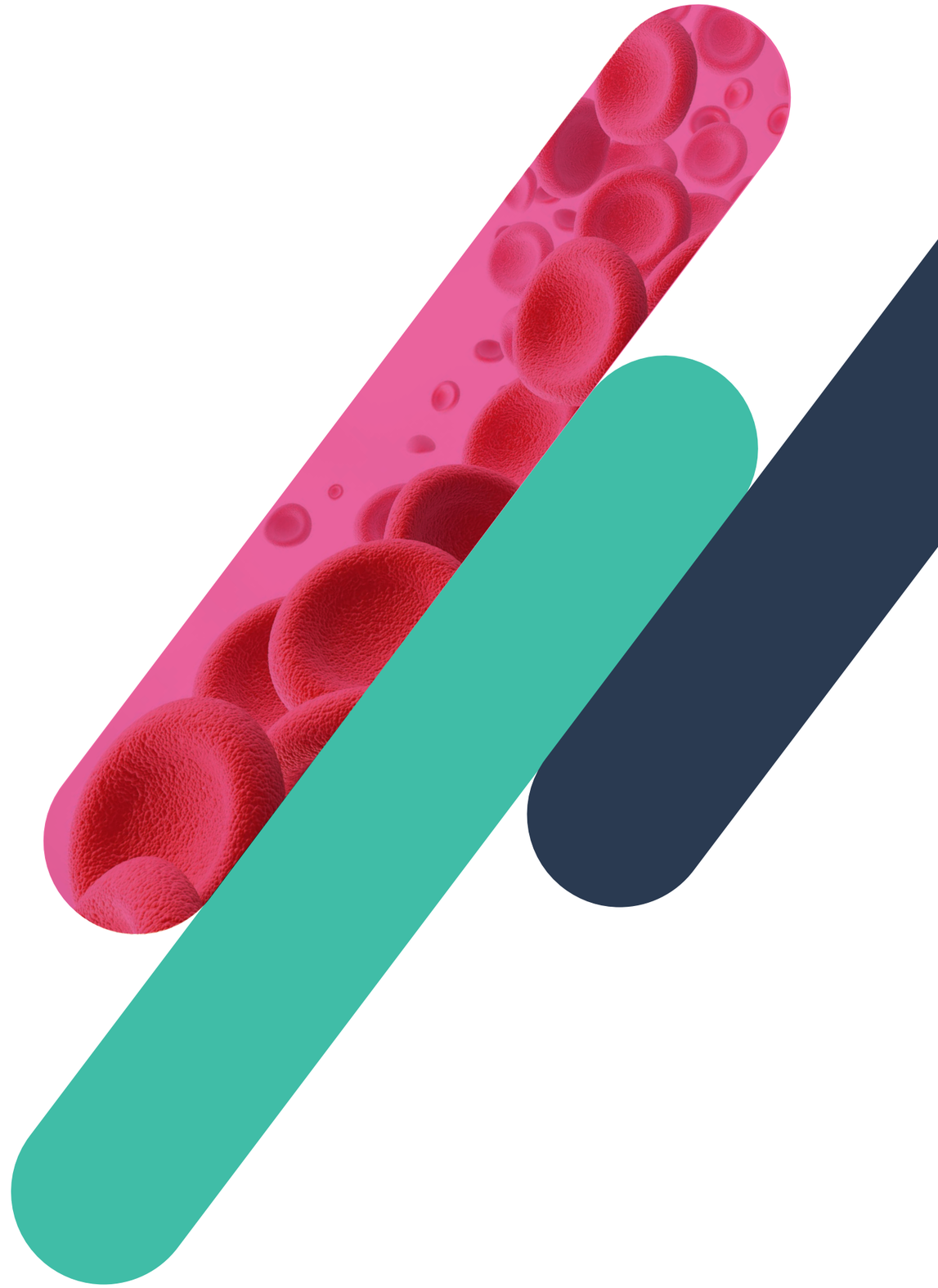
Optimising the UK transplant referral pathway for patients with haemoglobinopathies

WHITE PAPER

This white paper was produced under a collaborative working agreement, with funding and deliverables provided by Vertex.

See further details on page 3.



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FOREWORD

We are pleased to provide this foreword in support of *Optimising the UK transplant referral pathway for patients with haemoglobinopathies*.

Over the past decade, the therapeutic landscape for sickle cell disease and transfusion-dependent β-thalassaemia has evolved at an unprecedented rate. Allogeneic and autologous (gene edited) haematopoietic cell transplantation offer the possibility of a potential cure for selected individuals. Despite national commissioning frameworks and increasing clinical experience, access to these potentially transformative treatments remains variable across the UK.

The recommendations set out in this document offer a thoughtful and pragmatic framework to support greater consistency in referral practice in the UK. The recommendations are designed to strengthen collaboration between red cell and transplant teams and ensure that every eligible patient has equitable opportunity to be considered for potentially curative treatment.

We encourage all Haemoglobinopathy Coordinating Centres, Specialist Haemoglobinopathy Teams, transplant units and commissioners across the UK to review these recommendations and work collectively towards their implementation. As the therapeutic landscape continues to evolve, so must our systems of care. Through shared commitment and collaboration, meaningful progress is achievable.

Yours sincerely,

Professor Josu de la Fuente
Consultant Haematologist
Paediatric Bone Marrow Transplant Programme Director
Professor of Practice Gene and Cellular Therapy
Imperial College London, London, UK

Dr Victoria Potter
Consultant Haematologist
Transplant and Cell Therapy Programme Director
King's College Hospital, London, UK

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PROJECT DELIVERY TEAM

UK Forum for Haemoglobin Disorders (UKFHD) Steering Committee	Dr Rachel Kesse-Adu (Lead, Core committee) Consultant Haematologist, Guy's and St Thomas NHS Foundation Trust Dr Subarna Chakravorty (Core committee) Consultant Paediatric Haematologist, Kings College Hospital, NHS Foundation Trust Dr Amy Webster (Core committee) Consultant Haematologist, University Hospitals Leicester, NHS Foundation Trust Dr Beki James (Core committee) Consultant Paediatric Haematologist, The Leeds Teaching Hospital, NHS Trust Michelle Kenyon (Core committee) Consultant Nurse (BMT), Kings College Hospital, NHS Foundation Trust Dr Ben Carpenter (Core committee) Consultant Haematologist (Transplant specialist), University College London Hospital Dr Shivan Pancham (Core committee) Consultant Haematologist, Sandwell and West Birmingham, NHS Trust Dr Louisa McIlwaine (Core committee) Consultant Haematologist, Glasgow Royal Infirmary, NHS Trust Dr Indu Thakur (Core committee) Consultant Paediatric Haematologist, Children's Hospital for Wales, NHS Trust
Vertex Pharmaceuticals (Europe) Limited	Chloé Bost (Lead, Core committee, Project management) Associate Medical Director, Medical Affairs, Vertex

