

Abstract 11

A NOVEL AUTOSOMAL RECESSIVE MR/MCA SYNDROME COMPRISING SHORT STATURE, BRACHYDACTYLY AND PECTUS EXCAVATUM MAPS TO 5q23-q32

Kimia Kahrizi¹, Hossein Najmabadi¹, Masoud Garshasbi^{1,2}, H.-Hilger Ropers²,
Andreas W. Kuss², Andreas Tzschach²

¹Genetics Research Center, University of Social Welfare and Rehabilitation Sciences, Tehran, Iran

²Max Planck Institute for Molecular Genetics, Berlin, Germany

We report on a family in which three mentally retarded siblings suffer from an apparently novel autosomal recessive MR/MCA syndrome characterized by borderline mental retardation, short stature, brachydactyly of the feet and pectus excavatum. The patients – two brothers and a sister – were born to healthy Iranian parents who were first cousins. They had one healthy brother who was of normal height. Metabolic tests and chromosome analysis revealed no abnormalities in the patients. The pedigree structure suggested autosomal recessive inheritance, and homozygosity mapping using a 250K SNP array (Affymetrix) identified one large (24 Mb) interval in 5q23.1-q32 flanked by SNP markers rs1427941 and rs10036966. The LOD score for this interval was 2.5. A second interval in 3q13.11 had a size of only 0.5 Mb (flanked by SNPs rs6791670 and rs2961221; LOD score 2.5), but contained no known genes. We therefore presume that the large and gene-rich interval in 5q harbours the causative gene defect in this family, and screening of candidate genes will eventually elucidate the underlying mutation.