

Conditions tested for

TAY-SACHS DISEASE

affected children suffer blindness and paralysis – they usually die before their 5th birthday.
(1 in 28 Ashkenazi Jewish people are carriers)

NIEMANN-PICK DISEASE

similar to Tay-Sachs disease – children usually die in early childhood.
(1 in 70 Ashkenazi Jewish people are carriers)

CANAVAN DISEASE

similar to Tay-Sachs disease – children usually die in early childhood.
(1 in 40 Ashkenazi Jewish people are carriers)

CYSTIC FIBROSIS

affects lung and digestive function. Affected individuals require daily therapy and life expectancy is reduced.
(1 in 25 Ashkenazi Jewish people are carriers)

FANCONI ANAEMIA

affected individuals can have severe anaemia, immune system problems, malformations, kidney problems and leukaemia.
(1 in 90 Ashkenazi Jewish people are carriers)

FAMILIAL DYSAUTONOMIA

neurological condition characterised by feeding difficulties, vomiting, abnormal sweating, reduced pain sensation and low blood pressure.
(1 in 30 Ashkenazi Jewish people are carriers)

BLOOM SYNDROME

affected people have increased risk of cancer and infections which reduce life expectancy.
(1 in 100 Ashkenazi Jewish people are carriers)

MUCOLIPIDOSIS IV

affected people typically have intellectual disability, speech difficulties, trouble chewing and swallowing, problems with their muscles, and visual impairment. Life expectancy is reduced.
(1 in 96 Ashkenazi Jewish people are carriers)

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Genetic screening in the Jewish Community

Screening for Tay-Sachs disease & related conditions

Information for people with Sephardi Jewish ancestry

Each Sephardi community has different genetic conditions that are more common depending on the community's country of origin. This means that there is no single test recommendation for those of Sephardi Jewish ancestry.

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For more information visit: www.taysachs.net

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What is the test for?

Some rare but severe genetic conditions are more common in some ethnic groups than in others. These conditions can be passed from parents to children. This brochure provides information for people of Jewish ancestry who are at risk of having children with one of these conditions.

Most Jewish people in Australia have Ashkenazi (Eastern European) ancestry. Information for people with Sephardi Jewish ancestry is given on the back of this brochure.

There are 8 recessive genetic (inherited) conditions, more common in the Ashkenazi Jewish population for which screening is recommended:

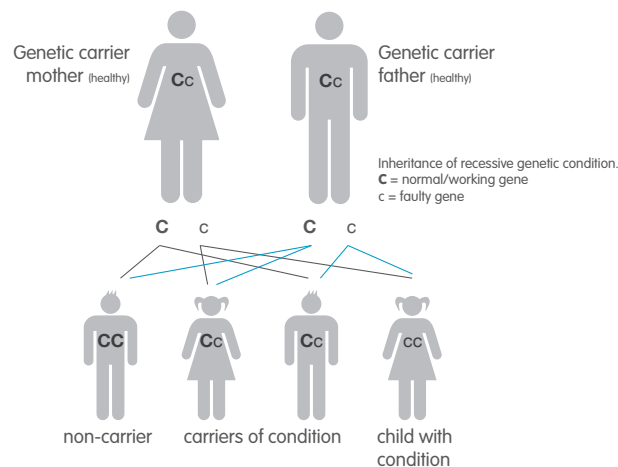
- Tay-Sachs disease
- Niemann-Pick disease
- Canavan disease
- Cystic fibrosis
- Fanconi anaemia
- Familial dysautonomia
- Bloom syndrome
- Mucopolysaccharidosis IV

Carrier testing

You can find out if you are at risk of having a child with one of these conditions by getting tested to see if you are a carrier of one or more conditions.

Carriers have one healthy copy of the gene and one altered copy of the gene. Carriers do not have the condition because a person needs to have two copies of the altered gene to be affected. If two people are carriers of the same condition, there is a 1 in 4 (25%) chance for each pregnancy of having a child affected by the condition.

People only know if they are carriers if they have a child with the condition or if they are tested.



• 1 in 4 chance that each child born may be affected. Males & females can be affected equally.

When should I get tested?

Testing is voluntary. You can be tested at any time but it is best to have carrier testing for these conditions before deciding to have children. This is to allow enough time to make decisions about family planning. Otherwise you can be tested in early pregnancy (ideally less than 12 weeks gestation). If both members of a couple are found to be carriers of the same condition, you can be referred to a genetic service to discuss your family planning options.

How is testing done?

Testing is either from a blood sample or saliva sample depending on the testing laboratory's requirements.

How do I arrange testing?

Contact your GP, obstetrician or local genetics service and they will be able to arrange testing.

Is there a Medicare rebate?

No, there is currently no Medicare rebate for Ashkenazi Jewish carrier screening.