The Hidden Struggle:

Confronting Sickle Cell Disease Inequalities

European Barometer

Commissioned by: Terumo Blood and Cell Technologies

Executive Summary

Sickle cell disease (SCD) is a hereditary blood disorder characterised by abnormal haemoglobin that causes red blood cells to become rigid and sickle-shaped, leading to chronic anaemia, severe pain episodes and potentially life-threatening complications affecting multiple organs.

This **Sickle Cell Disease Barometer** evaluates **how seven European countries (Belgium, France, Germany, Italy, the Netherlands, Portugal and Spain) address key priorities for people living with SCD.** Its **objective** is to provide a comparative overview of national policies and health system readiness, identify structural gaps and support informed dialogue between policymakers, healthcare professionals and patient organisations on improving access to equitable and high-quality care.
The analysis focuses on five core areas: access to care and treatment, workforce training, data and registries, newborn screening and stakeholder collaboration.

Access to care and treatment

Across countries, high-quality SCD care remains concentrated in specialised centres, typically within university hospitals. While children followed in these centres generally receive guideline-based care, transition to adult services remains a structural weakness. Adult patients often experience fragmented monitoring, lower uptake of disease-modifying therapies and reduced access to advanced interventions such as automated red blood cell exchange, transplantation and emerging gene-editing therapies.

Training and workforce capacity.

Paediatric expertise in SCD is generally strong across countries. However, healthcare professionals in adult care, primary care and emergency departments often receive limited structured training in SCD management. This contributes to variability in crisis management, delays in analgesia and inconsistent adherence to recommended care pathways.

Data and registries

Data systems remain fragmented. Although some countries maintain national registries, coverage is often voluntary, adult participation remains limited and systematic linkage between genotyping data and long-term clinical outcomes is inconsistent. These limitations restrict the ability of health systems to monitor disease burden, evaluate policy interventions and assess the impact of new therapies.

Newborn screening

Newborn screening represents one of the most significant areas of variation between countries. Universal screening programmes enable early diagnosis and timely referral to specialist care, whereas targeted or regional approaches can lead to delayed identification and avoidable complications. Even where screening programmes exist, integration with long-term follow-up and registry systems is often incomplete.

Stakeholder collaboration and governance

Strategic governance of SCD is typically embedded within broader rare disease frameworks. While collaboration between clinicians, policymakers and patient organisations exists in many countries, few have established dedicated national implementation measures or formal governance structures focused specifically on SCD.

Overall, the findings reveal a common European pattern: centres of excellence delivering high-quality care alongside systemic inequities in access, adult continuity of care, data integration and national coordination.

Methodology

This **Sickle Cell Disease Barometer** assesses how national health systems address key policy and care priorities for people living with sickle cell disease (SCD). The analysis covers seven European countries: **Belgium, France, Germany, Italy, the Netherlands, Portugal and Spain.** These countries were selected because they represent some of the highest SCD prevalence levels in the European Union and therefore face particular policy and healthcare system challenges related to the diagnosis, treatment and long-term management of the disease.

The assessment is based on a structured framework of policy goals and Key Performance Indicators (KPIs). Five policy areas were examined: **equitable access to SCD care and treatment, workforce training and expertise, data and registry infrastructure, newborn screening and stakeholder coordination.** These areas were selected because they represent core components of effective SCD care systems. Together, they reflect key stages of the patient pathway, from early detection and diagnosis to long-term clinical management, data collection and health system governance.

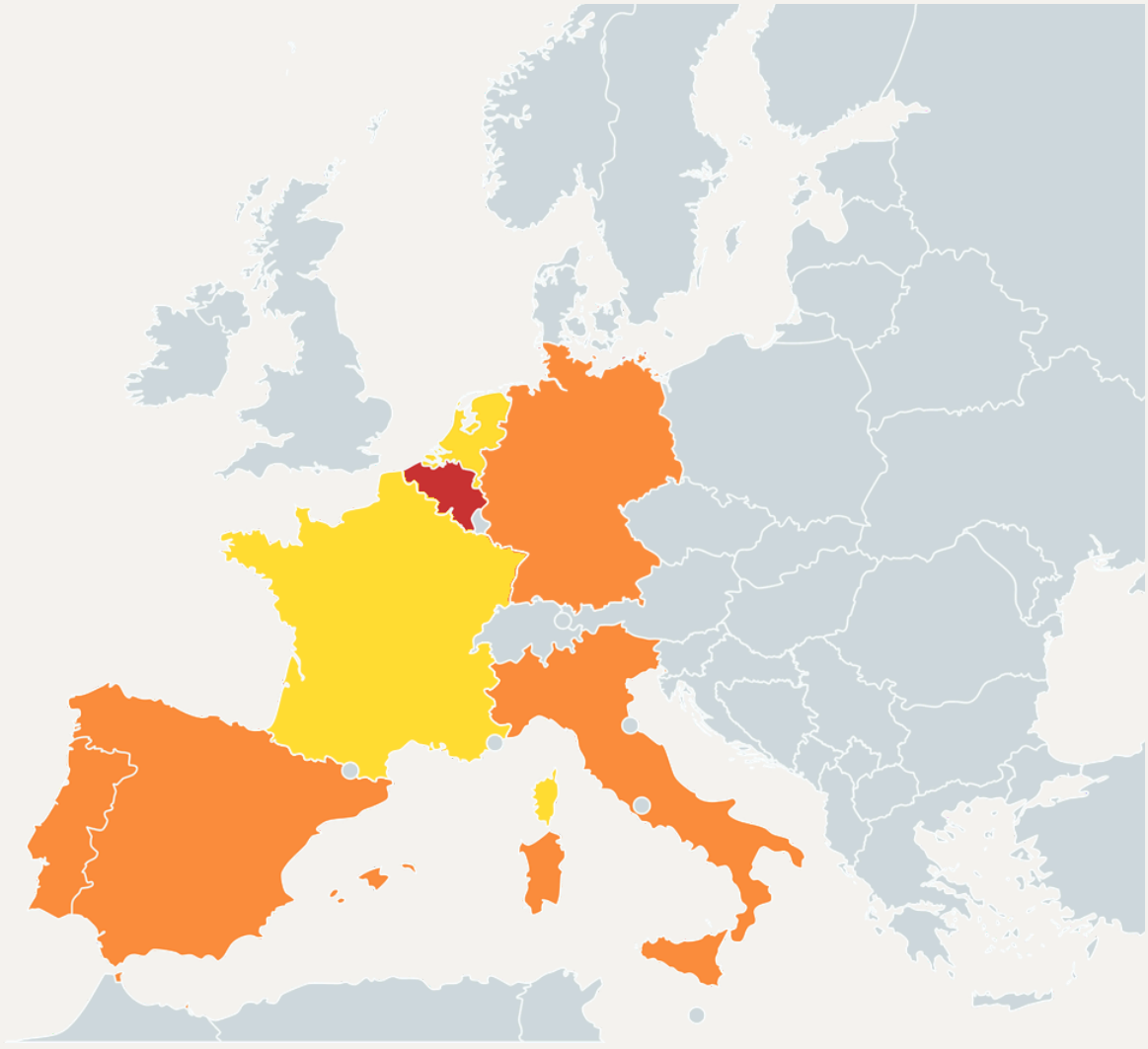
Within each policy area, specific KPIs were used to evaluate the presence, implementation and effectiveness of relevant policies, services and governance mechanisms. The analysis is based on publicly available sources, including national health strategies, rare disease plans, clinical guidelines registry reports, newborn screening programmes and peer-reviewed publications. Information from patient organisations, professional societies and institutional reports were also considered.

Each KPI was assessed using a standardised scoring system reflecting the level of implementation observed in each country. Scores range from 1 (low implementation) to 5 (high implementation) and take into account factors such as the existence of national policies, the degree of nationwide coverage and the consistency of implementation across the healthcare system.

In the Barometer visuals, results are represented using a colour gradient from red to yellow to green. Red indicates limited or absent implementation, yellow reflects partial implementation or emerging policies, and green represents strong or well-established implementation.

The Barometer is not intended to rank countries but to highlight policy strengths, identify structural gaps and support informed dialogue between policymakers, healthcare professionals and patient organisations on improving care and outcomes for people living with sickle cell disease.

Overview of selected countries



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Belgium
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

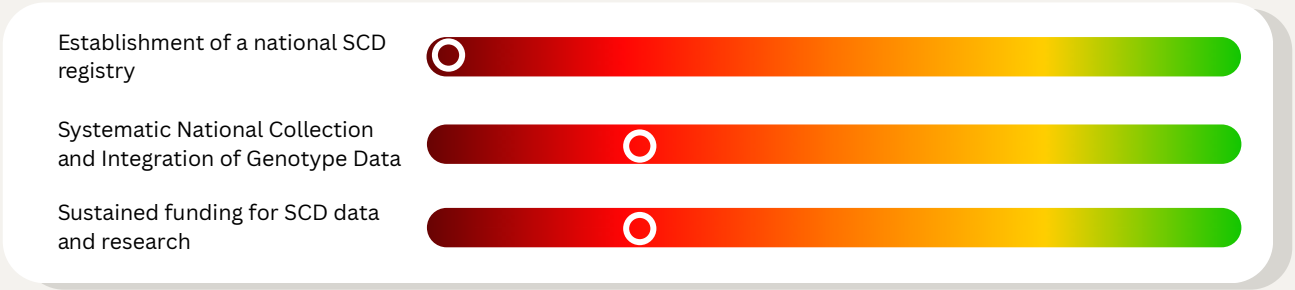
Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment	●	●	●	●
2. Strengthened National Workforce Capacity and Preparedness for SCD	●	●		
3. Comprehensive National Data, Registry and Research Infrastructure	●	●	●	
4. Early detection through newborn screening	●	●	●	
5. Coordinated policy and patient partnership	●	●		

Key Performance Indicator (KPI) Overview

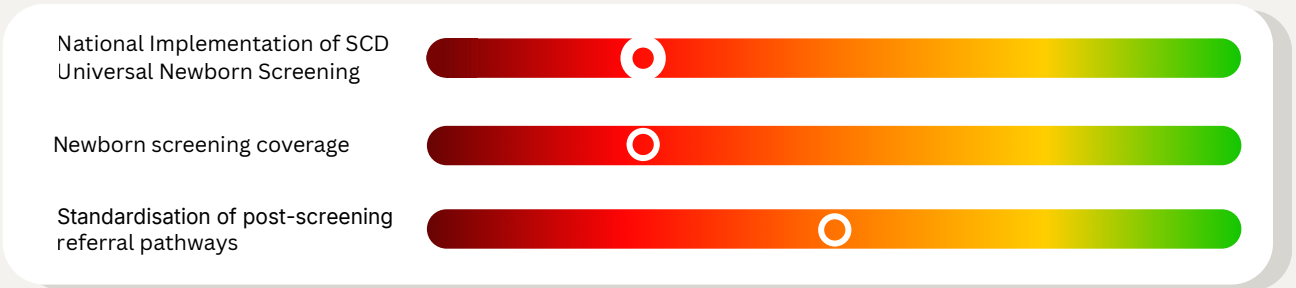
1. Equitable access to SCD care and treatment

2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure



4. Early detection through newborn screening



5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	Belgium has a Rare Diseases Plan but no SCD-specific plan with implementation measures; governance fragmentation across federal/regional levels creates gaps, especially at transition to adult care.	
Availability and Access to SCD Treatment	HU is reimbursed and widely available; transfusion programmes exist but adult follow-up and iron chelation are inconsistent; HSCT mainly paediatric; CASGEVY authorised at EU level, not yet implemented nationally; Adakveo and Oxbryta withdrawn in EU.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX available primarily in academic centres (e.g., UZ Leuven); many regional hospitals lack apheresis capacity; standardised protocols and training needed to mitigate geographic inequities.	
Access to curative treatment	HSCT offered in few university centres (e.g., Erasme); donor availability and transplant risk constrain access; procedures mostly paediatric, adult uptake minimal.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Strong paediatric expertise in university hospitals, but SCD not consistently embedded in broader curricula (adult haematology, GP, internal medicine); CME pilot exists but no national standardisation.	
Emergency care training and protocols for SCD crises	ED responses vary; local protocols (e.g., CHR Liège) exist, but no national standard; patients report delays in analgesia and variable crisis management outside specialised centres.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	No comprehensive national registry; Sciensano pilot and EU contributions (RADeep/EuroBloodNet) exist, but adult data underrepresented and no mandatory reporting.	
Systematic National Collection and Integration of Genotype Data	Genotyping performed in academic care, but not systematically collected or integrated nationally; limits genotype–phenotype analyses and stratified follow-up.	
Sustained funding for SCD data and research	Publications and projects rely on academic/hospital funds; no dedicated recurring federal stream for SCD registries/long-term outcomes.	
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	SCD joined the FWB systematic screening programme (ONE) in January 2023; implementation across maternities is uneven and Flanders does not screen, leading to regional inequities and delayed diagnosis.	
Newborn screening coverage	Coverage is limited to newborns in maternities participating in the FWB programme; a large proportion of Belgian newborns are not screened at birth, especially in Flanders.	
Standardisation of post-screening referral pathways	Where screening is performed, referrals to ULB Erasme/CHR Liège are structured, enabling early education/prophylaxis; where it is not, diagnosis after crises delays access.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	No formal multi-stakeholder national platform; coordination is ad hoc though Belgium participates in ERN-EuroBloodNet/RADeep.	
Inclusion of Patient Organisations in decision-making	ADB plays a central advocacy role, but formal inclusion in policy-making is limited; engagement often mediated through broader rare-disease structures.	

The Hidden Struggle: SCD in Belgium

I. Equitable access to SCD care and treatment

Belgium provides access to most evidence-based therapies for Sickle Cell Disease (SCD), but access is uneven across age groups, regions, and care settings. Paediatric patients followed in specialist university centres generally receive guideline-based care, whereas adults face fragmented provision, limited experience among local providers, and geographic disparities in advanced treatments.[1]

1. **National plan for rare diseases and SCD-specific measures**

SCD is recognised as a rare disease in Belgium, but there is no SCD-specific national plan with concrete implementation measures for diagnosis, treatment, and long-term follow-up. Instead, care depends on a combination of the Belgian Rare Diseases Plan, regional expertise (notably in Brussels and Liège), and local initiatives. The lack of a dedicated implementation framework contributes to gaps at transition to adult care, where responsibilities shift to haematologists or general practitioners who may have limited SCD experience. This results in discontinuity of care and underuse of hydroxyurea among adults.[2]

2. **Availability and Access to SCD Treatment**

Hydroxyurea (HU) is reimbursed and available as a first-line disease-modifying therapy under Belgium's compulsory health insurance.[3] Registry and clinical data show high uptake in children managed in university hospitals, but lower use in adults, highlighting a major drop-off after transition from paediatric services.[4] Chronic transfusion programmes are used for high-risk paediatric patients, but coverage and long-term iron chelation require better follow-up in adults.[5] Access to novel disease-modifying drugs is limited: crizanlizumab (Adakveo) and voxelotor (Oxbryta) were withdrawn at the EU level in 2023–2024.[6] [7]Gene-editing therapies (CASGEVY) have received EU authorisation for severe SCD, but Belgium must establish ATMP centres and reimbursement pathways before routine access becomes a reality.[8]

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[1] Association Drépano Belgique (ADB). (2023). Rapport sur le parcours de soins des patients adultes drépanocytaires en Belgique. Retrieved from https://www.drepanobelgique.be/reports/2023-adult-pathway-report
[2] Sciensano & SPF Santé Publique. (2023). Plan national maladies rares – suivi des actions sur la drépanocytose. Retrieved from https://www.health.belgium.be/fr/plan-national-maladies-rares-2023
[3] RIZIV/INAMI. (2024). List of reimbursable medicines – Hydroxycarbamide for sickle cell disease.
[4] Vandenplas, Y., & Rozen, L. (2021). Management of haemoglobinopathies in Belgian paediatric centres.
[5] Service du Sang de la Croix-Rouge de Belgique. (2025). Drépanocytose et don de sang.
[6] EMA. (2024a). Withdrawal assessment report: Adakveo (crizanlizumab).
[7] EMA. (2024b). Withdrawal assessment report: Oxbryta (voxelotor).
[8] EMA. (2023). CASGEVY (exagamglogene autotemcel) – European Public Assessment Report.

