

Frequently Asked Questions

1. Do I need to be tested every time I have a baby?

NO. If you are not a carrier, you remain at very low risk of having a child with the condition. If you are a carrier and you have a new partner, your new partner will need to be tested.

2. If no gene change is found, could I still be a carrier?

YES. The current carrier test cannot detect every gene change that causes these conditions. If no gene change is found, you are not a carrier of the most common gene changes, but there is still a small risk that you may be a carrier.

3. If I am a carrier, can I develop one of these conditions?

NO. If you are a carrier, you have a gene change in one of your two gene copies. The other copy of the gene works properly and there is no effect on your own health.

4. What if I have a relative who is a carrier of one of these conditions?

YOUR CHANCE of being a carrier is greater than most people and you and your partner should consider testing.

5. If I have no family history of these conditions can I still have an affected child?

YES. In more than 95% of families where a child is born with one of these conditions there is no family history of the condition.

6. Does this guarantee our baby will be healthy?

NO, this test is only for the conditions you have nominated to be tested for.

7. If I am not of Ashkenazi Jewish Ancestry, can I still have these tests?

If you are not of Ashkenazi Jewish Ancestry these tests may not be the most appropriate tests for you. Please consult our service before considering testing.

Contact Details

Genetic Health Services Victoria

10th Floor
Royal Children's Hospital
Flemington Road
Parkville VIC 3052
Ph: (03) 8341 6201
<http://www.genetichealthvic.net.au/>

More detailed information can be found at:
<http://www.taysachs.net>

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carrier testing for
Tay Sachs disease
and
related conditions



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What conditions are common to people of Ashkenazi Jewish Ancestry?

There are a number of inherited conditions that are common to people of Ashkenazi Jewish ancestry. Ashkenazi Jews are those with Eastern European ancestry. If you are not of Ashkenazi Jewish ancestry please consult our service before considering testing, as these tests may not be the most appropriate for you.

The conditions fall into the following categories:

Tay Sachs disease (TSD), Canavan disease (CD) and Niemann Pick disease (NP) are all serious conditions causing degeneration of the nervous system and generally result in death in early childhood.

Bloom syndrome (BS) and Fanconi anaemia (FA) predispose to cancer.

Cystic fibrosis (CF) affects the lungs and the digestive system and results in a reduced lifespan. Familial dysautonomia (FD) affects blood pressure and can result in episodes of severe illness.

Is our baby at risk?

Genes may have changes, known as mutations, which affect their function. People who have one copy of the gene change for the conditions outlined above are known as **healthy** carriers.

Babies inherit one copy of each gene from each parent.

A baby will have one of these conditions only if they inherit a gene change from **both** parents. This is only possible if **both** parents are carriers of a gene change for that condition.

What are the risks of having a child with one of these conditions?

The table below shows the chance of an individual of Ashkenazi Jewish ancestry being a carrier or having a child with any of these conditions.

		The risk of a child being affected by each condition is dependant on the couple's test results. Below are the risks for different testing scenarios					
Condition	Carrier Frequency	Both Not Tested	Both Non Carriers	1 Non carrier, 1 Not tested	1 Carrier, 1 Not tested	1 Carrier, 1 Non carrier	Both Carriers
TSD	1 in 28	1 in 3,000	LESS THAN 1 in 2 million	1 in 151,000	1 in 112	1 in 5,400	1 in 4
CD	1 in 40	1 in 6,400		1 in 312,000	1 in 160	1 in 7,800	1 in 4
NP	1 in 70	1 in 19,600		1 in 387,000	1 in 280	1 in 5,500	1 in 4
BS	1 in 100	1 in 40,000		1 in 990,000	1 in 400	1 in 9,900	1 in 4
FA	1 in 90	1 in 32,000		1 in 3,200,000	1 in 360	1 in 35,600	1 in 4
CF	1 in 25	1 in 2,500		1 in 80,000	1 in 100	1 in 3,200	1 in 4
FD	1 in 30	1 in 3,600		1 in 350,000	1 in 120	1 in 11,600	1 in 4

When should we have carrier testing?

It is up to you whether you choose to be tested. It is best to have carrier testing for these conditions either before pregnancy, or in early pregnancy, to make sure you have enough time to make decisions about family planning.

How do we get tested?

A simple and painless cheekbrush sample is all that is needed.

Speak to your Obstetrician or Midwife. As yet there is no Medicare rebate for this test and so there will be a charge.

What do the results mean?

There are two possible results from the test for each condition; that you are a carrier or you are a non-carrier.

Carrier

If your test shows that you have one copy of the gene change, you are a carrier. Your partner's

result will determine your risk as a couple of having a child with the condition.

Non-Carrier

This means that you do not have one of the common gene changes and so your risk of being a carrier is greatly reduced. However there is still a very small risk that you have a gene change because we cannot test for all possible changes.

What if we are both carriers?

Two people who are carriers of the same condition have a 1 in 4 (25%) chance of having a child with the condition in each pregnancy. If you are both carriers you will be provided with genetic counselling. Following counselling you may choose to have tests to identify whether your baby has the condition. If the tests diagnose any of these conditions you have a choice about whether to continue or terminate the pregnancy. If you are identified as being carriers prior to a pregnancy, you also have the option of preimplantation genetic diagnosis with invitro fertilisation.