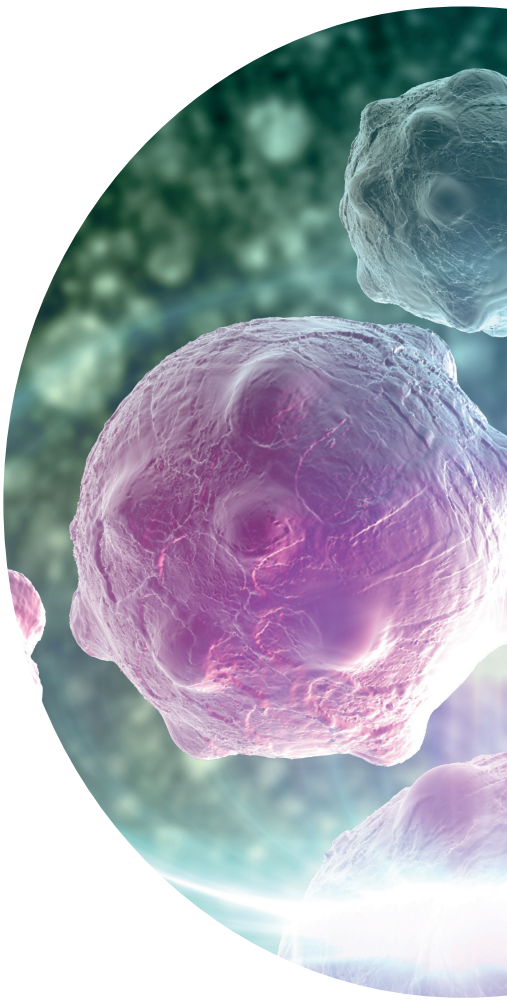
Yourgene® DPYD

DPYD Genotyping

5-fluorouracil (5-FU) is a chemotherapy agent used to treat a range of cancers including colorectal, head and neck, breast, pancreatic and stomach cancer. 5-FU is metabolized by the dihydropyrimidine dehydrogenase enzyme (DPD) which is encoded by the *DPYD* gene.
Several variants within *DPYD* have been described that lead to reduced or abolished DPD activity. Patients with these variants are at an increased risk of severe or fatal 5-FU toxicity. Therefore, implementation of DPD deficiency screening by genotyping will allow a more accurate prediction of toxicity and chemotherapeutic response.

Allelic Variant	rsID	Nucleotide Change	Protein Change	Allele Function
*2A	rs3918290	c.1905+1G>A	N/A	No Function
*13	rs55886062	c.1679T>G	p.I560S	No Function
N/A	rs67376798	c.2846A>T	p.D949V	Decreased
HapB3	rs75017182 rs56038477 rs56276561	c.1129-5923C>G c.1236G>A c.483+18G>A	N/A p.E412E N/A	Decreased Decreased Decreased



Key Benefits

Two tube assay:

- Tube 1 detects mutant sequences for the 6 SNPs in addition to the
- wildtype sequence for the *2A allele to allow determination of zygosity.
- Tube 2 detects the wildtype sequences for the 5 remaining wildtype alleles.
- STR Markers are included in each tube to aid sample identification.

6 Clinically Relevant Mutations inline with CPIC Guidelines

Ready to use reagents:

- Simply add DNA to the PCR reaction mix.

Rapid Turnaround Time:

- Minimal hands on time – 1 day turnaround:
- PCR: ~3 hours.
- Capillary Electrophoresis: ~1 hour.

CE-IVD ensures reliability

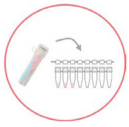
- Reliable and Accurate Detection

Simple data interpretation:

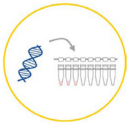
- Software analysis files for GeneMapper™ and GeneMarker® are provided by Yourgene Health.

- The CPIC guideline provides supplementary information to aid the clinical interpretation of *DPYD* allelic variants, with additional information regarding fluoropyrimidine dose.

Yourgene® DPYD Protocol Overview



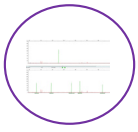
1. Reaction Mix
Prepare and dispense the *DPYD* reaction mixes into 0.2ml PCR tubes or a 96 well plate.



