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Pwyllgor Gwasanaethau Iechyd Arbenigol Cymru (PGIAC)

Welsh Health Specialised Services Committee (WHSSC)

Specialised Services
Service Specification: CP179

Sickle Cell Disorders, Thalassaemia Disorders and other Rare Hereditary Anaemias: all ages

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Description	NHS Wales will routinely commission this specialised service in accordance with the criteria described in this policy
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Contents

Statement

Welsh Health Specialised Services Committee (WHSSC) commission services for patients of all ages with sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias in accordance with the criteria outlined in this document.

In creating this document WHSSC has reviewed the requirements and standards of care that are expected to deliver this service.

Disclaimer

WHSSC assumes that healthcare professionals will use their clinical judgment, knowledge and expertise when deciding whether it is appropriate to apply this document.

This document may not be clinically appropriate for use in all situations and does not override the responsibility of healthcare professionals to make decisions appropriate to the circumstances of the individual patient, in consultation with the patient and/or their carer or guardian.

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1. Introduction

This policy has been developed as the Service Specification Proposal to enable the delivery of services for people of all ages with Sickle Cell Disorders, Thalassaemia Disorders and other Rare Hereditary Anaemias resident in Wales. This service will only be commissioned by the Welsh Health Specialised Services Committee (WHSSC) and applies to all children and adults with sickle cell disorders, thalassaemia disorders and certain other rare hereditary anaemias.

Within this policy the term Hereditary Anaemia is used to describe sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias.

1.1 Background

Plain Language Summary

Anaemia is a condition where the number of red blood cells or the amount of haemoglobin in red blood cells is less than normal. Haemoglobin is the substance that makes blood red. Its main purpose is to carry oxygen around the body. If the body does not receive enough oxygen, various symptoms occur. These include tiredness, weakness and lack of energy.

Sickle cell and thalassaemia are hereditary disorders of the haemoglobin in red blood cells. They belong to a group of red cell disorders called haemoglobin disorders, or haemoglobinopathies, of which they are the most common types. Rarer types of haemoglobin disorder occur that may or may not share the same symptoms and often have a different pattern of inheritance to the common types.

People born with a sickle cell disorder or a beta thalassaemia disorder become symptomatic at varying time points after birth. Alpha thalassaemia disorder affects foetal development and will be present at birth. These disorders are not contagious; they are usually inherited from both parents, each having passed on a sickle cell or thalassaemia gene to their child. Rare forms of both, thalassaemia in particular, can be inherited from only one parent, or occur from new changes in haemoglobin genes (called ‘de novo’) during embryonic development. Both common and rare types occur in the population of Wales.

It is possible to be a carrier of a sickle cell or thalassaemia disorder and thereby have an increased risk of having a child with the disorder, without having any symptoms of the disorder itself. This is known as ‘being a carrier’ or ‘having the trait’.

Sickle cell disorder

The main symptoms of sickle cell disorder are anaemia and episodes of pain usually treated at home but requiring hospital admission when particularly severe or accompanied by certain additional symptoms. The pain occurs

when the cells change shape (or 'sickle') after oxygen has been released. The distorted red blood cells then stick together, or to the blood vessel wall, causing blockages in the blood vessels and tissue damage from lack of oxygen. These painful episodes are referred to as sickle cell crisis. The severe pain is treated with strong painkillers such as morphine to control the pain.

People with sickle cell disorder are at risk of complications such as sepsis, sudden severe anaemia caused by sickling and pooling of blood in the spleen, stroke, acute chest syndrome, blindness, bone damage and priapism (a persistent, painful erection of the penis). Over time the sickling-induced damage to organs such as the spleen, kidney, liver, brain, eyes, joints, lungs, and heart can lead to chronic disease and varying degrees of disability. Death can also result from complications of the disorder.

Treatment of sickle cell disorder focuses on preventing and treating complications alongside education, first of parents about signs requiring urgent admission to hospital, and later of patients about prevention and coping strategies. Some patients with a particularly severe illness or who show a propensity for a life-threatening or severely-disabling complication require regular blood transfusions. Additional medication and health monitoring are then required by these patients to prevent excess iron that results from regular blood transfusion from building up in the body. The only possible cure for the disorder is bone marrow transplant but this is only possible for a limited number of affected individuals who have a suitable donor. A medicine called Hydroxycarbamide (or hydroxyurea), can significantly reduce the number of painful crises. Potential new treatments of various types are undergoing clinical trials including several different types of gene therapy.

Thalassaemia disorder

The haemoglobin in red blood cells of adults consists of two main parts bound tightly together called alpha and beta. They are normally produced in equal amounts during red blood cell development in the bone marrow because unbound (free) alpha or beta can damage the red blood cell.

Thalassaemia disorders arise from an imbalance in the production of these two parts of haemoglobin. In alpha thalassaemia there is a reduction or total loss of the alpha part leading to an excess of free beta; in beta thalassaemia there is a reduction or total loss of the beta part leading to an excess of free alpha. The resulting damage to red blood cells enhances their destruction and causes anaemia. There is a large range of severity depending on the types of genetic variation involved. Thalassaemia major is the most severe, requiring life-long regular blood transfusion (usually monthly) for survival. Less severe types, that do not require regular blood transfusions, are called beta- or alpha- thalassaemia intermedia. Alpha thalassaemia intermedia is also known as haemoglobin H disorder or

haemoglobin H disease). Patients with thalassaemia intermedia may require an occasional blood transfusion or short periods of regular blood transfusion when the anaemia is a problem such as during puberty, pregnancy or during surgery.

The main problems associated with thalassaemia arise from the anaemia and its treatment. Problems arising from the anaemia include delayed growth, enlargement of the spleen and liver, expansion of the bone marrow with concomitant increased bone fragility and deformities, enlargement of the heart, gall stones, increased pressure in the blood vessels of the lungs and increased absorption of iron from food that may eventually cause excess body iron that will lead to a condition known as 'iron overload'. In those patients with thalassaemia major who receive regular blood transfusions many problems are ameliorated or possibly avoided altogether except for the excess iron that occurs at a much faster rate because of the large amount of iron contained in each unit of transfused blood.

