Introduction

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There are two main types of Thalassaemia: Alpha and Beta. Alpha Thalassaemia is the reduction or absence of alpha-globin protein production. It is generally the milder form of the condition.

A healthy person will usually inherit four alpha globin genes for the production of alpha globin protein in haemoglobin. A carrier, who is considered healthy, may only have two or three of the normal four alpha globin genes. They are, however, at higher risk of having a child affected by Haemoglobin H Disease or Bart's Hydrops Fetalis if their partner is also a carrier. When an individual only has one alpha globin gene, they have Haemoglobin H Disease and require medical care, likely experiencing lifelong mild or moderate anemia. In more severe cases, when an individual is born without any alpha globin genes, it results in Bart's Hydrops Fetalis, ultimately leading to foetal death or death shortly after birth.

Beta Thalassaemia is the reduction or absence of beta-globin protein. A healthy person will usually inherit two beta globin genes for the production of the beta globin protein in haemoglobin. A healthy carrier, also known as having c Thalassaemia Minor, may have alterations in one of their two beta globin genes. Carriers may be at risk of having a child affected by Beta Thalassaemia Major if their partner is also a carrier of ß Thalassaemia. Beta Thalassaemia Major results from an individual having alterations in both of their ß globin genes. They require lifelong medical treatments.

If two individuals are carriers of Thalassaemia they have a 1 in 4 chance of having a child who suffers from a severe blood disorder. If one individual is a carrier and the other is a non-carrier, they have a 2 in 4 chance of having a child who will be a healthy carrier.

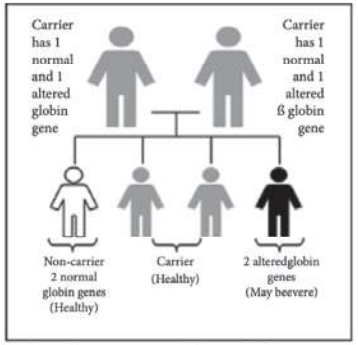
Similarly to Beta Thalassaemia, Sickle Cell Disease affects ß globin genes. A Sickle Cell carrier would have an alteration in one of their two ß globin genes, giving them a 1 in 4 chance of having a child with Sickle Cell disease if their partner is also a carrier, or a 2 in 4 chance of having a child who is also a carrier if their partner is not a carrier. When a person has the Sickle Cell alteration in one of their ß globin genes and a certain alteration in their other ß globin gene, they may have Sickle Cell Disease.

In 1976, a group of dedicated medical professionals came together to found the Thalassaemia Society of Victoria (TSV), driven by a shared vision: to secure government support for medical research, improved treatment facilities and to support patients and families affected by Thalassaemia and related genetic haemoglobinopathies. Over the decades, that early effort evolved into what is now known as Thalassaemia and Sickle Cell Australia (TASCA) – a purpose-driven, not-for-profit organisation committed to advocacy, community support and education across Australia.

Chances of having a child affected by a genetic blood disorder

Figure 1:

Both parents are carriers

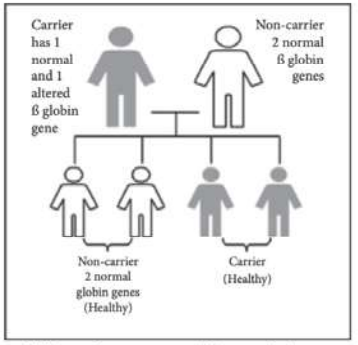


With **each** pregnancy, this couple has a:

- 1 in 4 chance of having a child who inherits
- 2 normal copies of the globin gene and is a non-carrier.
- 2 in 4 chance of having a healthy carrier, like its parents. This child has one normal copy of the globin gene and one altered copy of the globin gene.
- 1 in 4 chance of having a child which may have a severe genetic blood disorder. This child has inherited 2 altered copies of the globin gene.

Figure 2:

Only one parent is a carrier



With **each** pregnancy, this couple has a:

- 2 in 4 chance of having a child who inherits
- 2 normal copies of the globin gene and is a non-carrier.
- 2 in 4 chance of having a child who is a healthy carrier - like its parent. This child has one normal copy of the globin gene and one altered copy of the globin gene.

*Note: (ref: Modell B, Darlison (2008) Global epidemiology of haemoglobin disorders and derived service indicators - Bulletin of the World Health Organisation Vol 86, number 6, June 2008: 417-496)

As the organisation reaches its 50th anniversary, we pause to reflect on an extraordinary journey–one marked by compassion, resilience and unwavering dedication. From humble beginnings amid linguistic and cultural barriers–particularly in migrant and refugee communities–to becoming a national beacon of support, TASCA has always stood by Australians living with Thalassaemia and Sickle Cell Disease.

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From left to right: Jenny Hatyinicolaou (Mother of patient, James), Maria Hatyinicolaou (patient), Alex Pervritis (patient), Rosa Calabro (patient), Maria Cuccaro (patient) Sofia Koukouravas (patient, front holding baby)

Back row, left to right: Libby Reid, Dr. Bowden, Tarlie (former Thalassaemia Centre staff), Di Pantazis (mother of Eugenia), the late Sotirios Katakouzinos. Front row, left to right: Rhonda Kurzel, the late Dr. Mattheus, the late Audrey Jacovou (first Thalassaemia patient in Melbourne to have a child) and her son, Nicholas Jacovou