



Contents

Original articles

Dominant X linked subcortical laminar heterotopia and lissencephaly syndrome (XSCLH/LIS): evidence for the occurrence of mutation in males and mapping of a potential locus in Xq22

V des Portes, J M Pinard, D Smadja, J Motte, O Boespflug-Tanguy, M L Moutard, I Desguerre, P Billuart, A Carrie, T Bienvenu, M C Vinet, L Bachner, C Beldjord, O Dulac, A Kahn, G Ponsot, J Chelly

Hereditary spinal neurofibromatosis: a rare form of NF1? M Poyhonen, E-L Leisti, S Kytölä, J Leisti

Recurrence risk figures for isolated tetralogy of Fallot after screening for 22q11 microdeletion

M C Digilio, B Marino, A Giannotti, A Toscano, B Dallapiccola

Rieger syndrome locus: a new reciprocal translocation t(4;12)(q25;q15) and a deletion

del(4)(q25q27) both break between markers D4S2945 and D4S193 R H Flomen, P A Gorman, R Vatcheva, J Groet, I Barišić, I Ligutić, D Sheer, D Nižetić

A variant of the Nijmegen breakage syndrome with unusual cytogenetic features and intermediate cellular radiosensitivity R Tupler, G L Marseglia, M Stefanini, E Prosperi, L Chessa, T Nardo, A Marchi, P Maraschio

Submicroscopic deletions at 16p13.3 in Rubinstein-Taybi syndrome: frequency and clinical manifestations in a North American population R Wallerstein, C E Anderson, B Hay, P Gupta, L Gibas, K Ansari, F S Cowchock, V Weinblatt, C Reid, A Levitas, L Jackson

A FISH approach to defining the extent and possible clinical significance of deletions at the WAGR locus J A Crolla, J E Cawdery, C A Oley, I D Young, J Gray, J Fantès, V van Heyningen

Localisation of a 10q breakpoint within the PAX2 gene in a patient with a de novo t(10;13) translocation and optic nerve coloboma-renal disease K Narahara, E Baker, S Ito, Y Yokoyama, S Yu, D Hewitt, G R Sutherland, M R Eccles, R I Richards

Inherited DNA amplification of the proximal 15q region: cytogenetic and molecular studies C Mignon, F Parente, C Stavropoulou, P Collignon, A Moncla, C Turc-Carel, M-G Mattei

Characterisation of CAH alleles with non-radioactive DNA single strand conformation

polymorphism analysis of the CYP21 gene A Bobba, A Iolascon, S Giannattasio, M Albrizio, A Sinisi, F Prisco, F Schettini, E Marra

Mild myotonic dystrophy is associated with memory impairment in the context of normal general intelligence J S Rubinsztein, D C Rubinsztein, P J McKenna, S Goodburn, A J Holland

Non-Mendelian transmission at the Machado-Joseph disease locus in normal females: preferential transmission of alleles with smaller CAG repeats D C Rubinsztein, J Leggo

Genetic counselling: the psychological impact of meeting patients' expectations S Michie, T M Marteau, M Bobrow

Short reports

De novo der(X)t(X;10)(q26;q21) with features of distal trisomy 10q: case report of paternal origin identified by late replication with BrdU and the human androgen receptor assay (HAR)

J Garcia-Heras, J A Martin, S F Witchel, P Scacheri

Karyotype 69,XXX/47,XX,+15 in a 2½ year old child J Dean, G Cohen, J Kemp, L Robson, V Tembe, J Hasselaar, B Webster, A Lammi, A Smith

Rapid antibody test for prenatal diagnosis of fragile X syndrome on amniotic fluid cells: a new appraisal R Willemsen, F Los, S Mohkamsing, A van den Ouweland, W Deelen, H Galjaard, B Oostra

Cebocephaly, alobar holoprosencephaly, spina bifida, and sirenomelia in a stillbirth C-P Chen, S-L Shih, F-F Liu, S-W Jan

Tuberous sclerosis complex: neonatal deaths in three of four children of consanguineous, non-expressing parents M Ruggieri, C Carbonara, G Magro, N Migone, S Grasso, A Tinè, L Pavone, M R Gomez

Further evidence for preaxial hallucal polydactyly as a marker of diabetic embryopathy J Slee, J Goldblatt

Letter to the Editor

Avoiding errors in the diagnosis of (CAG)_n expansion in the huntingtin gene C Holzmann, A M M V Saecker, J T Epplen, O Riess

5764 X693 10
08•17•99 MAR