



Understanding Genetic Risk

Why it's important to understand your risk

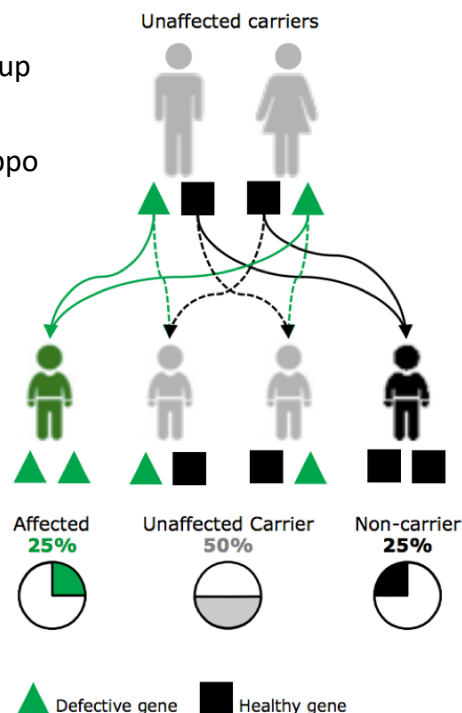
For most couples, the chances of having a child with Sanfilippo are slim – about **1 in every 70,000** babies are born with Sanfilippo. However, if there is Sanfilippo in your family tree, the risk could be up to **180 times higher**.

Couples can be screened for the genetic changes that cause Sanfilippo before they fall pregnant, and if there is a high risk, family planning options can be considered, such as IVF with genetic screening to select embryos that do not have the genetic change.

How is Sanfilippo inherited?

Everybody has two copies of each gene, one inherited from their mother, and one from their father (with some exceptions). Sanfilippo is an 'autosomal recessive disorder'. This means that for a child to inherit Sanfilippo, he or she must get one faulty gene from each parent.

If both parents have one copy of the faulty Sanfilippo gene, then for each pregnancy there is a one-in-four chance of having a child with Sanfilippo.



What are carriers?

Carriers are people who have one copy of the faulty gene and one healthy gene. They do not have any symptoms and usually don't know that they are a carrier.

If we take Sanfilippo type A as an example, approximately **1 in every 169 people** in the general population are carriers of the faulty gene. The chance of two people getting together who both have the same faulty gene and having a baby with Sanfilippo Type A is about **1 in 114,000**.

However, the risk increases if there is any family history of Sanfilippo, and this risk varies depending on where in the family tree you and the affected child are. Please see the table below for examples. A genetic counsellor can also calculate and explain the risk to you.

Sanfilippo type	Type A	Type B	Type C	Type D
Risk for general population	1 in 114,000	1 in 211,000	1 in 1.4 million	1 in 1.1 million
Risk if one parent's sibling has a child with Sanfilippo	1 in 1272	1 in 1840	1 in 4736	1 in 4112
Risk if one parent is a known carrier	1 in 636	1 in 920	1 in 2368	1 in 2056

Table 1: Risk of having a child with Sanfilippo Syndrome

(Reference: Meikle PJ, Hopwood JJ, Clague AE and Carey WF. Prevalence of lysosomal storage disorders. JAMA. 1999 Jan 20;281(3):249-54.)



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How do I get tested?

To arrange for testing you will need to see a genetic counsellor, which will require a referral, usually from your GP. The genetic counsellor will help you understand the risk and organise for tests to be done.

To find a genetic counsellor in your area you can search on this website:

<https://www.hgsa.org.au/asgc/find-a-genetic-counsellor>

Alternatively, private genetic testing called the [Counsyl - Foresight Carrier Screen](#) can be arranged by calling Counsyl Australia on [1300 268679](tel:1300268679). Currently testing includes Sanfilippo Types A, B and C and costs approximately \$600.

If you have trouble accessing genetic testing, please contact us: research@sanfilippo.org.au

If there is a family history of Sanfilippo we urge you to PLEASE seek genetic counselling

Read one family's story



Please print or email this downloadable PDF and distribute to relatives of children with Sanfilippo so they understand their genetic risk!



One Australian family has experienced Sanfilippo across two generations and were given inadequate genetic counselling. You can read Matilda's story at www.hopeformatilda.com.au

"We have already heard the phrase 'one in a million'... 'it's so rare'... 'no need to worry' a lot in our journey," says Matilda's mum Kate.

Matilda's parents Kate and Lachlan were told "not to worry", and that the chances of Kate also being a carrier were so slim. Sadly, Matilda has inherited Sanfilippo, the same disease which took the life of Matilda's uncle in the 1990s.