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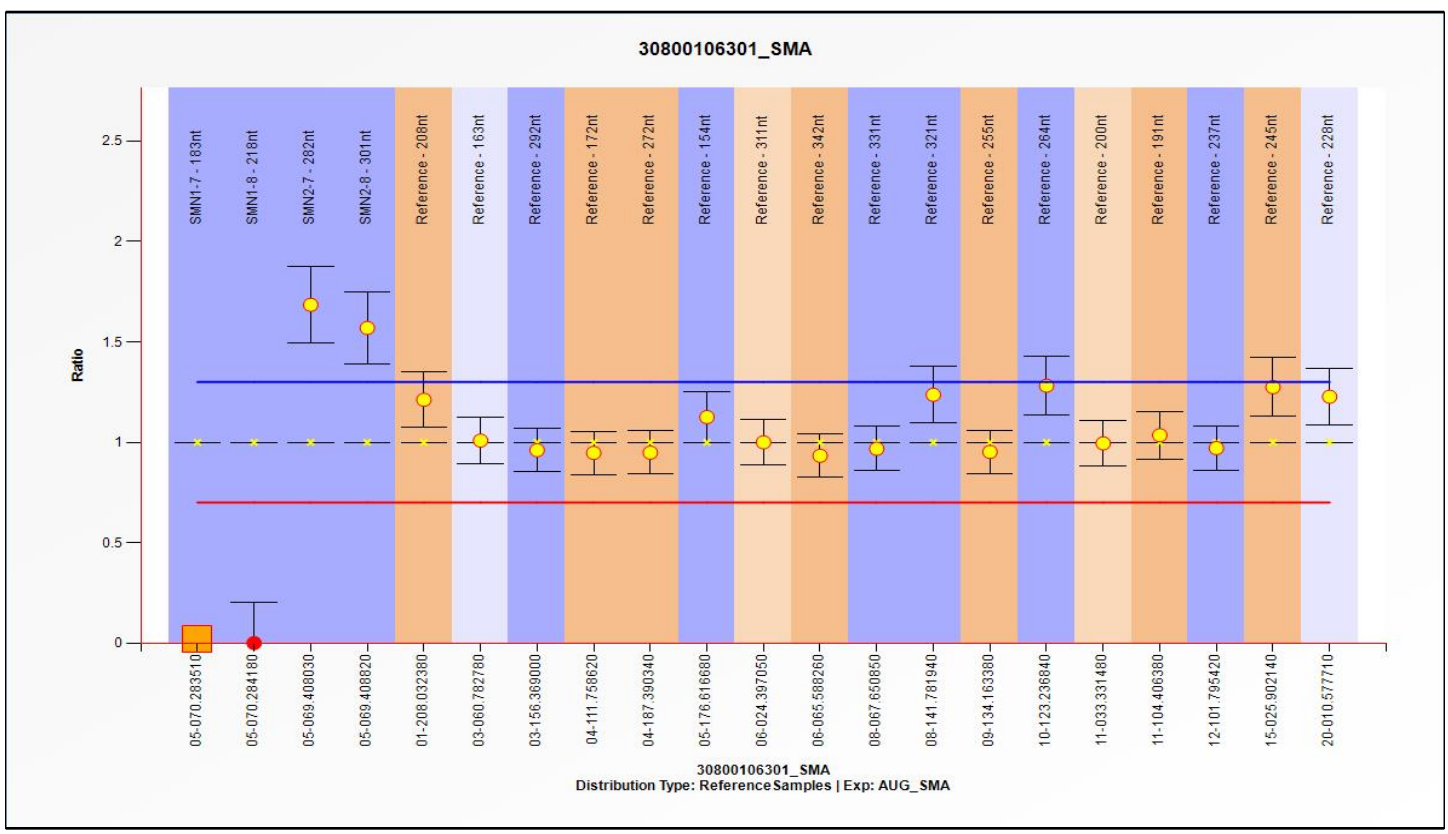
CIN - U85100TN2022PTC150858
support@mfine.co, www.mfine.co
Toll Free Number - 990 0599005

Name	: **Master. NIVANSH S (11189316)**	Lab ID	: KT30800106301	**CRM: 223001556272**
		Sample Collection Date	: 09-08-2023 16:00	
Age	: 7 Months	Sample Receipt Date	: 10-08-2023 12:45	
DOB	:	Reporting Date	: 17-08-2023 14:15	
Gender	: MALE	Location	: BANGALORE	
Referring Physician	: Dr. VINU			
Hospital Name	: Manipal Health Enterprises Private Limited			