3. **Automated Red Blood Cell Exchange (aRBCX)**

Red blood cell exchange (RBCx) is recognised as essential for stroke prevention, recurrent vaso-occlusive crises, and severe anaemia. In Belgium, automated RBCx is available primarily in specialised academic centres (e.g., UZ Leuven), where apheresis infrastructure and trained staff are concentrated, but some smaller hospitals also have aRBCX available. Many regional hospitals, however, lack the capacity to offer automated exchange, resulting in geographic inequities in access to optimal transfusion strategies. Standardisation of transfusion protocols and wider training on aRBCX are necessary to ensure more equitable provision.[9]

4. **Access to curative treatment**

Curative options remain restricted to a small subset of patients. HSCT is available at a limited number of university centres (e.g., Hôpital Erasme), and eligibility is constrained by donor availability and transplant risk. Procedures are mainly performed in paediatrics, with adult uptake remaining very limited.[10][11]

Belgium offers core therapies for SCD, including hydroxyurea and transfusion programmes, but **faces major gaps in equitable access.** Automated RBC exchange and curative HSCT are restricted to a few academic centres, while gene therapy is emerging but not yet implemented. **The absence of an SCD-specific national plan and fragmented governance exacerbate disparities**, particularly for adults and patients outside major cities. Addressing these gaps requires s**tructured transition care, expanded apheresis capacity, and clear reimbursement pathways for advanced therapies.**

II. Strengthened National Workforce Capacity and Preparedness for SCD

The level of SCD-specific expertise in Belgium is high in paediatric specialist centres, but much more variable in adult haematology, general practice, and emergency medicine. This uneven training profile directly affects access to guideline-based care, timely pain management, and prevention of complication. [12][13]

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[9] University Hospital Leuven. (2025, July 1). Red blood cell exchange. Retrieved from https://www.uzleuven.be/en/haematology/diseases-and-treatments/red-blood-cell-exchange

[10] Hôpital Erasme–ULB. (2022). Activité de greffe et indications pour les hémoglobinopathies.

[11] Vandenplas, Y., & Rozen, L. (2021). Management of haemoglobinopathies in Belgian paediatric centres.

[12] Association Drépano Belgique (ADB). (2022). Recommandations pour la formation des urgences et médecins généralistes. Retrieved from https://www.drepanobelgique.be/resources/2022-training-recommendations

[13] Ibid.

[14] Vandenplas, Y., & Rozen, L. (2021). Management of haemoglobinopathies in Belgian paediatric centres. Belgian Journal of Paediatrics, 23(2), 81–86. Retrieved from https://www.bjp-paediatrics.be/article/view/haemoglobinopathies-management-belgium

1. **Integration of SCD into routine clinical training**

In paediatric haematology, SCD is well integrated in specialist care and supported by structured protocols in university hospitals.[14] Paediatric centres frequently provide hydroxyurea therapy, transfusion programmes, and TCD screening, reinforcing clinical competence and adherence to best practice.

However, **SCD is not consistently embedded in broader medical curricula, including general internal medicine, family medicine, and adult haematology.** Many adult haematologists and general practitioners encounter relatively few SCD patients and may be less familiar with modern disease-modifying therapies, transfusion strategies, and long-term organ protection. As a result, adult patients frequently report gaps in knowledge, delayed referrals, and inconsistent monitoring outside centres of excellence [15][16]

2. **Emergency care training and protocols for SCD crises**

Emergency departments are critical touchpoints for SCD, but responses are often inconsistent. **Patients report delays in pain relief and variable understanding of acute complications** such as acute chest syndrome or severe infections. The absence of nationally standardised emergency protocols for SCD means that **practice differs significantly between hospitals, from triage and analgesia timing to antibiotic use and escalation of care.** Local protocols developed by CHR Liège and Erasme Hospital could serve as national models, but mandatory inclusion in emergency medicine training is still missing. [17][18]

Belgium's **training landscape for SCD is strong in paediatric specialist centres but weak in adult care and emergency medicine,** creating inequities in timely and effective management. **National standardisation of curricula and emergency protocols, combined with structured CME programmes**, is essential to improve knowledge among general practitioners, adult haematologists, and ED teams—ultimately reducing preventable complications and improving patient outcomes.

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[15] Association Drépano Belgique (ADB). (2023). Rapport sur le parcours de soins des patients adultes drépanocytaires en Belgique. Retrieved from https://www.drepanobelgique.be/reports/2023-adult-pathway-report
[16] Association Drépano Belgique (ADB). (2022). Ibid supra.
[17] Université de Liège–CHR Liège. (2022). Protocoles urgences – drépanocytose chez l'adulte et l'enfant. Retrieved from https://www.chr-liege.be/fr/professionnels/protocoles-urgence-drepanocytose
[18] Association Drépano Belgique (ADB). (2022). Recommandations pour la formation des urgences et médecins généralistes. Retrieved from https://www.drepanobelgique.be/resources/2022-training-recommendations

III. Comprehensive National Data, Registry and Research Infrastructure

Belgium contributes to European registry initiatives through platforms such as RADeep and ERN-EuroBloodNet, which aim to harmonise data collection on rare anaemias across member states. However, Belgium's participation is limited by the absence of a comprehensive national registry, resulting in fragmented datasets and underrepresentation of adult patients. This constrains Belgium's ability to monitor outcomes, complications, and costs at both national and EU levels.[19]

1. **Establishment of a national SCD registry**

Belgium **does not yet have a national SCD registry.** Data are fragmented across hospital databases and rare disease networks, with partial contributions to Sciensano's pilot registry and European initiatives. Current estimates suggest several hundred patients, concentrated in Brussels, Antwerp, and Liège, but lack of mandatory reporting means policymakers and clinicians lack comprehensive epidemiological visibility. This limits the ability to track long-term outcomes, transition-related gaps, and mortality in early adulthood.[20][21]

2. **Systematic National Collection and Integration of Genotype Data**

Belgium's current registry efforts **do not systematically capture genotyping data for all patients and integration with hospital information systems remains incomplete.** This limits genotype–phenotype analyses and the ability to stratify patients for advanced therapies or targeted follow-up. Academic centres perform genotyping for clinical care, but data are not centralised nationally, reducing opportunities for research and personalised medicine.[22][23]

3. **Sustained funding for SCD data and research**

Belgium has produced important publications on haemoglobinopathies and SCD epidemiology, supported mainly by academic and hospital-based research funds.[24]

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[19] EuroBloodNet & RADeep Consortium. (2023). European Rare Anaemia Database Annual Report 2023 – Belgium country summary. Retrieved from https://eurobloodnet.eu/research/radeep/annual-reports

[20] Sciensano. (2022). National Hemoglobinopathies Monitoring – Pilot Registry Report 2022. Retrieved from https://www.sciensano.be/en/reports/hemoglobinopathies-registry-pilot-belgium

[21] Aguilar Martinez, P., et al. (2014). Haemoglobinopathies in Europe: health and migration policy perspectives. Orphanet Journal of Rare Diseases, 9, 97.

[22] Sciensano. (2022). National Hemoglobinopathies Monitoring – Pilot Registry Report 2022.

[23] EuroBloodNet & RADeep Consortium. (2023). European Rare Anaemia Database Annual Report 2023 – Belgium country summary.

[24] Aguilar Martinez, P., et al. (2014). Haemoglobinopathies in Europe: health and migration policy perspectives.

[25]However, **there is no dedicated, large-scale, recurring federal funding stream for SCD registries or long-term outcome studies,** unlike some other rare diseases. Registry expansion and linkage to health insurance and hospital data would significantly enhance Belgium's contribution to EU-wide monitoring and comparative policy evaluation.[26]

Belgium's contribution to EU-wide SCD monitoring is **limited by the absence of a national registry and systematic genotyping data collection.** Research output exists but relies on ad hoc academic funding, with no sustained federal investment. **Developing a centralised registry, integrating genotypic data, and securing dedicated funding streams** would strengthen Belgium's role in European research and improve national policy planning.

IV. Early detection through newborn screening

Belgium does not have nationwide newborn screening for SCD. Since January 2023, SCD is part of the FWB systematic programme run by ONE, but implementation is uneven and SCD is absent from the Flemish panel, creating inequities and delayed diagnoses. Nationwide screening would enable early detection and improve outcomes.

1. **National Implementation of SCD Universal Newborn Screening**

Belgium does not have a nationwide universal newborn screening (NBS) programme for SCD. Since January 2023, SCD is included in the systematic FWB screening programme run by ONE, building on earlier universal screening in Brussels and Liège maternities that achieved strong results in early detection. However, implementation is not yet consistent across all maternities, and Flanders has not included SCD in its panel, creating significant regional inequities and delayed diagnoses after severe crises. [27][28][29]

2. **Newborn screening coverage**

Current coverage is determined by geography rather than by risk criteria. Newborns in maternities participating in the FWB programme are screened, while newborns in Flanders are not, and implementation within Brussels and Wallonia is not yet uniform. This leaves a large proportion of affected children undiagnosed at birth, missing

[25] Vandenplas, Y., & Rozen, L. (2021). Management of haemoglobinopathies in Belgian paediatric centres.
[26] Association Drépano Belgique (ADB). (2024). Rapport d'activités et recommandations politiques. Retrieved from https://www.drepanobelgique.be/reports/2024-annual-report
[27] Caillet, P., Gulbis, B., Ketelslegers, O., et al. (2022). Newborn screening for sickle cell disease in Europe: Current status and future perspectives. International Journal of Neonatal Screening, 8(4), 63. https://doi.org/10.3390/ijns8040063
[28] Sciensano. (2023). Programme de dépistage néonatal en Belgique – Rapport annuel. Retrieved from https://www.sciensano.be/en/projects/neonatal-screening-programme-belgium
[29] Association Drépano Belgique (ADB). (2024). Plaidoyer pour un dépistage néonatal universel de la drépanocytose en Belgique. Retrieved from https://www.drepanobelgique.be/declarations/2024-advocacy-nb-screening

opportunities for early prophylaxis and parental counselling. Coverage remains below 50 per cent nationally, with near-complete absence in Flanders.[30][31]

3. **Standardisation of post-screening referral pathways**

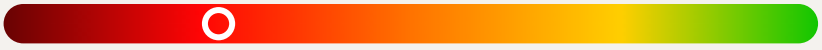
Where screening is implemented, **referral pathways are well structured:** newborns identified through the FWB programme are referred promptly to specialist paediatric centres such as ULB Erasme (Brussels) and CHR Liège. These centres provide early family education, infection prevention, and initiation of hydroxyurea when indicated, reducing hospitalisations and severe complications. However, where screening is not performed, diagnosis often occurs only after a severe crisis, delaying access to specialist care.[32] [33]

Belgium's newborn screening for SCD is fragmented and regionally limited, leaving many children undiagnosed until severe complications occur. Universal NBS, combined with structured referral pathways and early intervention, would align Belgium with best practices in Europe and significantly reduce preventable morbidity in early childhood.

V. Coordinated policy and patient partnership

Strategic dialogue on SCD in Belgium is fragmented, with various actors—academic centres, patient organisations, and public health authorities—contributing to awareness and policy discussions, but without a formal national platform dedicated exclusively to SCD.

1. **Formal national multi-stakeholder SCD coordination platform**



Belgium currently lacks a structured multi-stakeholder governance mechanism for SCD. Activities are driven by academic hospitals and rare disease networks rather than a dedicated national council or working group under the Ministry of Health. While Belgian

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[30] ONE – Dépistage néonatal FWB. (2025). Rapport sur les données 2023. https://www.depistageneonatal.be
[31] Caillet et al. (2022). Newborn screening for sickle cell disease in Europe.
[32] Vandenplas, Y., & Rozen, L. (2021). Management of haemoglobinopathies in Belgian paediatric centres. Belgian Journal of Paediatrics, 23(2), 81–86.
[33] ULB Erasme. (n.d.). Clinique de drépanocytose – Hôpital Erasme. Retrieved from https://www.erasme.ulb.ac.be/fr/sickle-cell-clinic
[34] Sciensano & SPF Santé Publique, 2023.

Belgian experts participate in European initiatives such as ERN-EuroBloodNet and RADeep, which aim to harmonise care and build registries, national coordination remains ad hoc and fragmented.[35] [36]

2. **Inclusion of patient organisations in decision-making**

Association Drépano Belgique (ADB) plays a vital role in advocacy, patient education, and community support, but formal involvement in policy-making is limited. Participation in national planning and rare disease strategies tends to be informal and mediated through broader haemoglobinopathy or rare disease networks, rather than through a dedicated SCD policy platform.[37] Strengthening patient representation in structured governance would improve service design and data integration.

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[35] EuroBloodNet & RADeep Consortium. (2023). European Rare Anaemia Database Annual Report 2023 – Belgium country summary. Retrieved from https://eurobloodnet.eu/research/radeep/annual-reports

[36] Sciensano & SPF Santé Publique. (2023). Plan national maladies rares – suivi des actions sur la drépanocytose. Retrieved from https://www.health.belgium.be/fr/plan-national-maladies-rares-2023

[37] Association Drépano Belgique (ADB). (2024). Rapport d'activités et recommandations politiques. Retrieved from https://www.drepanobelgique.be/reports/2024-annual-report

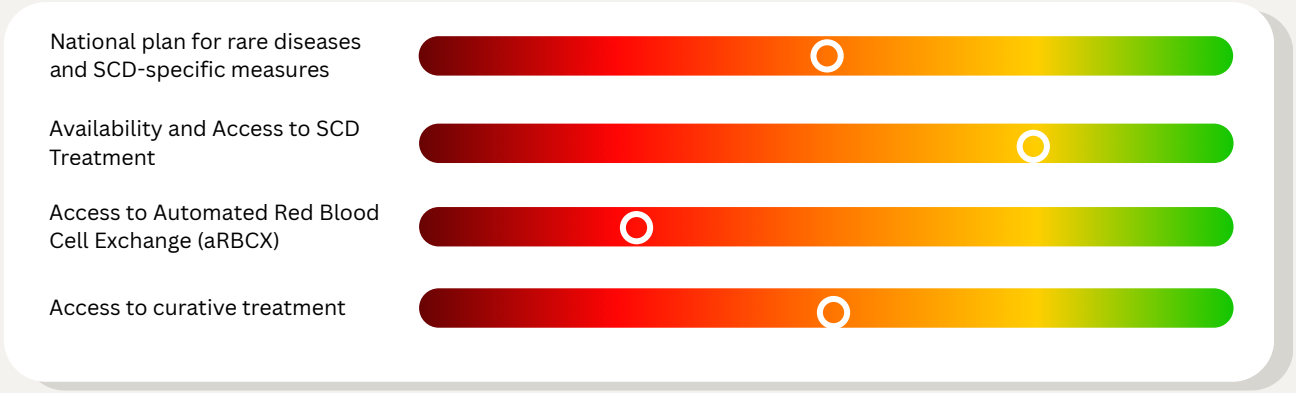
France
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment				
2. Strengthened National Workforce Capacity and Preparedness for SCD				
3. Comprehensive National Data, Registry and Research Infrastructure				
4. Early detection through newborn screening				
5. Coordinated policy and patient partnership				

Key Performance Indicator (KPI) Overview

1. Equitable access to SCD care and treatment



2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure

4. Early detection through newborn screening

5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	France integrates SCD into its rare disease strategy via MCGRE network and national protocols, but lacks a dedicated SCD-specific plan with formal governance.	
Availability and Access to SCD Treatment	Hydroxyurea widely available; HSCT offered for severe cases; CASGEVY approved but reimbursement and centre designation pending. Advanced therapies cluster in few centres, creating geographic inequities.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX is recognised in French expert consensus for selected indications, but centre-level variability in apheresis infrastructure and trained staff results in unequal access.	
Access to curative treatment	HSCT available for severe paediatric cases; donor matching and cost constraints limit broader access.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric teams well-trained; adult haematology, primary care, and ED staff need structured CME and protocol-linked education.	
Emergency care training and protocols for SCD crises	Emergency departments lack consistent SCD-specific protocols; delays in analgesia and uneven acute care persist.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	France maintains strong registry coverage through reference centres; prevalence data robust, but public dashboards and unified outcome reporting remain limited.	
Systematic National Collection and Integration of Genotype Data	Genotyping data collected but not systematically integrated across all centres; limits advanced therapy stratification.	
Sustained funding for SCD data and research	Research supported by Inserm and AFM-Téléthon; no large-scale recurring funding stream comparable to other rare diseases.	
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	Nationwide universal NBS implemented November 2024; integrated into national panel.	
Newborn screening coverage	Coverage expected near 100% due to integration into national neonatal screening programme.	
Standardisation of post-screening referral pathways	Infants referred within weeks to MCGRE centres; prophylaxis and stroke-risk monitoring initiated per HAS guidelines.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	Collaboration occurs via MCGRE and patient organisations; no formal national SCD-specific platform for multi-stakeholder governance	
Inclusion of Patient Organisations in decision-making	AFD and SOS Globi actively engage in advocacy and awareness campaigns; formal integration into policy-making and registry governance remains limited.	

