Sgrinio Smotyn Gwaed
Newydd-anedig Cymru
Newborn Bloodspot
Screening Wales

Sickle Cell Disorder (SCD) Clinical Referral Guidelines for Newborn Bloodspot Screening Wales

Version 2

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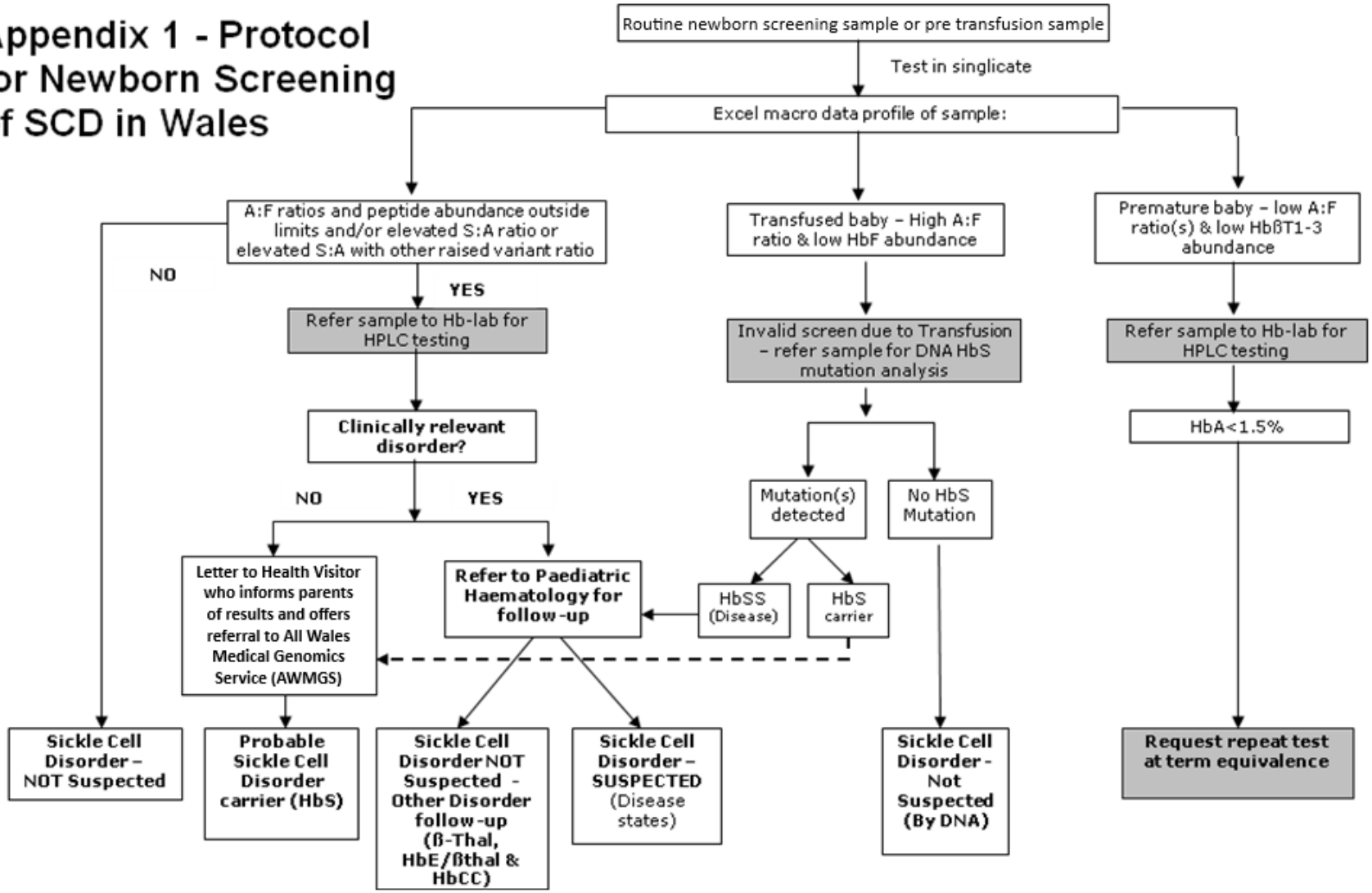
Appendix 1 - Protocol for Newborn Screening of SCD in Wales



Newborn Bloodspot Screening for Sickle Cell Disorder in Wales	The method of screening for Sickle Cell Disorders in Wales is by Tandem Mass Spectrometry. The aim of screening is to identify only Sickle Cell Disorders and not to identify carriers of a Sickle Cell Disorder. Cut offs have been based on ratios between the abnormal to normal haemoglobin peptide abundances ie HbS/HbA, HbC/HbA, HbE/HbA, HbD^{Punjab}/HbA, HbO^{Arab}/HbA, and HbA/HbF (ß-thalassaemia). A post analytical data analysis protocol is used to identify only the clinically relevant Sickle Cell Disorders and the aim is that carrier infants are not identified. Those haemoglobin (Hb) variants for which there is evidence that early intervention is likely to be beneficial and which are therefore specified as part of the UK National Screening programme are the following: Sickle cell anaemia (HbSS) Hb S/ß-thalassaemia* Hb S/HPFH Hb S/C disease HbS/D^{Punjab} , HbS/E, Hb S/O^{Arab}. *This is inclusive of Hb $S/ß^{+}$, Hb $S/ß^{0}$, HbS/δß and Hb S/Lepore. The screening method and data analysis protocol has been approved by the UK NSC to be implemented in Wales.

