M L P A Test

Patient		Sample		Clinician	
Name	Dummy	**SampleType**	Blood	**Name**	NA
Gender	NA	**Sample ID/ Specimen ID**	NA	**Hospital**	NA
DOB/Age	DD-MM-YYYY	**Date and Time of Sample Collection**	DD-MM-YYYY	**Address**	NA
Place	NA	**Date and Time of Sample Received**	DD-MM-YYYY	**Indication**	XYZ advised to get tested for DMD by MLPA.
Phone No. / EmailID	NA	**Date and Time of Sample Reported**	DD-MM-YYYY		

CLINICAL DIAGNOSIS/SYMPTOMS

NA

RESULT

Hemizygous deletion is detected in Exon 45 in DMD gene.

INTERPRETATION

Hemizygous deletion is detected in Exon 45 in the DMD gene in Dummy. Sample from Dummy 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:00000000_ DUMMY_RATIO CHART: DMD probemix1

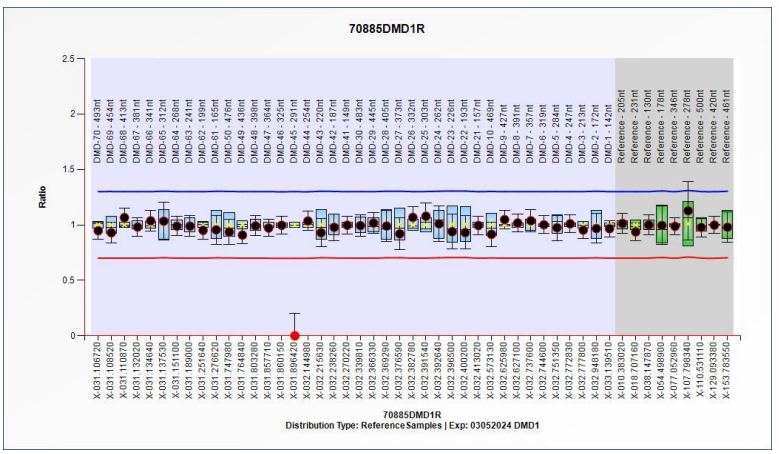

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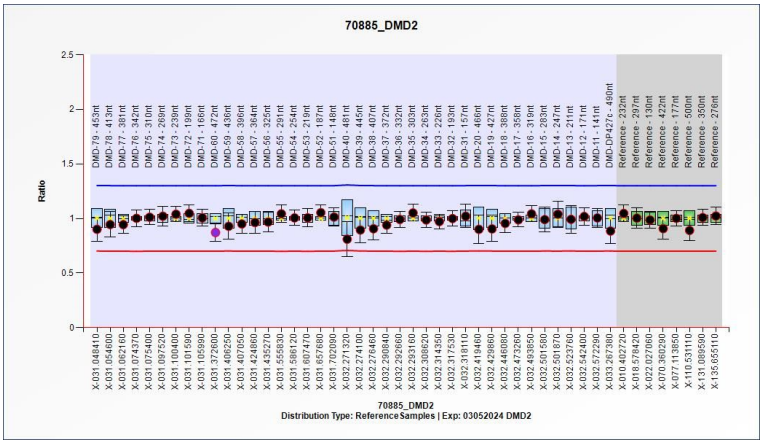
Fig-2: ID:000000000_ DUMMY_RATIO CHART: DMD probemix2



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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Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.