FUNDING

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EXECUTIVE SUMMARY

Sickle cell disease (SCD) and transfusion-dependent β-thalassaemia (TDT) are inherited disorders of β-globin chain synthesis, characterised by a range of debilitating symptoms and complications, which profoundly impact quality of life. Despite current standard of care, morbidity and mortality remain high for individuals living with haemoglobinopathies compared with the general population. Allogeneic haematopoietic stem cell therapy (HSCT) or autologous gene editing are potentially curative treatment options for some individuals living with haemoglobinopathies.

In the UK, according to the National Haemoglobinopathy Register, there are approximately 15,000 registered individuals living with SCD and 2000 individuals living with β-thalassaemia. Despite National Health Service frameworks and commissioning policies, the process of identification and referral of patients for HSCT is complex, and practice varies across the UK, resulting in inequity of access to these potentially curative options. The National Haemoglobinopathy Panel Annual Report from 2023/2024 indicated that 20% of referrals were not approved. From 2020 to 2024, 76 referrals have been approved, with 24% of these not proceeding to HSCT. Therefore, there is a pressing need to understand and address the potential barriers to transplant for patients living with haemoglobinopathies.

Members of the UK Forum on Haemoglobin Disorders, including paediatric and adult haematologists, consultant nurse experts, and transplant physicians' participated in a survey and then discussed the current SCD and TDT patient transplant referral pathways in a series of three structured virtual meetings to identify potential barriers. Twelve recommendations for improving referral practice were developed, reviewed and revised, and mapped to a proposed optimal patient referral pathway, focussing on regular follow-up, eligibility assessment, tissue typing coordination, patient and family discussions, fertility referral, psychologist referral, benefits/social care assessment, and patient and family consent.

Integration of these recommendations by red cell and transplant teams will ensure that the service for patients with haemoglobinopathies in the UK is increasingly equitable, efficient, and of high quality. To ensure we are successful, this project aims to assess the uptake, implementation, and impact of these recommendations on optimising the referral pathway to transplant.

RECOMMENDATIONS

1. All potentially eligible individuals with haemoglobinopathies and their families should be made aware of the option of HSCT and the eligibility criteria for their age group. They should be offered more information or an initial meeting with a transplant specialist
2. All annual reviews of individuals with haemoglobinopathies should include consideration of HSCT as a therapeutic option; the NHR annual review proforma should be updated
3. HCCs should have a mechanism by which all relevant HCPs in SHTs are aware of, and trained on, the latest eligibility criteria and referral guidelines for individuals with haemoglobinopathies in line with current commissioning policies
4. HCCs should ensure there is a tissue typing lead in all SHTs. SHTs should ensure this lead has appropriate training and dedicated time to screen individuals with haemoglobinopathies and coordinate patient/donor tissue typing
5. Tissue typing should be systematically performed in all individuals with haemoglobinopathies who meet indication criteria, coordinated by the nominated tissue typing lead
6. All individuals being considered for HSCT and their families should have sufficient time to process information, reflect, and make an informed decision through multiple appointments with the red cell and transplant teams
7. HCCs should have a mechanism by which all relevant HCPs in SHTs are able to signpost to fertility preservation services and, where possible, are trained to have discussions around fertility preservation with individuals with haemoglobinopathies considering HSCT
8. There should be clear pathways to fertility preservation services for all individuals with haemoglobinopathies undergoing allogeneic HSCT or autologous gene editing, coordinated by transplant teams, that are accessible long term
9. In line with current recommendations, all SHTs should provide support from a psychologist with specialist interest in haemoglobinopathies, and all individuals being considered for HSCT should be referred for pre-treatment psychological assessment
10. The financial impact of treatment, lengthy hospital stays, and recovery for individuals undergoing HSCT should be considered. SHTs should work with their local benefits/ social worker network or available services to ensure all those being considered for HSCT are appropriately assessed
11. Educational resources should be developed in a range of formats and offered to all individuals with haemoglobinopathies who are considering HSCT in order to support them in making an informed decision
12. Individuals with haemoglobinopathies who are considering HSCT should be referred to peer-to peer support groups, if available, or meetings to discuss HSCT with patients and families who have been through the process

CURRENT LANDSCAPE

Current care pathways for patients with haemoglobinopathies in the UK

In England, specialist and non-specialist haemoglobinopathy services are delivered to adults and children with sickle cell disease (SCD) and β-thalassaemia through a nationally coordinated, tiered model [The National Haemoglobinopathies Project]. This model comprises Local Haemoglobinopathy Teams (LHTs), Specialist Haemoglobinopathy Teams (SHTs), and Haemoglobinopathy Coordinating Centres (HCCs).

LHTs offer both routine and urgent care for patients as close to where they live as possible, including consultant-led reviews, hydroxycarbamide clinics, routine outpatient appointments for bloods, crossmatching, regular transfusions, and emergency presentations and admissions. SHTs provide specialist care across the region as well as managing routine local patient caseloads. SHTs provide annual reviews and specialist services including iron overload scans and input from other specialties, for example endocrine clinics. HCCs organise, run, and lead networks for haemoglobinopathies in each region, ensuring each patient's care is reviewed annually and that there is access to specialist services. The aim of this structure is to improve the experience of all haemoglobinopathy patients, reduce inequities, and improve timely access to high quality expert care [Service Specifications – SHTs, Specialist haemoglobinopathy services (adults and children]].

Transplant therapies for patients with haemoglobinopathies

Established by NHS England in 2020, the National Haemoglobinopathy Panel (NHP) provides national oversight of complex and high-cost care decisions. It hosts monthly national multidisciplinary teams (MDTs) where expert panels review complex patients and referrals for transplant therapies, such as allogeneic haematopoietic stem cell therapy (HSCT) or autologous gene editing [NHP]. Allogeneic HSCT is commissioned nationally in England as a specialised service for specific paediatric and adult patients with haemoglobinopathies meeting certain criteria [Appendix 1, Table 1]. Where required, cases are reviewed by the NHP before a referral is accepted by a transplant centre [Clinical Commissioning Policy HSCT].

