AWMGS
All Wales Medical Genomics Service

Fanconi Anemia (FA) Service

Background

Fanconi anemia (FA) is an autosomal recessive heterogeneous condition with a broad clinical presentation. It is the most commonly inherited bone marrow failure syndrome and has predisposition to leukemia and solid tumors in addition to presenting physical congenital abnormalities.

Over 20 FA genes have been identified, all of which play a role in DNA repair (Cheung and Taniguchi, 2017).

Test information

Samples for the investigation of FA are tested by the NHS Grampian Medical Genetics Service in Aberdeen.

Referral criteria

Samples must be sent directly to the NHS Grampian Medical Genetics Service.

The NHS Grampian Medical Genetics Service general request form (GEN FORM 066), must be completed and sent with the FA breakage studies sample. Please provide as much relevant clinical information as possible.

Where possible, please also send an anonymised control blood (5ml in lithium heparin), complete the 'Control Sample for Fanconi Anemia' referral form (GEN FORM 213) and post this with the patient sample.

Sample Requirements

5ml venous blood in a Lithium Heparin tube
Please label samples with three identifiers and date of collection
All samples must be accompanied by a completed request form
Consent for testing and sample storage is assumed when the request is received – it is the responsibility of the referring clinician to ensure that appropriate consent has been obtained
Incomplete or illegible referral forms may lead to sample rejection and a delay in testing

Please send FA samples to the following address:
C/O Duty Scientist,
Department of Medical Genetics
Polwarth Building, 2nd Floor
Foresterhill
Aberdeen
AB25 2ZD

Please see www.nhsgrampian.org/medicalgenetics for further sample requirements

TAT (Calendar days)

TAT for routine blood referrals	28CD

Contact Details

The AWMGL must be notified of any samples sent to ensure that the tests are appropriately invoiced.

All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW

Tel: 029 2074 2641
Fax: 029 2074 4043

Haematology.Genetics.cav@wales.nhs.uk

lab.genetics@wales.nhs.uk
www.wales.nhs.uk/AWMGS/
Accredited to ISO 15189:2012

References

Cheung, R.S. and Taniguchi, T. (2017). Recent insights into the molecular basis of Fanconi anemia: genes, modifiers, and drivers. *International Journal of Hematology*, 106(3), pp.335–344.