Excess iron in the body will cause problems with the heart, liver and hormone levels that could lead to serious illness such as cardiac failure, liver cirrhosis, diabetes, hypothyroidism and infertility if its accumulation over time is not prevented by treatment. Chelation therapy is a treatment with medications that prevents the continued build-up of excess iron in the body either from regular blood transfusions or from years of chronic anaemia. It is also used at a higher dose, or in combination with a different iron chelator, to remove excess iron if it is already present in life-threatening amounts. Experience and care are required to get the treatment right for each patient. Regular health monitoring to prevent unnecessary complications and to respond rapidly to those that do occur is essential for keeping patients well. Various different additional medications will be required depending on the state of the individual patient.

Bone disease remains a prominent cause of illness, even in patients with well-managed disorder, and gives rise to considerable pain and disability. The only possible cure for the disorder is bone marrow transplant but this is only possible for a limited number of affected individuals who have a suitable donor and is a hazardous procedure for anyone to undergo. Potential new treatments of various types are undergoing clinical trials including several different types of gene therapy.

Other Rare Hereditary Anaemias

Other hereditary anaemias can be divided into several different groups:

- hereditary anaemias with increased red cell fragility and rapid loss from the circulation[1] including those caused by defective red blood

[1] causal features shared with the sickle cell and thalassaemia disorders

cell metabolism (e.g. Pyruvate Kinase deficiency) and those caused by a defective red cell wall (e.g. hereditary spherocytosis)

- a hereditary inability to produce enough, or any, developing red blood cells (e.g. Diamond Blackfan anaemia, Fanconi Anaemia, and Shwachman-Diamond Syndrome)[2]
- congenital anaemias associated with defective development of red blood cells in the bone marrow[3] the congenital dyserythropoietic anaemias
- hereditary anaemias associated with an abnormal accumulation of iron destined for haemoglobin production in energy-producing particles called mitochondria within the developing red cells of the bone marrow- the sideroblastic anaemias[4]
- hereditary non-sideroblastic anaemias due to defects that prevent iron required for haemoglobin production from getting to, or being used properly by, the developing red blood cells[4]
- hereditary defects depriving developing red blood cells of folic acid or vitamin B12 essential for their division and amplification in the bone marrow
- hereditary/congenital anaemias due to rare complex mechanisms

and

- familial/congenital anaemias of unknown origin.

The predominant causes of these other hereditary anaemias are a shortened red cell life span and damaged red cell development, as occurs with sickle cell and thalassaemia disorders, enabling many of the standards of care for the latter conditions to be applied and expertise to be shared. In others, the deficient production of red cell precursors in the bone marrow or altered handling of elemental iron, an essential component of haemoglobin, requires careful evaluation of treatment options and health surveillance in particular to avoid iron overload, as occurs for the thalassaemias, that not only limits life expectancy but will also limit the choice of treatment for some.

In summary, sickle cell disorders (SCD), thalassaemia disorders and other rare hereditary anaemias, are lifelong chronic diseases that cause complex multi-system medical problems that can be associated with diverse clinical presentations and significant social and psychological challenges. This leads to variable care pathways across the life course, and the need to work with large numbers of different practitioners.

[2] indicates different cause but risk of transfusional iron overload
[3] causal features shared with the sickle cell and thalassaemia disorders
[4] indicates different cause but risk of iron overload

Care and support provided by a multi-disciplinary team, working across sector and agency boundaries is required. This will include health and social care provision, community nursing care, primary health care and secondary/tertiary care in specialist centres.

Epidemiology

Carriers of sickle cell disorders and thalassaemia disorders are protected against death from falciparum malaria and have increased in frequency in parts of the world where malaria is or was endemic[5]. In Wales, the sickle cell disorders and thalassaemia disorders affect primarily people of black and minority ethnicity but also occur at a lower frequency within all other populations including the indigenous British.

Sickle cell disorder is one of the most common single gene disorders in the UK with the number of patients registered and a birth rate both similar to those expected for cystic fibrosis; around 250 babies with sickle cell disorders and 20-30 with thalassaemia disorders are born each year[6]. In England, the National Haemoglobinopathy Registry (NHR) and the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) hold records of most individuals with a sickle cell disorder or a thalassaemia disorder. The January 2019 NHR dataset showed 13,592 living patients altogether: 11,643 with sickle cell disorders, 1,579 with thalassaemia disorders and 370 with other hereditary anaemias[7].

The 2011 UK Census data indicates that people of Black and Minority Ethnic (BME) backgrounds represented 4.4% of Wales' population, an increase from 2.1% in 2001. The relatively young age of that population (in 2011 it accounted for 7.9% of 0-4yr olds), and the sanctuary offered to refugees by many parts of Wales, Cardiff, Newport, Wrexham and Swansea in particular, mean that the number of patients with a sickle cell disorder or a thalassaemia disorder is likely to continue to rise. Moreover, the increasing life expectancy of individuals with these conditions means that services will need to develop to meet the needs of older patients with the additional comorbidities encountered with age. The patient population is widely but unevenly distributed across Wales with the highest numbers in the combined Cardiff and Vale districts which has 12.2% ethnic minority population and 22.4% ethnic minority births[8]. They include some of the poorest and most vulnerable.

[5] Weatherall D, Akinyanju O, Fucharoen S, Olivieri N, Musgrove P (2006), "Inherited disorders of haemoglobin". In: Jamison DT, Breman JG, Measham AR, Alleyne G, Claeson M, et al. (2006) Disease control priorities in developing countries (2nd Edition). New York: Oxford University Press. pp 663–680. https://www.ncbi.nlm.nih.gov/books/NBK11727/

[6] https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/713120/SCT_data_report_2016_to_2017.pdf

[7] 21st January 2019 http://nhr.mdsas.com/wp-content/uploads/2019/01/NumberPatientsDiagnosis.pdf

[8] Calculated from the Census 2011 data tables of population by age and ethnicity

There are about 17 adults and 15 children with thalassaemia and 44 adults and 38 children with Sickle Cell Disorder (SCD) in Wales. Around 3-4 babies could be born in Wales each year with SCD, and 1 baby every two years with thalassaemia[9]. Nearly all SCD affected children born in Wales, and the majority with thalassaemia, will be identified by neonatal testing or newborn bloodspot screening programmes[10]. Other new patients may present through immigration or late diagnosis.

Most other rare hereditary anaemias[11] seem to be distributed across all populations. A notable exception is hereditary spherocytosis, said to be most common in people of north European origins (1 in 5000 according to Orphanet) and rarely seen in people of non-white ethnicity.

