# M L P A Test

| Patient | | Sample | | Clinician | |
|---|---|---|---|---|---|
| Name | | SampleType | | Name | |
| Gender | | Sample ID/ Specimen ID | | Hospital | |
| DOB/Age | | Date and Time of Sample Collection | | Address | |
| Place | | Date and Time of Sample Received | | Indication | |
| Phone No. / EmailID | | Date and Time of Sample Reported | | | |

## CLINICAL DIAGNOSIS/SYMPTOMS

Akilesh advised to get tested for DMD by MLPA. Child brought with complaints of difficulty in getting up from sitting position, noted since 05 years associated with difficulty in climbing stairs at started walking with abnormal difficulty walking past 1and 1/5 years also difficulty in getting up from supine position. others: Calf hypertrophy, Valley sign, exaggerated lumbar lordosis, gowers sign waddling gait and QP (proximal>distal).

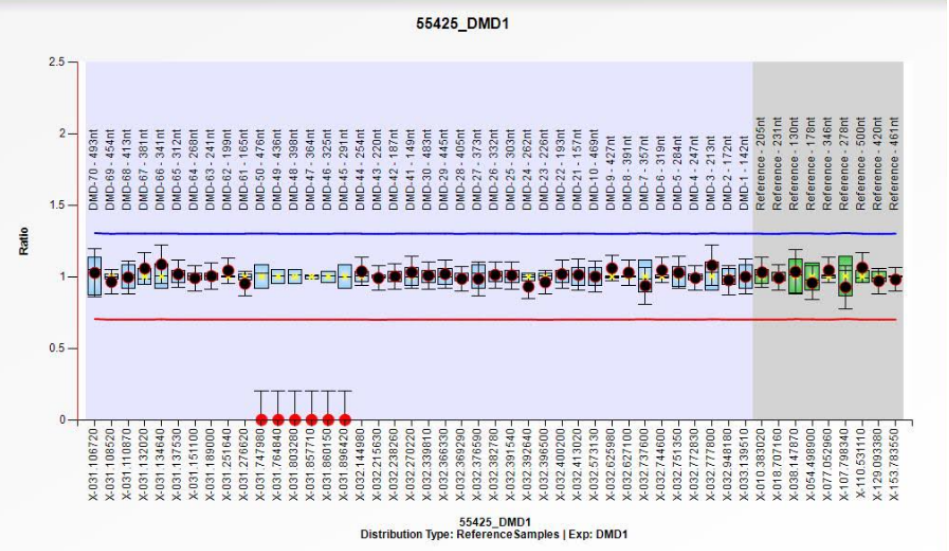
## RESULT

**Hemizygous deletion is detected in Exons 45, 46, 47, 48, 49, 50, 51, 52, 53 and 54 in DMD gene.**

## INTERPRETATION

Hemizygous deletion is detected in Exons 45, 46, 47, 48, 49, 50, 51, 52, 53 and 54 in the DMD gene in Akilesh. Sample from Akilesh was referred to our laboratory for molecular testing for Duchenne muscular dystrophy. Duchenne muscular dystrophy(DMD) is an X-linked muscular disease that result from abnormalities of the dystrophin protein.

Fig-1: ID:5133001/MD055425_Akilesh_RATIO CHART: DMDprobemix1



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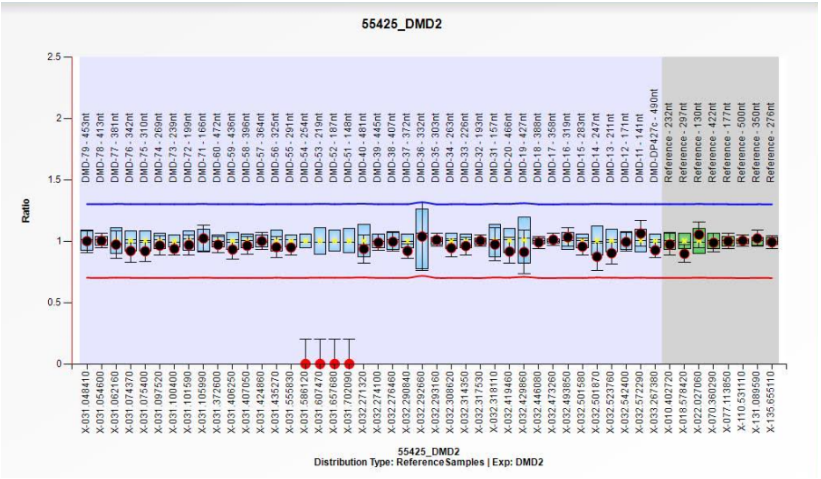
Name: ID:! Date

Fig-2: ID:5133001/MD055425_Akilesh_RATIO CHART: DMDprobemix2




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

**Genetic counselling is advised.**

Note: Smaller deletions, duplications and point mutation in the *DMD* gene are not detected by this technique. Only the coding regions of the *DMD* gene 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 P034-DMD-1 and P035-*DMD-2* kit for *DMD* gene (NM_004014). Analysis was done by Coffalyser (designed by MRC-Holland).

## DISCLAIMER

- The MLPA test will not detect the point mutations in the *DMD* gene, which are the second most common cause of genetic defects in the *DMD* gene. It is therefore recommended to use MLPA in combination with sequence analysis.
- 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 change in that gene or chromosomal region do exist but remain undetected.

#this test is not under NABL scope.

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