The Hidden Struggle: SCD in France

I. Equitable access to SCD care and treatment

France combines universal newborn screening (from 1 Nov 2024), reference-centre coordination (MCGRE), and PNDS national guidance to deliver robust paediatric pathways; however, adult care remains more variable across regions and overseas territories, with documented gaps in HU optimisation, timely analgesia in EDs, and long-term organ-protection monitoring.[1][2] Large, national-scale analyses confirm substantial disease burden and heterogeneous resource use, underscoring the need for stronger adult implementation and outcome transparency.[3]

1. National plan for rare diseases and SCD-specific measures

SCD is embedded in France's rare-disease architecture through **reference centres**, the MCGRE **filière**, and **PNDS protocols**, ensuring standardised paediatric management and rapid referral from **universal NBS (effective 1 Nov 2024)**; [4] implementation strengths are clear in early prevention and structured pathways.[5] [6] However, **adult service harmonisation and public outcome dashboards** could be improved to reduce regional variability and support policy monitoring.[7]

2. Availability and Access to SCD Treatment

Hydroxyurea (HU) remains the backbone therapy in the PNDS, **yet adult uptake is suboptimal** despite robust efficacy/safety data from clinical practice analyses.[8] [9] **Transfusions** (acute and chronic) and **automated RBC exchange (aRBCX)** are available but **heterogeneously implemented** across centres; experts highlight the need for clearer integration and standardisation.[10]

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[1] Filière de Santé MCGRE. (2024). Organisation nationale de la prise en charge de la drépanocytose en France. https://www.filiere-mcgre.fr
[2] Haute Autorité de Santé (HAS). (2023). Protocole national de diagnostic et de soins (PNDS) – Drépanocytose de l'enfant et de l'adulte. https://www.has-sante.fr
[3] Brousse, V., Arnaud, C., et al. (2023). Severity and burden of sickle cell disease in France: A nationwide study. Haematologica, 108(9), 2421–2432. https://doi.org/10.3324/haematol.2022.282050
[4] Ministère de la Santé et de la Prévention. (2024, October 23). Le programme national du dépistage néonatal évolue : tous les nouveau-nés seront dépistés pour la drépanocytose à partir du 1er novembre 2024. https://sante.gouv.fr
[5] Filière de Santé MCGRE. (2024). Organisation nationale de la prise en charge de la drépanocytose en France. https://www.filiere-mcgre.fr
[6] Haute Autorité de Santé (HAS). (2023). Protocole national de diagnostic et de soins (PNDS) – Drépanocytose de l'enfant et de l'adulte. https://www.has-sante.fr
[7] Beillat, M., et al. (2023). Prevalence and cost of sickle cell disease in France: A population-based study. Frontiers in Public Health, 11, 1113853. https://doi.org/10.3389/fpubh.2023.1113853
[8] Haute Autorité de Santé (HAS). (2023). PNDS – Drépanocytose. https://www.has-sante.fr
[9] Inserm & Assistance Publique–Hôpitaux de Paris (AP-HP). (2022). Hydroxycarbamide in SCD: efficacy, safety, and underuse in adult practice. Bulletin de l'Académie Nationale de Médecine, 206(8), 1365–1373. https://doi.org/10.1016/j.banm.2022.08.002
[10] Benmoussa, K., Bernaudin, F., Connes, P., Héquet, O., Joseph, L., Beraud, M., & Bah, A. (2024). Position paper on advancing SCD management in France by bridging clinical practices and guidelines. Transfusion and Apheresis Science, 63(5), 103988. https://doi.org/10.1016/j.transci.2024.103988

HSCT is the established curative option, generally reserved for severe paediatric cases given risk and donor constraints.[11] **CASGEVY (exa-cel)** holds EU authorisation; in France, **HAS Transparency** and **CEESP** opinions (2024) have been issued, with **pricing/reimbursement and centre designation** the next steps for equitable rollout.[12] [13] **Crizanlizumab** and **voxelotor** have been withdrawn/suspended at EU level, narrowing non-HU options.[14] France also **advances gene-therapy R&D** through AFM-Téléthon initiatives.[15]

3. **Automated Red Blood Cell Exchange (aRBCX)**

French expert consensus recognises **aRBCX** as valuable for selected indications (e.g., recurrent VOCs, ACS prevention, iron burden control), but **centre-level variability** in apheresis infrastructure and trained staff results in **unequal access;** the field would benefit from **nationally standardised protocols and targeted workforce development**.[16]

4. **Access to curative treatment**

HSCT remains **the only established curative option** in France and is typically **reserved for severe paediatric phenotypes** due to risk, complexity, and donor-matching constraints; adult curative access is therefore limited and should be guided by **clear referral criteria and long-term outcome tracking**.[17] [18]

II. Strengthened National Workforce Capacity and Preparedness for SCD

France's paediatric reference centres provide strong, protocol-based expertise, **but adult haematology, primary care, and emergency departments lack systematic training**, resulting in uneven care quality and delayed crisis management.[19] [20] Embedding SCD content in medical curricula and linking CME to PNDS protocols would improve adult outcomes and reduce regional variation.

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[11] Inserm. (n.d.). Drépanocytose. https://www.inserm.fr/dossier/drepanocytose/
[12] Haute Autorité de Santé (HAS). (2024a, June). Casgevy (exa-cel): Avis de la Commission de la Transparence. https://www.has-sante.fr/jcms/p_3418130
[13] Haute Autorité de Santé (HAS). (2024b, July). Casgevy (exa-cel): Avis de la CEESP (évaluation économique). https://www.has-sante.fr/jcms/p_3419505
[14] Centre Régional de Pharmacovigilance d'Île-de-France. (n.d.). Suspension d'AMM pour Oxbryta® (voxelotor). https://www.pharmacovigilance-iledefrance.fr/d%C3%A9tails-dune-alerte/suspension-d-amm-pour-oxbryta-r-voxelotor
[15] AFM-Téléthon. (n.d.). Drépanocytose : vers un essai de thérapie génique soutenu par l'AFM-Téléthon. https://www.afm-telethon.fr/fr/actualites/drepanocytose-vers-un-essai-de-therapie-genique-soutenu-par-lafm-telethon
[16] Benmoussa, K., Bernaudin, F., Connes, P., Héquet, O., Joseph, L., Beraud, M., & Bah, A. (2024). Position paper on advancing SCD management in France by bridging clinical practices and guidelines. Transfusion and Apheresis Science, 63(5), 103988. https://doi.org/10.1016/j.transci.2024.103988
[17] Inserm. (n.d.). Drépanocytose. https://www.inserm.fr/dossier/drepanocytose/
[18] Haute Autorité de Santé (HAS). (2023). PNDS – Drépanocytose. https://www.has-sante.fr
[19] Haute Autorité de Santé (HAS). (2023). Protocole national de diagnostic et de soins (PNDS) – Drépanocytose de l'enfant et de l'adulte. Retrieved from https://www.has-sante.fr
[20] Filière de Santé MCGRE. (2024). Organisation nationale de la prise en charge de la drépanocytose en France. Retrieved from https://www.filiere-mcgre.fr

1. **Integration of SCD into routine clinical training**

SCD is well integrated into paediatric specialist training, supported by national guidelines and MCGRE coordination. However, **adult haematologists and general practitioners often lack exposure to modern disease-modifying therapies, transfusion strategies, and organ-protection protocols**, leading to gaps in monitoring and delayed referrals.[21]

2. **Emergency care training and protocols for SCD crises**

Emergency departments are critical touchpoints, but **responses remain inconsistent**, with delays in analgesia and variable awareness of acute complications such as ACS or severe infections.[22] Wider roll-out of standardised ED protocols and mandatory CME would improve crisis care and patient experience.[23]

Training in France is strong for paediatric care but **weak for adult and emergency settings**, creating inequities in timely and effective management. **National standardisation of curricula and ED protocols, combined with structured CME**, is essential to improve knowledge and reduce preventable complications.

III. Comprehensive National Data, Registry and Research Infrastructure

France contributes actively to **European haemoglobinopathy initiatives** through **EuroBloodNet**, supporting registry harmonisation and research collaboration.[24] National datasets and cost-of-illness studies provide strong epidemiological visibility, but **public dashboards and unified outcome reporting** would further strengthen benchmarking and policy evaluation.[25]

1. **Establishment of a national SCD registry**

France leverages **reference-centre datasets** and health-system data to generate **nationwide estimates** (~20,000–22,000 individuals living with SCD between 2016 and 2018), with the highest prevalence in Île-de-France and overseas territories.[26] To strengthen policy and care evaluation, **adult outcomes and transition-related data** should be more visible in routine reporting and linked to service use and mortality trends.[27]

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[21] Inserm & Assistance Publique–Hôpitaux de Paris (AP-HP). (2022). Hydroxycarbamide in sickle cell disease: efficacy, safety, and underuse in adult practice. Bulletin de l'Académie Nationale de Médecine, 206(8), 1365–1373. https://doi.org/10.1016/j.banm.2022.08.002

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[23] Haute Autorité de Santé (HAS). (2023). PNDS – Drépanocytose. https://www.has-sante.fr

[24] European Reference Network EuroBloodNet. (2023). Activities and registries on haemoglobinopathies in Europe. Retrieved from https://eurobloodnet.eu/activities/research/registries

[25] Beillat, M., et al. (2023). Prevalence and cost of sickle cell disease in France: A population-based study. Frontiers in Public Health, 11, 1113853. https://doi.org/10.3389/fpubh.2023.1113853

[26] Brousse, V., Arnaud, C., et al. (2023). Severity and burden of sickle cell disease in France: A nationwide study. Haematologica, 108(9), 2421–2432. https://doi.org/10.3324/haematol.2022.282050

[27] Assistance Publique–Hôpitaux de Paris (AP-HP). (2023). Programme national de dépistage néonatal et registres associés. Retrieved from https://www.aphp.fr/actualite/depistage-neonatal-dr%25C3%25A9panocytose

2. **Systematic National Collection and Integration of Genotype Data**

Reference centres routinely perform **diagnostic genotyping**, but **systematic national capture and integration with outcome data** remain incomplete, limiting genotype–phenotype analyses and risk-stratified follow-up.[28] [29]

3. **Sustained funding for SCD data and research**

France has produced important publications on SCD epidemiology and cost-of-illness, supported mainly by academic and hospital-based research funds, and remains active in gene-therapy research via AFM-Téléthon initiatives.[30] However, there is no dedicated, large-scale, recurring federal funding stream for SCD registries or long-term outcome studies; expanding registry infrastructure and linking to insurance/hospital data would enhance EU-wide monitoring and comparative policy evaluation.[31] [32] [33]

IV. Early detection through newborn screening

Early identification of sickle cell disease (SCD) is a cornerstone of effective prevention and care. Newborn screening (NBS) enables timely diagnosis, initiation of prophylaxis, and family education, reducing life-threatening complications in infancy. Countries with universal NBS have demonstrated significant improvements in survival and quality of life, making this intervention a critical public health priority. France's recent move to nationwide screening marks a major step toward equity and comprehensive care for affected children.

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[28] Assistance Publique–Hôpitaux de Paris (AP-HP). (2023). Programme national de dépistage néonatal et registres associés. Retrieved from https://www.aphp.fr/actualite/depistage-neonatal-dr%25C3%25A9panocytose
[29] European Reference Network EuroBloodNet. (2023). Activities and registries on haemoglobinopathies in Europe. Retrieved from https://eurobloodnet.eu/activities/research/registries
[30] AFM-Téléthon. (n.d.). Drépanocytose : vers un essai de thérapie génique soutenu par l'AFM-Téléthon. Retrieved from https://www.afm-telethon.fr/fr/actualites/drepanocytose-vers-un-essai-de-therapie-genique-soutenu-par-lafm-telethon
[31] Beillat, M., et al. (2023). Prevalence and cost of sickle cell disease in France: A population-based study. Frontiers in Public Health, 11, 1113853. https://doi.org/10.3389/fpubh.2023.1113853
[32] Brousse, V., Arnaud, C., et al. (2023). Severity and burden of sickle cell disease in France: A nationwide study. Haematologica, 108(9), 2421–2432. https://doi.org/10.3324/haematol.2022.282050
[33] European Reference Network EuroBloodNet. (2023). Activities and registries on haemoglobinopathies in Europe. Retrieved from https://eurobloodnet.eu/activities/research/registries
[34] Ministère de la Santé et de la Prévention. (2024, October 23). Le programme national du dépistage néonatal évolue : tous les nouveau-nés seront dépistés pour la drépanocytose à partir du 1er novembre 2024. Gouvernement.fr. Retrieved from https://sante.gouv.fr/actualites/actualites-du-ministere/article/le-programme-national-du-depistage-neonatal-evolue-tous-les-nouveau-nes-seront
[35] Brousse, V., Arnaud, C., Kamdem, A., & Hau, I. (2021). Newborn screening for sickle cell disease in France. Archives de Pédiatrie, 28(7), 567–572. https://doi.org/10.1016/j.arcped.2021.07.001

1. **National Implementation of SCD Universal Newborn Screening**

France introduced **nationwide universal newborn screening (NBS) for SCD on 1 November 2024**, after years of targeted screening in high-prevalence areas.[34] This policy ensures that all newborns are screened regardless of ethnic background, enabling early identification and intervention.[35]

2. **Newborn screening coverage**

Coverage is expected to reach **near 100%**, as the programme is integrated into the national neonatal screening panel alongside other conditions.[36] Previous targeted screening already demonstrated high feasibility and clinical benefit, reducing severe infections and improving early family education.[37]

3. **Standardisation of post-screening referral pathways**

Infants identified through NBS are referred within weeks to specialised centres under the MCGRE **rare disease network**, where prophylactic penicillin, vaccination, and stroke-risk monitoring are initiated according to **Haute Autorité de Santé (HAS)** guidelines.[38] This structured pathway has significantly improved paediatric outcomes, though adult transition remains a challenge.

France's universal NBS programme for SCD represents a major public health advance, ensuring early diagnosis and linkage to specialist care. Strong referral pathways and national protocols have improved paediatric outcomes, but monitoring of adult transition and regional equity remains essential for long-term success.[39]

V. Coordinated policy and patient partnership

Strategic dialogue and stakeholder collaboration are essential for aligning clinical practice, policy, and patient needs in rare diseases such as sickle cell disease (SCD). Effective governance frameworks ensure that healthcare providers, policymakers, researchers, and patient organisations work together to improve access, equity, and long-term outcomes. In France, collaboration exists through rare disease networks and advocacy groups, but formal national structures dedicated to SCD remain limited.

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[36] Assistance Publique–Hôpitaux de Paris (AP-HP). (2023). Programme national de dépistage néonatal et registres associés. Retrieved from https://www.aphp.fr/actualite/depistage-neonatal-dr%25C3%25A9panocytose
[37] Brousse, V., Arnaud, C., Kamdem, A., & Hau, I. (2021). Newborn screening for sickle cell disease in France. Archives de Pédiatrie, 28(7), 567–572. https://doi.org/10.1016/j.arcped.2021.07.001
[38] Filière de Santé MCGRE. (2024). Organisation nationale de la prise en charge de la drépanocytose en France. Retrieved from https://www.filiere-mcgre.fr
[39] Haute Autorité de Santé (HAS). (2023). Protocole national de diagnostic et de soins (PNDS) – Drépanocytose de l'enfant et de l'adulte. Retrieved from https://www.has-sante.fr

1. F**ormal national multi-stakeholder SCD coordination platform**

France does not yet have a **formal national platform dedicated exclusively to SCD**, but collaboration occurs through rare disease networks such as MCGRE, professional societies, and patient organisations.[40] [41] These networks facilitate clinical coordination, guideline dissemination, and awareness campaigns. However, governance remains fragmented, and multi-stakeholder dialogue is often project-based rather than institutionalised.

