

Haemoglobinopathies

Guide to the completion of the EBMT data collection form: Haemoglobinopathies_v2.0

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EBMT Registry

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Introduction

Please make sure you have already checked the **Introduction to the EBMT Registry Completion Guidelines** document latest version available under *Manuals and Reference Documents* section on [EBMT website](#).

Haemoglobinopathies

Haemoglobinopathies are a heterogeneous group of inherited diseases characterised by alteration of haemoglobin production. Adult haemoglobin is composed of 2 α and 2 β chains (tetramer 22). Lack of production of β chains characterises β -thalassemia (here and after referred to as Beta thalassemia), lack of production of α chains characterises α -thalassemia (which is a homozygous condition not compatible with life, Haemoglobin Bart's).

Sickle Cell Disease is a homozygous congenital disease where a single point mutation characterised by the production of an altered β -chain. The heterozygous carrier state is called sickle cell trait (SCT).

Main haemoglobinopathies treated with stem cell transplantation are:

- Beta thalassemia
- Sickle cell disease
- Other haemoglobinopathies

Disease

Date of diagnosis/date of first event

Enter the date of diagnosis of the disease or the date of the first haemoglobinopathy event.

Classification

Select the class that is appropriate for the haemoglobinopathy and check the box next to it. If the class is not listed, check the box 'Other haemoglobinopathy' and specify it in the textbox.

Thalassaemia

In case of thalassaemia, indicate the sub-class.

Beta thalassaemia subclasses

β^0 / β^0 thalassaemia refers to the absence of production of beta globin. When patients are homozygous for a beta 0 thalassaemia gene, they cannot make any normal beta chains (haemoglobin A). This mutation is transmitted by both parents who are carriers for thalassaemia.

β^0 / β^+ thalassaemia indicates a mutation that presents decreased but not absent production of beta globin. Thalassaemia patients in which one or both of their beta thalassaemia mutations are beta+ mutations make some haemoglobin A, and the disorder may be less severe.

β^0 / β^E thalassaemia is a type of thalassaemia disease that is prevalent in southeast Asia and parts of the Indian subcontinent, and it accounts for almost half of all patients with severe thalassaemia worldwide. Hb E is due to a point mutation that inserts a splice site in the β -globin gene and results in decreased production of Hb E.³ In the homozygous state (Hb EE) the clinical symptoms are similar to a mild β -thalassaemia.

β^+ / β^+ thalassaemia (Haemoglobin S-beta-thalassaemia disease) is a haemoglobinopathy that causes symptoms similar to those of sickle cell disease. Beta S thalassaemia are to be reported in the Sickle Cell Disease section, as SB+ or SB0.

Sickle cell disease

Indicate the sub-class of sickle cell disease.

Sickle Cell Disease (SCD) sub-classes include:

(synonym: Sickle cell anaemia, falciform anaemia, drepanocytosis) is a hereditary disease characterised by the presence of an altered haemoglobin (haemoglobin S = HbS). There are the following sub-classes:

SS - haemoglobin SS disease is the most common type of sickle cell disease. It occurs when a person inherits copies of the haemoglobin S gene from both parents. This forms haemoglobin known as Hb SS. As the most severe form of SCD, individuals with this form also experience the symptoms at a higher rate.

SC - haemoglobin SC disease is the second most common type of sickle cell disease. It occurs when a person inherits the Hb C gene from one parent and the Hb S gene from the other. Individuals with Hb SC have similar symptoms to individuals with Hb SS. However, the anaemia is less severe.

SB+ - haemoglobin SB+ (beta) thalassaemia affects beta globin gene production. The size of the red blood cell is reduced because less beta protein is made. If inherited with the Hb S gene, the person will have haemoglobin S beta thalassaemia. Symptoms are not as severe.

SBO - sickle beta-zero thalassaemia is the fourth type of sickle cell disease. It also involves the beta globin gene. It has similar symptoms to Hb SS anaemia. However, sometimes the symptoms of beta zero thalassaemia are more severe. It is associated with a poorer prognosis.

Other haemoglobinopathy; specify

Choose this answer option if the haemoglobinopathy of the patient that was diagnosed is not listed in the table. Please, describe the diagnosis in the textbox in English.