Patients with rare hereditary anaemias are scattered throughout Wales and are often diagnosed late and managed by local haematologists and paediatricians. The numbers of patients in this population are unknown but expected to be small.

Population covered

This specification applies to all children and adults with Hereditary Anaemia who require specialist review. This includes patients with relatively common disorders which generally can be managed in General Haematology clinics, but who, from time to time, may need specialist input. In the latter category, for instance, patients with glucose-6-phosphate dehydrogenase (G6PD) deficiency, hereditary spherocytosis, hereditary elliptocytosis, hereditary ovalocytosis will generally not be seen in the specialist service.

Current service

There is currently no nationally commissioned service for these patients. However, a limited service is provided by Cardiff and Vale principally for its resident population and by Liverpool for the north Wales population. A review carried out in 2016 considered that specialist commissioning of haemoglobinopathy services in Wales would help to ensure equity of access to specialist care for all patients of all ages with haemoglobinopathies in Wales[12].

[9] These figures were provided to WHSSC by the UHW haematology service in November 2018

[10] http://www.newbornbloodspotscreening.wales.nhs.uk/home

[11] Orphanet: https://www.orpha.net/consor/cgi-bin/Disease_Search_Simple.php?lng=EN ; ENERCA: https://www.enerca.org/anaemias/index.php & https://www.enerca.org/activities-news/news/106/the-enerca-white-book-recommendations-for-centres-of-expertise-in-rare-anaemias ; OMIM: https://www.omim.org/ ; Gene Reviews: https://www.ncbi.nlm.nih.gov/books/NBK1116/ .

[12] West Midlands Quality Review Service (WMQRS) and the UK Forum on Haemoglobin Disorders (June 2016) Health Services for People with Haemoglobin Disorders, Wales: Cardiff & Vale University Health Board.

Proposed service

The proposed service will ensure equity of access to specialist care for patients of all ages with Hereditary Anaemia. The specialised service will be part of a holistic model of care (see Annex ii).

1.2 Aims and objectives

The aim of this service specification is to define the requirements and standard of care essential for delivering specialised services for people with sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias.

The objectives are to:

- improve timely access to high quality expert care
- reduce levels of morbidity and mortality
- develop further an integrated model of care, and underpinning systems and processes
- improve the care, support and quality of life of patients with sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias
- ensure equitable access to the specialised service
- embed opportunities for patient feedback and wider community involvement in co-designing service developments.

The purpose of this specification is to outline the responsibilities of both the specialist team and the Hereditary Anaemia MDT.

1.3 Relationship with other documents

This document should be read in conjunction with the following documents:

- **NHS Wales**
  - All Wales Policy: Making Decisions in Individual Patient Funding requests (IPFR).
  - Health and Care Standards Digital ISBN: 978-1-4734-3358-8 WG 24729 April 2015
  - NHS Wales Delivery Framework and Reporting Guidance 2018-2019 © Crown copyright 2018 WG33812 Digital ISBN 978-1-78903-167-6A Healthier Wales: our Plan for Health and Social Care
  - Public Health Wales | Strategic Plan 2017-2020
  - Welsh Government. 2015. Welsh Implementation Plan for rare Diseases.

- **WHSSC policies and service specifications**
  - WHSSC Referral Management Policy CPL-008

- **National Institute of Health and Care Excellence (NICE) guidance**
  - Sickle cell disease: managing acute painful episodes in hospital Clinical guideline [CG143] Published: 27 June 2012
  - Spectra Optia for automatic red blood cell exchange in patients with sickle cell disease Medical technologies guidance [MTG28] Published date: March 2016

- **Relevant NHS England policies**
  - Specialised Services for Haemoglobinopathy Care (All Ages)
  - Clinical Commissioning policy: Treatment of iron overload for transfused and non-transfused patients with chronic inherited anaemia 2016

- **Other published documents**
  - UK Thalassaemia Society, Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK. 3rd Edition 2016.
  - Sickle Cell Society, Standards for the Clinical care of Adults with Sickle Cell Disease in the UK, 2nd edition, 2018.
  - Transcranial Doppler Scanning for Children with Sickle Cell Disease Standards and Guidance Second Edition September 2016. UK Forum on Haemoglobin Disorders and NHS England
  - Sickle Cell Disease In Childhood: Standards And Guidelines For Clinical Care 2nd edition October 2010 NHS Sickle Cell and Thalassaemia Screening Programme and the Sickle Cell Society ISBN : 0-9554319-7-2
  - Public Health Wales (2017) Newborn Bloodspot Screening Wales Annual Statistical Report 2016-2017.
  - Anionwu E, Tangayi S & Streetly A (2011) Caring for people with sickle cell disease and thalassaemia syndromes. A framework for nursing staff
  - West Midlands Quality Review Service (WMQRS) (2018) Quality Standards: health services for people with Haemoglobin Disorders (Adult and Child) V3.1
  - Equality and Human Rights Commission (EHRC) (October 2017): A roadmap to racial equality.
  - Better health Briefing 17. The social consequences of Sickle Cell and Thalassaemia: improving the quality of support A Race Equality Foundation Briefing Paper. February 2012

- **Other references**
  - Well-being of Future Generation (Wales) Act 2015
  - Social Services and Well-being (Wales) Act 2014
  - Is Wales Fairer in 2018?
  - Bevan Commission – A New Way of Doing: Delivering a Prudent Model of Health & Care: "The Prudent Keys to Success"

2. Service Delivery

2.1 Access Criteria

The Welsh Health Specialised Services Committee will commission a specialised service for adults and children of all ages with Sickle Cell Disorders, Thalassaemia Disorders and other Rare Hereditary Anaemias that require specialist review. This will include patients with relatively common disorders which can usually be managed in General Haematology clinics, but who may, from time to time, need specialist input as determined by the specialist team. In the latter category, for instance, patients with glucose-6-phosphate dehydrogenase (G6PD) deficiency, hereditary spherocytosis, hereditary elliptocytosis, hereditary ovalocytosis will normally not be seen in the specialist service.

2.2 Service Delivery

Specialised services for sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias include all care provided by specialist teams including inpatient care where the cause of admission is related to sickle cell disorders, thalassaemia disorders and other rare hereditary anaemias, and outreach care when delivered as part of a provider network.

