DPYD Genotyping

Molecular analysis of variants associated with DPD enzyme activity

THE DPYD GENE

The *DPYD* gene encodes the dihydropyrimidine dehydrogenase (DPD) enzyme, which plays a key role in pyrimidine metabolism. Genetic variants may lead to reduced or absent enzymatic activity, with consequent effects on the patient's metabolic profile.

THE TEST

The kit enables targeted detection of selected *DPYD* gene variants by Real-time PCR. A reliable solution for genetic characterization and support of specialized diagnostic pathways.

CONGENITAL DPD DEFICIENCY

Congenital DPD deficiency is a pyrimidine metabolism disorder associated with reduced or absent enzymatic activity.

Patients may present with:

- neonatal or early-onset seizures
- global developmental delay / intellectual disability
- hypotonia
- microcephaly

DPYD gene analysis supports genetic diagnosis and subsequent specialized metabolic and clinical assessment.

FLUOROPYRIMIDINE

DPYD and fluoropyrimidine metabolism

The DPD enzyme represents the rate-limiting step in fluoropyrimidine catabolism (5-FU, capecitabine).
Variants in the *DPYD* gene are associated with reduced DPD enzymatic activity and consequently altered metabolic capacity for these compounds.
Reduced enzymatic activity has been associated with an increased risk of toxicity during fluoropyrimidine treatment.

The role of *DPYD* is widely recognized in fluoropyrimidine pharmacogenetics.

MAIN FEATURES

- Targeted detection of DPYD gene variants
- Multiplex Real-time PCR approach
- Quality and compliance according to IVDR (EU) 2017/746

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IVD *In Vitro* Diagnostic Medical Device
IVDR (EU) 2017/746

DPYD (5 gene mutations)

(REF: RT-104)

Detection of *DPYD* gene variants by Multiplex Real-time PCR

TARGETS ANALYZED

Detection of 5 *DPYD* gene polymorphisms. The test uses 3 reaction mixes for identification of the following polymorphisms:

- **c.1905+1G>A**
- **c.1129-5923C>G**
- **c.2846A>T**
- **c.1679T>G**
- **c.2194G>A**

TECHNOLOGY AND WORKFLOW

- Multiplex qPCR amplification assay performed on DNA extracted from EDTA peripheral whole blood samples
- Allele-specific technology
- 3 optimized reaction mixes
- *Wild-type* and mutated positive controls included

SAMPLE AND PREPARATION

- DNA extracted from EDTA peripheral whole blood samples
- **Manual extraction:** QIAamp DNA Mini Kit (QIAGEN)
- **Automated extraction:** CloNext 12 or CloNext 24 (Ref. OP01018 / OP01019)

INSTRUMENTATION

The kit has been validated on the following instruments:

- Applied Biosystems 7500 Fast (Thermo Fisher Scientific)
- Rotor-Gene Q MDx (QIAGEN)
- CFX96 Real-Time PCR Detection System (Bio-Rad)
- ELITe InGenius® System and ELITe BeGenius® (ELITechGroup S.p.A.)

OPERATIONAL FEATURES

- Total assay time: ~1 hour 40 minutes
- Format: 24 tests
- Storage and transport: -20 °C

VARIANT SELECTION

The variants included in the panel have been selected based on their relevance reported in scientific literature and major international reference sources.

INFORMATION

DESCRIPTION	CODE	REGULATORY STATUS	FORMAT
DPYD (5 gene mutations)	RT-104	CE IVD 0123	24 tests

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