

SANGER SEQUENCING ANALYSIS

Patient ID	NA	Gender	NA	Location	NA
Patient Name	DUMMY	Clinician Name	NA	Sample Collected	DDMMYYYY
DOB	NA	GA/LMP Date	NA	Sample Received	DDMMYYYY
Age	NA	Hospital Name	NA	Report Released	DDMMYYYY

Test Requested:- Glucose 6 Phosphate Ddehydrogenase (G6PD) Gene Mutation	Sample Type:- NA	Sample Quality:- Acceptable
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RESULT

Fig No.	Patient Name	Gene Name	Variant Reported in the Index Patient	Variant Status	Inheritance
1	Dummy	G6PD	c.131C>G (p.Ala44Gly)	No variant identified	X-linked
2		G6PD	c.563C>T (p.Ser188Phe)	No variant identified	X-linked
3		G6PD	c.949G>A (p.Glu317Lys)	No variant identified	X-linked

TEST INFORMATION

This assay test for the confirmation of variant in Dummy for G6PD gene. Analysis is performed only for variant at c.131C>G (p.Ala44Gly), c.563C>T (p.Ser188Phe) and c.949G>A (p.Glu317Lys) in G6PD 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.

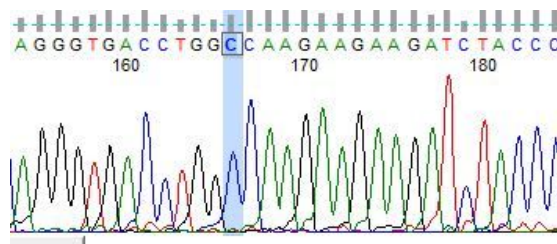


Fig 1: Sanger sequencing data (electropherogram) for the provided sample no showing nucleotide change at c.131C>G (p.Ala44Gly) in G6PD gene.

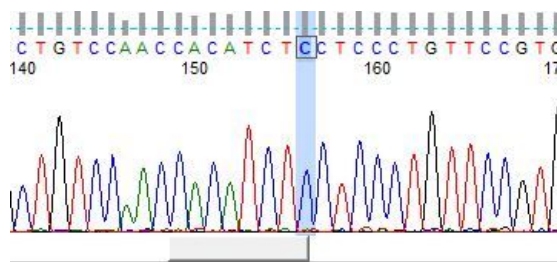


Fig 2: Sanger sequencing data (electropherogram) for the provided sample showing no nucleotide change at c.563C>T (p.Ser188Phe) in G6PD gene.

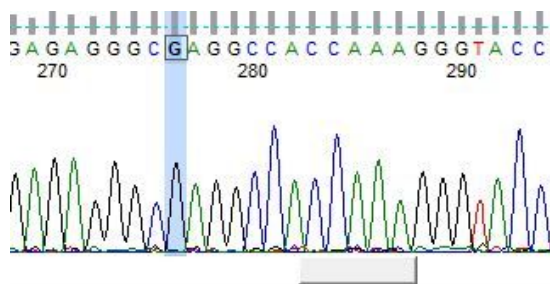


Fig 3: Sanger sequencing data (electropherogram) for the provided sample showing no nucleotide change at c.949G>A (p.Glu317Lys) in G6PD gene.

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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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