

# SANGER SEQUENCING ANALYSIS

<b>Patient ID</b>		<b>Gender</b>	Female	<b>Location</b>	
<b>Patient Name</b>		<b>Clinician Name</b>		<b>Sample Collected</b>	12-11-2024
<b>DOB</b>	NA	<b>GA/LMP Date</b>		<b>Sample Received</b>	13-11-2024
<b>Age</b>	27 Years	<b>Hospital Name</b>		<b>Report Released</b>	29-11-2024

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

Fig No.	Patient Name	Gene Name	Variant Reported in the Index Patient	Variant Status	Inheritance
1	Mrs. Dummy	LRBA	c.5149G>A (p.Val1717Met)	No variant identified	Autosomal Recessive
2		LRBA	c.2540T>C (p.Ile847Thr)	Heterozygous	Autosomal Recessive
3		PRSS1	c.47C>T (p.Ala16Val)	No variant identified	Autosomal Dominant
4		PRSS1	c.86A>T (p.Asn29Ile)	Heterozygous	Autosomal Dominant

## TEST INFORMATION

This assay tests for the confirmation of variant in Mrs. Seenath which has been detected in LRBA and PRSS1 gene in Fathima Hayza. Analysis is performed only for variant at c.5149G>A (p.Val1717Met), c.2540T>C (p.Ile847Thr) in LRBA gene and c.47C>T (p.Ala16Val), c.86A>T (p.Asn29Ile) in PRSS1 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 >>

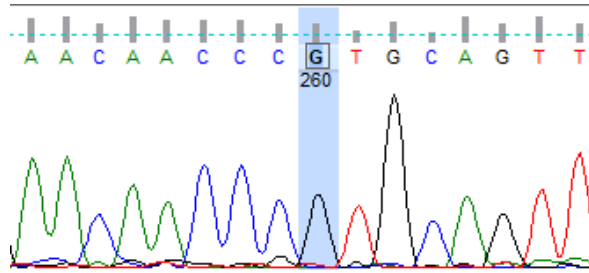


Fig 1: Sanger sequencing data (electropherogram) for the provided sample showing no nucleotide change at c.5149G>A (p.Val1717Met) in LRBA gene

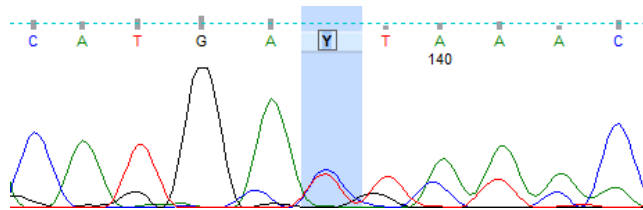


Fig 2: Sanger sequencing data (electropherogram) for the provided sample showing nucleotide change at c.2540T>C (p.Ile847Thr) in LRBA gene

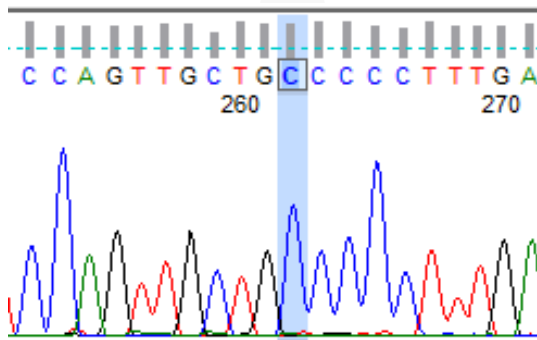


Fig 3: Sanger sequencing data (electropherogram) for the provided sample showing no nucleotide change at c.47C>T (p.Ala16Val) in PRSS1 gene.

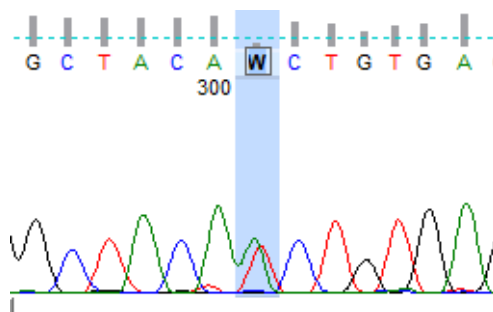


Fig 4: Sanger sequencing data (electropherogram) for the provided sample showing nucleotide change c.86A>T (p.Asn29Ile) in PRSS1 gene.

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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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