The Specialist Team

The Specialist Team are the red cell specialist consultant haematologists (adult and paediatric) and clinical nurse specialists (adult and paediatric) who review patients with Hereditary Anaemias.

The Hereditary Anaemia MDT

The Hereditary Anaemia MDT should include all members of the Specialist Team but also those who will provide certain aspects of service delivery in the community or hospital setting. In addition to its specialist role, the MDT will provide a coordinated leadership function, liaising with local teams in the delivery of clinical care and promoting a system-wide networked approach. The MDT will be expected to have clear Terms of Reference.

The MDT will be composed of:

- Red cell medical lead (adult and paediatric consultant haematologists)
- Red cell specialist nurse (adult and paediatric)
- General haematologist
- Haemoglobinopathy laboratory lead or deputy scientist
- Genetic counsellor
- Molecular geneticist
- Psychologist
- Social Worker
- Pain team representative
- Community nurse

See applicable standards A3 Expert Multidisciplinary Care and B1 Access to a Comprehensive range of Clinical Specialists Experienced in Treating Patients with Hereditary Anaemia in section 3.1.

An MDT co-ordinator will be required to co-ordinate the Hereditary Anaemia MDT, and provide essential administrative support, which includes, registration of patients onto the National Haemoglobinopathy Register.

The Hereditary Anaemia MDT will:

- agree and monitor compliance with network care pathways and treatment protocols (elective and emergency) for its caseload of patients.
- support the provision of coordinated expert care and advice within the network.
- provide 24/7 specialist advice for any other clinical teams both within the hospital and to other local hospitals. This may be either directly or as part of a shared-care arrangement with other Hereditary Anaemia MDTs, as the aim is to provide equitable access to specialised care.
- support the provision of routine, non-complex care and will be responsible for ensuring that all patients have an annual review of their care and treatment plan and their condition. Wherever the care is provided, it will follow a consistent approach through network, regional and national collaboration.
- demonstrate close working with WHSSC and other providers to capitalise on the expertise available outside of the Hereditary Anaemia MDT (including secondary, primary, community and voluntary) when designing the care pathways and including discharge planning.
- oversee and support the production of a training and development plan for all healthcare staff involved in the delivery of Hereditary Anaemia care in its network area (see also standard A12 Education and Research in section 3.1). The responsibility for resourcing appropriate training for healthcare staff remains with the employing organisations.
- ensure all consented patients are registered on the National Haemoglobinopathy Registry (NHR). The Hereditary Anaemia MDT and newborn screening laboratory will be responsible for ensuring all children identified by the neonatal screening programme are incorporated into the care system via the NHR. The Hereditary Anaemia MDT will make sure that individual records are complete and up to date.
- publish pathways which describe how a patient can access any interdependent medical or non-medical service including social

services, local authorities, public health, educational services, social welfare and voluntary organisations to provide additional support.

- demonstrate that opportunities are provided for receiving feedback from patients, carers and staff involved in all aspects of care (see standard A11 Patient and Carer Engagement in section 3.1). This will include ensuring links with service user groups and community advocacy are in place.
- establish the referral pathway to the National Haemoglobinopathy Panel.

National Haemoglobinopathy Panel (NHP)

The Hereditary Anaemia MDT will have access to the NHP (based in England) which will provide national expert clinical opinion with regard to the treatment of complex patients.

National Haemoglobinopathy Registry (NHR)

This model will be supported by the NHR. This will act as a national repository for:

- data on patient management including annual review
- patient information including details of local Hereditary Anaemia services and contacts
- guidelines and protocols
- educational materials

Hereditary Anaemia MDTs are responsible for entering data on their own patients into the Registry and for data quality.

Community Nursing

According to local provision, and prevalence, specialised community nursing support may be provided at a specialist community centre where there is access to specialist nurses (adult and paediatric)/health visitors, nurse counsellors, outreach nursing teams, and social services. Such a centre may be based in the periphery or at the hospital where the specialist team is based.

The role of the paediatric community nurse would include:

- informing parents/carers in a sensitive manner of the diagnosis to provide accurate information and timely access to prophylactic treatment
- providing ongoing medical and emotional support, educating the family regarding treatment pathway, management of the patient's condition and information on how to prevent illness and complications

- liaising with primary care team and hospital services to facilitate access to community health services, social services, educational services and welfare benefits
- providing guidance, training and support to the patient's family to provide clinical care at home e.g. desferal treatment
- attending the patient's home for blood tests for dosing hydroxycarbamide and pre transfusion requirements
- development of school care plans-educate teachers and pupils on conditions
- support children and families in home management of mild sickle cell crises
- provide supervision after discharge from hospital and frequent hospital admissions.

The role of the adult community nurse would include:

- liaising with the primary care team and hospital services to facilitate access to community health services, social services and welfare benefits
- providing results in the community with new patients to the area
- working with patients and carers at home i.e. providing support to help keep the patient at home when in crisis and when discharged from hospital
- working with GP surgeries to ensure easy access to the specialised service is available to all patients
- working with genetic counsellors regarding genetic and family planning advice
- taking bloods at home if required
- providing teaching in schools, universities, health care events and community support groups
- working closely with community support groups
- liaising with acute nurse in hospital to work on service improvement projects
- being the point of contact for advice about benefits, immigration, social support services.

Please also see A16 Community Nursing in section 3.1

2.3 Acceptance Criteria

The service specification is for patients ordinarily resident in Wales, or otherwise the commissioning responsibility of the NHS in Wales. This excludes patients who whilst resident in Wales, are registered with a GP practice in England, but includes patients resident in England who are registered with a GP Practice in Wales.

2.4 Patient Pathway (Annex i)

Routes into paediatric and adult specialist care include, but are not limited to, ante-natal and neonatal screening, referral from a GP, local and acute hospitals sickle cell and thalassaemia community centres or transfer from other Hereditary Anaemia MDTs. Patients with difficult anaemias will be discussed at MDT before accessing clinics.

Hereditary Anaemias are lifelong conditions and patients will access both on-going routine, as well as specialist, care throughout their lifetime. Patients' care will be delivered as close to home as possible. Whilst rare and sometimes complex, the ongoing and routine monitoring and treatment of patients with Hereditary Anaemia can be managed with the use of protocols, pathways and access to specialist expertise at the Hereditary Anaemia MDT.

The configuration of care provision will be based on networks linked to local prevalence, expertise and availability of service providers, this may include hospitals, community care, primary care and the voluntary sector.