2. DNA
Add patient DNA to the dispensed *DPYD* reaction mixes.

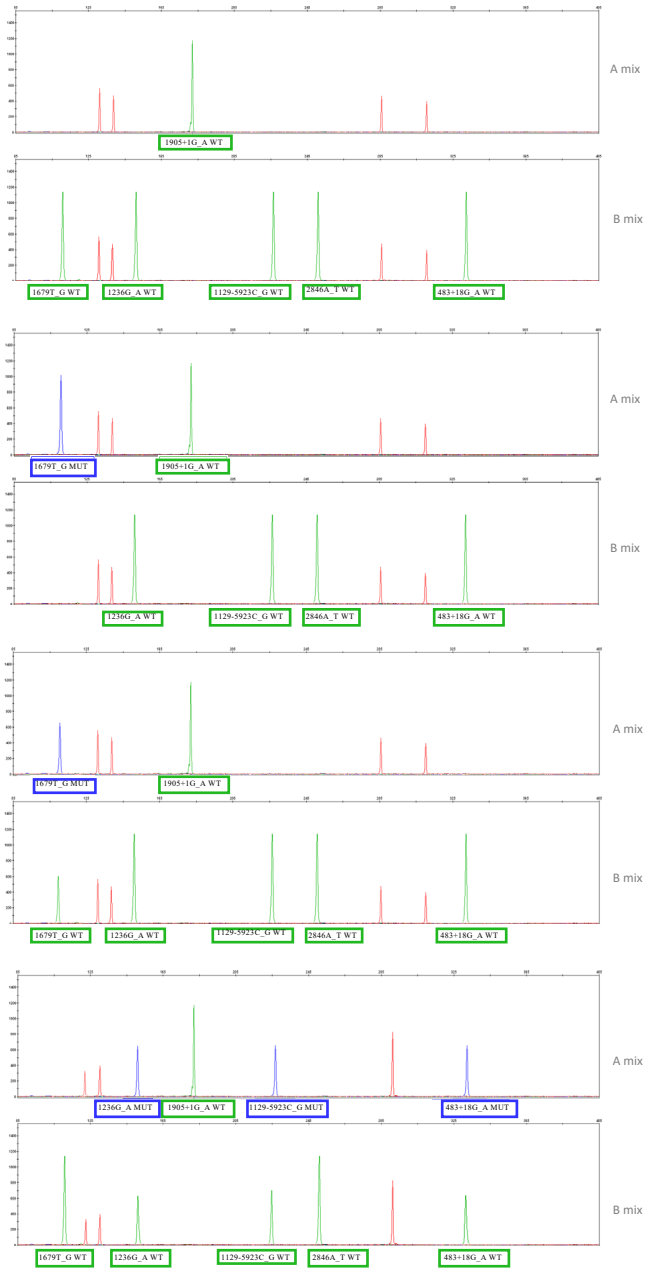
3. PCR
Run thermal cycling program.

4. Capillary Electrophoresis.
No post-PCR modifications required.
Compatible with ABI 3*** Genetic Analysers.



5. Analysis
Simple analysis and interpretation of results using GeneMapper™ or GeneMarker® software.

Example Results



Normal Result

In a normal result, all alleles detected are homozygous wildtype. In this example, the sample is wildtype for all markers detected:
*2A – green WT peak present in the A mix.
*13, c.2846A>T, HapB3 – Green WT peaks present in the B mix

Homozygous Result

In an individual homozygous for a specific mutation, the marker will only display a single mutant peak and no wildtype. In this example, the sample is homozygous mutant for 1679 T>G (blue peak), with the remaining markers showing a homozygous wildtype genotype

Heterozygous Result

If an individual is heterozygous for a marker, two peaks will be detected. In this example, the sample is heterozygous mutant for c.1679 T>G, with the remaining markers showing a homozygous wildtype genotype

Compound Heterozygous Result

If an individual is heterozygous for a marker, two peaks will be detected. In this example, the sample is heterozygous mutant for all haplotype B3 markers (c.1129-5923 C>G, c.1236 G>A, c.483+18 G>A), with the other remaining markers showing a homozygous wildtype genotype

Ordering Information

Assay	Catalogue Number	Description	Kit Size
Yourgene® DPYD, CE-IVD	ONDYDB1	*DPYD* screen testing for *2A, *13, c.2846A>T, HapB3	25 Tests

Yourgene® DPYD previously known as Elucigene® DPYD

References

- *DPYD* genotype-guided dose individualisation of fluoropyrimidine therapy in patients with cancer: a prospective safety analysis (Henricks et al., 2019)
- A cost analysis of upfront DPYD genotype guided dose individualisation in fluoropyrimidine-based anticancer therapy (Henricks et al., 2019)

About YOURGENE HEALTH

Yourgene Health is an international molecular diagnostics group which develops integrated genomic technologies and services enabling genomic medicine.

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