

**GENETIC TEST:****Dihydropyrimidine dehydrogenase deficiency/5-fluorouracil toxicity - Pharmacogenetics  
(4 variants: DPYD\*2A, DPYD\*13, c.2846A>T, HapB3)**

<b>FULL NAME:</b>	Dihydropyrimidine dehydrogenase deficiency/5-fluorouracil toxicity - Pharmacogenetics (4 variants: DPYD*2A, DPYD*13, c.2846A>T, HapB3)
<b>DESCRIPTION:</b>	Using Sanger sequencing and/or SALSA MLPA, the presence of the following four variants in the DPYD gene is investigated: - c.1236G>A (rs56038477) or c.1129-5923C>G (rs75017182) - c.1905+1G>A (rs3918290) - c.1679T>G (rs55886062) - c.2846A>T (rs67376798) The c.1236 G>A variant is in linkage disequilibrium with the c.1129-5923C>G variant.
<b>TEST TYPE:</b>	Clinical
<b>TEST SPECIALTY:</b>	Molecular Genetics
<b>TEST PURPOSE:</b>	Therapeutic Management
<b>SPECIMEN:</b>	Peripheral (whole) blood on EDTA
<b>METHOD CATEGORY:</b>	Targeted variant analysis
<b>METHOD TECHNIQUE:</b>	Bi-directional Sanger Sequence analysis MLPA based techniques
<b>RIZIV CODE:</b>	565390-565401

<b>ACCREDITATION (ISO 15189):</b>	2021-07-08 / 2026-02-02
<b>EQA:</b>	<ul style="list-style-type: none"><li>• Dihydropyrimidine dehydrogenase deficiency,</li><li>• Dihydropyrimidine dehydrogenase deficiency</li></ul>
<b>TURNAROUND TIME (MAXIMUM):</b>	7 calendar days
<b>CREATED:</b>	23 Dec 2019 - 12:24
<b>CHANGED:</b>	01 Mar 2023 - 14:47
<b>URL:</b>	<a href="https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/12619">https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/12619</a>

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- dihydropyrimidine dehydrogenase

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