Table 1. Transplant options for patients with haemoglobinopathies in the UK

Table 1. Transplant options for patients with haemoglobinopathies in the UK

Transfusion-dependent ß-thalassaemia (TDT)

Transplant type	<12 years	12–18 years	≥19 years
Matched related donor	✓[1]	✓[1]	✓[2]
Haploidentical donor	✓[1]*	✓[1]*	X[2]
Unrelated donor	✓[1]	✓[1]	X[2]
Autologous gene editing	Clinical trial[3†]	✓[4]	✓[4]

Sickle cell disease (SCD)

Transplant type	<12 years	12–18 years	≥19 years
Matched related donor	✓[1]	✓[1]	✓[5]
Haploidentical donor	✓[1]	✓[1]	Clinical trial[6]
Unrelated donor	✓[1]*	✓[1]*	X[5]
Autologous gene editing	Clinical trial[3]	✓[4]	✓[4]

✓: Current option; X: Not currently an option.
*Clinical option requires careful assessment of risks and benefits.[1]
Alternative donor transplants need to be discussed in the NHP meeting.

†Patients ages 2 to 11 years.

1. Currently active BSBMTCT table of indications – paediatric https://bsbmtct.org/wp-content/uploads/2023/02/Paeds-BMT-indications-2022.pdf.
2. NHS England criteria for adults with TDT https://www.england.nhs.uk/publication/clinical-commissioning-policy-allogeneic-haematopoietic-stem-cell-transplantation.
3. CLIMB-141 (NCT03745287) and CLIMB-151 (NCT03655678)
4. Exagamglogene autotemcel Summary of Product Characteristics.
5. NHS England criteria for adults with SCD https://www.england.nhs.uk/publication/allogeneic-haematopoietic-stem-cell-transplantation-for-adults-with-sickle-cell-disease.
6. REDRESS Study. https://www.redresstrial.co.uk.

HOW ARE WE DOING WITH ALLOGENEIC HSCT IN THE UK?

In the UK, there were 15,031 registered patients with SCD, as well as 2143 registered patients with thalassaemia, according to the National Haemoglobinopathy Register (NHR) Annual Report for 2023/2024. According to the UK Forum on Haemoglobin Disorders (UKFHD) peer-reviewed data in 2024/2025, only 64% of patients registered with SCD were reported to receive an annual review, with a higher proportion of reviews in the paediatric population (64% overall; 56% adult; 78% paediatric). A similar picture is reported for thalassaemia (55% overall; 50% adult; 70% paediatric). In addition, current referral pathways for haemoglobinopathies are complex, with the likelihood of multiple barriers reducing the number of potentially eligible patients with access to transplant. According to the 2023/2024 NHP Annual Report, 80% of referrals were approved. In this report, it was found that paediatric referrals were more likely to be rejected than adult referrals. Despite making up about half of the total referrals, paediatric referrals accounted for 75% of those declined and 100% of those delayed for possible later consideration during the financial year. In the period 2020–2024, only 76 referral cases have been approved where transplant was the main request. For those cases where feedback was obtained (n=49), only 76% proceeded to HSCT.

These data underpin the need to understand and address the potential barriers in the treatment of haemoglobinopathies. These barriers mean few patients are given the opportunity to pursue potentially curative treatment, despite the establishment of NHS frameworks and commissioning policies, and almost 25% of those who are successfully referred for transplantation do not proceed to HSCT.

METHODOLOGY

To map the current landscape, this project aimed to generate a consolidated view of current SCD and TDT patient transplant referral pathways through a survey as well as three virtual steering committee meetings with nine members of the UKFHD from England, Scotland, and Wales, representing paediatric and adult haematologists, consultant nurse, and transplant physicians. The objective of this white paper is to provide a consensus of the key challenges facing healthcare professionals (HCPs) and patients for SCD and TDT transplant referral in the UK, enabling the creation of a consolidated list of recommendations to help address these, linked to a proposed optimal patient referral pathway. Considerations of the challenges and optimal pathway during or after transplantation are beyond the scope of this project.

Based on the survey and these meetings with members of the UKFHD, we present 12 expert-informed recommendations for improving referral practice.

Barriers to referral

As previously outlined, the current landscape and data indicate there are likely to be significant barriers to referral for and completion of HSCT, with few patients receiving potentially curative treatment. Following discussion, the key barriers identified by the UKFHD panel can be split into four categories:

1 | Patient-specific barriers

The UKFHD panel indicated that many people with SCD and TDT, and their families, are not fully informed about what HSCT or emerging therapies might offer. As well as this lack of information, it was suggested that variable disease and treatment literacy means the idea of a ‘cure’ is often delayed in the patient journey. Nurse conversations in clinics have highlighted that, when the topic is raised, it is often accompanied by fears about infertility, transplant-related mortality, and long-term graft versus host disease (GvHD). Beyond these concerns, the UKFHD panel stated that these anxieties are magnified for individuals who live far from specialist centres, juggle severe comorbidities, or rely on extended family networks that may have different viewpoints on the need for transplantation. Language barriers and deep-rooted cultural beliefs further complicate truly informed shared decision-making.

2 | Challenges in day-to-day clinical care

Discussions with non-transplant specialists often indicate that there is a lack of awareness of potentially curative options among HCPs. This results in varying confidence when having these conversations, sometimes leading to avoiding the topic altogether. Similarly, some HCPs have preconceived or outdated opinions on curative therapies, filtering out patients perceived as ineligible, who may benefit from such options. Even motivated clinicians may experience confusion around the changing eligibility criteria, treatments, and outcomes. Additionally, they may have limited exposure to complex transplant cases when they practice outside tertiary haemoglobinopathy centres. This results in patchy, location-dependent care.

3 | System capacity and resourcing shortfalls

A number of experts indicated that, even when a patient expresses interest, bottlenecks can quickly appear. Clinical nurse specialist (CNS) and transplant-coordinator teams are overburdened; psychologists, social workers, and fertility services are under-resourced; and transplant beds can be de-prioritised in favour of malignant conditions. The time allocated for annual reviews is insufficient to discuss all therapy options, including potentially curative options.