2. I**nclusion of Patient organisations in decision-making**

Patient organisations play an active role in advocacy and education. Groups such as APIPD and the federation SOS Globi collaborate with reference centres and participate in awareness campaigns.[42] [43] Despite this
engagement, formal integration into policy-making and registry governance is limited, reducing the potential impact of patient voices on national planning and resource allocation.

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[40] Filière de Santé MCGRE. (2024). Les associations de patients partenaires. Retrieved from https://www.filiere-mcgre.fr

[41] EuroBloodNet. (2023). Activities and registries on haemoglobinopathies in Europe. Retrieved from https://eurobloodnet.eu/activities/research/registries

[42] APIPD – Association pour l'Information et la Prévention de la Drépanocytose. (2024). Nos actions. https://apipd.fr

[43] SOS Globi. (2024). Nos missions et partenariats. Retrieved from https://www.sosglobi.fr

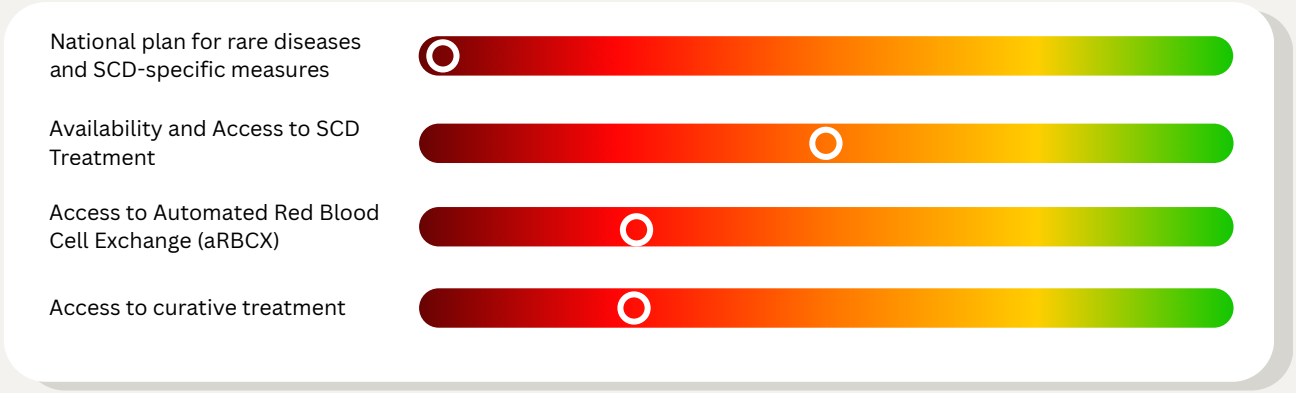
Germany
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment	●	●	●	●
2. Strengthened National Workforce Capacity and Preparedness for SCD	●	●		
3. Comprehensive National Data, Registry and Research Infrastructure	●	●	●	
4. Early detection through newborn screening	●	●	●	
5. Coordinated policy and patient partnership	●	●		

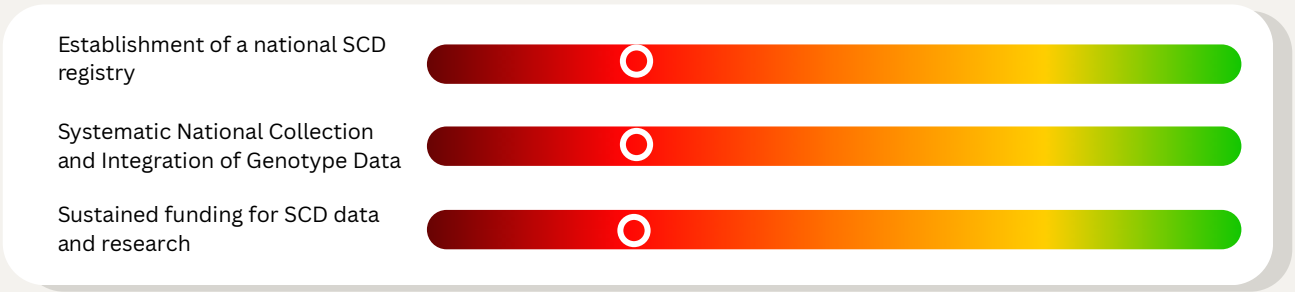
Key Performance Indicator (KPI) Overview

1. Equitable access to SCD care and treatment



2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure



4. Early detection through newborn screening

5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	Germany lacks a national rare disease plan with concrete implementation measures for SCD. Care fragmentation, especially during transition to adulthood, leads to inequitable access to essential services.	
Availability and Access to SCD Treatment	HU is reimbursed and widely available; chronic transfusion programmes exist but are inconsistent; HSCT is limited to specific centres; the G-BA completed its benefit assessment of gene-editing therapy in July 2025 and a reimbursement agreement is in place.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX is available mainly in specialised centres (Berlin, Ulm, Frankfurt). Limited staffing and apheresis capacity restrict national implementation.	
Access to curative treatment	HSCT is offered in a small number of university hospitals (Essen, Hamburg, Ulm). Donor availability and regional variation limit equitable access.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric haematologists are well-trained, but adult haematologists, GPs, and emergency physicians often lack SCD-specific training, resulting in significant regional disparities in care quality.	
Emergency care training and protocols for SCD crises	Emergency departments frequently lack SCD-specific training and standardised crisis protocols, leading to delayed analgesia and inconsistent management of acute episodes.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	Germany maintains a voluntary national SCD registry primarily populated by paediatric centres; adult patient data remain significantly underrepresented.	
Systematic National Collection and Integration of Genotype Data	Genotyping data are collected inconsistently; lack of integration with national hospital systems or insurance datasets limits comprehensiveness.	
Sustained funding for SCD data and research	Dedicated federal funding for SCD research is minimal; registry development and studies rely mainly on paediatric centres and academic collaborations.	
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	SCD was added to the national newborn screening panel (Kinder-Richtlinie) in October 2021 after a positive G-BA assessment, making screening universal for all newborns.	
Newborn screening coverage	With SCD in the national panel, coverage should approach the very high levels of other screened conditions, though real-world SCD coverage data remain limited.	
Standardisation of post-screening referral pathways	Referral systems function well where screening identifies infants early: 95.6% receive penicillin prophylaxis. The focus is now on linking all screened newborns to care.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	Germany lacks a structured national platform for multi-stakeholder engagement on SCD. Collaboration remains fragmented across federal bodies, paediatric centres, and patient groups.	
Inclusion of Patient Organisations in decision-making	Patient organisations contribute actively but are not systematically embedded in policy processes. Engagement occurs through ad hoc interactions rather than formal governance structures.	

The Hidden Struggle: SCD in Germany

I. Equitable access to SCD care and treatment

Germany provides access to most evidence-based therapies for Sickle Cell Disease (SCD), but access is **uneven across age groups, regions and care settings**. Paediatric patients followed in specialist centres generally receive guideline-based care[1], whereas adults are more exposed to fragmented provision, limited experience among local providers, and geographic disparities in access to advanced treatments[2].

1. **National plan for rare diseases and SCD-specific measures**

SCD is recognised as a rare disease in Germany, but there is **no SCD-specific national plan with concrete implementation measures** for diagnosis, treatment and long-term follow-up[3]. Instead, care depends on a combination of national treatment guidelines, regional expertise (particularly in paediatric university hospitals), and local initiatives. The lack of a dedicated implementation framework contributes to gaps at transition to adult care, where responsibilities shift to haematologists or general practitioners who may have limited SCD experience. This results in discontinuity of care, lower hydroxyurea uptake in adults, and preventable complications[4].

2. **Availability and Access to SCD Treatment**

Hydroxyurea (HU) is reimbursed and available as a **first-line disease-modifying therapy**[5]. Registry data show high uptake in children (around 70–90% of eligible paediatric patients) and markedly lower use in adults, highlighting a major drop-off after transition from paediatric services[6][7]. Chronic transfusion programmes are used for high-risk paediatric patients, but coverage is inconsistent and long-term iron chelation requires better follow-up in adults[8]. Curative treatment with **haematopoietic stem cell transplantation (HSCT)** is available only in selected university hospitals (e.g. Essen, Hamburg, Ulm), with eligibility constrained by donor availability and centre capacity[9].

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[1] sichelzellerkrankung.de. (2024). Informationsportal zur Sichelzellkrankheit. https://sichelzellerkrankung.de/

[2] Ibid.

[3] Ibid.

[4] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/

[5] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit [Sickle cell disease guideline]. https://www.dgho.de/

[6] Kunz, J. B., et al.; German Sickle Cell Disease Registry. (2020). Sickle cell disease in Germany: Results from a national registry. Pediatric Blood and Cancer, 67(4), e28130.

[7] Ibid.

[8] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/

[9] Ibid.

3. **Automated Red Blood Cell Exchange (aRBCX)**

·Red blood cell exchange (RBCx) is recognised as essential for stroke prevention, recurrent vaso-occlusive crises, and severe anaemia[10]. In Germany, **automated RBCx is available primarily in specialised haematology or paediatric centres** (e.g. Charité Berlin, Ulm, Frankfurt), where apheresis infrastructure and trained staff are concentrated[11]. Many regional hospitals lack the capacity to offer automated exchange, resulting in **geographic inequities** in access to optimal transfusion strategies[12]. Standardisation of transfusion protocols and wider training on aRBCX are necessary to ensure more equitable provision[13].

4. **Access to curative treatment**

Curative options remain restricted to a **small subset of patients**[14]. HSCT is available at a limited number of university centres and is constrained by donor matching and transplant risk[15]. Gene-editing therapy offers the prospect of a one-time functional cure; the G-BA completed its benefit assessment in July 2025 and a reimbursement agreement with statutory health insurance is in place, so uptake now depends on the capacity of qualified treatment centres[16][17].

Overall, **access in Germany is best for children in specialised centres and weakest for adults and those living far from university hospitals**, which aligns with inequities seen in other rare diseases.

II. Strengthened National Workforce Capacity and Preparedness for SCD

The level of SCD-specific expertise in Germany is **high in paediatric specialist centres**, but much more variable in adult haematology, general practice, and emergency medicine[1]. This uneven training profile directly affects access to guideline-based care, timely pain management and prevention of complications[2].

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[10] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[11] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[12] Ibid.
[13] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[14] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[15] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[16] G-BA – Gemeinsamer Bundesausschuss. (2024). Dossierverfahren für neuartige Therapien [Dossier procedure for advanced therapies]. https://www.g-ba.de/
[17] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[18] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[19] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/

1. **Integration of SCD into routine clinical training**

In paediatric haematology–oncology, SCD is well integrated in specialist training and supported by national guidelines (e.g. GPOH and DGHO recommendations)[20]. Paediatric centres frequently participate in registry projects and structured disease management programmes, which reinforce clinical competence and adherence to best practice (penicillin prophylaxis, vaccination, HU use, TCD screening)[21].

However, **SCD is not consistently embedded in broader medical curricula** (e.g. general internal medicine, family medicine, emergency medicine)[22]. Many adult haematologists and general practitioners encounter relatively few SCD patients and may be less familiar with modern disease-modifying therapies, transfusion strategies, and long-term organ protection[23]. As a result, adult patients frequently report gaps in knowledge, delayed referrals and inconsistent monitoring outside centres of excellence[24].

2. **Emergency care training and protocols for SCD crises**

·Emergency departments are **critical touchpoints** for SCD, but responses are often inconsistent[25]. Patients report delays in pain relief and variable understanding of acute complications such as acute chest syndrome or severe infections[26].

The absence of **nationally standardised emergency protocols** for SCD means that practice differs significantly between hospitals, from triage and analgesia timing to antibiotic use and escalation of care.

III. Comprehensive National Data, Registry and Research Infrastructure

Germany has one of the **most established paediatric SCD registries** in Europe, but adult enrolment and data integration remain limited. Consequently, national monitoring of outcomes, complications and costs is incomplete[27].

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[20] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[21] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[22] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[23] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[24] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[25] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[26] Ibid.
[27] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/

1. **Establishment of a national SCD registry**

A nationwide SCD registry was developed under the German Society for Paediatric Oncology and Haematology (GPOH) to collect retrospective and prospective data on affected children and adolescents[28]. The registry has generated robust information on treatment patterns, HU uptake, TCD screening rates and complications, which has informed national guidelines and disease management programmes[29]. However, participation is voluntary and **centred primarily on paediatric university hospitals**, resulting in underrepresentation of adult patients and those treated outside specialist centres[30]. Adult care is therefore less visible in national datasets, limiting the ability to track long-term outcomes, transition-related gaps and mortality in early adulthood[31].

2. **Systematic National Collection and Integration of Genotype Data**

The registry captures clinical and some genotypic information, but genotyping data are **not yet systematically collected for all patients** and are not fully integrated with routine laboratory or hospital information systems[32]. This constrains genotype–phenotype analyses and the ability to stratify patients for advanced therapies or targeted follow-up[33].

3. **Sustained funding for SCD data and research**

·Several important publications on SCD epidemiology, registry findings and disease management in Germany have been supported by academic and society-based research funds[34].

However, **there is no dedicated, large-scale, recurring federal funding stream** for SCD registries or long-term outcome studies comparable to those for some cancers or more prevalent chronic diseases[35]. Registry expansion and linkage to health insurance and hospital data would significantly enhance Germany's contribution to EU-wide monitoring and comparative policy evaluation[36].

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[28] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[29] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[30] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[31] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[32] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[33] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[34] GPOH – Gesellschaft für Pädiatrische Onkologie und Hämatologie. (2020). Sichelzellkrankheit Register: Jahresbericht 2020. https://www.kinderkrebsinfo.de/
[35] Kuerten, D., Overfeld, J., & Milde-Busch, A. (2022). National funding mechanisms for rare disease research in Germany: A policy analysis. Orphanet Journal of Rare Diseases, 17, 34–45. https://ojrd.biomedcentral.com/articles/10.1186/s13023-022-02265-5
[36] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/

IV. Early detection through newborn screening

Germany has moved from pilot newborn screening for SCD in selected regions to nationwide inclusion of SCD in the national newborn screening (NBS) panel, but implementation and follow-up still face challenges[37].

1. **National Implementation of SCD Universal Newborn Screening**

Historically, Germany relied on targeted or pilot projects in urban regions, which identified SCD birth prevalence between ~1:2,127 and 1:4,154 and estimated a national birth prevalence of roughly 1:5,000–7,500 (100–160 affected newborns per year)[38].

These pilots demonstrated the feasibility and clinical value of NBS: early diagnosis enabled penicillin prophylaxis, parental counselling and timely stroke prevention measures (e.g. TCD screening)[39]. In October 2021, SCD was **formally added to the national NBS programme** (Kinder-Richtlinie) as one of the official target diseases, following a positive assessment by the G-BA[40]. This represents a major policy advance and aligns Germany with international recommendations for universal NBS for SCD[41].

2. **Newborn screening coverage**

With SCD now included in the national panel, coverage should approach that of other NBS conditions (i.e. very high national coverage)[42]. However, available reports focus primarily on implementation and technical aspects; detailed published data on **real-world SCD screening coverage and regional variation** remain limited[43].

3. **Standardisation of post-screening referral pathways**

Where early diagnosis occurs, referral pathways are **strong and clinically effective**: registry data show that 95.6% of young children under six receive penicillin prophylaxis when diagnosed early, and high proportions receive guideline-recommended vaccination and TCD screening[44].

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[37] Kunz, J. B., Cario, H., Grosse, R., Jarisch, A., Lobitz, S., and Kulozik, A. E. (2017). The epidemiology of sickle cell disease in Germany following recent large-scale immigration. Pediatric Blood and Cancer, 64(7), e26550.
[38] Ibid.
[39] Ibid.
[40] G-BA – Gemeinsamer Bundesausschuss. (2021). Beschluss zur Aufnahme der Sichelzellanämie in das Neugeborenen-Screening. https://www.g-ba.de/
[41] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[42] IQTIG – Institut für Qualitätssicherung und Transparenz im Gesundheitswesen. (2022). Qualitätsbericht Neugeborenen-Screening 2022. https://iqtig.org/
[43] Grosse, R., Lukacs, Z., Cobos, P. N., et al. (2016). The prevalence of sickle cell disease and its implication for newborn screening in Germany (Hamburg metropolitan area). Pediatric Blood and Cancer, 63, 168-170.
[44] Kunz, J. B., et al.; German Sickle Cell Disease Registry. (2020). Sickle cell disease in Germany: Results from a national registry. Pediatric Blood and Cancer, 67(4), e28130.

