

## Referral guidelines for antenatal screening specimens

Biological mother carrier state	Biological father carrier state	Further studies by DNA analysis
No abnormalities detected	Testing of baby's biological father not required	None required
Any abnormal Hb or thalassaemia	No abnormality detected	None required
HbS	HbS or HbC	None required until PND
HbS	HbO <sup>Arab</sup> , D <sup>Punjab</sup> , E, Lepore, $\beta$ thalassaemia or $\delta\beta$ thalassaemia	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available
HbS	HPFH	Send bloods for mutation confirmation. PND is not usually indicated when HPFH has been confirmed
HbS + $\alpha$ thalassaemia	Assess risk as per HbS alone, unless family origins indicate a high risk of $\alpha^0$ thalassaemia	Assess risk as per HbS alone, unless family origins indicate a high risk of $\alpha^0$ thalassaemia
HbC	HbS	None required until PND
HbD	HbS	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available
HbO <sup>Arab</sup>	HbS, $\beta$ thalassaemia or $\delta\beta$ thalassaemia	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available
Hb Lepore	HbS, E, O <sup>Arab</sup> , Lepore, $\beta$ thalassaemia or $\delta\beta$ thalassaemia	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available.
HbE	$\beta$ thalassaemia, Hb Lepore, $\delta\beta$ thalassaemia, HbS	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available
HbE	$\alpha$ thalassaemia (MCH < 25pg)	Send bloods for mutation confirmation if high risk family origins for $\alpha^0$ thalassaemia, consider impact on options available

β or δβ thalassaemia	HbS, E, O <sup>Arab</sup> , Lepore, β thalassaemia or δβ thalassaemia	If PND is being considered send bloods for mutation confirmation, provided it does not limit options available
β or δβ thalassaemia	α thalassaemia (high risk family origins)	Send bloods for mutation confirmation if high risk family origins for α <sup>0</sup> thalassaemia, consider impact on options available
HPFH	HbS, E, O <sup>Arab</sup> , Lepore, β or δβ thalassaemia	Send bloods for mutation confirmation. PND is not usually indicated when HPFH has been confirmed
α thalassaemia (MCH < 27pg but ≥ 25pg)	Testing of baby's biological father not required	None required

### α thalassaemia (MCH < 25pg)

1. Low risk α <sup>0</sup> thal family origins in either biological parent	Testing of baby's biological father not required	None required
2. High risk α <sup>0</sup> thal family origins in both biological parents or unknown	Test baby's biological father and if MCH < 25pg irrespective of any other phenotype detected	Send maternal and paternal bloods for mutation confirmation, consider impact on options available.