4 | Fragmented referral

UKFHD panel consensus indicated that, once clinicians and patients agree to proceed, clear, nationwide referral criteria are still lacking. There is no universally adopted protocol stipulating who should be tissue typed, when, and by whom. Turnaround times vary widely, and the process can stall while trying to reach and tissue type potential related donors living overseas. In addition, it was outlined that roles across local hospitals, SHTs, and transplant centres blur, and there is a divergence in the eligibility criteria for children and adults, resulting in complexity as patients transition into adulthood.

In summary, despite the establishment of NHS frameworks and commissioning policies, variation still persists in the timing, process, and frequency with which allogeneic HSCT and autologous gene editing procedures are offered to patients with haemoglobinopathies in the UK. While agreed clinical eligibility criteria exist, their application remains inconsistent across regions and treatment centres. Referral decisions are often influenced by the experience and confidence of individual clinicians within SHTs, as well as the availability and engagement of transplant centres. This variability results in marked inequalities in patient access to potentially curative therapies.

Viewed collectively, these findings paint a clear picture: even a centre with resources will struggle to provide optimal care unless patients and families receive timely, culturally attuned information that empowers them to pursue a potentially curative therapy. Despite an appetite from clinicians for these treatments, there are still systemic barriers such as hesitancy or bias, under-resourced services, and ambiguous referral rules. Addressing any single barrier in isolation is unlikely to move the needle. Therefore, a system-wide response that tackles education, professional practice, resourcing, and national governance is required if more people with haemoglobinopathies are to be given the opportunity to access potentially curative treatment in a timely, equitable manner. As such, there is an important need for a standardised approach to HSCT referral pathways in patients with haemoglobinopathies.

Recommendations

The following sections of this white paper aim to outline the referral pathway for HSCT in haemoglobinopathies and provide recommendations to overcome system-level barriers to enable equitable access to HSCT for patients with haemoglobinopathies in the UK.

The following 12 recommendations are mapped to key steps in the patient referral journey from the identification of eligible patients and recognition of HSCT as an option to transplant readiness and pre-HSCT planning (Figure 1). By improving clarity, consistency, and coordination between red cell and transplant teams, we aim to ensure that all eligible patients, regardless of location, are offered an equitable opportunity to benefit from potentially curative treatment.

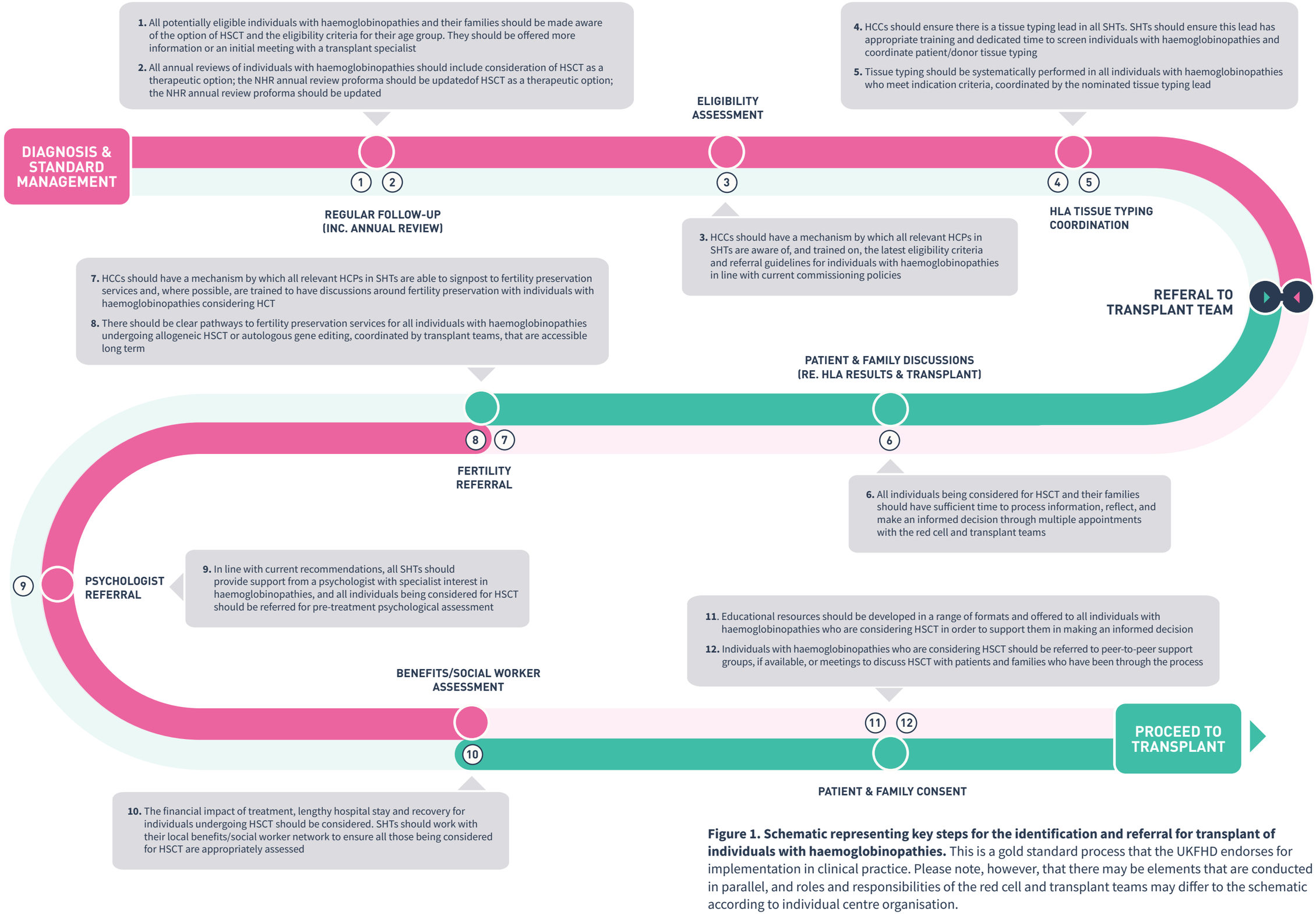


Figure 1. Schematic representing key steps for the identification and referral for transplant of individuals with haemoglobinopathies. This is a gold standard process that the UKFHD endorses for implementation in clinical practice. Please note, however, that there may be elements that are conducted in parallel, and roles and responsibilities of the red cell and transplant teams may differ to the schematic according to individual centre organisation.

