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SNP Microarray Characterization and Genotype-Phenotype Analysis in a Patient with a Ring Chromosome 22

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ABSTRACT Ring chromosome 22 is a rare cytogenetic anomaly. The aim of this study was to present a case carrying ring chromosome 22 in a 9-year-old Chinese girl with long face, thick eyebrows, large and low-set ears, mental retardation, severe speech delay, autistic disorders and talipes equinovarus (TEV). A chromosome analysis of the proband revealed a de novo 46,XX,r(22)(p13;q13) karyotype and the deleted region was confirmed by means of SNP microarray analysis showing deletion range from 22q13.31 to 22q13.33 (3.8Mb). The present report describes the case of a patient with a ring chromosome 22 abnormality completely characterized by SNP microarray which provided additional information for genotype-phenotype studies.