The main challenge is therefore not the quality of care after diagnosis, but ensuring that **all affected newborns are identified and linked to specialist care**, and that families receive structured counselling and support[45].

Going forward, Germany will need robust **monitoring of NBS performance indicators** (uptake, timeliness, loss to follow-up) and formal integration of NBS data with the national SCD registry to fully realise the benefits of universal screening[46].

V. Coordinated policy and patient partnership

Strategic dialogue on SCD in Germany is **emerging but fragmented**, with various actors – specialist clinicians, scientific societies, patient organisations and industry – contributing to awareness and policy debates, but without a single, formal national platform dedicated exclusively to SCD[47].

1. **Formal national multi-stakeholder SCD coordination platform**

Professional societies such as the **German Society for Haematology and Medical Oncology (DGHO)** and paediatric haematology networks (e.g. GPOH) have played a leading role in developing guidelines, advocating for newborn screening, and providing expert input to the G-BA on SCD-related decisions[48].

However, these activities are **disease-area initiatives within broader oncology/haematology structures**, rather than a dedicated SCD policy platform that regularly convenes all relevant stakeholders (clinicians, payers, policymakers, patient organisations, and industry)[49]. At the European level, German experts and patient representatives participate in initiatives such as **ERN-EuroBloodNet** and the **European Sickle Cell Federation**, which aim to harmonise care, build registries and strengthen advocacy[50].

Nevertheless, at the national level, structured multi-stakeholder governance for SCD (e.g. a national SCD council or working group under the Ministry of Health) is not yet firmly established[51].

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[45] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/
[46] IQTIG – Institut für Qualitätssicherung und Transparenz im Gesundheitswesen. (2022). Qualitätsbericht Neugeborenen-Screening 2022. https://iqtig.org/
[47] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[48] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[49] DGHO – Deutsche Gesellschaft für Hämatologie und Medizinische Onkologie. (2021). Leitlinie Sichelzellkrankheit. https://www.dgho.de/
[50] European Sickle Cell Federation. (2023). Annual report 2023. https://www.escfederation.eu/
[51] ERN-EuroBloodNet. (2023). European Reference Network on Rare Haematological Diseases: Annual activity report. https://eurobloodnet.eu/

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2. **Inclusion of patient organisations in decision-making**

There are active patient organisations and support groups relevant to SCD, including **SAM Deutschland e.V. (Association for Rare Anaemias in Germany)**[52] and community platforms such as sichelzellerkrankung.de[53] and Realtalk Sichelzellkrankheit, which provide information, peer support and advocacy[54]. These organisations are increasingly visible in awareness campaigns and educational initiatives, but **formal, systematic involvement in national policy development remains limited**. Participation in G-BA consultations and guideline processes tends to be ad hoc and mediated through broader rare disease or haemoglobinopathy networks[55].

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[52] SAM Deutschland e.V. (2024). Seltene Anämien und Sichelzellkrankheit – Patientenorganisation Deutschland. https://www.seltene-anaemien-deutschland.de/

[53] sichelzellerkrankung.de. (2024). Informationsportal für Sichelzellkrankheit. https://www.sichelzellerkrankung.de/

[54] Realtalk Sichelzellkrankheit. (2024). Community initiative for sickle cell disease awareness. https://www.realtalk-sichelzellkrankheit.de/

[55] SAM Deutschland e.V. (2024). Seltene Anämien und Sichelzellkrankheit – Patientenorganisation Deutschland. https://www.seltene-anaemien-deutschland.de/

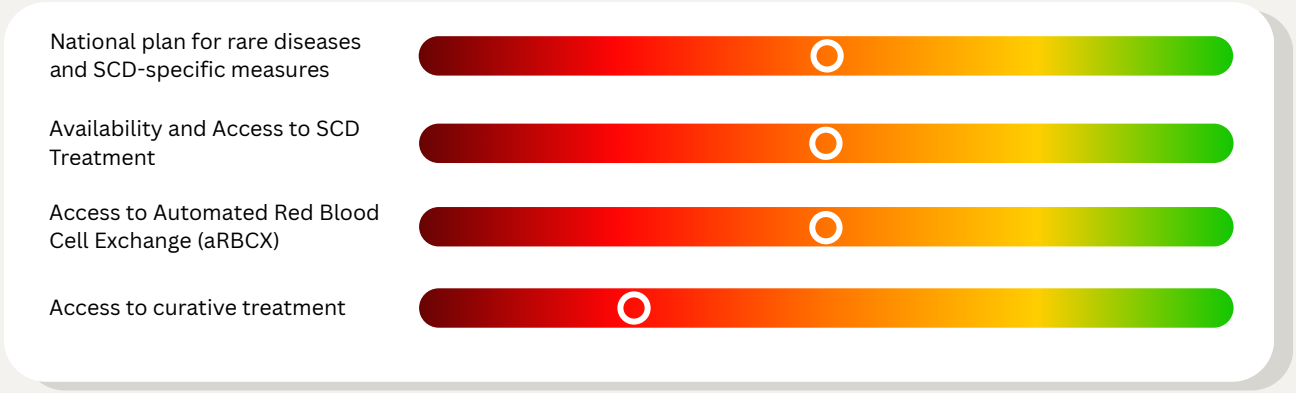
Italy
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment				
2. Strengthened National Workforce Capacity and Preparedness for SCD				
3. Comprehensive National Data, Registry and Research Infrastructure				
4. Early detection through newborn screening				
5. Coordinated policy and patient partnership				

Key Performance Indicator (KPI) Overview

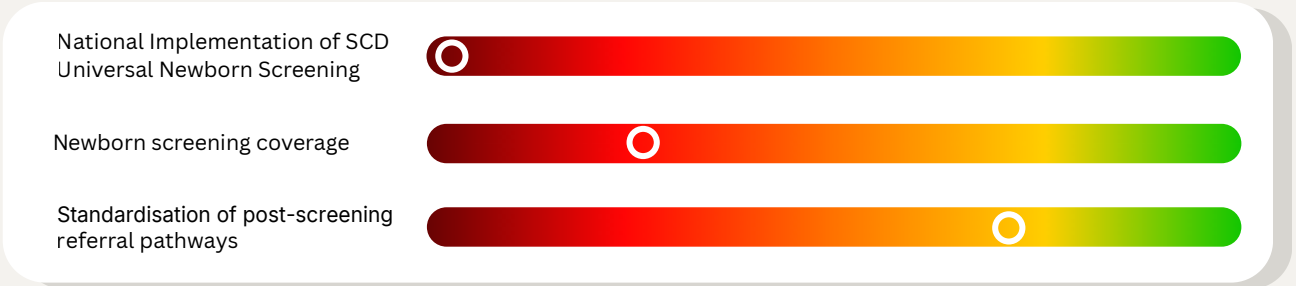
1. Equitable access to SCD care and treatment



2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure

4. Early detection through newborn screening



5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	Haemoglobinopathies are included in rare disease frameworks and registry initiatives, but there is **no SCD-specific national plan** with structured implementation measures.	
Availability and Access to SCD Treatment	Claims data show only ~6–7% of patients on SCD-specific drug therapy; transfusion programmes vary regionally. CASGEVY reimbursed since 2025, with access limited to a small number of specialised centres.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX is available only in select centres; there is **no national strategy** for equitable access.	
Access to curative treatment	HSCT offered for severe paediatric cases and selected adults, but donor availability and transplant risk limit broader access.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric hubs are well-trained, but adult haematology and primary care lack structured CME. Training gaps contribute to under-treatment and poor transition outcomes.	
Emergency care training and protocols for SCD crises	Emergency departments lack standardised protocols; delays in analgesia and inconsistent management persist despite targeted education initiatives.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	National Haemoglobinopathy Register covers >100 centres, but participation is uneven and data completeness varies, limiting planning and monitoring.	
Systematic National Collection and Integration of Genotype Data	Genotyping performed in many centres but **not systematically integrated** into the national registry, limiting genotype–phenotype analysis and personalised care.	
Sustained funding for SCD data and research	Research supported by academic collaborations and patient associations, but **no recurring national funding stream** comparable to other rare diseases.	
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	Italy lacks universal NBS; screening occurs only through regional pilots and voluntary programmes, leaving many infants undiagnosed until emergency presentations.	
Newborn screening coverage	Coverage is uneven and limited to regions with pilot programmes; participation in voluntary screening remains poor.	
Standardisation of post-screening referral pathways	Where screening occurs, referral pathways are effective: infants receive prophylaxis, vaccination, and stroke-risk monitoring per guidelines.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	Coordination occurs through CNS and scientific societies (SITE, SIE), but there is **no formal national platform** dedicated exclusively to SCD.	
Inclusion of Patient Organisations in decision-making	Patient organisations (UNITED Onlus, Associazione Piera Cutino) advocate for screening and therapy access, but formal integration into policy-making and registry governance remains limited.	

The Hidden Struggle: SCD in Italy

I. Equitable access to SCD care and treatment

Access to comprehensive care for **sickle cell disease (SCD)** in Italy remains **uneven**, reflecting regional disparities and fragmented adult services. While **paediatric hubs** deliver high-quality care, adult patients often face gaps in treatment, delayed crisis management, and limited access to advanced therapies. Addressing these inequities requires **coordinated policy**, structured referral systems, and investment in training and infrastructure.[1] [2]

1. **National plan for rare diseases and SCD-specific measures**

Italy does not have an **SCD-specific national plan**, but haemoglobinopathies are included in broader rare disease frameworks and registry initiatives. The **National Haemoglobinopathy Register**, launched in 2017, is a major step forward, yet participation varies by region, limiting its effectiveness for planning and monitoring outcomes.[3] A formal implementation strategy for SCD care, including **newborn screening** and transition protocols, is still lacking.

2. **Availability and Access to SCD Treatment**

Hydroxyurea remains under-prescribed: only ~6–7% receive SCD-specific drug therapy in claims data, far below recommendations.[4] Transfusion programmes are available and widely used for acute and chronic indications, supported by national guidelines.[5] Innovative therapies are emerging: **CASGEVY (exa-cel)** received **AIFA reimbursement approval** in September 2025, positioning Italy among the first EU countries to offer **gene-**editing therapy.[6] However, access is currently limited to a few highly specialised centres, creating a risk of geographic inequity.[7] [8]

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[1] De Franceschi, L., et al. (2022). Real-world evidence on disease burden and economic impact of sickle cell disease in Italy. Journal of Clinical Medicine, 12(1), 117. https://doi.org/10.3390/jcm12010117

[2] Russo, G., De Franceschi, L., Cappellini, M. D., et al. (2019). Current challenges in the management of patients with sickle cell disease in Italy. Journal of Clinical Medicine, 8(10), 1757. https://doi.org/10.3390/jcm8101757

3] Italian National Blood Centre (CNS). (2017). National Haemoglobinopathy Register. Retrieved from https://www.centronazionalesangue.it/en/national-haemoglobinopathy-register/

[4] De Franceschi, L., et al. (2022). Real-world evidence on disease burden and economic impact of sickle cell disease in Italy. Journal of Clinical Medicine, 12(1), 117. https://doi.org/10.3390/jcm12010117

[5] Graziadei, G., et al. (2022). Transfusional approach in multiethnic sickle cell patients: Real-world practice data from a multicenter survey in Italy. Frontiers in Medicine, 9, 832154. https://hdl.handle.net/11573/1650750

[6] AIFA. (2025, September 17). The NHS will reimburse the first gene editing therapy for β-thalassemia and sickle cell anaemia (CASGEVY). Retrieved from https://www.aifa.gov.it/en/-/sar%C3%A0-rimborsata-dal-ssn-la-prima-terapia-di-editing-genetico-per-la-%CE%B2-talassemia-e-l-anemia-falciforme

[7] Agenzia Italiana del Farmaco (AIFA). (2025, January). Elenco delle strutture autorizzate alla somministrazione delle terapie avanzate per l'anemia falciforme e le emoglobinopatie. Retrieved from https://www.aifa.gov.it

[8] Osservatorio Malattie Rare. (2023, November 14). Anemia falciforme: tra terapie convenzionali e terapie curative. Retrieved from https://www.osservatoriomalattierare.it/malattie-rare/anemia-falciforme/20325-anemia-falciforme-tra-terapie-convenzionali-e-terapie-curative

3. **Automated Red Blood Cell Exchange (aRBCX)**

Automated RBC exchange is available in select Italian centres, primarily those managing multi-ethnic populations and severe cases.[9] However, it is not uniformly implemented nationwide, and there is no structured national strategy to ensure equitable access. This limits its role in **stroke prevention** and management of severe complications.

4. **Access to curative treatment**

Haematopoietic stem cell transplantation (HSCT) is offered for severe paediatric cases and selected adults, but eligibility is constrained by donor availability and transplant risk.[10] While HSCT remains the only established curative option, **gene-editing therapies** like CASGEVY represent a transformative advance. Ensuring **equitable access** to these therapies will require referral networks, financial support for families, and training for multidisciplinary teams.

Italy has made progress in **registry development** and access to advanced therapies, but systemic gaps persist. Underuse of hydroxyurea, fragmented adult care, and regional disparities in access to curative options undermine equity. A **national implementation plan**, combined with structured referral systems and workforce training, is essential to translate scientific advances into real-world benefits for all patients..[11] [12]

II. Strengthened National Workforce Capacity and Preparedness for SCD

Effective management of **sickle cell disease (SCD)** requires a well-trained workforce across all care settings. While Italy has strong **paediatric haematology hubs**, adult care and emergency services often lack structured education on SCD, contributing to **under-treatment**, delayed crisis management, and poor transition outcomes.[13] [14]

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[9] Osservatorio Malattie Rare. (2023, November 14). Anemia falciforme: tra terapie convenzionali e terapie curative. Retrieved from https://www.osservatoriomalattierare.it/malattie-rare/anemia-falciforme/20325-anemia-falciforme-tra-terapie-convenzionali-e-terapie-curative

[10] AIL – Associazione Italiana contro le Leucemie, Linfomi e Mieloma. (n.d.). Anemia Falciforme. Retrieved October 24, 2025, from https://www.ail.it/informati-sulla-malattia/patologie-ematologiche/ail-anemie/ail-anemia-falciforme

[11] Russo, G., De Franceschi, L., Cappellini, M. D., et al. (2019). Current challenges in the management of patients with sickle cell disease in Italy. Journal of Clinical Medicine, 8(10), 1757. https://doi.org/10.3390/jcm8101757

[12] De Franceschi, L., et al. (2022). Real-world evidence on disease burden and economic impact of sickle cell disease in Italy. Journal of Clinical Medicine, 12(1), 117. https://doi.org/10.3390/jcm12010117

[13] Russo, G., De Franceschi, L., Colombatti, R., et al. (2019). Current challenges in the management of patients with sickle cell disease – The Italian experience. Orphanet Journal of Rare Diseases, 14(1), 120. https://doi.org/10.1186/s13023-019-1099-0

[14] Società Italiana di Ematologia (SIE). (2023). Documento di consenso: Gestione delle emoglobinopatie nell'adulto. Retrieved from https://www.siematologia.it/documenti

1. **Integration of SCD into routine clinical training**

In Italy, **paediatric haematology teams** generally follow international best practices, supported by national guidelines.[15] However, **adult haematology and primary care** show significant gaps in knowledge, particularly regarding **hydroxyurea use**, chronic complication monitoring, and transition protocols. Claims data linking these gaps to very low SCD-specific drug use (~6–7%) suggest uptake far below recommended levels.[16] [17] The **Società Italiana di Ematologia (SIE)** has issued consensus documents, but implementation remains inconsistent across regions.[18]

2. **Emergency care training and protocols for SCD crises**

Emergency departments are critical for managing **acute pain crises** and complications such as **acute chest syndrome**, yet Italian data show persistent delays in analgesia and inconsistent adherence to protocols.[19] [20]Targeted staff education reduced average wait times for pain relief from **87 to 64 minutes**, but this remains far from best practice. A **national CME programme** linked to ED protocols and registry participation could standardise care and reduce regional disparities.