Regular follow-up

Recommendation 1. All potentially eligible individuals with haemoglobinopathies and their families should be made aware of the option of HSCT and the eligibility criteria for their age group. They should be offered more information or an initial meeting with a transplant specialist if appropriate.

It is essential that red cell teams initiate proactive, structured conversations about HSCT with all potentially eligible individuals or families who have a child with a haemoglobinopathy during routine clinical follow-up. Discussions should focus on detailing the various age-appropriate eligibility criteria for HSCT. Engaging patients and families early in transparent conversations around risks and benefits, and the evolving treatment options, will help to promote equity in care. Paediatric patients should be included in the discussions in an age-appropriate manner, and this may require inclusion of experienced play specialists, psychologists, and CNS teams. Coordination with transplant teams may be appropriate where more detailed discussions are required, and this could be facilitated through joint red cell and transplant team clinics or MDTs. To support red cell teams with early conversations regarding HSCT, a FAQ document has been developed (Appendix 2).

Recommendation 2. All annual reviews of individuals with haemoglobinopathies should include consideration of HSCT as a therapeutic option; the NHR annual review proforma should be updated

Individuals with haemoglobinopathies are reviewed in regular red cell outpatient clinics and additionally have a comprehensive annual review. Annual reviews are a key element of best-practice care and serve various purposes, including comprehensive health monitoring; review of disease management, laboratory, and imaging investigations; psychosocial and educational support; and care planning and coordination. Discussion of HSCT as a treatment option should be incorporated into the mandated patient annual reviews to facilitate a structured, consistent approach to patient referral. All HCCs should update the annual review proforma to include questions on whether a discussion about HSCT has taken place with the patient and family. Where appropriate, this should also record whether tissue typing has been performed, whether a donor search has been initiated, and whether a donor has been identified. A suggested proforma update has been developed (Appendix 3).

Eligibility assessment

Recommendation 3. HCCs should have a mechanism by which all relevant HCPs in SHTs are aware of, and trained on, the latest eligibility criteria and referral guidelines for individuals with haemoglobinopathies in line with current commissioning policies.

HSCT is commissioned nationally in the UK for selected patients with haemoglobinopathies. Eligibility criteria for HSCT should be clearly defined, readily accessible, and straightforward for clinicians to assess during patient evaluation. All relevant HCPs in SHTs should have awareness of the NHS England/Wales commissioning guidelines and the British Society of Blood and Marrow Transplantation and Cellular Therapy (BSBMTCT) indication tables (Appendix 1). Training on eligibility criteria should be provided to all appropriate HCPs in SHTs who will be having discussions on HSCT with patients and families. Further, all SHTs should have formal processes in place within the network, as agreed by the HCC to discuss and refer patients for HSCT via the NHP (if required). All relevant HCPs in SHTs should be aware of the channels for referral for HSCT in their network [Service Specifications – SHTs].

Tissue typing coordination

Recommendation 4. HCCs should ensure there is a tissue typing lead in all SHTs. SHTs should ensure this lead has appropriate training and dedicated time to screen individuals with haemoglobinopathies and coordinate patient/donor tissue typing.

A nominated team member from each SHT should take responsibility for tissue typing coordination for individuals with haemoglobinopathies. This nominated team member could be a haemoglobinopathies CNS working in collaboration with existing coordinators or it may be appropriate to create a new CNS role focused on pre-transplant work-up. Following appropriate training, and under supervision of a consultant, the nominated lead should be responsible for ensuring all potentially eligible patients have been made aware of HSCT as a treatment option, as per recommendation 1, and that the process of tissue typing is initiated for all appropriate patients and potential donors. SHTs should facilitate the dedication of appropriate time for the designated lead to be trained and to fulfil these responsibilities.

Recommendation 5. Tissue typing should be systematically performed in all individuals with haemoglobinopathies who meet indication criteria, coordinated by the nominated tissue typing lead.

Tissue typing is the process of identifying human leukocyte antigen (HLA) proteins. A recipient and donor must be HLA-matched to minimise the risk of acute graft rejection or acute GvHD and to optimise patient outcomes [Geo 2024]. Tissue typing should be coordinated by the nominated lead in all eligible patients with their consent (as per recommendation 4), and the process of contacting and coordinating potential donors should be initiated. Red cell and transplant teams should discuss and agree who will deliver the results to patients and all tissue-typed relatives or potential donors. It may be appropriate to coordinate monthly joint red cell and transplant team meetings to discuss tissue typing results and next steps. Subsequent detailed discussion of transplant options is the remit of the transplant teams.

Patient & family discussions

Recommendation 6. All individuals being considered for HSCT and their families should have sufficient time to process information, reflect, and make an informed decision through multiple appointments with the red cell and transplant teams.

Patients and families must understand the goals, processes, risks and benefits of HSCT, and weigh these factors against their usual standard-of-care therapy or alternative transplant options, as appropriate [D'Souza 2015]. The decision to proceed with HSCT is complex and differs significantly for patients with malignant disorders compared with those with non-malignant haematological disorders [Tolar 2015]. It is important that individuals with haemoglobinopathies and their families understand the associated lengthy hospital stay, the number of examinations and procedures involved, the intense period of conditioning for HSCT, and the monitoring and management requirements of early and long-term effects of treatment, which they will not be used to [Pawełczak-Szastok 2025]. To facilitate the patient's decision to proceed with HSCT, there should be multiple touchpoints and ongoing dialogue between patients and families and the red cell and transplant teams [Cusatis 2023]. These touchpoints should include time for patients and families to ask questions and access multiformat, age-appropriate educational materials and allow time for the clinical teams to check understanding.

Fertility referral

Recommendation 7. HCCs should have a mechanism by which all relevant HCPs in SHTs are able to signpost to fertility preservation services and, where possible, are trained to have discussions around fertility preservation with individuals with haemoglobinopathies considering HSCT.

Many of the conditioning regimens used for HSCT contain chemotherapeutic agents with known gonadotoxicity, with the risk varying depending on age, sex, and the intensity of chemotherapy [Kenyon EBMT Textbook, Bedrick 2022]. For many individuals with haemoglobinopathies, fertility is a deeply personal and important aspect of future quality of life. As such, all relevant red cell team members should be trained to discuss and answer questions from patients and families on the risks of infertility due to the disease itself or due to therapy, including conditioning regimens required for HSCT. The NHS service specification for SHTs states that, at a minimum, patients and their families should be offered written information, or written guidance on where to access information, covering implications for fertility [Service Specifications – SHTs].

