SMN1, SMN2 Deletion/Duplication Test Report

Personal Information	Specimen Information	Test Information
Name: Jane Doe	**Sample ID:** 20211102-971-0000	**Test reported:** 2021-12-01
Date of Birth: 2018-09-06	**Medical record No:**	**Ordering physician:** Dr.Smith
Sex: Female	**Date received:** 2021-11-02	**Institution:** Hospital A

TEST PERFORMED

SMN1, SMN2 GENE DOSAGE ANALYSIS REPORT

RESULT Detected

SMN1 Gene Deletion/Duplication: Detected

Gene	Exon	Copy
SMN1	7	0
SMN1	8	0
SMN2	7	3
SMN2	8	3

INTERPRETATION

The MLPA test was conducted to determine the deletion/duplication of the SMN1/SMN2 genes. The result showed a homozygous deletion of the SMN1 gene (SMN1 Gene: 0 Copy). Additionally, the SMN2 gene was confirmed to have 3 copies.

TEST INFORMATION

Specimen: Genomic DNA isolated from peripheral blood leukocytes
Target Disease: Spinal muscular atrophy
Analysed Gene: SMN1 on Chromosome 5q13.2
SMN2 on Chromosome 5q13.2
Mode of inheritance: Autosomal Recessive
Methods: MLPA (Multiplex Ligation-dependent Probe Amplification)

Tested by: M-K Lee M.T(20058) Confirmed by: Sae-Mi Lee M.D(1067) Chang-ahn Seol M.D(1037)

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COMMENTS

1. Spinal muscular atrophy is a group of genetic neuromuscular disorders that inherited in an autosomal recessive manner. Spinal muscular atrophy is characterized by muscle weakness and atrophy resulting from progressive degeneration and irreversible loss of the anterior horn cells in the spinal cord and the brain stem nuclei. Spinal muscular atrophy is caused by biallelic pathogenic variants in the SMN1 gene. 95% of the patients are homozygous for SMN1 deletion, and the remaining 5%are compound heterozygous for an intragenic pathogenic variant and an SMN1 deletion. The number of copies (dosage) of SMN2 is used to predict clinical severity.

2. This test identifies the deletion and duplication of exons 7-8 of SMN1 and SMN2 genes using MLPA (Multiplex Ligation-dependent Probe Amplification) method.

3. The limitations of this test are as following.
   1) If there is a nucleotide sequence mutation at the MLPA probe attachment site, a false-positive result may be shown.
   2) It is impossible to distinguish the normal individuals who have one SMN 1 gene in both alleles (1+1) normally and the

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