



Contents

Editorial

- Mutation databases on the web** A I Wacey, E G D Tuddenham 529

Original articles

- Molecular screening for proximal 15q abnormalities in a mentally retarded population** J Jacobsen, B H King, B L Leventhal, S L Christian, D H Ledbetter, E H Cook Jr 534

- Uniparental and functional X disomy in Turner syndrome patients with unexplained mental retardation and X derived marker chromosomes** T Yorifuji, J Muroi, M Kawai, A Uematsu, H Sasaki, T Momoi, M Kaji, C Yamanaka, K Furusho 539

- Analysis of a familial three way translocation involving chromosomes 3q, 6q, and 15q by high resolution banding and fluorescent in situ hybridisation (FISH) shows two different unbalanced karyotypes in sibs** D Wiczorek, H Engels, R Viersbach, B Henke, G Schwanzitz, E Passarge 545

- Choroid plexus cysts and aneuploidy** D Peleg, J Yankowitz 554

- Smith-Lemli-Opitz syndrome: a variable clinical and biochemical phenotype** A K Ryan, K Bartlett, P Clayton, S Eaton, L Mills, D Donnai, R M Winter, J Burn 558

- A distinct form of spondyloepimetaphyseal dysplasia with multiple dislocations** C M Hall, N H Elçioglu, D G Shaw 566

- Identification of a common low density lipoprotein receptor mutation (C163Y) in the west of Scotland** W K Lee, L Haddad, M J Macleod, A M Dorrance, D J Wilson, D Gaffney, M H Dominiczak, C J D Packard, I N M Day, S E Humphries, A F Dominiczak 573

Syndrome of the month

- The fragile X syndrome** B B A de Vries, D J J Halley, B A Oostra, M F Niermeijer 579

Short reports

- Mutation analysis of the nerve specific promoter of the peripheral myelin protein 22 gene in CMT1 disease and HNPP** E Nelis, P De Jonghe, E De Vriendt, P I Patel, J J Martin, C Van Broeckhoven 590

- Linkage disequilibrium between the M470V variant and the IVS8 polyT alleles of the CFTR gene in CBAVD** A de Meeus, C Guittard, M Desgeorges, S Carles, J Demaille, M Claustres 594

- Acampomelic campomelic dysplasia with de novo 5q;17q reciprocal translocation and severe phenotype** R Savarirayan, A Bankier 597

- Mosaicism for a tandem duplication dup(1)(q12q22) in an 18 year old female** D de Silva, D Massie, J Drummond, D Couzin, J C S Dean 600

- Two cases of partial trisomy 8p and partial monosomy 21q in a family with a reciprocal translocation (8;21)(p21.1;q22.3)** A S Plomp, J J M Engelen, J C M Albrechts, C E M de Die-Smulders, A J H Hamers 604

- Extensive form of aplasia cutis congenita: a new syndrome?** M-S Park, S-H Hahn, C-H Hong, J-S Kim, H-S Kim 609

Letters to the Editor

- Holoprosencephaly in deletions of proximal chromosome 14q** K Devriendt, J-P Fryns, C-P Chen 612

- No evidence for heterogeneity in oculopharyngeal muscular dystrophy** W Kress, B Halliger-Keller, T Grimm, H Porschke, A Engelhardt, H-H Goebel, B Müller-Mysok 613

- Another holoprosencephaly locus at 7q21.2?** J-P Fryns 614

Book review

- Gene Therapy for Neurological Disorders and Brain Tumours** 615

- Notice to contributors** 616