Hereditary recurrent fevers

Also known as: Periodic fevers, Familial Mediterranean fever (FMF) screen, MEFV, Familial Hibernian fever, TRAPS screen, mevalonate kinase deficiency, cryopyrin-associated periodic fevers syndromes

Test category:

Familial - Hereditary recurrent fever; Immunology - Hereditary recurrent fever

Use of test

Purpose:

Hereditary recurrent fever syndromes are a wide group of autoinflammatory diseases. Molecular analysis of causative genes can dramatically improve patient quality of life by allowing early and accurate diagnosis and classification and the administration of appropriate treatments.

MEFV, MVK, NLRP3 and TNFRSF1A are sequenced to help confirm the diagnosis of specific recurrent fevers.

Utility:

In an affected person the identification of mutation(s) in the MEFV, MVK, NLRP3 and TNFRSF1A genes can suggest or confirm the diagnosis of familial Mediterranean fever (FMF), mevalonate kinase deficiency (MVK), cryopyrin-associated periodic syndromes (CAPS), and tumour necrosis factor receptor-associated periodic syndrome (TRAPS, or familial Hibernian fever) respectively.

The absence of mutations makes it less likely that the patient has FMF, TRAPS, MKD or CAPS, but does not exclude these clinical diagnoses, and so diagnosis and medical management should be guided by clinical criteria and judgement.

Ethical considerations:

This is an assay for heritable mutations. It may raise issues of ethics or consent that are different from most other investigations ordered in the routine care of a patient. Use of the Sonic Genetics consent form (below) is not mandated, but could be used to guide the discussion with the patient.

In the case of carrier testing of an unaffected child, national regulations require that the laboratory has evidence of pre-test counselling by a genetics professional and written consent.

Consent form:

Download the Sonic Genetics consent form

Methodology:

Sequencing of the MEFV, MVK, NLRP3 and TNFRSF1A genes.

Requesting the test

Ordering:

This test can be requested by any medical practitioner.

Sample required:

4 mL blood in EDTA (separate tube required). Specimens may be collected by the requesting practitioner or at any Sonic Healthcare collection centre (see link below).

To help ensure the quality of the test, a genetic test should be done with a dedicated sample whenever possible i.e. a sample collected specifically for that test rather than a sample that is used for multiple tests.

We recommend that the patient or another adult check the labelling of request forms and sample tubes.

>Link to Sonic Healthcare collection centres

Turnaround time:

4 weeks.

Price:

$580.

Rebate:

This test is not rebated by Medicare. The laboratory assumes that the patient or client has provided informed financial consent for the test.