Italy's training landscape is **fragmented**, with strong paediatric expertise but major gaps in adult and emergency care. Embedding **SCD-specific content** in medical curricula and implementing **mandatory CME programmes** for ED and adult haematology teams are essential to improve outcomes and ensure equity.[20] [21]

III. Comprehensive National Data, Registry and Research Infrastructure

Robust data collection is essential for improving outcomes and guiding policy in **sickle cell disease (SCD)**. Italy has made progress with its **National Haemoglobinopathy Register**, but challenges remain in achieving **complete coverage**, integrating **genotyping data**, and securing sustainable research funding. These gaps limit Italy's contribution to EU-wide monitoring and comparative policy evaluation. [22] [23]

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[15] Società Italiana di Talassemie ed Emoglobinopatie (SITE). (2023). Linee guida per la gestione della drepanocitosi in età pediatrica. Retrieved from https://www.site-italia.org/documenti/linee-guida-drepanocitosi-2023.pdf
[16] De Franceschi, L., et al. (2022). Real-world evidence on disease burden and economic impact of sickle cell disease in Italy. Journal of Clinical Medicine, 12(1), 117. https://doi.org/10.3390/jcm12010117
[17] Russo, G., De Franceschi, L., Colombatti, R., et al. (2019). Current challenges in the management of patients with sickle cell disease – The Italian experience. Orphanet Journal of Rare Diseases, 14(1), 120. https://doi.org/10.1186/s13023-019-1099-0
[18] SIE, 2023, op cit.
[19] Po', C., Colombatti, R., Cirigliano, A., et al. (2013). The management of sickle cell pain in the emergency department: A priority for health systems. The Clinical Journal of Pain, 29(1), 60–63. https://pubmed.ncbi.nlm.nih.gov/22751027/
[20] Russo, G., De Franceschi, L., Colombatti, R., et al. (2019). Current challenges in the management of patients with sickle cell disease – The Italian experience. Orphanet Journal of Rare Diseases, 14(1), 120. https://doi.org/10.1186/s13023-019-1099-0
[21] SIE, 2023, op cit ; Russo et al., 2019, op cit.
[22] Italian National Blood Centre (CNS). (2017). National Haemoglobinopathy Register. Retrieved from https://www.centronazionalesangue.it/en/national-haemoglobinopathy-register/
[23] Istituto Superiore di Sanità (ISS). (2022). Le emoglobinopatie in Italia: Rapporto epidemiologico nazionale 2022. Retrieved from https://www.iss.it/emoglobinopatie

1. **Establishment of a national SCD registry**

Italy established the **National Haemoglobinopathy Register** in 2017, coordinated by the **National Blood Centre (CNS)**. It covers both SCD and thalassemia and involves more than **100 centres nationwide**. While this is a major achievement, **data completeness varies**, with stronger participation in the North than in the South and Islands. Without uniform data, it is difficult to plan services, monitor outcomes, or assess the impact of new therapies.[24] [25] [26]

2. **Systematic National Collection and Integration of Genotype Data**

Genotyping is performed in many centres, but **integration into the national registry is inconsistent**. This limits the ability to conduct **genotype–phenotype analyses**, stratify patients for advanced therapies, and support personalised care. A structured approach to link **laboratory data with registry records** is needed to strengthen Italy's research and clinical capacity.[27] [28]

3. **Sustained funding for SCD data and research**

Research on SCD in Italy has been supported by **academic collaborations, patient associations**, and targeted projects, but there is **no large-scale recurring national funding stream** comparable to other rare diseases. Studies on **real-world burden, economic impact**, and **newborn screening feasibility** have been published, yet registry expansion and linkage to health system data remain underfunded.[29] [30] [31] Sustainable funding is critical to maintain Italy's role in **EU-wide initiatives** and accelerate innovation.

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[24] Italian National Blood Centre (CNS). (2017). National Haemoglobinopathy Register. Retrieved from https://www.centronazionalesangue.it/en/national-haemoglobinopathy-register/

[25] Istituto Superiore di Sanità (ISS). (2022). Le emoglobinopatie in Italia: Rapporto epidemiologico nazionale 2022. Retrieved from https://www.iss.it/emoglobinopatie

[26] Centro Nazionale Sangue & SIMTI. (2023). Registro Nazionale Talassemie e Drepanocitosi – Dati 2023. Retrieved from https://www.centronazionalesangue.it

[27] Società Italiana di Talassemie ed Emoglobinopatie (SITE). (2023). Linee guida per la gestione della drepanocitosi in età pediatrica. Retrieved from https://www.site-italia.org/documenti/linee-guida-drepanocitosi-2023.pdf

[28] Russo, G., De Franceschi, L., Colombatti, R., et al. (2019). Current challenges in the management of patients with sickle cell disease – The Italian experience. Orphanet Journal of Rare Diseases, 14(1), 120. https://doi.org/10.1186/s13023-019-1099-0

[29] De Franceschi, L., et al. (2022). Real-world evidence on disease burden and economic impact of sickle cell disease in Italy. Journal of Clinical Medicine, 12(1), 117. https://doi.org/10.3390/jcm12010117

[30] Associazione Piera Cutino. (2023). Progetti e attività per la drepanocitosi e le emoglobinopatie. Retrieved from https://www.pieracutino.it

[31] UNITED Onlus. (2024). Chi siamo e le nostre attività. Retrieved from https://www.unitedonlus.org

IV. Early detection through newborn screening

Early identification of **sickle cell disease (SCD)** through **newborn screening (NBS)** is critical to prevent severe complications and improve survival. Countries with universal NBS have demonstrated significant reductions in morbidity and mortality. In Italy, however, **universal screening** is still absent, leaving many infants undiagnosed until emergency presentations.[32] [33]

1. **National Implementation of SCD Universal Newborn Screening**

Italy does **not yet have nationwide universal NBS** for SCD. Screening is currently offered through **regional pilots** and voluntary programmes, despite evidence of feasibility and clinical benefit. Multicentre studies have shown that incidence justifies inclusion in the national panel, but policy implementation remains pending.

2. **Newborn screening coverage**

·Coverage is **low and uneven**, limited to regions that have adopted pilot programmes or point-of-care initiatives. Recent projects identified **0.3% of children with SCD** and up to **10% carriers**, but participation was poor when screening was voluntary.[34] Without national integration, many affected newborns miss early prophylaxis and family counselling.

3. **Standardisation of post-screening referral pathways**

Where screening occurs, referral pathways are **strong and clinically effective**: infants diagnosed early are linked to specialist centres for **penicillin prophylaxis**, vaccination, and **stroke-risk monitoring** in line with national guidelines.[35] However, the absence of universal screening means these benefits are not consistently delivered nationwide.

Italy's reliance on **regional pilots** and voluntary screening leaves significant gaps in early diagnosis. Implementing **universal NBS** and integrating it into the national panel is essential to ensure equity and prevent avoidable complications. Strong referral systems already exist and could be scaled nationally to maximise impact.

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[32] Mosca, A., et al. (2024). Screening for sickle cell disease: Focus on newborn screening in Italy. Biochimica Clinica, 48(3), 204–213. https://biochimicaclinica.it/wp-content/uploads/2024/06/BC-046-24_Mosca_Eng_dp.pdf

[33] Colombatti, R., Casale, M., Masera, N., et al. (2019). Results of a multicenter universal newborn screening program for sickle cell disease in Italy. Pediatric Blood & Cancer, 66(4), e27579. https://doi.org/10.1002/pbc.27579

[34] Casale, M., et al. (2025). Screening for sickle cell disease by point-of-care tests in Italy. Italian Journal of Pediatrics, 51, 12. https://pubmed.ncbi.nlm.nih.gov/39875760/

[35] Società Italiana di Talassemie ed Emoglobinopatie (SITE). (2023). Linee guida per la gestione della drepanocitosi in età pediatrica. Retrieved from https://www.site-italia.org/documenti/linee-guida-drepanocitosi-2023.pdf

V. Coordinated policy and patient partnership

Effective **multi-stakeholder collaboration** is essential to address systemic gaps in **sickle cell disease (SCD)** care. In Italy, dialogue between clinicians, policymakers, and patient organisations exists but remains **fragmented**, limiting progress on national screening, equitable access to therapies, and standardised emergency protocols.[36] [37]

1. **Formal national multi-stakeholder SCD coordination platform**

Italy lacks a **formal national platform dedicated exclusively to SCD**, relying instead on broader haemoglobinopathy networks and rare disease initiatives. Coordination occurs through the **National Blood Centre (CNS)** and scientific societies such as **SITE** and **SIE**, which issue guidelines and advocate for improved care. However, governance is **project-based rather than institutionalised**, and there is no structured mechanism for integrating patient voices into policy decisions.[38] [39] [40]

2. **Inclusion of patient organisations in decision-making**

Patient organisations play a **critical role** in advocacy and awareness. Groups such as **UNITED Onlus** and **Associazione Piera Cutino** lead campaigns for **universal newborn screening**, improved registry data, and equitable access to advanced therapies. Despite their efforts, **formal integration into national planning and registry governance remains limited**, reducing the potential impact of patient perspectives on resource allocation and policy.[41]

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[36] Russo, G., De Franceschi, L., Colombatti, R., et al. (2019). Current challenges in the management of patients with sickle cell disease – The Italian experience. Orphanet Journal of Rare Diseases, 14(1), 120. https://doi.org/10.1186/s13023-019-1099-0

[37] Associazione Piera Cutino. (2023). Progetti e attività per la drepanocitosi e le emoglobinopatie. Retrieved from https://www.pieracutino.it

[38] Centro Nazionale Sangue & SIMTI. (2023). Registro Nazionale Talassemie e Drepanocitosi – Dati 2023. Retrieved from https://www.centronazionalesangue.it

[39] Società Italiana di Ematologia (SIE). (2023). Documento di consenso: Gestione delle emoglobinopatie nell'adulto. Retrieved from https://www.siematologia.it/documenti

[40] Società Italiana di Talassemie ed Emoglobinopatie (SITE). (2023). Linee guida per la gestione della drepanocitosi in età pediatrica. Retrieved from https://www.site-italia.org/documenti/linee-guida-drepanocitosi-2023.pdf

[41] UNITED Onlus. (2024). Chi siamo e le nostre attività. Retrieved from https://www.unitedonlus.org

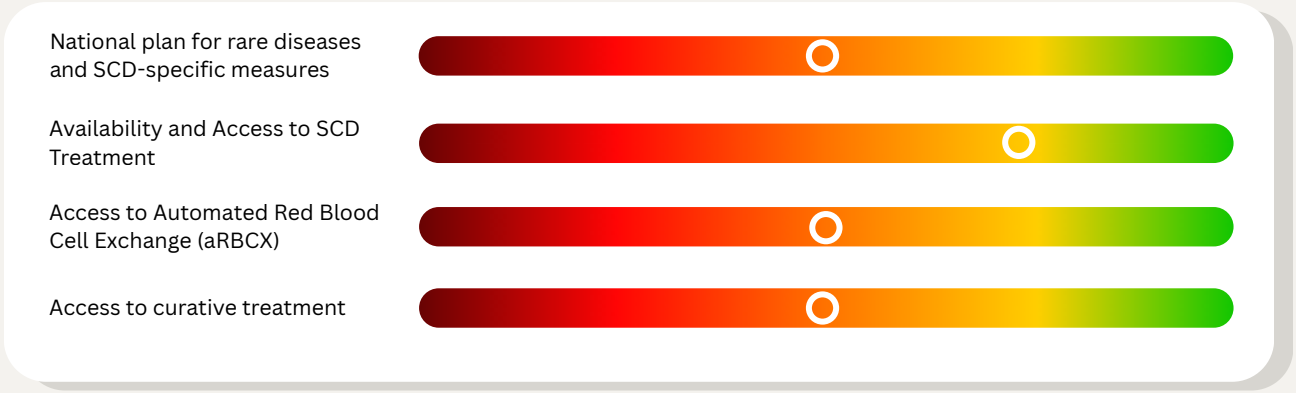
The Netherlands
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

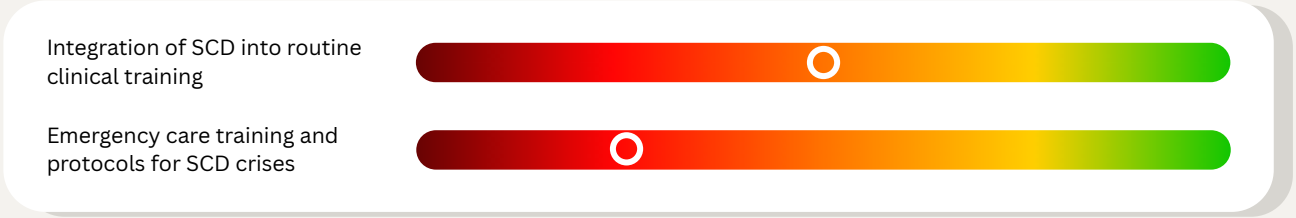
Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment				
2. Strengthened National Workforce Capacity and Preparedness for SCD				
3. Comprehensive National Data, Registry and Research Infrastructure				
4. Early detection through newborn screening				
5. Coordinated policy and patient partnership				

Key Performance Indicator (KPI) Overview

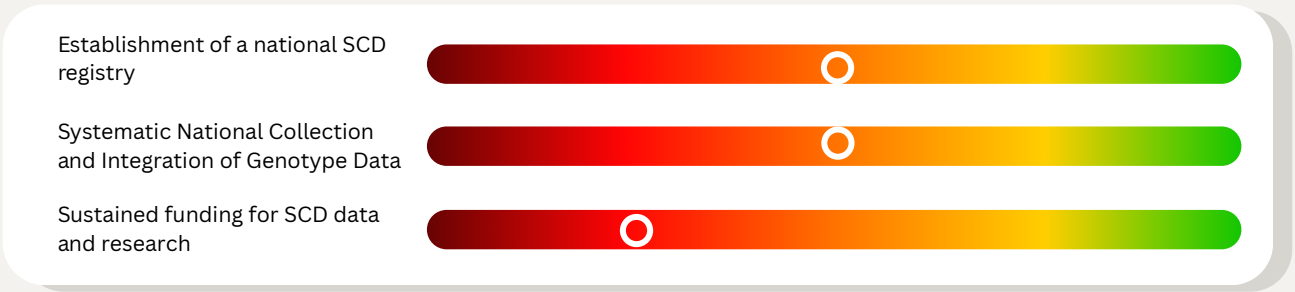
1. Equitable access to SCD care and treatment



2. Strengthened National Workforce Capacity and Preparedness for SCD



3. Comprehensive National Data, Registry and Research Infrastructure



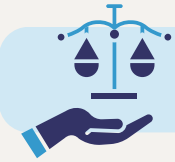
4. Early detection through newborn screening

5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	The Netherlands has a National Plan for Rare Diseases, which recognises rare conditions and provides a strategic framework, but it does not contain sickle cell-specific implementation measures. Regional implementation and attention to haemoglobinopathies differ between university centres and smaller hospitals.	
Availability and Access to SCD Treatment	Hydroxyurea is widely available and routinely used in paediatric and major university hospitals. Zorginstituut Nederland issued a positive reimbursement advice for gene editing therapy in February 2026, with access now being implemented after price negotiation.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX is available in universities and large teaching hospitals, particularly in the Randstad, but capacity is limited in smaller regional hospitals, which may rely on simple transfusion or referral. This leads to geographic variation in access to optimal transfusion strategies.	
Access to curative treatment	HSCT for sickle cell disease is performed in a small number of transplant centres with outcomes comparable to other EU countries. However, donor availability, strict eligibility criteria and centre capacity limit access for most patients.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric and adult haematologists in university centres have strong expertise and participate in national haemoglobinopathy networks. However, sickle cell disease is not consistently embedded in broader internal medicine and general practice training, leading to variable awareness outside centres of excellence.	
Emergency care training and protocols for SCD crises	Specialist centres have local emergency protocols and training for vaso-occlusive crises and acute chest syndrome. Smaller hospitals report less familiarity and occasional delays in analgesia and escalation, as dedicated sickle cell training is not standardised nationwide.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	The SCORE (Sickle Cell Outcome Research) initiative forms a national research registry linking all Dutch sickle cell centres and collecting longitudinal data on outcomes. Participation is strong in university hospitals, but registration is not yet a mandatory national administrative registry.	
Systematic National Collection and Integration of Genotype Data	Specialist centres and the national haemoglobinopathy expert laboratory routinely collect genotyping information, but systematic linkage of genotype data to clinical outcomes across all centres is still developing.	
Sustained funding for SCD data and research	Registry and outcomes research rely largely on project-based funding, foundations and academic grants. There is no permanent dedicated national funding stream for sickle cell disease registries and long-term observational research.	
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	The Netherlands was among the early adopters of newborn blood spot screening for haemoglobinopathies, and sickle cell disease is included in the national heel prick programme.	
Newborn screening coverage	National monitoring reports show very high coverage of the newborn blood spot programme. However, children born abroad and arriving after the neonatal period are not captured by screening and may be diagnosed later, which introduces some inequities.	