Sickle Cell Disorder is suspected or other clinically relevant disorder is suspected

Stage of Process	Guidelines
Defining a positive screening result:	If a sample from a baby is found to have an A:F or F:A ratio and peptide abundances outside the limits and/or an elevated S:A peptide ratio the sample is referred to the Cardiff Haemoglobinopathy Laboratory for further testing. If the Haemoglobinopathy Laboratory identifies a clinically relevant Sickle Cell Disorder then **sickle cell disorder is suspected.** If the Haemoglobinopathy Laboratory identifies a clinically relevant by-product e.g. ß-thalassaemia; HbE/ß-thalassaemia or HbCC disease then **sickle cell disorder is Not suspected – other disorder follow-up is reported.** If a sample from a baby that has had a transfusion (either noted on the card or a high A:F ratio and low HbF abundance is identified) and there is no pre-transfusion sample to test then the sample is sent for DNA HbS mutation testing. If DNA HbS mutation testing identifies a clinically relevant Sickle Cell Disorder then **sickle cell disorder is suspected.**

Referral of babies with a positive screening result: **Sickle Cell Disorder is suspected**.	Wales Newborn Screening Laboratory notifies the Paediatric Haematology Team in UHW that a baby has a positive screening result within **one working day** of the result being reported by the Haemoglobinopathy or DNA laboratory. The clinical referral must be both verbally (by telephone) and by an email using a standard letter.

First family contact	It is the responsibility of a consultant/specialist nurse from the Paediatric Haematology Team at UHW to organise the referral of the baby to the appropriate specialist haemoglobinopathy team. **For babies resident in South Wales the paediatric haematology team is based in University Hospital of Wales, Cardiff.** **For babies resident in Betsi Cadwaladr Health Board the paediatric haematology team is based in Liverpool with shared care at Ysbyty Gwynedd.** **For babies resident in mid Wales the paediatric haematology team is based in Birmingham.** It is the responsibility of a consultant/specialist nurse from the Paediatric Haematology Team to organise or undertake the initial contact with the family. This initial contact must be made within **three working days** of the result being reported to the Paediatric Haematology Team. The first contact should be verbally by a consultant/specialist nurse from the Paediatric Haematology team by telephone to explain the positive screening result to the family, to give them initial advice, to reassure and to arrange a face to face follow up meeting. The family must be provided with the 'Sickle Cell Disorder is suspected' leaflet via email or web link. It is the responsibility of a consultant/specialist nurse to make contact with the baby's GP and Health Visitor to inform them of the screening results.

First family face to face review with haematology team (including confirmatory tests)	This appointment should be arranged within **twenty eight working days.** Confirmatory diagnostic samples should be collected at this visit. Samples for diagnostic testing should be sent to a specialist laboratory with expertise in haemoglobinopathy analysis in the newborn period. Premature babies with no HbA (<1.5%) need repeat testing. Penicillin prophylaxis should be started while waiting for clarification of diagnosis. Pneumococcal vaccine is recommended at 12 weeks of age. Pre-diagnosis management should include: • Explanation of the condition • Ensure family have received – specialist team contact details • Ensure family have received available written information including 'SCD is suspected' or other disorders leaflet • Ensure family have received SCD GP letter • Details of the time and location of the next appointment Confirmation of diagnosis is the responsibility of the specialist haemoglobinopathy team. Outcome and results of any further tests undertaken MUST be communicated to the Newborn Screening Laboratory. A medical specialist team for Sickle Cell Disorder should comprise: • A consultant Paediatric Haematologist (with cover in event of annual leave) • A specialist paediatric haematology nurse • Be part of a haemoglobinopathy Co-ordinating centre (HCC) network.

Probable Sickle cell disorder carrier (HbS)/other haemoglobin variant carrier

These are cases that have been identified as a by-product of sickle cell disorder screening.

Probable Sickle Cell Disorder Carrier/ other haemoglobin variant carrier	If a sample from a baby is found to have an A:F or F:A ratio and peptide abundances outside the limits and/or an elevated S:A peptide ratio with or without another raised variant ratio, the sample is referred to the Cardiff Haemoglobinopathy Laboratory for further testing. If the Haemoglobinopathy laboratory identifies the baby as a probable carrier of a sickle cell disorder then **Probable sickle cell disorder carrier is reported**. If the Haemoglobinopathy laboratory identifies the baby as a probable carrier of another haemoglobin variant disorder then **Probable other haemoglobin variant carrier is reported**.
Referral of babies with probable sickle cell disorder carrier/other haemoglobin variant carrier results	Wales Newborn Screening Laboratory is to communicate with the Newborn Bloodspot Screening Coordinators within **two working days** of the result being reported by the Haemoglobinopathy or DNA laboratory.

Family Contact	Parents of babies who are found to be a probable sickle cell disorder (SCD) carrier/other haemoglobin variant carrier are informed of this and the other bloodspot screening results within 6 weeks of sample collection. When the NBSW programme co-ordinators are informed by the laboratory they will notify the health visitor by phone and ask the health visitor to inform the parents of the results. The following information is emailed to the health visitor:- • Health visitor cover letter • Information leaflet for health visitor • Results letter for the parents • Information leaflet for parents The information leaflet for the Health Visitor includes contact details of the All Wales Medical Genomics Service (AWMGS) that parents can be referred to if wanting further information or advice. The Health visitor should offer the parents referral to this service, and if wanted by parents, should make the referral according to the guidance in the information leaflet.