

# SANGER SEQUENCING ANALYSIS

Patient ID	NA	Gender	NA	Location	XYZ
Patient Name	DUMMY	Clinician Name	NA	Sample Collected	DD-MM-YYYY
DOB	NA	GA/LMP Date	NA	Sample Received	DD-MM-YYYY
Age	NA	Hospital Name	NA	Report Released	DD-MM-YYYY

Test Requested:- Sanger Sequencing	Sample Type:- Blood	Sample Quality:- Acceptable
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## RESULT >>

Fig No.	Sample Name	Gene Name	Variant Reported in the Index Patient	Variant Status	Inheritance
1.	DUMMY	LTBP2	c.1756dup (p.Val586GlyfsTer17)	Homozygous	Autosomal Recessive

## TEST INFORMATION >>

This assay test for the confirmation of variant in the DUMMY which has been detected in LTBP2 gene in her WES. Analysis is performed only for variant at c.1756dup (p.Val586GlyfsTer17) in LTBP2 gene.

**RECOMMENDATION >>** Please correlate clinically and genetic counselling is recommended.

## TECHNOLOGY >>

Targeted sequencing and mutation analysis was performed by Polymerase Chain Reaction (PCR) followed by automated DNA sequencing of the amplicon using BigDye ABI Genetic Analyzer 3500XL platform. The raw data obtained is subsequently analyzed for the nucleotide variants

## DISCLAIMER >>

This test is designed to detect mutations in the above-mentioned regions only. Sequences surrounding the regions of interest are analysed but not reported. In rare cases because of allele dropout, heterozygosity may be reported as homozygosity. This assay is unable to differentiate between cis and trans mutations. Though oligos are designed specifically to parent gene using bioinformatics tool, Interference of pseudogene sequence cannot be ruled out completely. Any change in primer binding site can result and interfere with the results and allele dropout cannot be ruled out using this experiment.

ANNEXURE >>

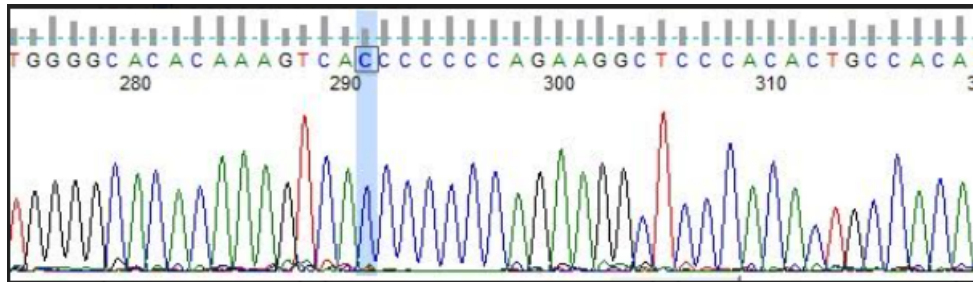


Fig 1: Sanger sequencing data (electropherogram) for the provided sample showing nucleotide change at c.1756dup (p.Val586GlyfsTer17) in LTBP2 gene.



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.

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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

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