Pathways for paediatric and adult Hereditary Anaemias feature both routine and specialised care.

New patients enter the service either as babies (including those notified through the NHS neonatal Sickle Cell and Thalassaemia Screening Programme[13]), later diagnoses or as new arrivals to Wales notified via GPs, community services, emergency departments and other clinical specialities.

2.5 Service providers/Designated Centres

- University Hospital Wales
  Heath Park Way
  Cardiff
  CF14 4XW

- Children's Hospital for Wales
  University Hospital Wales
  Heath Park Way
  Cardiff
  CF14 4XW

- Royal Liverpool University Hospital
  Prescot Street
  Liverpool
  L7 8XP

[13] Newborn Bloodspot Screening Wales | Sickle Cell Disorders

- Alder Hey Hospital Children's Hospital
  East Prescot Road
  Liverpool
  L14 5AB

2.6 Exceptions

If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If the patient wishes to be referred to a provider outside of the agreed pathway, and IPFR should be submitted.

Further information on making IPFR requests can be found at: Welsh Health Specialised Services Committee (WHSSC) | Individual Patient Funding Requests

3. Quality and Patient Safety

The provider must work to written quality standards and provide monitoring information to the lead commissioner. The quality management systems must be externally audited and accredited.

The centre must enable the patients', carers' and advocates' informed participation and be able to demonstrate this. Provision should be made for patients with communication difficulties and for children, teenagers and young adults.

3.1 Quality Indicators (Standards)

Section A: Core Standards (mandatory)

These are standards that the Hereditary Anaemia MDT must meet directly, i.e. they must have the clinical expertise and facilities within their organisation.

A1 Clinical Leadership

- Medical Leadership – the Hereditary Anaemia MDT will have a named medical lead at consultant level. This must be a haematologist/paediatric haematologist or a paediatrician with expertise in haemoglobinopathies. Dependent on configuration of acute care there may be two medical leads to cover paediatric and adult care.
- The Hereditary Anaemia MDT will have a named medical deputy at consultant level responsible for Hereditary Anaemia care. There may be two deputies i.e. one each for paediatric and adult care.
- Nursing leadership – the Hereditary Anaemia MDT will identify a lead nurse. There may be two deputies i.e. one each for paediatric and adult care. The lead nurse will support all nurses across the Hereditary Anaemia MDT and linked hospitals
- The Hereditary Anaemia MDT will be responsible for data, audit and outcome monitoring for all the patients under its care.

A2 Prevention and management of neurological complications of SCD through transcranial Doppler (TCD) scanning in childhood; specialised neuro- radiology, neurology and neuropsychology services.

The Hereditary Anaemia MDT must be able to demonstrate responsibility for:

- The coordination of access to TCD screening for all eligible children.
- The expert clinical management of those children and adults identified at risk of stroke and other neurological impairment to minimise the risk.

- The multidisciplinary team management of complex neurological abnormalities.
- Compliance with training and quality assurance schemes established to support continuous quality improvement.

A3 Expert Multidisciplinary Care

Expert multidisciplinary care will include, but is not limited to, the following:

- Paediatric and adult out-patient review and care; annual reviews; referral for specialist diagnostic investigations; discussion of disease modifying treatments; discussion of new treatments and new trials; and neurocognitive assessment and review.
- Transition care from paediatric to adult services
- Specialist support and advice on conditions such as transfusion reactions; severe or recurrent painful vaso-occlusive episodes; sickle acute chest syndrome and aplastic crisis
- Multidisciplinary team will review and oversee the overall progress of all patients with clinical complexities to optimise overall care. Note: for children this will include growth, development and academic achievement.
- The MDT should include the following professionals:
    - Red cell medical lead, nursing representation (acute and community). This should be for adult and paediatric.
    - Psychologist
    - Haemoglobinopathy laboratory lead or deputy scientist
    - Genetic counsellor/Molecular geneticist
    - Pain team representative
- The MDT may require input from physiotherapy, neurology, cardiology, general haematology, radiographer and sonographer.
- The MDT should have systems in place for input from a social worker.

The Hereditary Anaemia Specialist Team is responsible for the management of complex patients using a multidisciplinary team approach. Indicators of complexity include but are not limited to:

- Multi-system disease including organ damage.
- Psychological and psychosocial problems.
- Surgery (see standard A8).
- Orthopaedic issues.
- Infection prevention and control requirements.

- Initiation of hydroxycarbamide treatment, blood monitoring and dose escalation as appropriate
- Acute organ failure
- Transfusion management including decisions regarding initiation and cessation of elective transfusion programmes
- Prescription and routine monitoring of iron chelating drugs
- Neurocognitive assessment and review (see standard A2)
- MRI assessment of liver and cardiac iron
- Management of complications related to iron overload and management of endocrine and growth
- Management of complex patients and those with co-morbidities. This may entail working with other Hereditary Anaemia MDTs/HCC to establish expert clinics, such as renal and cardiac
- Advice and referral for stem cell transplant, novel and curative therapies
- Specialist advice for the management of pregnancy in conjunction with expert obstetric teams (see standard A9).

A4 Initiation, Modification and Cessation of Long-Term Transfusion Regimens and Preventative Therapy in Hereditary Anaemias

This standard is associated with standard A5

- The initiation, modification and cessation of long-term blood transfusion regimens should be under the responsibility of the Hereditary Anaemia MDT.
- Regular administration and monitoring of transfusions should be carried out locally wherever possible and sessions should be designed to fit around the requirements of patients wherever possible (e.g. evening clinics and out of hours transfusion).
- Access to automated red cell exchange transfusion for sickle cell patients needing long term transfusion therapy should be available

A5 Initiation, Modification and Cessation of Long-Term Iron Chelation. Monitoring of Complications of Chelation

This standard is associated with standard A5

- The initiation and amendment of long-term iron chelation regimen is the responsibility of the Hereditary Anaemia Specialist Team.
- The regular administration of iron chelation regimen can be carried out locally wherever possible.

- The Hereditary Anaemia Specialist Team will have access to cardiac and liver magnetic resonance scanning (this does not necessarily need to be on site).
- The Hereditary Anaemia Specialist Team will have access to neuro-psychological, psychosocial and social worker support for patients that struggle with adherence.

