BAP1 Tumour Predisposition Syndrome (BAP1 TPDS)

A Guide for Patients & Families

MESOTHELIOMA UK

www.mesothelioma.uk.com

0800 169 2409

Introduction

BAP1 tumour predisposition syndrome (BAP1 TPDS) is an inherited disorder which increases the risk of developing certain types of cancer.

What is BAP1?

Genes carry instructions for making the proteins that determine how we grow and develop. We each have approximately 25,000 genes, one of which is the BAP1 gene.

The body reads the BAP1 gene and makes the BAP1 protein. Under usual circumstances this protein can **protect** the individual from developing specific types of cancer.

It does this by controlling cell growth and division, and by repairing DNA in our cells when it is damaged through day to day 'wear and tear' or environmental factors such as smoking.

What is BAP1 TPDS?

As with all genes we have two copies of the BAP1 gene, one copy inherited from our mother and one from our father. In individuals with BAP1 TPDS, one copy of the BAP1 gene carries a 'spelling mistake' called a mutation.

As a result, the body cannot read that copy of the gene properly and the individual loses some protection against developing cancer.

As the BAP1 gene mutation has been inherited, it is present in every cell in the body and is called a germline mutation.

Having a germline mutation of the BAP1 gene increases the risk of developing specific types of cancers, including certain types of melanoma, kidney cancers and mesothelioma. It is important to remember that not all people who have BAP1 TPDS will develop cancer, and the risk of cancer may be influenced by other genetic and environmental factors as well.

The exact incidence of BAP1 TPDS is currently unknown although it is thought to be rare. Research is continuing to try and establish a clearer understanding of this gene and its relation to mesothelioma.

"It is important to remember that not all people who have BAP1 TPDS will develop cancer"

Why have I been given this information?

You have been given this information because you have been diagnosed with mesothelioma and your doctor or nurse has identified that you may have BAP1 TPDS.

Your doctor or nurse might suspect this because either you or a first degree relative (parent, sibling, child) have had one or more of the cancers associated with BAP1 TPDS.

What happens now?

Your doctor will offer the option of referring you to the nearest genetics team, which may not be at your local hospital. The genetics team will take a detailed family history from you and help you decide whether you should have genetic testing for BAP1 TPDS.

What does the test involve?

If you decide to proceed with the test a small blood sample will be taken and sent for analysis to determine if you have the condition.

Why should I have the test?

Knowing whether you have BAP1 TPDS can help you and your healthcare team make decisions about your future care. As BAP1 TPDS is known to run in families (inherited), if the diagnosis is confirmed in you, it will

allow your relatives to make decisions about having genetic testing to see if they too have the condition.

What if I don't want to have the test?

You will be given support and counselling by the genetics team to help you decide whether to have the test or not. If you decide not to have the test the care and treatment you are offered for your mesothelioma will not change.

If you have any further questions, please contact your consultant or specialist nurse.

Mesothelioma UK provides specialist, impartial, up to date support and information.

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Email | info@mesothelioma.uk.com

Freephone | 0800 169 2409

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