KPI	Overview	Score
4. Early detection through newborn screening		
Standardisation of post-screening referral pathways	Abnormal heel prick results are promptly referred to paediatric haematology centres, where confirmatory testing, prophylaxis and counselling are provided. Pathways function well, although systematic linkage of screening data with long-term registry data is still being optimised.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	The Netherlands benefits from close collaboration between university centres, national haemoglobinopathy expertise networks and governmental agencies. However, there is no single national governance body dedicated exclusively to sickle cell disease policy, and coordination remains embedded in broader rare disease structures.	
Inclusion of Patient Organisations in decision-making	Patient organisations such as OSCAR Nederland, IXL Sickle Cell Awareness and the Sikkelcelfonds are active in awareness, advocacy and co-creation of tools such as PROMs. Their involvement in formal decision-making and national planning is increasing, but not yet fully systematic across all policy processes.	

The Hidden Struggle: SCD in the Netherlands



I. Equitable access to SCD care and treatment

1. **National plan for rare diseases and SCD-specific measures**

The Netherlands has a National Plan for Rare Diseases that provides a strategic framework for improving diagnosis, treatment and integrated care for rare conditions, although it does not contain sickle cell specific implementation measures[1]. Implementation of rare disease policies is strongly linked to university medical centres, which act as expertise hubs, and this creates variation between academic hospitals and smaller regional hospitals in access to multidisciplinary care[2]. As a result, access to specialist sickle cell disease services and advanced therapies depends on geographical proximity to centres of expertise and on regional referral practices[3].

2. **Availability and Access to SCD Treatment**

Hydroxyurea is widely available in the Netherlands and is routinely used in specialist paediatric and adult haematology centres, where clinical practice follows national and European guidance[4]. Chronic transfusion programmes are available in all major university hospitals and are used for stroke prevention and severe disease, although the frequency of follow-up and access to specialist transfusion services differ between university and regional settings[5]. New gene editing therapies for sickle cell disease have been authorised at the European level. Following assessment within the ZIN sluis procedure, Zorginstituut Nederland issued a positive reimbursement advice in February 2026, and access is now being implemented after national price negotiation[6].

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[1] Ministry of Health, Welfare and Sport. (2013). National Plan for Rare Diseases (Nationaal Plan Zeldzame Ziekten). https://www.rijksoverheid.nl/documenten/rapporten/2013/10/10/nationaal-plan-zeldzame-ziekten
[2] Dutch Federation for University Medical Centres (NFU). (2020). Expertise centres for rare diseases. https://www.nfu.nl/kwaliteit/expertisecentra-zeldzame-aandoeningen
[3] Dutch Sickle Cell Disease Expert Group / SCORE-NL. (2022). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net
[4] Dutch Society for Haematology (NVvH). (2021). Richtlijn hemoglobinopathieën. https://richtlijnendatabase.nl/richtlijn/hemoglobinopathieen
[5] Netherlands Sickle Cell Expert Centres (Amsterdam UMC, LUMC, Erasmus MC). (2023). Clinical care pathways for haemoglobinopathies in the Netherlands. https://www.amsterdamumc.org/en/research/haemoglobinopathies
[6] Zorginstituut Nederland. (2026). Pakketadvies exagamglogene autotemcel (Casgevy) - sluisprocedure. https://www.zorginstituutnederland.nl

3. **Automated Red Blood Cell Exchange (aRBCX)**

Automated red blood cell exchange is available in university and large teaching hospitals, particularly within the Randstad region, and is used for stroke prevention and management of severe complications[7]. In addition to in-house capacity in academic and larger centres, apheresis services for smaller regional hospitals are partly supported by Sanquin, which operates mobile apheresis capacity to provide services where devices are not available on-site. Smaller regional hospitals otherwise rely on simple transfusion or referral to academic centres, which introduces variation in access to optimal transfusion strategies across regions. Awareness of aRBCX as a treatment option in regional hospitals may also vary[8]. Smaller regional hospitals lack apheresis units and rely on simple transfusion or referral to academic centres, which introduces variation in access to optimal transfusion strategies across regions[9].

4. **Access to curative treatment**

Haematopoietic stem cell transplantation for sickle cell disease is performed in specialised transplant centres in the Netherlands, mainly for paediatric patients with severe disease who have a suitable donor[10]. Outcomes are comparable to other European countries, but donor availability, strict eligibility criteria and limited transplant capacity restrict access to this curative treatment option for most patients[11].

II. Strengthened National Workforce Capacity and Preparedness for SCD

1. **Integration of SCD into routine clinical training**

Paediatric and adult haematologists in Dutch university medical centres have strong expertise in haemoglobinopathies and follow national and European guidance on sickle cell disease. Participation in national initiatives such as the SCORE sickle cell outcome research infrastructure and concentration of care in recognised expertise centres help maintain a high level of knowledge in these hospitals[12].

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[7] Netherlands Sickle Cell Expert Centres (Amsterdam UMC, LUMC, Erasmus MC). (2023). Clinical care pathways for haemoglobinopathies in the Netherlands. https://www.amsterdamumc.org/en/research/haemoglobinopathies
[8] Sanquin Blood Supply. (n.d.). Our expertise and knowledge: Therapeutic apheresis services. Sanquin. Retrieved January 19, 2026, from https://www.sanquin.org/products-and-services/immunomonitoring-services/expertise-and-knowledge/index
[9] Dutch Sickle Cell Disease Expert Group / SCORE-NL. (2022). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net
[10] European Society for Blood and Marrow Transplantation. (2023). EBMT transplant activity report: Netherlands. https://www.ebmt.org/ebmt/documents/ebmt-activity-survey
[11] Ibid.
[12] Dutch Society for Haematology (NVvH). (2021). Richtlijn hemoglobinopathieën. https://richtlijnendatabase.nl/richtlijn/hemoglobinopathieen

However, sickle cell disease is not systematically integrated into training for general internal medicine, general practice and other non-specialist disciplines, which results in variable awareness and confidence outside university centres[13][14]. European rare disease education programmes provide additional opportunities for Dutch clinicians to develop skills in the diagnosis and management of rare conditions, but participation is voluntary and not specific to sickle cell disease[15].

2. **Emergency care training and protocols for SCD crises**

Emergency departments in university centres have developed local protocols for vaso-occlusive crises and have implemented multimodal pain management pathways to improve timeliness and quality of analgesia[16]. Despite these advances, there is no nationwide mandatory emergency department training standard for sickle cell disease, and smaller hospitals report less familiarity with specific protocols for acute chest syndrome and severe pain[17][18]. Overall, emergency care quality still depends on the level of local experience and the presence of formal collaboration with haemoglobinopathy expertise centres[19].

III. Comprehensive National Data, Registry and Research Infrastructure

1. **Establishment of a national SCD registry**

The Netherlands has developed a national research registry for sickle cell disease through the SCORE initiative, which links all major sickle cell disease centres and collects longitudinal clinical data[20]. SCORE participation is strong in university hospitals, and the registry captures information on complications, organ damage, treatments and outcomes[21][22]. However, registration is not yet part of a formal nationwide administrative registry system, and participation outside university centres is more limited, which reduces the visibility of patients treated in regional hospitals[23].

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[13] UMC Utrecht. (2024). (Inter)National research collaborations: SCORE (Sickle Cell Outcome Research). https://www.umcutrecht.nl/nl/inter-nationale-onderzoekssamenwerkingen
[14] Erasmus MC. (2024). Expertisecentrum voor sikkelcelziekte en thalassemie. https://www.erasmusmc.nl/nl-nl/patientenzorg/centra/sikkelcel-of-hemoglobinopathie
[15] Innovation for Rare Diseases Study Group. (2021). Innovation for rare diseases: How does the Netherlands compare to other European countries? Copernicus Institute, Utrecht University. https://www.uu.nl/en/research/copernicus-institute-of-sustainable-development/innovation-for-rare-diseases-how-does-the-netherlands-compare-to-other-european-countries
[16] European Joint Programme on Rare Diseases (EJP RD). (2023). Training and empowerment programme. https://www.ejprarediseases.org/general-information-training/
[17] Ibid.
[18] Erasmus MC. (2024). Expertisecentrum voor sikkelcelziekte en thalassemie. https://www.erasmusmc.nl/nl-nl/patientenzorg/centra/sikkelcel-of-hemoglobinopathie
[19] UMC Utrecht. (2024). (Inter)National research collaborations: SCORE (Sickle Cell Outcome Research). https://www.umcutrecht.nl/nl/inter-nationale-onderzoekssamenwerkingen
[20] UMC Utrecht. (2024). (Inter)National research collaborations: SCORE (Sickle Cell Outcome Research). https://www.umcutrecht.nl/nl/inter-nationale-onderzoekssamenwerkingen
[21] Ibid.
[22] Erasmus MC. (2024). Expertisecentrum voor sikkelcelziekte en thalassemie. https://www.erasmusmc.nl/nl-nl/patientenzorg/centra/sikkelcel-of-hemoglobinopathie
[23] Dutch Sickle Cell Expert Group. (2023). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net

2. **Systematic National Collection and Integration of Genotype Data**

Specialist laboratories in the Netherlands routinely perform genotyping for haemoglobinopathies and support the national network of expertise centres[24]. Although genotyping is widely available, linkage of genotype data to longitudinal outcomes across all centres is still being developed and is not yet fully integrated into the SCORE registry[25] [26].

3. **Sustained funding for SCD data and research**

Sickle cell disease registry and outcomes research in the Netherlands is primarily supported by academic grants, foundations and temporary project-based funding[27]. There is currently no permanent national funding mechanism dedicated specifically to sickle cell disease registries or long-term patient follow-up, which limits the capacity to expand data collection and conduct nationwide analyses[28].

IV. Early detection through newborn screening

1. **National Implementation of SCD Universal Newborn Screening**

The Netherlands includes sickle cell disease in its national newborn blood spot screening programme, which screens all newborns through the heel prick test[29]. Sickle cell disease was added to the national panel following evidence from pilot studies demonstrating that early diagnosis enables timely prophylaxis and reduces the risk of severe complications[30]. Coverage of the newborn screening programme is consistently high across the country, with participation approaching the levels seen for other long-standing screening conditions[31].

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[24] Erasmus MC. (2024). Expertisecentrum voor sikkelcelziekte en thalassemie. https://www.erasmusmc.nl/nl-nl/patientenzorg/centra/sikkelcel-of-hemoglobinopathie
[25] UMC Utrecht. (2024). (Inter)National research collaborations: SCORE (Sickle Cell Outcome Research). https://www.umcutrecht.nl/nl/inter-nationale-onderzoekssamenwerkingen
[26] Dutch Sickle Cell Expert Group. (2023). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net
[27] ZonMw. (2023). Haemoglobinopathies research and innovation funding overview. https://www.zonmw.nl/en/programmes/rare-diseases
[28] Ibid.
[29] RIVM. (2024). Neonatal heel prick screening programme (Hielprikprogramma). https://www.rivm.nl/hielprik
[30] Loeber, J. G., et al. (2012). Neonatal screening in Europe: The situation in 2007. Journal of Inherited Metabolic Disease, 35(3), 603–615. https://link.springer.com/article/10.1007/s10545-012-9493-0
[31] RIVM. (2024). Neonatal heel prick screening programme (Hielprikprogramma). https://www.rivm.nl/hielprik

2. **Newborn screening coverage**

Monitoring reports indicate that screening coverage is above ninety-nine per cent nationwide, although children born abroad who arrive in the Netherlands later in infancy are not captured by the heel prick programme[32].

Regional analyses confirm that newborn screening enables early detection of sickle cell disease and supports prompt referral to specialist care centres[33].

3. **Standardisation of post-screening referral pathways**

Referral pathways for abnormal newborn screening results are well established, with rapid communication between screening laboratories and paediatric haematology teams[34].

Confirmatory testing and the initiation of penicillin prophylaxis, vaccination schedules and transcranial Doppler screening follow national and European clinical recommendations[35]. However, the full integration of newborn screening data with long-term clinical registries such as SCORE is not yet complete, which limits the ability to track outcomes into adolescence and adulthood[36].

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[24] Erasmus MC. (2024). Expertisecentrum voor sikkelcelziekte en thalassemie. https://www.erasmusmc.nl/nl-nl/patientenzorg/centra/sikkelcel-of-hemoglobinopathie
[25] UMC Utrecht. (2024). (Inter)National research collaborations: SCORE (Sickle Cell Outcome Research). https://www.umcutrecht.nl/nl/inter-nationale-onderzoekssamenwerkingen
[26] Dutch Sickle Cell Expert Group. (2023). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net
[27] ZonMw. (2023). Haemoglobinopathies research and innovation funding overview. https://www.zonmw.nl/en/programmes/rare-diseases
[28] Ibid.
[29] RIVM. (2024). Neonatal heel prick screening programme (Hielprikprogramma). https://www.rivm.nl/hielprik
[30] Loeber, J. G., et al. (2012). Neonatal screening in Europe: The situation in 2007. Journal of Inherited Metabolic Disease, 35(3), 603–615. https://link.springer.com/article/10.1007/s10545-012-9493-0
[31] RIVM. (2024). Neonatal heel prick screening programme (Hielprikprogramma). https://www.rivm.nl/hielprik
[32] RIVM. (2023). Neonatale hielprikscreening: Monitor 2023. https://www.rivm.nl/hielprik/monitoring
[33] Loeber, J. G., et al. (2012). Neonatal screening in Europe: The situation in 2007. Journal of Inherited Metabolic Disease, 35(3), 603–615. https://link.springer.com/article/10.1007/s10545-012-9493-0
[34] RIVM. (2024). Neonatal heel prick screening programme (Hielprikprogramma). https://www.rivm.nl/hielprik
[35] Dutch Society for Haematology (NVvH). (2021). Richtlijn hemoglobinopathieën. https://richtlijnendatabase.nl/richtlijn/hemoglobinopathieen
[36] Dutch Sickle Cell Expert Group. (2023). SCORE-NL national sickle cell disease research infrastructure overview. https://sikkelcel-en-thalassemie-expertise.net

V. Coordinated policy and patient partnership

1. Formal national multi-stakeholder SCD coordination platform

Strategic dialogue on sickle cell disease in the Netherlands is supported by close cooperation between university medical centres, the national haemoglobinopathy expertise network and patient organisations, but there is no single formal national council dedicated exclusively to sickle cell policy[37].

The national expertise network for sickle cell disease and thalassaemia brings together clinicians, laboratories, researchers and patient representatives and supports joint projects such as the SCORE outcome research initiative[38][39]. These structures provide a basis for multi-stakeholder collaboration on care pathways, research and information, but formalised, government-led governance for sickle cell disease remains limited[40].

2. Inclusion of patient organisations in decision-making

Patient organisations play a visible role in awareness raising, peer support and advocacy for people with sickle cell disease in the Netherlands[41][42]. OSCAR Nederland acts as a multi ethnic patient organisation for people with sickle cell disease and thalassaemia, providing information, support groups and advice to patients and healthcare professionals[43].

IXL Sickle Cell Awareness focuses on empowerment, public awareness and community outreach, including collaboration with the national blood bank Sanquin to promote blood donation among communities at higher genetic risk[44]. The Sikkelcelfonds supports research funding and public campaigns to increase recognition of sickle cell disease, and its activities complement those of the patient organisations. While these organisations are increasingly involved in consultations with expertise centres and participate in network meetings, their participation in formal national health planning and decision-making processes is still mostly project-based rather than embedded in a structured national sickle cell governance framework[45].