A6 Acute Management of Severe and Life Threatening Complications of Hereditary Anaemias

The Hereditary Anaemia Specialist Team will develop guidelines to implement the NICE guidance on the management of acute painful episodes. The Hereditary Anaemia Specialist Team will be able to clinically manage or have referral pathways for the following range of complications for SCD:

- Fulminant sepsis.
- Acute sickle lung syndrome.
- Acute splenic or hepatic sequestration.
- Ischaemic and haemorrhage stroke.
- Subarachnoid haemorrhage.
- Acute renal failure.
- Multi-organ failure.
- Billiary obstruction.
- Fulminant priapism.
- Post-transfusion hyperhaemolysis and severe delayed haemolytic transfusion reactions
- Acute ophthalmological complications (for example complications of sickle retinopathy/central retinal artery occlusion).
- Osteonecrosis of major joints (for example hip, shoulder).

The Hereditary Anaemia Specialist Team will be able to manage or have agreed referral pathways the following complications for thalassaemia and rare hereditary anaemias:

- Heart failure and cardiac arrhythmias.
- Infection prevention and control.
- Post-splenectomy sepsis.
- Acute endocrine disturbances (for example hypocalcaemic tetany).
- Acute hepatic decompensation.

The Hereditary Anaemia MDT will offer formal liaison support to any acute provider within its network area.

A7 Long-Term Specific Therapy for Severe Complicated Hereditary Anaemias (Complex Long-Term Conditions Management)

This standard links to standard A3 relating to annual reviews and multidisciplinary team management of complex patients.

Hereditary Anaemia Specialist Team will be able to clinically manage or have referral pathways within the network a range of progressive and often irreversible complications in both outpatient and in-patient settings. In SCD, these include:

- Stroke.
- Chronic sickle lung syndrome.
- Pulmonary hypertension.
- Chronic renal impairment.
- Avascular necrosis of the hips, spine and shoulders.
- Retinopathy.
- Chronic leg ulceration.
- Chronic pain.

In thalassaemia major and intermedia and certain other rare hereditary anaemias, these complications include:

- Endocrine dysfunction (growth hormone deficiency), hypogonadotrophic hypogonadism, hypothyroidism, hypoparathyroidism, diabetes, (which may require insulin treatment).
- Cardiac dysfunction.
- Chronic liver disease (cirrhosis portal hypertension, hepatic failure, hepatocellular carcinoma, often associated with transfusion-transmitted hepatitis B or C).
- Bone problems (avascular necrosis, osteoporotic fractures of the hips and spine, disc disease).
- Gallstones.
- Leg ulceration.
- Iron overload.
- Pulmonary hypertension.
- Thrombosis.
- Retinal damage.
- Pseudoxanthoma
- Chronic pain.

The Hereditary Anaemia Specialist Team must be able to initiate, modify and cease long-term medication regimens. For instance, to prevent or

mitigate sickle painful episodes. The monitoring of such drug regimens is not a specialised function but any modification based on the outcomes of that monitoring remains specialised.

The Hereditary Anaemia Specialist Team must be able to provide psycho-social/psycho-neurological support to complex patients struggling to manage their condition.

A8 Peri-Operative Management Hereditary Anaemias Patients Requiring Surgery

In principle all elective surgery, and where possible all emergency surgery, should be carried out where there is access to the Hereditary Anaemia Specialist Team. For practical purposes this may not be possible or desirable and it will be for the Hereditary Anaemia Specialist Team to agree surgical pathways.

The Hereditary Anaemia Specialist Team will demonstrate close liaison between haematologists, paediatricians, surgeons and anaesthetists.

Where a local acute provider is required to deliver an emergency operation, it should liaise with the Specialist Team.

The Hereditary Anaemia Specialist Team is required to have pathways in place to manage emergency scenarios.

A9 Management of Pregnant Women with Hereditary Anaemias

All women of child bearing age should receive personalised pre-pregnancy and maternity care planning from specialised services. Therefore, women with Hereditary Anaemias must be referred immediately once they are pregnant to a high risk obstetric clinic. In addition the patient must be reviewed early on within their first trimester or at first presentation in accordance with Antenatal Screening Wales protocols by the Hereditary Anaemia MDT to disorder/disease specific aspects of pregnancy management; this must include specialist advice regarding contraception. The individualised care plan must cover the antenatal, intrapartum and postnatal periods. It must include clear instructions for shared care with secondary services, including escalation and transfer protocols and clear guidelines for planned and emergency delivery.

A10 Clinical Governance and Audit

The Hereditary Anaemia MDT will adopt a clinical governance and leadership function. This will include:

- Reporting significant adverse events to WHSSC and the National Haemoglobinopathy Registry (NHR)
- Reporting adverse transfusion events to SABRE/SHOT. Where this happens outside of the specialist centre, the local team will ensure the specialist treating centre is also notified.

- Undertaking an agreed number of clinical/quality audits as per the requirements of UK National Guidelines
- Participating in any peer review process.
- Ensuring compliance with network clinical guidelines and protocols
- Submission of data to support local and national benchmarking (e.g. NHR)

A11 Patient and Carer Engagement

The Hereditary Anaemia MDT will ensure public and patient engagement (PPE):

- User or user group representation at stakeholder engagement meetings
- User involvement in service planning and development

To promote user feedback and engagement with all healthcare providers.

A12 Education and Research

- The Hereditary Anaemia MDT will be able to provide practical training to all relevant clinical staff including junior doctors and nurses and other allied health professionals
- Training for nurses should meet a recognised competency framework for nursing Hereditary Anaemia patients.
- All counsellors or healthcare professionals who counsel couples at risk of an affected pregnancy should have undertaken the relevant training.

Hereditary Anaemia MDT must demonstrate a research portfolio possibly linked to clinical and cost effectiveness of certain aspects of care.

A13 Timely Access to Critical Care

The Hereditary Anaemia MDT must have an Intensive Therapy Unit (ITU) on site – applicable to adults & children.

A14 Transition

The Hereditary Anaemia MDT should develop, provide and oversee a protocol for adolescents transitioning between paediatric and adult services with adequate facilities and staff trained to be sensitive to the special needs of this group of patients and which meets national guidance

A15 Laboratory services

- UKAS accredited laboratory services with satisfactory performance in the relevant NEQAS schemes and MHRA compliance for transfusion should be available at all times (other than genetics services which can be provided in working hours only). Hereditary Anaemia MDT

must be able to access the range of laboratory tests and transfusion support to manage elective and emergency patients.

