Appendix 6 – Sickle cell disease

Genotype	Anaemia	**Splenomegaly**	**Clinical features**	**Clinical severity**	**Remarks/treatment**
Sickle Cell Anaemia (Hb SS)	Severe	Yes Usually auto-splenectomy by age 5-7 years	Vaso-occlusive episodes (painful crisis) Bone and joint infarcts Hepato-renal complications Bacterial infections If Hb F is elevated then condition may be milder than usual	Severe	Progressive disability common. Usually shortened life span. Bone marrow transplant may be considered (in children) if there is severe disease and an HLA matched sibling is available. **Treatment:** Oral Penicillin is routinely prescribed for children but is optional for adults Pneumococcal vaccine (adults & children) Folic acid depending on diet and/or local protocol
Haemoglobin SC Disorder **(Hb SC)**	Mild to moderate Hb level is just below normal	Common	Intermittent painful crises Bone and joint infarcts (less common than in SS) Retinopathy in adults May have aseptic necrosis of joints	**Variable**	Severity often increased during pregnancy May present with any of the same symptoms as Sickle Cell Anaemia but condition is usually milder. **Treatment:** Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococ*cal vaccine (adults & children)* Folic acid depending on diet and/or local protocol
S/β thalassaemia This is inclusive of Hb S/β^+, Hb S/δβ, Hb S/γδβ and Hb S/Lepore	Mild to moderate	Occasional	Intermittent pain crises Bone and joint infarcts	**Variable**	**Treatment:** Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococcal vaccine *(adults & children)* Folic acid depending on diet and/or local protocol

Genotype	Anaemia	**Splenomegaly**	**Clinical features**	**Clinical severity**	**Remarks/treatment**
S/β^0 Thalassaemia	Moderate to severe	Common	Intermittent pain crises Bone and joint infarcts Severity depends on type of beta (β) thalassaemia gene inherited and % of Hb F produced	**Moderate to severe**	May be as severe as Sickle Cell Anaemia, depends on the thalassaemia mutation inherited. No normal Hb A. Treatment: Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococcal vaccine (adults & children), Folic acid depending on diet and/or local protocol
Haemoglobin S/D^{Punjab} Disorder ($HbSD^{Punjab}$)	Mild to moderate haemolytic anaemia	Common	Severity of clinical picture is variable but usually milder than sickle cell anaemia	**Variable**	Treatment: Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococcal vaccine (adults & children), Folic acid depending on diet and/or local protocol
Haemoglobin S/O^{Arab} Disorder ($HbSO^{Arab}$)	Mild to moderate haemolytic anaemia		Usually mild to moderate condition	**Variable**	Treatment: Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococcal vaccine (adults & children), Folic acid depending on diet and/or local protocol
Haemoglobin S/E Disorder (Hb SE)	May have normal haemoglobin levels		Usually asymptomatic or very mild	Variable	Treatment: Oral Penicillin is *routinely prescribed* for children but *is optional for adults* Pneumococcal vaccine (adults & children), Folic acid depending on diet and/or local protocol

Genotype	Anaemia	**Splenomegaly**	**Clinical features**	**Clinical severity**	**Remarks/treatment**
Haemoglobin S/HPFH	Mild haemolytic anaemia but haemoglobin level is usually normal		Usually asymptomatic or very mild		**Not distinguishable on newborn screening from** S/βeta^0 thalassaemia or Sickle Cell Anaemia (Hb SS) on initial newborn screen **Usually no regular treatment required** once diagnosis is confirmed Regularity of monitoring by Specialist varies dependent on local protocol

Notes

It is not possible at birth to differentiate with certainty between sickle cell anaemia (HbSS), Hb S/β^0 thalassaemia and Hb S with hereditary persistence of fetal haemoglobin (Hb S/HPFH), since all of these conditions produce only Hb F and HbS on routine analysis.

- Offer genetic counselling and prenatal diagnosis to women/couples at risk of having a baby with any of the above conditions (except S/HPFH)
- Individuals with these conditions should be registered with and followed up regularly by a Haematology Clinic
- Pregnant women with any of the above sickle cell disorders should be considered as being “high risk” and should be followed up by a Haematologist and Obstetrician during pregnancy with booked hospital delivery.

References

Bain BJ (2006) *Sickle cell haemoglobin and its interactions with other various haemoglobins and with thalassaemia.* Haemoglobinopathy Diagnosis, 2nd Edition Blackwall Publishing Ltd

Brent Sickle Cell & Thalassaemia Centre (2012) *A Parent’s Guide to Managing Sickle Cell Disease* https://www.gov.uk/government/publications/sickle-cell-disease-managing-the-condition

NHS Sickle Cell & Thalassaemia Screening Programme (2012) *Handbook for Laboratories.4th Edition.* https://www.gov.uk/government/publications/sickle-cell-and-thalassaemia-screening-handbook-for-laboratories

Thalassaemia International Federation (TIF) *About Sickle Cell Disorders*.http://www.thalassaemia.org.cy/