

SANGER SEQUENCING ANALYSIS

DOB	NA	GA/LMP Date	NA	Sample Received	07-02-2024
Age	31 Years	Hospital Name	PGIMER	Report Released	15-02-2024

Test Requested:- Sanger Sequencing	Sample Type:- Amniotic Fluid	Sample Quality:- Acceptable
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RESULT

Fig No.	Sample Name	Gene Name	Variant Reported in the Index Patient	Variant Status	Inheritance
1	AF of	DYNC2L1	c.3133G>A (p.Asp1045Asn)	Heterozygous	Autosomal Recessive
2		POP1	c.2057+2T>C (p.?)	No variant Identified	Autosomal Recessive
3		KIAA0753	c.328C>T (p.Arg110Ter)	Heterozygous	Autosomal Recessive

TEST INFORMATION

This assay tests for the confirmation of variant in the AF of Sudesh Kaur which has been detected in DYNC2L1, POP1 and KIAA0753 gene in parents carrier screening report. Analysis is performed only for variant at c.3133G>A (p.Asp1045Asn) in DYNC2L1 gene, c.2057+2T>C (p.?) in POP1 gene and c.328C>T (p.Arg110Ter) in KIAA0753 gene.

Note: No significant maternal cell contamination is detected in AF of Sudesh Kaur. Maternal cell contamination (MCC) was ruled out using STR markers

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

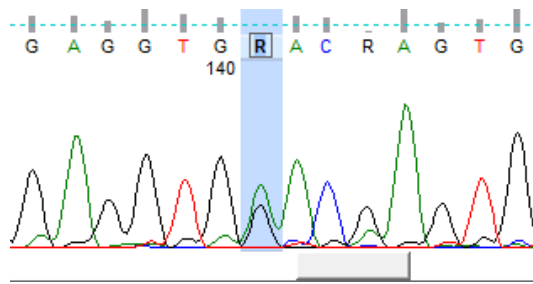


Fig 1: Sanger sequencing data (electropherogram) for the provided sample showing nucleotide change at c.3133G>A (p.Asp1045Asn) in DYNC2L1 gene

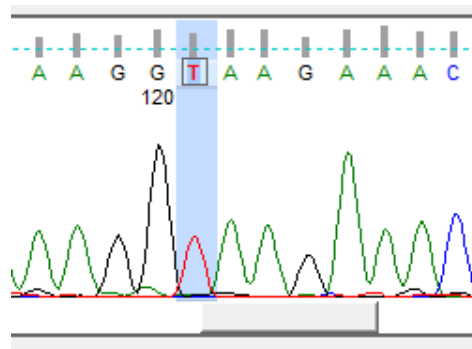


Fig 2: Sanger sequencing data (electropherogram) for the provided sample showing no nucleotide change at c.2057+2T>C (p.?) in POP1 gene.

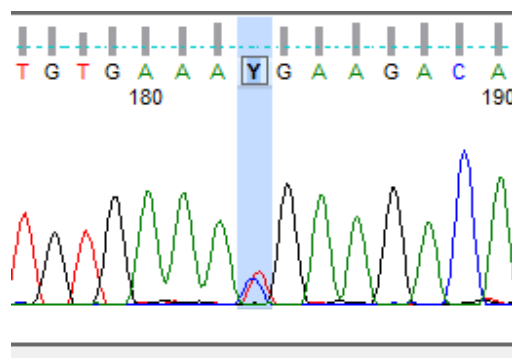


Fig 3: Sanger sequencing data (electropherogram) for the provided sample showing nucleotide change at c.328C>T (p.Arg110Ter) in KIAA0753 gene.

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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: 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.