- Transfusion laboratories must be aware of the special requirements of Hereditary Anaemia patients and ensure that relevant national guidelines have been incorporated into their transfusion guidelines and standard operating procedures.

A16 Community Nursing

- All patients with Hereditary Anaemias should have access to community nursing support.
- The number and case mix of specialist nurses in the community should be regularly evaluated to ensure that services have adequate staffing levels in line with the duties they are undertaking.
- Specialist community nurses should receive appropriate training, supported by certification and competencies to be evaluated as part of their annual professional practice review.
- Clear arrangements for shared care between the community team and local hospital should be in place. This should include multidisciplinary team meetings.

Section B: Collaborative Standards (mandatory)

These are standards that the Hereditary Anaemia MDT may deliver in collaboration with other Hereditary Anaemia MDTs to ensure clinical and cost effectiveness. In addition, some elements will be super-specialised and will be limited to a very small number of providers nationally.

B1 Access to a Comprehensive range of Clinical Specialists Experienced in Treating Patients with Hereditary Anaemia

The Hereditary Anaemia MDT must have demonstrable arrangements in place that recognise the challenges that patients face in travelling long distances, for patient access to the following specialists:

- Experienced nurse specialising in the conditions
- Acute and chronic pain team
- Consultant cardiologist
- Consultant respiratory physician
- Consultant teams with experience in managing pulmonary hypertension
- Consultant nephrologist and access to renal replacement therapy and transplant
- Consultant hepatologist

- Consultant urologist with expertise in managing priapism, erectile dysfunction
- Consultant neurologist and acute stroke service
- Consultant ophthalmologist
- Consultant endocrinologist
- Contraception and sexual health services
- Genetic counselling and fertility services
- Consultant obstetrician
- Consultant general surgeon
- Tissue viability service/leg ulcer clinic
- A psychological care team (e.g. practitioner psychologists (child and adult) and those with neuropsychological expertise) and other mental health services.

B2 Access to Bone Marrow and Stem Cell Transplantation

Both of these interventions are deemed super-specialised and will be available at only a few centres nationally. The Hereditary Anaemia MDT will have formal processes in place to consider patients for such clinical interventions and to refer to the National Panel.

3.2 National Standards

- UK Thalassaemia Society, Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK. 3rd Edition 2016.
- Sickle Cell Society, Standards for the Clinical care of Adults with Sickle Cell Disease in the UK, 2nd edition, 2018.
- Sickle Cell Disease in Childhood: Standards and Guidelines for Clinical Care 2nd edition October 2010 NHS Sickle Cell and Thalassaemia Screening Programme and the Sickle Cell Society ISBN : 0-9554319-7-2
- WMQRS (2018) Quality Standards: health services for people with Haemoglobin Disorders (Adult and Child) V4
- The National Haemoglobinopathy Project: A guide to Effectively Commissioning High Quality Sickle cell and Thalassaemia Services (2011), East Midlands Specialised Commissioning Group.
- WMQRS (2018) Quality Standards: health services for people with Haemoglobin Disorders (Adult and Child) V4
- Royal College of Nursing – Caring for people with sickle cell disease and thalassaemia syndromes – a framework for nursing staff (2011)
- Trans-cranial Doppler Scanning for Children with Sickle Cell Disease –standards and guidance 2nd Edition 2016

- Specialised Services National Definitions Set (SSNDS) 3rd edition – specialised haemoglobinopathy services (all ages) – Definition No. 38 (2009)
- NHS Sickle Cell and Thalassaemia Screening Programme – Handbook for New-born Laboratories January 2017, Handbook for antenatal laboratories Nov 2017
- UK Thalassaemia Society, Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK. 3rd Edition 2016.
- Sickle Cell Society, Standards for the Clinical care of Adults with Sickle Cell Disease in the UK, 2nd edition, 2018.
- Sickle Cell Disease in Childhood: Standards and Guidelines for Clinical Care 2nd edition October 2010 NHS Sickle Cell and Thalassaemia Screening Programme and the Sickle Cell Society ISBN : 0-9554319-7-2
- Standards for the Linked Antenatal and New-born Screening Programme Second Edition (2011), NHS Sickle Cell and Thalassaemia Screening Programme
- Sickle cell disease: managing acute painful episodes in hospital, NICE (2012)
- The National Confidential Enquiry into Patient Outcome and Death
- Understanding the Contribution of sickle cell and thalassaemia specialist nurses: a summary report (2012), NHS Sickle Cell and Thalassaemia Screening Programme
- Transition: Getting it Right for Young People, improving the transition of young people with long term conditions (2006), Department of Health. Gateway reference 5914
- Spectra Optia for automatic red blood cell exchange in patients with sickle cell disease, NICE Medical technologies guidance [MTG28], March 2016

4. Performance monitoring and Information Requirement

4.1 Performance Monitoring

WHSSC will be responsible for commissioning services in line with this service specification. This will include agreeing appropriate information and procedures to monitor the performance of organisations.

For the services defined in this specification the following approach will be adopted:

- Service providers to evidence quality and performance controls
- Service providers to evidence compliance with standards of care

WHSSC will conduct performance and quality reviews on an annual basis.

4.2 Key Performance Indicators

The providers will be expected to monitor against the full list of Quality Indicators derived from the service delivery components described in Section 2.2.

The provider should also monitor the appropriateness of referrals into the service and provide regular feedback to referrers on inappropriate referrals, identifying any trends or potential educational needs.

In particular, the provider will be expected to monitor against the following target outcomes:

- The clinical directorates of adult and paediatric haematology will demonstrate that sufficient medical and specialist nurse time is given to deliver the specification for the benefit of patients with Hereditary Anaemia.
- The Service will provide anonymised minutes of MDT meetings annually. The MDT will be expected to meet every 8 (eight) weeks, although this may vary with incidence of complex cases requiring discussion.
- The Service will arrange educational events for haematology medical trainees, nursing staff and for other departments in its host Health Board or Trust, and other Health Boards, as required.
- The Service will participate in regular peer review cycles as currently governed by the West Midlands Quality Review Service.
- The Service will produce an annual audit plan to include emergency admissions, patient reported outcomes and access to specialist scanning including Trans-Cranial Doppler, and will provide an annual summary to the commissioners.
- The Service will ensure >85% of patients have a documented annual review.
- The Service will deliver an improvement plan to reduce the number of emergency admissions.