Recommendation 8. There should be clear pathways to fertility preservation services for all individuals with haemoglobinopathies undergoing allogeneic HSCT or autologous gene editing, coordinated by transplant teams, that are accessible long term.

Fertility counselling and preservation are important elements of contemporary comprehensive care for patients with haemoglobinopathies undergoing HSCT. The NHS service specification for SHTs states that all patients should be provided access to a comprehensive range of clinical specialists, including fertility specialists [Service Specifications – SHTs]. Coordination and referral of patients with haemoglobinopathies undergoing HSCT to fertility specialists is the remit of the transplant teams within the framework of the current national hub-and-spoke model. Further, the provision of fertility counselling and preservation services for patients with haemoglobinopathies should be the same as those for malignant diseases in accordance with the NICE Quality Standard [NICE Quality Standard]. The emphasis should be on early referral and access to counselling to ensure informed decision-making about fertility preservation options, including sperm, testicular, egg, or ovarian tissue cryopreservation, depending on the patient's age and gender, ahead of proceeding to transplant.

Psychologist referral

Recommendation 9. In line with current recommendations, all SHTs should provide support from a psychologist with specialist interest in haemoglobinopathies, and all individuals being considered for HSCT should be referred for pre-treatment psychological assessment.

Individuals with haemoglobinopathies undergoing HSCT will face significant psychological challenges before, during, and after treatment. All SHTs should invest in psychology support for all patients with haemoglobinopathies as per current Quality Standards for psychology staffing [Quality Standards, Chakravorty 2024]. The recommendation from the British Psychology Society Special Interest Group for Psychologists working in Sickle Cell and Thalassaemia suggests one whole-time equivalent psychologist for every 300 patients [Quality Standards]. In addition, educational support should be provided to all specialist psychologists to ensure that they are up to date with the advances in transplantation for patients with haemoglobinopathies and the complex medical, emotional, and social needs of patients and families undergoing HSCT. Pre-transplant assessment by a psychologist is essential to ensure optimal outcomes, and there should be a protocol in place for appropriate continuous psychological support for patients throughout the HSCT process, from referral through to post-transplant rehabilitation.

Benefits/social worker assessment

Recommendation 10. The financial impact of treatment, lengthy hospital stays and recovery for individuals undergoing HSCT should be considered. SHTs should work with their local benefits/social worker network or available services to ensure all those being considered for HSCT are appropriately assessed.

HSCT can have a significant financial burden on patients and their families, including indirect out-of-pocket expenses for travel and accommodation as well as loss of income or employment disruption, which can be compounded by the length of hospitalisation [Biddell 2022]. Early financial assessment and referral to the appropriate social worker network, if available, or benefit support services are essential to support families in managing these pressures. It is imperative that continued support and investment are provided to sustain and enhance social welfare and benefits programmes to ensure that individuals with haemoglobinopathies receive the necessary resources and support to navigate the HSCT process and recovery [UKTS and SCS Standards of Care].

Patient & family consent

Recommendation 11. Educational resources should be developed in a range of formats and offered to all individuals with haemoglobinopathies who are considering HSCT in order to support them in making an informed decision.

The UK Sickle Cell Society guidelines state that education of adults with SCD should include increased patient involvement, knowledge, and confidence. [SCS Standards of Care]. Similarly, the UK Thalassaemia Society standards state that education should be delivered to individuals and their families to nurture and empower independence [UKTS Standards of Care]. Concise, age-appropriate, and culturally sensitive information empowers families to make informed decisions, improves treatment adherence, reduces psychological stress and enhances outcomes. [Krist 2017, Hickmann 2022]. Education also fosters collaboration between red cell and transplant teams and families, ensuring that the psychosocial, medical, and practical needs are addressed throughout the HSCT process. A variety of easily accessible, culturally sensitive educational tools in different formats are required to support individuals with haemoglobinopathies and their families, e.g. videos, interactive tools, and printable documents. Existing tools should be utilised where appropriate (Appendix 4); however, additional educational materials will need to be developed to supplement these. Educational tools could be hosted at HCC level, and on the NHS or the NHR website, which could act as a national repository.

Recommendation 12. Individuals with haemoglobinopathies who are considering HSCT should be referred to peer-to-peer support groups, if available, or meetings to discuss HSCT with patients and families who have been through the process.

Peer support programmes are generally more established and utilised in the context of malignancies than other disease areas; they involve the sharing of emotional, educational, and practical help based on lived experience [Amonoo 2022]. For individuals with haemoglobinopathies undergoing HSCT, peer-support meetings may offer experiential empathy, reframe the risk-benefit appraisal and provide a different perspective than can be provided by their clinical teams. SHTs should consider hosting annual peer-to-peer patient support meetings if possible. It may be appropriate in some cases to utilise and refer patients to existing forums e.g. Rare Disease Centre Wales, Sickle Cell Society Support Groups.

FUNDING

HCCs should work with commissioners to agree to a policy for funding tissue typing in all eligible patients (recommendation 5). HCCs should work with commissioners to agree to a policy for funding appropriate psychological support (recommendation 9).

MEASUREMENT

In order to assess the impact of these recommendations accurately, it is essential to understand how they impact the care pathway outlined above. Using currently available data it is not possible to quantify why, out of approximately 17,500 patients registered with SCD or TDT, so few are successfully referred for transplant. Neither is it possible to determine where these patients are being 'lost' in the care pathway. Measuring patient eligibility allows for an evidence-based approach to identify where care gaps exist as well as whether these recommendations have addressed them in a meaningful way at a population level. Figure 2 outlines which datapoints, including those already captured by UKFHD in their Peer Review, we intend to measure to gain a complete understanding of care gaps impacting patients with haemoglobinopathies ahead of transplantation.

