

Patient Fact Sheet: BRAF Testing

What is BRAF?

Cell growth in the body is controlled by a complex set of communications between proteins. These communications, collectively described as “cell signalling” occur in a carefully controlled and ordered way along “pathways”.

BRAF is one of the proteins in the cell signalling pathway that controls cell growth in a number of different tissues in the body. In normal cells, BRAF is tightly controlled and only becomes active when the cell receives the right signals to grow.

What is a BRAF Mutation?

The BRAF gene contains the genetic(coding) information required to make the B-Raf protein. Errors, or mutations, in the BRAF gene result in unregulated production of B-Raf proteins, causing rapid uncontrolled cell growth, allowing tumours to grow.

In melanoma, approximately 50% of patients have tumours which carry an activating mutation somewhere on the BRAF gene. The most common mutation, which accounts for around 80% of mutations of the BRAF gene is called V600E. A mutation at this site causes BRAF to become overactive, thus activating the growth pathways and causing tumour growth. BRAF mutations are also found in significant proportions of colorectal and thyroid cancers and less commonly other tumour types.

How can BRAF Mutations be detected?

A molecular test can be performed on the tumour tissue that has been excised from the patient to determine whether the patient’s tumour contains this mutation. The test specifically looks for the V600E mutation. The test is performed at Pathlab using well established molecular technology called PCR. The test result may take up to a week to become available.

What implications does the test result have for my treatment?

Patients who test positive for the BRAF V600E mutation may be suitable for drugs which specifically inhibit BRAF containing the V600E mutation. There is now well documented research evidence to show that if the mutation is present, then these drugs prolong life expectancy in patients with advanced melanoma.

Who should have this test?

Patients should discuss their suitability for this test with their oncologist or dermatologist. The specialist will be able to explain whether the test is appropriate for the patient, and what the treatment options are likely to be when the test results are known. The specialist should also be able to answer other questions regarding the test including the costs, as well as possible side effects and likely benefits of any ensuing treatment.

For further information on the BRAF test, please contact your physician or email our molecular team molecular.testing@pathlab.co.nz