# 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** | NA | **Date and Time of Sample Collection** | DD-MM-YYYY | **Address** | NA |
| **Place** | NA | **Date and Time of Sample Received** | DD-MM-YYYY | **Indication** | Dummy advised to get tested for F7, F8 and F9 genes by MLPA. |
| **Phone No. / EmailID** | NA | **Date and Time of Sample Reported** | DD-MM-YYYY | | |

**TEST REQUESTED: Deletion and duplication analysis of VII deficiency, *F8* in hemophilia A, and *F9* in hemophilia B by MLPA.**

## CLINICAL DIAGNOSIS/SYMPTOMS

NA

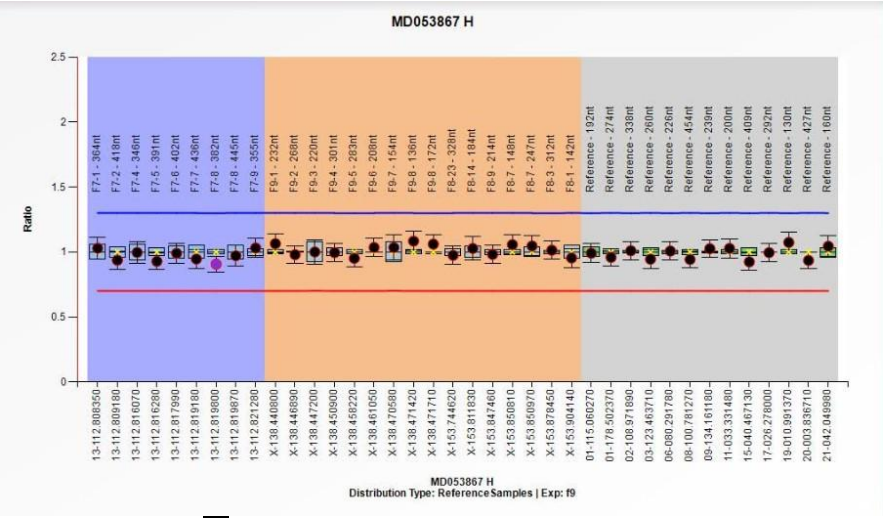
## RESULT

**No deletion and duplication is detected in exons of *F7*, *F8* and *F9* genes, encoding coagulation factors factor VII, factor VIII and factor IX, respectively.**

## INTERPRETATION

No deletion and duplication is detected in exons of *F7, F8* and *F9* genes, encoding coagulation factors factor VII, factor VIII and factor IX, respectively. Sample from Niranjan Singh was referred to our laboratory for molecular testing for Factor VII deficiency, Hemophilia A and Hemophilia B. It is an X-linked disease that results from abnormalities of the coagulation factors factor VII, factor VIII and factor IX.

**Fig-1: ID:DUMMY_RATIO CHART: F7, F8 and F9 genes Probemix**




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



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LABORATORY REPORT

Name: DUMMY ID:NA Date:DD-MM-YYY

## RECOMMENDATION

**Genetic counselling is advised.**

## TEST BACKGROUND

Coagulation factors play an important role in bleeding control and vessel repair. Deficiency in any of these genes may results in a bleeding disorders with different clinical manifestation: F7 is involved in factor VII deficiency, F8 in hemophilia A, and F9 in hemophilia B. Both hemophilia A and B are inherited in an X-linked manner, while factor VII deficiency is inherited in an autosomal recessive fashion.

## METHODOLOGY

Mutational analysis has been performed on genomic DNA by multiplex ligation probe dependent amplification (MLPA, MRC Holland) using SALSA MLPA probe mix P207 F9 kit for *F7* (NM_000131.4), *F8* (NM_000132.4) and *F9* (NM_000133.4) genes. Analysis was done by Coffalyser (designed by MRC-Holland).

## INTERPRETATION GUIDELINES

| Copy number status | | Final ratio (FR) |
|---|---|---|
| **Autosomal sequences and X chromosome sequences in females** | **X chromosome sequences in males** | |
| Normal | Normal | 0.80 < FR < 1.20 |
| Homozygous deletion | Deletion | FR=0 |
| Heterozygous deletion | | 0.40 < FR < 0.65 |
| Heterozygous duplication | | 1.30 < FR < 1.65 |
| Heterozygous triplication/homozygous duplication | Duplication | 1.75 < FR < 2.15 |
| Ambiguous copy number | | All other values |

## DISCLAIMER

- Smaller deletions, duplications and point mutation in the F7, F8 and F9 genes are not detected by this technique. Only the coding regions of the F7, F8 and F9 genes were examined. Changes further into the introns or in other non-coding regions of the gene would not be detected. 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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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

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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.**

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