# What variants of sickle cell disease, beta-thalassaemia, and other haemoglobinopathies may be detected in Australia’s newborn screening (NBS) programs?

There are a number of different variants (or types) of sickle cell disease, beta-thalassaemia or other haemoglobinopathies that may be detected in NBS programs. The full list of variants is below.

| **Haemoglobin disorder/ Variant** | **How this variant is detected** |
| --- | --- |
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| **Sickle cell disease** |
| Sickle cell anaemia / Hb S,S | Target  |
| Sickle cell, Haemoglobin C / Hb S,C | Target |
| Sickle cell, β-thalassaemia / Hb S,β-thalassaemia | Target |
| Sickle cell, haemoglobin D-Punjab / Hb S,D-Punjab | Target |
| Sickle cell, haemoglobin O-Arab / Hb S,O-Arab | Target |
| Sickle cell, haemoglobin Lepore / Hb S,Lepore | Target |
| Sickle cell, δβ thalassaemia / Hb S, δβ-thalassaemia | Target |
| Sickle cell, haemoglobin E / Hb S,E | Target |
| Haemoglobin S, variant / Hb S,Var | Target |
| Sickle cell, hereditary persistency of foetal haemoglobin / HbS, HPFH | Target |
| **Beta-thalassaemia** |
| β-thalassaemia major / Hb β-thalassaemia, β-thalassaemia | Non-target |
| β-thalassaemia intermedia / Hb β-thalassaemia, β-thalassaemia | Non-target |
| β thalassaemia minor / Hb β-thalassaemia | Non-target |
| Haemoglobin C, β-Thalassaemia / Hb C,β-thalassaemia | Non-target |
| Haemoglobin D, β-Thalassaemia / Hb D,β-thalassaemia | Non-target |
| Haemoglobin E β-Thalassaemia / Hb E,β-thalassaemia | Non-target |
| Haemoglobin E Delta-β-thalassaemia / Hb E, δ-β-thalassaemia | Non-target |
| Haemoglobin variant/ β-thalassaemia / Hb Var,β-thalassaemia | Non-target |
| **Other Haemoglobinopathies** |
| Beta-globin |
| Haemoglobin C disease / Hb C,C | Non-target |
| Haemoglobin D disease / Hb D,D | Non-target |
| Haemoglobin E disease / Hb E,E | Non-target |
| Haemoglobin variant, variant / Hb Var,Var | Non-target |
| Alpha thalassemia |
| Alpha Thalassaemia Major | Non-target |
| Haemoglobin H Disease  | Non-target |
| Hereditary Persistence of Fetal Hemoglobin |
| HPFH/HPFH | Non-target |
| **Carrier status**  |
| Sickle cell trait (carrier) / Hb A,S | Non-target |

NBS target conditions are intentionally screened for in Australia’s NBS programs. There is a specific and reliable test available to detect these conditions, the health outcomes of the condition are well understood and there is an available and effective treatment.

NBS non-target conditions **may** be incidentally detected when screening for a target condition. Although NBS is not specifically designed to detect these conditions, it may find babies with a non-target condition who may benefit from early detection. Abnormal findings from NBS (both target and non-target conditions) are reported and followed up as required.