DIAGNOSIS & STANDARD MANAGEMENT

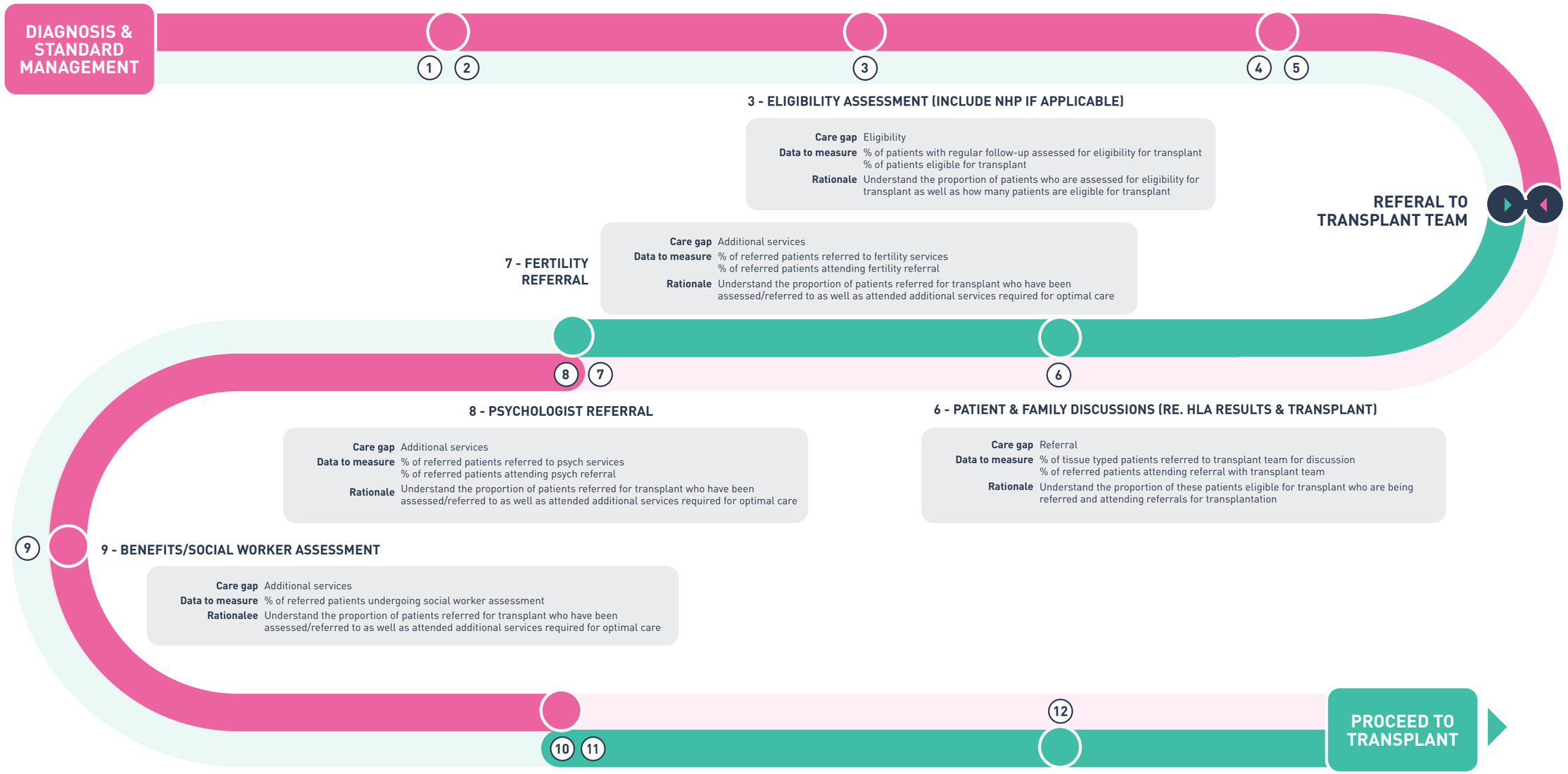
Care gap Diagnosis
Data to measure Size of total prevalent population % of patients diagnosed
Rationale Understand the size of the total population.
All data to be split by SCD/TDT and Adult/Paeds

1, 2 - REGULAR FOLLOW-UP (INCLUDING ANNUAL REVIEW)

Care gap Follow-up
Data to measure % of diagnosed patients with regular follow-up (including annual review)
Rationale Understand the proportion of patients that are receiving regular follow up

4, 5 - HLA TISSUE TYPING COORDINATION

Care gap HLA tissue typing
Data to measure % of eligible patients undergoing HLA tissue typing
Rationale Understand the proportion of patients eligible for transplant who have HLA tissue typing performed



3 - ELIGIBILITY ASSESSMENT (INCLUDE NHP IF APPLICABLE)

Care gap Eligibility
Data to measure % of patients with regular follow-up assessed for eligibility for transplant
% of patients eligible for transplant
Rationale Understand the proportion of patients who are assessed for eligibility for transplant as well as how many patients are eligible for transplant

7 - FERTILITY REFERRAL

Care gap Additional services
Data to measure % of referred patients referred to fertility services
% of referred patients attending fertility referral
Rationale Understand the proportion of patients referred for transplant who have been assessed/referred to as well as attended additional services required for optimal care

8 - PSYCHOLOGIST REFERRAL

Care gap Additional services
Data to measure % of referred patients referred to psych services
% of referred patients attending psych referral
Rationale Understand the proportion of patients referred for transplant who have been assessed/referred to as well as attended additional services required for optimal care

6 - PATIENT & FAMILY DISCUSSIONS (RE. HLA RESULTS & TRANSPLANT)

Care gap Referral
Data to measure % of tissue typed patients referred to transplant team for discussion
% of referred patients attending referral with transplant team
Rationale Understand the proportion of these patients eligible for transplant who are being referred and attending referrals for transplantation

9 - BENEFITS/SOCIAL WORKER ASSESSMENT

Care gap Additional services
Data to measure % of referred patients undergoing social worker assessment
Rationalee Understand the proportion of patients referred for transplant who have been assessed/referred to as well as attended additional services required for optimal care

10, 11 - PATIENT & FAMILY CONSENT

Care gap Transplant
Data to measure % of referred patients consenting to transplant
Reasons for not proceeding to transplant
Rationale Understand the proportion of patients who are consenting to transplant after undergoing all assessments and referrals, as well as the proportion of consenting parents receiving transplant

12 - PROCEED TO TRANSPLANT

Care gap Transplant
Data to measure % of referred patients receiving transplant
Rationale Understand the proportion of patients who are consenting to transplant after undergoing all assessments and referrals, as well as the proportion of consenting parents receiving transplant

Figure 2. Schematic representing key steps for the identification and referral of individuals with haemoglobinopathies for transplant and accompanying data to measure and quantify care gaps.

