Laboratory Investigation Report

Patient Name	: Mrs. Nitu Bakshi	Centre	: 1103 - Max Hospital Saket(East Block)
Age/Gender	: 64 Y 11 M 27 D /Female	OP/IP No/UHID	: SKCS4524218
MaxID/Lab ID	: SKDD.1052779/0832072408848	Collection Date/Time	: Jul 09, 2024, 07:23 PM
Ref By	: Dr.Bhuvan Chugh	Reporting Date/Time	: Jul 12, 2024, 09:22 AM

SIN No:DD0536521

TEST REQUESTED

DPD GENE MUTATIONS (5-Floro Uracil Toxicity)

METHOD USED

Sanger sequencing

RESULT

Mutation Not Detected

Interpretation	
Wild Type	An individual carrying two normal function alleles. Based on genotype, there is no indication to change dose or therapy
Heterozygous	An individual carrying one normal function allele plus one no function allele or one decreased function allele, or an individual carrying two-decreased function alleles. Reduce starting dose followed by titration of dose based on toxicity or therapeutic drug monitoring (if available)
Homozygous	An individual carrying two no function alleles or an individual carrying one no function plus one decreased function allele.

NOTE

1. This is an in-house developed assay and detects c.[2846A>T], c.[1905+1G>A] and c.[1679T>G]. Other mutations are not detected by this assay.
2. Clinical significance of the variant if any need to be evaluated by clinician in context with clinical findings, family history and other relevant laboratory data.
3. Genetic Counselling is recommended.
4. Presence of PCR inhibitors if any, might lead to amplification failure.
5. Indeterminate / Not detected results do not rule out the presence of mutations below the detection limit of detection of the assay.
6. Test conducted on Whole blood.
7. The method used is Sanger sequencing.

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COMMENTS

DPYD, the gene encoding dihydropyrimidine dehydrogenase (DPD), the rate-limiting enzyme for fluoropyrimidine catabolism. Numerous genetic variants in DPYD are known that alter the protein sequence or mRNA splicing. In the context of 5-fluorouracil (anticancer drug), decreased function DPYD variants are of primary relevance due to their population frequency and established impact on enzyme function and toxicity risk: c.190511G>A (rs3918290, also known as DPYD*2A, DPYD:IVS14 1 1G>A), c.1679T>G (rs55886062, DPYD *13, p.I560S), c.2846A>T (rs67376798, p.D949V). Of these variants, c.190511G>A and c.1679T>G have the most deleterious impact on DPD activity, whereas c.2846A>T result in moderately reduced DPD activity. The presence of decreased or no function variants does not always result in toxicity. Overall, 50% of decreased function DPYD variant carriers develop severe 5-fluorouracil-related toxicity with standard doses. At the same time, patients without a DPYD decreased/no function variant may still experience severe toxicity due to other genetic, environmental, or other factors.

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