ÇANAKKALE ONSEKIZ MART UNIVERSITY
IMPLEMENTATION AND RESEARCH HOSPITAL
MOLECULAR GENETICS LABORATORY

SPINAL MUSCULAR ATROPHY DNA Analysis Report

Ad-Soyadı	: MERVE TEKİN
Date of Birth	: 28.09.2018
Material	: Peripheric Blood-EDTA
Incoming Date	: 24.12.2018
Report Date	: 27.12.2018
Laboratory Code	: SMA- 2018- 075

No 02547
18 Şubat 2022

Dear My Colleague;
Your patient was examined by fragment analysis (P-060-B2) with multiplex PCR and capillary electrophoresis including the SMN 1 and SMN 2 genes in terms of mitochondrial deletion-duplications after DNA isolation from peripheral blood and, the following results were determined.

Result: Homozygous deletion was detected in the 7th and 8th exons of the SMN1 gene analyzed by MLPA in your patient and, SMN2 gene copy number (2 copies) was considered normal.

Comments: This finding is consistent with SMA. Family cooperation is recommended. Spinal muscular atrophy (SMA), is an autosomal recessive disease characterized by progressive degeneration of lower motor neuron and anterior horn cells. Homozygous deletions in the SMN1 gene are responsible for the etiology of the disease most frequently (95-98%), and point mutations are responsible for the second frequency (2-5%). SMN2 copy number has prognostic value in SMA patients.

Regards,

Prof. Dr. Fatma SILAN
Medical Geneticist

Prof. Dr. Öztürk ÖZDEMİR
Head of Department of Medical Genetics

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Tercüme edilmek üzere bana verilen Türkçe dilindeki işbu belgeyi İngilizce diline tam ve doğru olarak çevirdiğimi beyan ederim. ***Noter Yeminli Tercüman*** ***Mustafa DİNÇER***	*İşbu belgenin enin Noterliğimiz yeminli Tercümanı Mustafa DİNÇER tarafından Türkçeden İngilizceye tercüme edildiğini onaylarım.* *BEŞİKTAŞ 7. NOTERİ* *Veysi KAHRAMAN* *Yerine imzaya yetkili Başkatip* *AZİZE DÜNDAR AYTAÇ*