DISCUSSION

Despite NHS specifications and agreed eligibility criteria from NHS England, NHS Wales, and the BSBMTCT, referral rates for HSCT in individuals with haemoglobinopathies in the UK remain low relative to clinical need. Regional and centre-level variance have been reported in approach to patient selection for HSCT, clinician awareness, confidence, personal experience, referral pathways, and mechanisms to coordinate patients between red cell and transplant teams through the pre-transplant phase. Referral processes are further complicated by differences in local service capacity, unclear roles and responsibilities, and barriers related to funding. A more consistent and patient-centred referral process is essential to ensure that HSCT is considered and offered equitably to all eligible patients with haemoglobinopathies in line with the NHS Long Term Plan to tackle health inequalities [NHS Long Term Plan].

The 12 recommendations developed by the UKFHD panel are intended to streamline and optimise the current patient referral journey to HSCT for patients and families as well as the clinical MDTs involved in their care. Each recommendation has been designed to be practical, achievable, and aligned with existing NHS structures and commissioning arrangements.

It is acknowledged that the recommendations have been developed in the context of significant landscape changes in the management of haemoglobinopathies. The number of transplants performed from 2010 to 2021 based on data from the European Group for Blood and Marrow Transplantation (EBMT) database was analysed; it was found that HSCT for haemoglobinopathies has experienced a sustained growth in the last decade, driven by the number of procedures for SCD across all ages [De la Fuente 2024]. In the UK, the number of transplants undertaken in individuals with haemoglobinopathies is also increasing due to expanded eligibility criteria, increased experience with and patient demand for HSCT, and concerted efforts to improve overall patient quality of life.

CONCLUSION

It is now the time to optimise referral processes to support red cell and transplant teams. As more patients become eligible for potentially curative treatment options, our services must also adapt and evolve. Through integration of these recommendations, we aim to ensure that the service for patients with haemoglobinopathies in the UK remains equitable, efficient, and of high quality.

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ABBREVIATIONS

BSBMTCT	British Society of Blood and Marrow Transplantation and Cellular Therapy
CNS	Clinical Nurse Specialist
CRISPR	Clustered Regularly Interspaced Short Palindromic Repeats
EBMT	European Group for Blood and Marrow Transplantation
FAQ	Frequently asked questions
GvHD	Graft versus host disease
HCC	Haemoglobinopathy Coordinating Centre
HCP	Healthcare Professional
HLA	Human leukocyte antigen
HSCT	Haemopoietic stem cell transplantation
LHT	Local Haemoglobinopathy Team
MDT	Multidisciplinary teams
MHRA	Medicines and Healthcare products Regulatory Agency
NHP	National Haemoglobinopathy Panel
NHR	National Haemoglobinopathy Register
NHS	National Health Service
NICE	The National Institute for Health and Care Excellence
SCD	Sickle cell disease
SHT	Specialist Haemoglobinopathy Team
TDT	Transfusion-dependent β-thalassaemia
UKFHD	The UK Forum on Haemoglobin Disorders

APPENDIX

Appendix 1 – UK commissioning guidelines and indications for HSCT

Allogeneic HSCT is routinely commissioned in the UK for adults with SCD when there is a matched sibling donor and the patient meets eligibility criteria, including history of stroke, repeated acute chest syndrome, or recurrent veno-occlusive crises despite best medical therapy.

NHS England criteria for adults with SCD	https://www.england.nhs.uk/publication/allogeneic-haematopoietic-stem-cell-transplantation-for-adults-with-sickle-cell-disease/
NHS Wales criteria for patients of all ages with SCD (CP224)	https://jcc.nhs.wales/clinical-policies/cancer-and-blood/allogeneic-haematopoietic-stem-cell-transplant-hsct-for-people-of-all-ages-with-sickle-cell-disease-scd-commissioning-policy-cp224-march-2025-pdf-/?ts=1774460201324

Allogeneic HSCT is recommended to be made available as a routine commissioning treatment option for adults with TDT when there is a matched sibling donor and the patient is fit for transplant based on medical, psychological, and social assessment.

NHS England criteria for adults with TDT	https://www.england.nhs.uk/publication/clinical-commissioning-policy-allogeneic-haematopoietic-stem-cell-transplantation/

HSCT for paediatric patients with haemoglobinopathies are commissioned according to the BSBMTCT indication tables.

Currently active BSBMTCT table of indications – paediatric	https://bsbmtct.org/wp-content/uploads/2023/02/Paeds-BMT-indications-2022.pdf

Appendix 2 – Frequently Asked Questions: HSCT

Common questions asked by individuals with haemoglobinopathies and their families around transplant.

Understanding the basics

1. What is a stem cell transplant?
2. If I have a transplant, does that mean I won't carry the sickle or thalassaemia gene anymore?
3. How can a stem cell transplant cure my [sickle cell disease/transfusion-dependent β-thalassaemia]?

Who can have a transplant

4. Is my child [am I] suitable?
5. How do we check if there is a stem cell match in the family?
6. Does the donor need to be a sibling (brother or sister)?
7. What happens if there is no match?

Benefits and risks

8. What are the benefits?
9. What are the risks?
10. What do other patients say is/are the most challenging issue(s) with transplant?
11. Is it better to stay on my current treatment?

The treatment journey

12. What happens before the transplant?
13. What happens in the hospital during the transplant?
14. Why do I need chemotherapy? Will I be able to have children after transplant?
15. What side effects can I expect?
16. What happens after the transplant?
17. How long will my child [will I] be in hospital?

Appendix 3 – Annual review proforma suggested update

Haematopoietic stem cell transplant (mandatory fields)			
Discussion about HSCT option (age-appropriate eligibility criteria)	Yes	No	N/A (provide reason)
Patient medically eligible for HSCT	Yes	No (provide reason)	Unknown
Tissue typing on patient done	Yes	No	Unknown
Donor identified	Yes	No	Unknown
Has the patient accepted the offer of transplant?	Yes	No (provide reason)	Unknown

Appendix 4 – Signposts to existing educational resources

Anthony Nolan	SCD TDT
Sickle Cell Society	https://www.sicklecellsociety.org/resources/
UK Thalassaemia Society	https://ukts.org/resources-for-teachers/ https://ukts.org/healthcare-professionals/

Sources to support initial conversations

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Thank you

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See further details on page 3.