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Coexistence of SMN1 deletion and duplication in unrelated cases

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Spinal muscular atrophy (SMA) is the second most common autosomal recessive genetic disorder. About 95% of cases are characterized by homozygous deletion of exon 7 in SMN1 gene. Here we report an unusual presentation of SMA in cases that was referred to Dr. Zeinali Medical Genetic Lab.

SMA molecular diagnosis for cases with clinical impression of SMA was performed using specific MLPA kit P021B1, MRC-Holland. DNA was extracted and qualified. MLPA was performed according to the standard protocol including hybridization, ligation and amplification. PCR product was separated using ABI-3130 genetic analyzer. Raw data were analyzed by Gene Marker V1.95.

Through analysis of those referred to our Lab, we found 3 unrelated families with a deletion in one parent and normal MLPA pattern in another parent (i.e. two copy of SMN1 gene). Paternity and/or maternity were tested and confirmed. Pedigree analysis showed several consanguinity marriages in previous generations. Further investigation of the family and pedigree analysis showed that normal parents have received both deletion and duplication from their parents. So they had 2 copies of SMN1 gene in one chromosome (duplication) and no copy in another one. This feature couldn't be identified from normal cases by MLPA technique.

In spite of high prevalence of deletion in SMN gene, co-existence of a duplication in a normal people with family history of an affected child should be considered. Further investigation of other family members is recommended.

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