AWMGS
All Wales Medical Genomics Service

DPYD testing: Variant analysis for fluoropyrimidine toxicity

DPYD testing information

Germline variants in the dihydropyrimidine dehydrogenase (*DPYD*) gene can confer an increased risk of severe or fatal toxicity for cancer patients being treated with common fluoropyrimidine based chemotherapies, 5-fluoruracil, tegafur or capecitabine.

AWMGS uses an ARMS PCR based method to identify the following variants in the *DPYD* gene.

c.1905+1G>A
c.1679T>G
c.2846A>T
c.1129-5923C>G
c.1236G>A

Sample requirements

4ml blood in EDTA anticoagulant (purple top)

All samples must be accompanied by a completed request form. The general laboratory genetics request form (this is available on our website) with at least 3 patient identifiers, the relevant clinical information.

'DPYD' must be clearly marked on the request form.

Send samples to:

All Wales Medical Genomics Service
Institute of Medical Genetics

University Hospital of Wales

Heath Park Way

Cardiff, CF14 4XW

Referral criteria

Patients being considered for fluoropyrimidine based therapy should undergo pre-treatment pharmacogenomic screening for the five variants of DPYD associated with severe toxicity.

Turnaround Time

A report will be issued listing any variants identified, and the treatment recommendation based on the patient's genotype **within 5 working days.**

Contact details

Email: Lab.Genetics.CAV@wales.nhs.uk
Phone: 02921 842641

References

- Clinical Pharmacogenetics Implementation Consortium

https://cpicpgx.org/guidelines/guideline-for-fluoropyrimidines-and-dpyd/