MLPA Test

Patient		Sample		Clinician	
Name		SampleType		Name	
Gender		Sample ID		Hospital	
Age		Date and Time of Sample Collection		Address	
Place		Date and Time of Sample Received		Indication	
Phone No./ Email ID		Date and Time of Report Released			

CLINICAL INDICATION

Ibrahim s/o Bhurhan was evaluated to rule out spinal muscular atrophy for SMN gene.

SAMPLE DESCRIPTION

Sample quality is optimum for the test.

RESULT

Homozygous deletion is detected in Exon 7 and Exon 8 of the SMN1 gene.

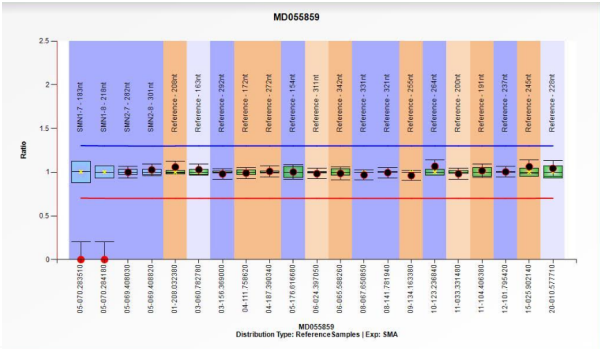
No deletion and No duplication detected in Exon 7 and Exon 8 of the SMN2 gene.

Sr. No.	Gene	Location	Deletion/Duplication	Final Ratio (FR)
1	*SMN1*	Exon7	Homozygous deletion	0
2	*SMN1*	Exon8	Homozygous deletion	0
3	*SMN2*	Exon7	No deletion and duplication	1
4	*SMN2*	Exon8	No deletion and duplication	1.03

INTERPRETATION

Homozygous deletion in exons 7 and exon 8 of SMN1 gene is detected in Ibrahim s/o Bhurhan. Sample from Ibrahim s/o Bhurhan was referred to our laboratory for molecular testing for Spinal Muscular Atrophy(SMA). Spinal Muscular Atrophy(SMA) is inherited in an autosomal recessive pattern that result from abnormalities of SMN protein.

Fig-1: 5254106/MD055859_ Ibrahim s/o Bhurhan_RATIO CHART: SMN1 and SMN2



Comment: The result must be interpreted in the context of the individual's clinical and biochemical profile.

Name : Date:

Interpretation Reference:	
Final Ratio [FR]	**Copy Number Status**
0.80 < FR< 1.20	**Normal**
FR = 0- < 0.10	Homozygous deletion
0.40 < FR <0.65	Heterozygous deletion
1.30 < FR <1.65	Heterozygous duplication
1.75 < FR <2.15	Heterozygous triplication/Homozygous duplication

Genetic counselling is advised.

Note: Smaller deletions, duplications and point mutation in the *SMN1* and *SMN2* genes are not detected by this technique. The coding regions of the *SMN1* and *SMN2* genes were examined. Changes further into the introns or in other non-coding regions of the gene would not be detected.

METHODOLOGY

Mutational analysis has been performed on genomic DNA by multiplex ligation probe dependent amplification (MLPA, MRC Holland) using SALSA MLPA probe mix P060-SMA Carrier kit for *SMN1 and SMN2* genes (NM_004014). Analysis was done by Coffalyser (designed by MRC - Holland).

DISCLAIMER

- Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder, with an incidence of approximately 1 in 10,000 births. The condition has variable severity and age of onset, and has been categorized into clinical types 0-IV. SMA I accounts for 60% of all SMA and has onset of symptoms in infancy.
- MLPA assay for the molecular diagnosis of SMA is based on a kit containing several probes for the SMA critical region, including specific probes for *SMN1* and *SMN2* genes, probes able to hybridize both genes and other probes for sequences mapped either within the SMA critical region or on other autosomal regions.
- A point mutation or polymorphism in the sequence detected by a probe, which results in reduced probe binding efficiency, can also cause a reduction in relative peak area. Therefore, single exon deletions detected by MLPA should always be confirmed by other methods like multiplex PCR or sequencing.
- MLPA cannot detect any changes that lie outside the target sequence of the probes and will not detect most inversions or translocations. Even when MLPA did not detect any aberrations, the possibility remains that biological changes in that gene or chromosomal region do exist but remain undetected.

REFERENCES

1. SchoutenJPetal.(2002) Relativequantificationof40nucleic acidsequences by multiplex ligation-dependent probe amplificationNucleic Acids Res 30, e57.
2. Redeker,E.J.,et al. (2008).Multiplex ligation-dependent probe amplification(MLPA) enhances the molecular diagnosisofaniridia and related disorders Mol Vis 14, 836-840.
3. 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.

#this test is not under NABL scope

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