

Contents

Review article

- Menkes disease: recent advances and new aspects** Z Tümer, N Horn 265

Original articles

- Global prevalence of putative haemochromatosis mutations** A T Merryweather-Clarke, J J Pointon, J D Shearman, K J H Robson 275

- The incidence of PAX6 mutation in patients with simple aniridia: an evaluation of mutation detection in 12 cases** R Axton, I Hanson, S Danes, G Sellar, V van Heyningen, J Prosser 279

- RDCI, the vasoactive intestinal peptide receptor: a candidate gene for the features of Albright hereditary osteodystrophy associated with deletion of 2q37** M M Power, R S James, J C K Barber, A M Fisher, P J Wood, B A Leatherdale, D E H Flanagan, E Hatchwell 287

- Detection of a novel mutation in the ryanodine receptor gene in an Irish malignant hyperthermia pedigree: correlation of the IVCT response with the affected and unaffected haplotypes** K E Keating, L Giblin, P J Lynch, K A Quane, M Lehane, J J A Heffron, T V McCarthy 291

- CFTR mutations and IVS8-5T variant in newborns with hypertrypsinaemia and normal sweat test** C Castellani, A Bonizzato, G Mastella 297

- Prenatal detection of fetal aneuploidies using transcervical cell samples** J Sherlock, A Halder, B Tutschek, J Delhanty, C Rodeck, M Adinolfi 302

- Fluorescent in situ hybridisation (FISH) for hemizygous deletion at the elastin locus in patients with isolated supraaortic stenosis** H Fryssira, R Palmer, K A Hallidie-Smith, J Taylor, D Donnai, W Reardon 306

- Detection of a de novo duplication of 1q32-qter by fluorescence in situ hybridisation in a boy with multiple malformations: further delineation of the trisomy 1q syndrome** H-C Duba, M Erdel, J Löffler, L Bereuther, H Fischer, B Utermann, G Utermann 309

- Molecular-cytogenetic detection of a deletion of 1p36.3** F Giraudeau, D Aubert, I Young, S Horsley, S Knight, L Kearney, G Vergnaud, J Flint 314

- Prader-Willi syndrome in a child with mosaic trisomy 15 and mosaic triplo-X: a molecular analysis** K Devriendt, G Matthijs, S Claes, E Legius, W Proesmans, J J Cassiman, J-P Fryns 318

Hypothesis

- The Y specific growth gene(s): how does it promote stature?** T Ogata, N Matsuo 323

Syndrome of the month

- Alport's syndrome** F Flinter 326

Short reports

- Impaired male sex development in an infant with molecularly defined partial 9p monosomy: implication for a testis forming gene(s) on 9p** T Ogata, K Muroya, N Matsuo, J Hata, Y Fukushima, Y Suzuki 331

- A 4 Mb cryptic deletion associated with inv(8)(q13.1q24.11) in a patient with trichorhinophalangeal syndrome type I** T Sasaki, H Tonoki, H Soejima, N Niikawa 335

- Proximal and distal spinal muscular atrophy in one family: molecular genetic studies provide further evidence for the non-allelic origin of both diseases** S Spranger, S Rudnik-Schöneborn, M Spranger, M Schächtele, K Zerres, B Wirth 340

- Physical localisation of the breakpoints of a constitutional translocation t(5;6)(q21;q21) in a child with bilateral Wilms' tumour** P R Hoban, R L Cowen, E L D Mitchell, D G Evans, M Kelly, P J Howard, J Highway 343

Conference report

- Report of the Fifth European Meeting on Psychosocial Aspects of Genetics** C Barnes, T Marteau, G Evers-Kiebooms 346

Medical genetics: advances in brief

- 349

Letters to the Editor

- FMR1 fully expanded mutation with minimal methylation in a high functioning fragile X male** D Z Loesch 350

- Reply** A K Taylor 350

- Raising the sensitivity of fetal RhD typing and sex determination from maternal blood** M Hengstschläger, G Hölzl, B Ulm, G Bernaschek 350

Book reviews

- 351

Notice to contributors

- 352