



A microdeletion syndrome at 9q21.11-q21.31 characterised by mental retardation, speech delay, epilepsy

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We report a case with significant developmental delay, intellectual disability and epilepsy with a 11 Mb large one copy loss on the long arm of chromosome 9 encompassing the chromosomal region 9q21.11-q21.31 by the analysis of the whole exome sequencing. The patient was a girl, five years old with delayed speech and language development, global developmental delay, epilepsy, inability to walk. She had four other sisters all healthy. It was proceeded to deletion analysis of the TRPM6 gene by qPCR, which is included in the deletion region. The TRPM6 resulted deleted supporting the result. The whole exome sequencing analysis of the parents was performed and no clinically relevant variant has been detected. The deletion of the TRPM6 gene has not been detected in the patient's parents, supporting a de novo origin of the detected deletion. It was recommended proceeding to chromosomal microarray (CMA) analysis of the patient to confirm the detected deletion and to determine the localization of the breakpoints (extension of the deletion). To our knowledge, the genotype-phenotype correlations of sixteen cases with 9q21 deletion having different points and variable length revealed common characteristic features including severe developmental delay, epilepsy, neuro-behavioural disorders and facial dysmorphism. The smallest overlapping deleted region in all defined cases to date including our case comprised four genes. Among these deleted genes, especially RORB is considered to be a strong candidate for neurological phenotype.

Key words: 9q21 deletion, intellectual disability, developmental delay, epilepsy, delayed speech

Literature

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