Appendix 4 – Thalassaemia conditions

Genotype	Anaemia	**Splenomegaly**	**Other clinical features**	**Clinical severity**	**Remarks/treatment**
Beta (β) **Thalassaemia Major** **(βthal/βthal)**	Severe Impaired red blood cell production	Common May require splenectomy	Endocrine dysfunction Cardiac failure Iron overload Hepatic dysfunction Increased risk of infections	Severe Shortened life span if untreated or inadequately treated	Decreased synthesis of βeta globin chains. Presents in infancy. **Treatment:** Blood transfusions and iron chelation therapy for life. Bone marrow transplant if HLA matched donor. **Offer counselling and PND to couples 'at risk' of having a baby with this condition.**
E/β Thalassaemia (Hb E/βthal)	Moderate to severe	Common	Iron overload Cardiac failure Hepatic dysfunction Endocrine dysfunction Cholelithiasis Increased risk of ↑ infections	Very variable may range from mild to severe	**May present as severe as** β Thalassaemia Major **(requiring blood transfusions and iron chelation for life) or as** β Thalassaemia Intermedia. **Folic acid supplements needed.** Bone marrow transplant may be indicated depending on severity of condition. **Offer counselling and PND to couples 'at risk' of having a baby with this condition.**
Thalassaemia Intermedia **(could be caused by a range of haemoglobinopathies– see carrier chart for further information)**	Mild to moderate	Common	Usually moderate to mild thalassaemic condition depending on genetic mutations inherited Usually presents after 2 years of age if not identified at birth Osteoporosis Renal calculi	Moderate Possible iron overload	DNA needed to confirm genotype/severity **Usually no regular treatment required but may need occasional blood transfusions for example during pregnancy or infection** **Folic acid supplements may be beneficial.** **Regular monitoring by specialist even if no treatment is required.** **Assessment by specialist if a couple are at risk of having a baby with this condition.**

Genotype	**Anaemia**	**Splenomegaly**	**Other clinical features**	**Clinical severity**	**Remarks/treatment**
Hb "H Disease" **(Alpha Thalassaemia 3 gene deletion --/-α)**	Mild to moderate	Occasional	Occasional haemolytic anaemia Gallstones	Mild to moderate	**No regular treatment required** but haemoglobin levels may fall during infection or episode of anaemia so occasional blood transfusion may be needed. Regular monitoring by Specialist is important. **Genetic counselling but PND not indicated**
Alpha (--/--) Thalassaemia Major	Severe intrauterine anaemia	Not applicable	Also known as Haemoglobin Barts Hydrops Fetalis Associated with maternal morbidity & mortality if undetected during pregnancy	Very severe Absence of all 4 alpha (α) globin genes	Incompatible with life in utero as no fetal haemoglobin is produced. The use of intra uterine blood transfusion has been used on occasion. Most common in Far East Asia. **Offer counselling and PND to couples 'at risk' of having a baby with this condition.**

Notes

- Individuals with these conditions (except alpha thalassaemia major) should be registered with and followed up regularly by a Haematology Clinic
- Pregnant women with any of the above disorders should be considered *"high risk"* and should be followed up by a Haematologist and Obstetrician during pregnancy with booked hospital delivery.

References

Eleftheriou A, Angastiniotis M About Alpha ThalassaemiaThalassaemia International Federation (TIF) www.thalassaemia.org.cy

Eleftheriou A, Angastiniotis M About Beta ThalassaemiaThalassaemia International Federation (TIF) www.thalassaemia.org.cy

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