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
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.