocal translocation 46,X,t(X;16)(q28;q11.2) M Masuno, K Imaizumi, Y Fukushima, Y Tanaka, T Ishii, M Nakamura, Y Kuroki 952

Diagnostic advances

Rapid detection of the major deletion in the Batten disease gene CLN3 by allele specific PCR P E M Taschner, N de Vos, M H Breuning 955

Abstracts

Medical genetics: advances in brief 957

Letters to the editor

Methionine synthase and neural tube defects K Morrison, Y H Edwards, S A Lynch, J Burn, F Hol, E Mariman 958

Limb-girdle muscular dystrophy or spinal muscular atrophy: a source of diagnostic confusion? R Pogue, T Jackson, B Sayli, A Curtis, K M D Bushby 958

Book reviews 959

Notices 960