

## Abstract 63

### A NOVEL AUTOSOMAL RECESSIVE SYNDROME OF MENTAL RETARDATION, KYPHOSCOLIOSIS AND HEART DEFECTS MAPS TO CHROMOSOME REGION 11q14-q23

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We report on three siblings – two brothers and a sister born to healthy consanguineous Iranian parents - with a novel mental retardation syndrome. All three patients suffered from moderate mental retardation, kyphoscoliosis which became apparent at the age of approximately 6 years, and heart defects. Body measurements were within the normal range, and metabolic tests, neurological investigations and chromosome analysis including array CGH revealed no abnormalities. The patients had three healthy siblings; their parents were first cousins. Assuming an autosomal recessive gene defect in this family, we performed genome-wide linkage analysis (homozygosity mapping) using a 10K SNP array (Affymetrix) and identified a single linkage interval on the long arm of chromosome 11. The homozygous interval was flanked by SNPs rs1391221 and rs1880206 and spans 28 Mb in 11q14.2-q23.2. The LOD score was 2.65. The similarity of the clinical features, the pedigree structure and the linkage data suggest that the patients in this family suffer from a novel autosomal recessive mental retardation/multiple congenital anomalies (MR/MCA) syndrome. Mutation analysis in candidate genes within the homozygous interval will eventually disclose the underlying genetic defect in this family.