Appendix 5 – Benign haemoglobin disorders

Genotype	Anaemia	Other clinical features	Comments
Haemoglobin C Disease **C/β^{+}·Thalassaemia** **C/β^{0}·Thalassaemia** or **Hb CC**	Mild haemolytic anaemia	Occasional intermittent abdominal pain Gallstones Fatigue Occasional jaundice Usually a mild condition Genetic counselling is essential during pregnancy for parents 'at risk' of having a child with this condition but prenatal diagnosis is not indicated.	**Register with haematology/ specialist clinic** decision regarding regularity of follow-up is made locally No regular treatment necessary, only required in relation to symptoms Genetic counselling and partner testing for individuals with this condition is important as they do not have any normal Hb A genes.
Haemoglobin D^{Punjab} Disease **Hb D^{Punjab}/D^{Punjab}** **D^{Punjab}/β^{0}·Thalassaemia** **D^{Punjab}/β^{+}·Thalassaemia**	Microcytosis Hypochromia Hb at lower end of normal	Occasional abdominal pain Symptoms related to haemolytic anaemia Gallstones Fatigue Occasional jaundice Iron medication not required unless iron deficient Genetic counselling is essential during pregnancy for parents 'at risk' of having a child with this condition but prenatal diagnosis is not indicated.	**Register with haematology/ specialist clinic** decision regarding regularity of follow-up is made locally No regular treatment necessary, only required in relation to symptoms Genetic counselling and partner testing for individuals with this condition is important as they do not have any normal Hb A genes
Haemoglobin E Disease (Hb EE) **(Hb E/β·Thalassaemia** may be clinically significant, please see information on thalassaemia disorders for further information**)**	Mild haemolytic anaemia Hypochromic	Very mild condition **Genetic counselling is essential during pregnancy for parents 'at risk' of having a child with this condition but prenatal diagnosis is not indicated.** Hb EE and Hb E/β^{0}·Thalassaemia will look similar on the initial screening test and further investigations will be needed for the conditions to be differentiated.	**Register with haematology/ specialist clinic** decision regarding regularity of follow-up is made locally No regular treatment necessary, only required in relation to symptoms Common in South East Asian populations.

Genotype	Anaemia	Other clinical features	Comments
			Genetic counselling and partner testing for individuals with this condition is important as they do not have any normal Hb A genes.

References

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