- The Service will document its regular meetings with service user groups.

4.3 Date of Review

This document is scheduled for review before TBC, where we will check if any new evidence is available.

If an update is carried out the service specification will remain extant until the revised service specification is published.

5. Equality Impact and Assessment

The Equality Impact Assessment (EQIA) process has been developed to help promote fair and equal treatment in the delivery of health services. It aims to enable Welsh Health Specialised Services Committee to identify and eliminate detrimental treatment caused by the adverse impact of health service policies upon groups and individuals for reasons of race, gender re-assignment, disability, sex, sexual orientation, age, religion and belief, marriage and civil partnership, pregnancy and maternity and language (Welsh).

This policy has been subjected to an Equality Impact Assessment.

The Assessment demonstrates the policy is robust and there is no potential for discrimination or adverse impact. All opportunities to promote equality have been taken.

6. Putting Things Right: Raising a Concern

6.1 Raising a Concern

Whilst every effort has been made to ensure that decisions made under this policy are robust and appropriate for the patient group, it is acknowledged that there may be occasions when the patient or their representative are not happy with decisions made or the treatment provided.

The patient or their representative should be guided by the clinician, or the member of NHS staff with whom the concern is raised, to the appropriate arrangements for management of their concern.

If a patient or their representative is unhappy with the care provided during the treatment or the clinical decision to withdraw treatment provided under this policy, the patient and/or their representative should be guided to the LHB for NHS Putting Things Right. For services provided outside NHS Wales the patient or their representative should be guided to the NHS Trust Concerns Procedure, with a copy of the concern being sent to WHSSC.

6.2 Individual Patient Funding Request (IPFR)

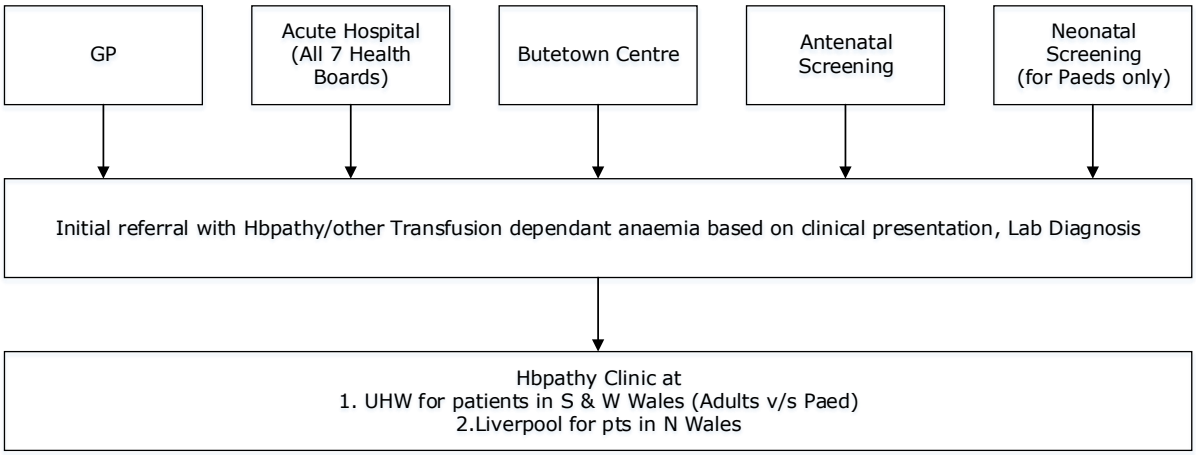
If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If an IPFR is declined by the Panel, a patient and/or their NHS clinician has the right to request information about how the decision was reached. If the patient and their NHS clinician feel the process has not been followed in accordance with this policy, arrangements can be made for an independent review of the process to be undertaken by the patient's Local Health Board. The ground for the review, which are detailed in the All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR), must be clearly stated

If the patient wishes to be referred to a provider outside of the agreed pathway, and IPFR should be submitted.

Further information on making IPFR requests can be found at: Welsh Health Specialised Services Committee (WHSSC) | Individual Patient Funding Requests

Annex i Patient Pathway



Annex ii Service Delivery

It is envisaged that the specialised service will be one element of a holistic care model:

Annex iii Abbreviations and Glossary

Abbreviations

G6PD	Glucose-6-phosphate dehydrogenase
EHRC	Equality and Human Rights Commission
HCC	Haemoglobinopathy Coordinating Centre
IPFR	Individual Patient Funding Request
MDT	Multi-Disciplinary Team
NEQAS	National External Quality Assessment Service
NHP	National Haemoglobinopathy Panel
NHR	National Haemoglobinopathy Registry
NICE	National Institute of Health and Care Excellence
SCD	Sickle Cell Disorder
SC&T	Sickle Cell & Thalassaemia
TCD	Transcranial Doppler
UKAS	United Kingdom Accreditation Service
WHSSC	Welsh Health Specialised Services Committee
WMQRS	West Midland Quality Review Service

Glossary

Individual Patient Funding Request (IPFR)

An IPFR is a request to Welsh Health Specialised Services Committee (WHSSC) to fund an intervention, device or treatment for patients that fall outside the range of services and treatments routinely provided across Wales.

Welsh Health Specialised Services Committee (WHSSC)

WHSSC is a joint committee of the seven local health boards in Wales. The purpose of WHSSC is to ensure that the population of Wales has fair and equitable access to the full range of Specialised Services and Tertiary Services. WHSSC ensures that specialised services are commissioned from providers that have the appropriate experience and expertise. They ensure that these providers are able to provide a robust, high quality and sustainable services, which are safe for patients and are cost effective for NHS Wales.

Haemoglobin

Haemoglobin is the substance in red blood cells that is responsible for the colour of the cell and for carrying oxygen around the body.

Anaemia
Anaemia is caused by a lack of haemoglobin. The symptoms are severe tiredness (fatigue), weakness, shortness of breath, noticeably pounding, fluttering or irregular heartbeats (palpitations), and pale skin.

Chelation Therapy
Treatment with medications to remove the excess iron from the body that builds up as a result of having regular blood transfusions, from iatrogenic iron overload or from having absorbed more iron than needed to compensate for anaemia caused by ineffective red cell production in the bone marrow.