Initial Report	☑	Duplicate Report	☐	Revised Report	☐	Version No	1

SPINAL MUSCULAR ATROPHY (SMA) REPORT RESULTS

Sample Type	:	Peripheral Venous Blood
Quality of Sample	:	Acceptable
Clinical Indication	:	Master. NIVANSH is suspected for SMA. Peripheral Venous Blood has been sent for MLPA testing to evaluate for SMA
Test Requested	:	Spinal Muscular Atrophy by MLPA



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Genes/Exon	Deletion/Duplication	Copy Number
SMN1 / EXON 7	Homozygous deletion	0
SMN1 / EXON 8	Homozygous deletion	0
SMN2 / EXON 7	Heterozygous duplication	3
SMN2 / EXON 8	Heterozygous duplication	3

Interpretation : Homozygous deletion in exon 7 and exon 8 of SMN1 gene in the clinical sample tested and this is indicative of SMA Affected. Heterozygous duplication detected in exon 7 and exon 8 of SMN2 gene.

NOTE:
1. Some individuals have two copies of SMN1 on just one chromosome and no copies of SMN1 on the second chromosome, this individual will be a carrier but this will not be detected by this test.

Recommendation : Correlate the results clinically and genetic counseling is recommended.

TABLE 1:

INTERPRETATION REFERENCE:

Copy Number Status	Dosage Quotient (DQ) Distribution
Normal	0.80 < DQ < 1.20
Homozygous deletion	DQ = 0
Homozygous duplication	1.75 < DQ < 2.15
Heterozygous deletion	0.40 < DQ < 0.65
Heterozygous duplication	1.30 < DQ < 1.65

TABLE 2:

INTERPRETATION FOR SMA CARRIERS:

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Finding	Conclusion	Explanation
No SMA symptoms SMN1 exon 7: 1 copy. SMN1 exon 8: 1 copy.	SMA carrier	One copy of SMN1 is absent, making the person a carrier. The absence of one copy of the SMN1 exon 8 sequence confirms this.
No SMA symptoms SMN1 exon 7: 1 copy. - A: SMN1 exon 8 copies > 1. - B: SMN1 exon 8 copies = 0	SMA carrier	One copy of SMN1 is absent, making the person a carrier. A: due to gene conversion, 1 (or more) copies of the characteristic SMN1 exon 8 have become incorporated in the SMN2 gene. B: an SMN2 exon 8 copy has replaced the characteristic SMN1 exon 8 copy.

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BACKGROUND

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Two nearly identical genes SMN1 and SMN2 located on 5q13 plays crucial role in SMA. Most individuals have two copies of each gene. SMN1 gene codes the survival of motor neuron protein (SMN) and plays a crucial role in survival of motor neurons. SMN2 is a complicated inverted repeat area displaying high instability, leading to frequent deletions and gene conversions. SMN1 and SMN2 can only be distinguished by two single nucleotide differences: one in exon 7 and one in exon 8. The single nucleotide difference in exon 7 of SMN2 affects mRNA splicing resulting in an alterec SMN protein with a limited half-life and function. The majority of SMA carriers can be identified by the presence of only one single SMN1 exon 7 copy.

METHODOLOGY

Multiplex Ligation-dependent probe amplification (MLPA) method is used for the detection of copy number changes of exons 7 and 8 of SMN1 and SMN2. Coffalyser software is used for data analysis. DNA isolated from the provided sample using commercial kit that works on silica-membrane-based DNA purification was subjected for MLPA analysis.

LIMITATIONS OF THE TEST

Cannot detect copy number neutral inversions, translocations and methylation changes. All possible causes of the syndromes included cannot be detected as the probemix has limited number of probes for each chromosomal region. The detection rate may vary between syndromes, depending on the heterogeneity of the disorder. Even when MLPA doesnot detect any aberrations, the possibility remains that biological changes in that gene or chromosomal region do exist but remain undetected. Sequence changes (e.g. SNPs, point mutations, small indels) in the target sequence or even when >20 nt from the probe ligation site detected by a probe can affect the results. This method cannot detect point mutation in the SMN1/SMN2 gene.

REFERENCES:

Alias L et al. (2014).Improving detection and genetic counseling in carriers of spinal muscular atrophy with two copies of the SMN1 gene.Clin Genet. 85:470-475.
Arkblad EL et al. (2006). Multiplex ligation-dependent probe amplification improves diagnostics in spinal muscular atrophy. NeuromusculDisord. 16:830-838.
Ben-Shachar S et al. (2011). Large-scale population screening for spinal muscular atrophy: clinical implications. Genet Med. 13:110-114

DISCLAIMER:

As per the PRE-NATAL DIAGNOSTIC TECHNIQUES (REGULATIONS & PREVENTION OF MISUSE) AMENDMENT ACT 2002, sex determination shall not be done for all prenatal samples.

DR.JAYAKRISHNA MSc. Ph.D

Senior Manager

DR. CHIRAYU PADHIAR, M.B.B.S..: DCP (G25442)

Lab Director

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