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[37] [38] Ibid.
[39] Sikkelcel en thalassemie expertise netwerk. (2022). SCORE – Landelijk onderzoek naar de gezondheidsuitkomsten van sikkelcelziekte. https://sikkelcel-en-thalassemie-expertise.net/research/score-landelijk-onderzoek-naar-de-gezondheidsuitkomsten-van-sikkelcelziekte-om-zo-de-behandeling-te-verbeteren/
[40] Ibid.
[41] Zicht op Zeldzaam. (2024). OSCAR Nederland: Multi-etnische stichting voor patiënten met sikkelcelanemie en thalassemie. https://zichtopzeldzaam.nl/organisaties/oscar-nederland/
[42] Sikkelcel en thalassemie expertise netwerk. (2024). Patiëntenorganisaties voor sikkelcelziekte en thalassemie. https://sikkelcel-en-thalassemie-expertise.net/voor-patienten/patientenorganisatie/
[43] Zicht op Zeldzaam. (2024). OSCAR Nederland: Multi-etnische stichting voor patiënten met sikkelcelanemie en thalassemie.
https://zichtopzeldzaam.nl/organisaties/oscar-nederland/
[44] Sanquin. (2021, June 23). Donors met Afrikaanse roots zijn hard nodig.
https://www.sanquin.nl/donors/actueel/donors-met-afrikaanse-roots-zijn-hard-nodig
[45] Sikkelcel en thalassemie expertise netwerk. (2024). Patiëntenorganisaties: OSCAR Nederland en IXL Sickle Cell Awareness. https://sikkelcel-en-thalassemie-expertise.net/voor-patienten/patientenorganisatie/

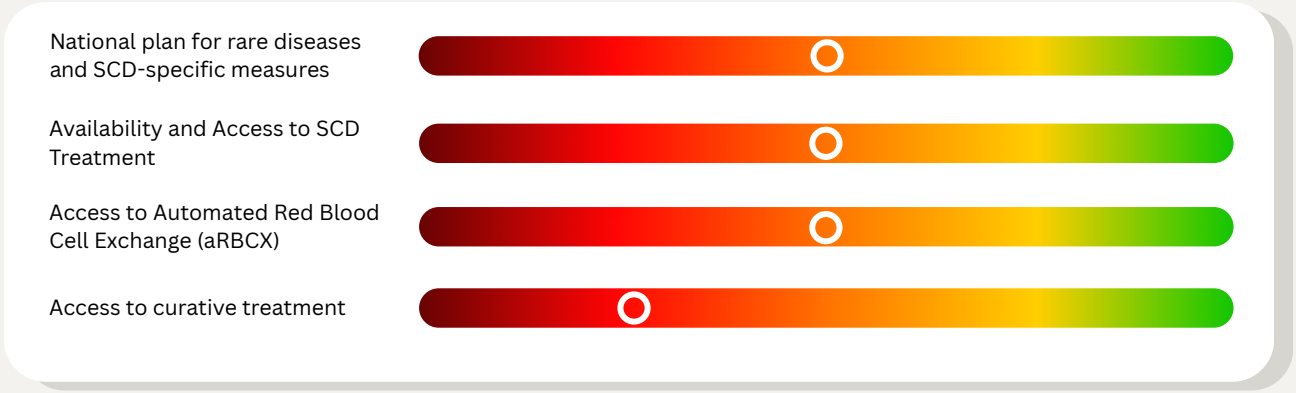
Portugal
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment				
2. Strengthened National Workforce Capacity and Preparedness for SCD				
3. Comprehensive National Data, Registry and Research Infrastructure				
4. Early detection through newborn screening				
5. Coordinated policy and patient partnership				

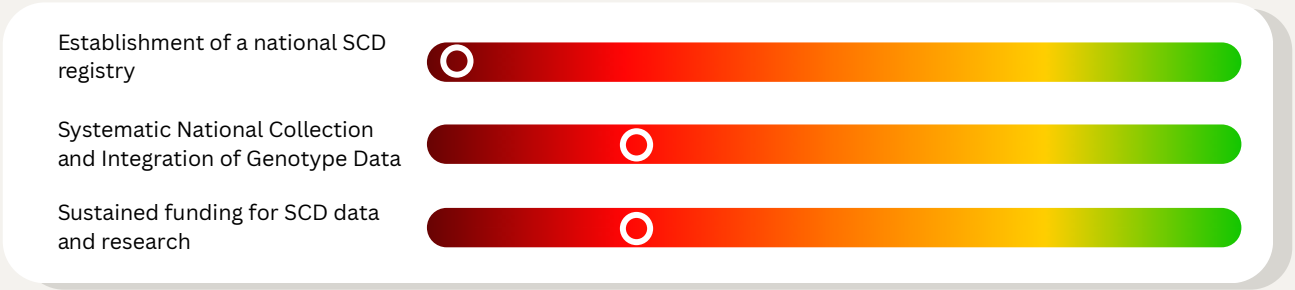
Key Performance Indicator (KPI) Overview

1. Equitable access to SCD care and treatment



2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure



4. Early detection through newborn screening

5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	Portugal has a National Programme for Rare Diseases and a new Action Plan for Rare Diseases 2025–2030, which provides a strategic framework for rare conditions. However, there are no sickle cell-specific implementation measures, and practical delivery depends heavily on a small number of university centres.	
Availability and Access to SCD Treatment	Core disease-modifying and supportive therapies, including hydroxyurea and chronic transfusion, are available and used in specialist centres in Lisbon and Porto, as well as the Coimbra University Hospital. Evidence from Portuguese cohort studies shows benefits of hydroxyurea in children, but access and optimal dosing remain more uneven outside these centres, and adult follow-up is less structured.	
Access to Automated Red Blood Cell Exchange (aRBCX)	Automated red blood cell exchange is available only in a few hospitals with apheresis capacity, mainly in Lisbon and Porto. Many hospitals rely on simple transfusion or must transfer patients, creating significant regional inequities in access to optimal transfusion strategies	
Access to curative treatment	Haematopoietic stem cell transplantation for sickle cell disease is performed in a small number of paediatric transplant centres, with strict eligibility criteria and limited donor availability. Adult transplantation is rare, and overall curative access remains restricted for the majority of patients.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric haematology teams in Lisbon and Porto are experienced and follow international recommendations, but sickle cell disease is not consistently embedded in general internal medicine, family medicine or primary care training. Knowledge and confidence are considerably weaker in smaller hospitals and in adult services.	
Emergency care training and protocols for SCD crises	Emergency departments frequently lack specific protocols for vaso-occlusive crises and acute chest syndrome. Reports from Portuguese cohorts and patient feedback highlight delays in analgesia and variable recognition of complications, and there is no national standard for emergency department training in sickle cell disease.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	Portugal does not yet have a dedicated national sickle cell disease registry. Available information comes from hospital based cohorts, cross sectional studies and pilot projects, which do not provide comprehensive nationwide coverage.	
Systematic National Collection and Integration of Genotype Data	Genotyping for haemoglobinopathies is routinely available in reference laboratories and used in clinical practice, but systematic linkage of genetic data with longitudinal clinical outcomes at national level is lacking.	
Sustained funding for SCD data and research	Research on sickle cell disease in Portugal, including adult characterisation and impact studies, relies largely on academic initiatives, European collaborations and time limited project funding. There is no permanent national funding stream dedicated specifically to sickle cell disease registries or long-term outcome research.	

KPI	Overview	Score
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	Portugal integrated sickle cell disease into the national newborn screening panel in 2023, following a national pilot in which around 100,000 newborns were screened between 2021 and 2022. This places Portugal among the first EU countries to include sickle cell disease in the national panel.	
Newborn screening coverage	The Portuguese newborn screening programme is nationally organised and achieves very high coverage, similar to other screened conditions. Pilot and early implementation data confirm that most newborns are tested, although children born abroad and arriving later are not captured.	
Standardisation of post-screening referral pathways	Pilot data show that newborn screening enables early diagnosis, penicillin prophylaxis and vaccination; however, before screening a substantial proportion of children were still diagnosed during hospitalisation after severe complications. Early implementation has strengthened referral pathways to paediatric haematology, but full integration with long-term follow-up and adult transition remains a work in progress.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	Strategic dialogue on sickle cell disease in Portugal is driven mainly by collaborations between the national newborn screening programme, university centres, APPDH and emerging networks such as ALUA and DrepaComunidade. Portugal participates in European initiatives, including ERN EuroBloodNet and the first international conference on sickle cell disease, but there is no dedicated national governance body focused solely on sickle cell disease policy.	
Inclusion of Patient Organisations in decision-making	Patient organisations such as APPDH and ALUA, and initiatives like DrepaComunidade, are highly active in awareness, community outreach and advocacy, and they played an important role in the implementation of newborn screening. Their involvement in formal decision making and long term planning is increasing but remains project based rather than embedded in a structured national sickle cell policy framework.	

The Hidden Struggle: SCD in Portugal

I. Equitable access to SCD care and treatment

1. **National plan for rare diseases and SCD-specific measures**

Portugal has a National Programme for Rare Diseases and a new Action Plan for Rare Diseases 2025–2030, which provides a strategic framework for rare conditions. However, there are no sickle cell-specific implementation measures, and practical delivery depends heavily on a small number of university centres.[1][2]

2. **Availability and Access to SCD Treatment**

Hydroxyurea is available and reimbursed in Portugal and is widely used in paediatric care, where cohort studies have shown reductions in hospitalisations and acute complications after treatment initiation[3].

Adult access to hydroxyurea is more variable, and long-term follow-up outside specialist centres remains inconsistent, which contributes to unequal uptake. Chronic transfusion programmes are available in major hospitals and are used for stroke prevention and severe disease, although access to coordinated transfusion care outside Lisbon and Porto is more limited[4].

Gene editing therapies for sickle cell disease have been approved at the European level, but access in Portugal will depend on evaluation and pricing decisions led by INFARMED, and these therapies are not yet available in routine practice[5].

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[1] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt/

[2] Marcão, A., et al. (2025). Newborn screening for sickle cell disease: Results from a pilot study in the Portuguese population. International Journal of Neonatal Screening, 11(1), 10. https://doi.org/10.3390/ijns11010010

[3] Alves, C., et al. (2020). Hydroxyurea in children with sickle cell disease in Portugal: Clinical outcomes from a national cohort. Acta Médica Portuguesa, 33(12), 789–796. https://www.actamedicaportuguesa.com/revista/index.php/amp/article/view/14053

[4] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

[5] INFARMED. (2024). Avaliação de terapias avançadas para doenças hereditárias do sangue. Autoridade Nacional do Medicamento e Produtos de Saúde. https://www.infarmed.pt/

3. **Automated Red Blood Cell Exchange (aRBCX)**

Automated red blood cell exchange is available only in a few hospitals with apheresis capacity in Lisbon and Porto, and is rarely accessible in regional settings. Hospitals without an apheresis capability rely on simple transfusion or must transfer patients to specialist centres, which creates substantial inequities in access to optimal transfusion strategies[6].

4. **Access to curative therapies (HSCT)**

Haematopoietic stem cell transplantation for sickle cell disease is performed in a small number of paediatric transplant centres, mainly for severe cases with an appropriate donor.

Transplant activity remains low due to donor limitations, strict eligibility criteria and limited national transplant capacity, which restrict access to curative treatment for most patients[7].

II. Strengthened National Workforce Capacity and Preparedness for SCD

1. **Integration of SCD into routine clinical training**

Specialist knowledge of sickle cell disease in Portugal is concentrated in paediatric haematology units in Lisbon and Porto, where clinicians follow European recommendations and participate in international networks[8].

Outside these centres, sickle cell disease is not consistently integrated into training for internal medicine, family medicine and general adult haematology, which leads to significant variation in clinical familiarity[9].

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[6] Marcão, A., et al. (2025). Newborn screening for sickle cell disease: Results from a pilot study in the Portuguese population. International Journal of Neonatal Screening, 11(1), 10. https://doi.org/10.3390/ijns11010010

[7] Portuguese Society of Haematology (SPH). (2023). Transplantation practices and indications in haemoglobinopathies in Portugal. https://sph.org.pt/

[8] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt/

[9] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

Studies examining rare disease preparedness in Portugal have noted that many healthcare professionals feel insufficiently trained to diagnose and manage rare conditions, including sickle cell disease, especially in non-specialist settings[10].
The lack of structured training pathways means that adult services often have limited experience in long-term management, which contributes to gaps in monitoring, treatment optimisation and transition from paediatric care[11].

2. Emergency care training and protocols for SCD crises

Emergency departments in Portugal frequently lack specific protocols for vaso-occlusive crises, and patient reports indicate delays in analgesia and inconsistent recognition of acute complications such as acute chest syndrome[12][13].

While specialist hospitals have internal guidelines for managing acute presentations, these have not been standardised nationally and do not reach all emergency care providers[14].
National rare disease policy acknowledges the need to strengthen clinical training, but no national emergency department training standard for sickle cell disease has yet been introduced[15].

III. Comprehensive National Data, Registry and Research Infrastructure

1. Establishment of a national SCD registry

Portugal does not yet have a dedicated national sickle cell disease registry, and most available information comes from hospital-based cohorts, single-centre series and multicentre studies[16][17].

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[10] Costa, E., et al. (2021). Professional training needs for rare diseases in Portugal: A national assessment. Orphanet Journal of Rare Diseases, 16(412). https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-02076-4

[11] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

[12] Ibid.

[13] Hospital de Santa Maria. (2022). Protocolo interno para crises de dor aguda e complicações da doença falciforme. Centro Hospitalar Universitário Lisboa Norte. https://www.chln.pt/

[14] Ibid.

[15] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt/

[16] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

[17] Brás, A., et al. (2022). Adult sickle cell disease in Portugal: Clinical profile and healthcare use in a multicentre cohort. Acta Médica Portuguesa, 35(6), 457–465. https://www.actamedicaportuguesa.com/revista/index.php/amp/article/view/17021

Existing datasets provide useful insight into the characteristics of Portuguese sickle cell disease populations and patterns of care, but they do not offer comprehensive nationwide coverage or long-term follow-up for all patients[18][19].

The absence of a national registry limits the ability to monitor outcomes systematically, to assess regional inequalities in care and to generate robust evidence for planning services and evaluating new therapies[20].

2. **Systematic National Collection and Integration of Genotype Data**

Genotyping for haemoglobinopathies is routinely available in reference laboratories in Portugal and is used to confirm diagnoses and inform family counselling[21]. However, genetic results are not yet systematically linked to clinical outcome data at the national level, and information on genotype-phenotype correlations is mainly derived from research cohorts rather than from an integrated registry[22][23].

3. **Sustained funding for SCD data and research**

Research on sickle cell disease in Portugal, including studies characterising adult and paediatric cohorts, is mainly driven by academic centres and supported by competitive research grants and European funding rather than by a permanent national programme.

The broader rare disease community in Portugal has highlighted the need for more stable and predictable funding for registries and data infrastructures that can support long-term clinical research and health service planning[24].

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[18] Ibid.

[19] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

[20] European Haematology Association. (2023). Country review: Sickle cell disease care in Southern Europe. https://ehaweb.org/

[21] Centro Hospitalar Universitário Lisboa Norte. (2022). Unidade de Hemoglobinopatias: Actividade assistencial e laboratorial. Hospital de Santa Maria. https://www.chln.pt/

[22] Ibid.

[23] Barometer country desk research, Portugal (2025), consolidating publicly available hospital, regulatory and patient organisation information.

[24] Direção-Geral da Saúde. (2021). Programa Nacional para as Doenças Raras: Avaliação intermédia e prioridades de desenvolvimento. Ministério da Saúde, Portugal. https://www.dgs.pt/

IV. Early detection through newborn screening

1. **National Implementation of SCD Universal Newborn Screening**

Portugal integrated sickle cell disease into its universal newborn screening panel in 2023, following a national pilot phase in which around 100,000 newborns were screened between May 2021 and December 2022. The pilot demonstrated high feasibility and confirmed that early diagnosis through newborn screening improves access to prophylaxis and specialist follow-up.
With this integration, Portugal became one of the first EU member states to add sickle cell disease to its universal screening panel, aligning the programme with international recommendations[25][26].

2. **Newborn screening coverage**

Portugal's newborn screening system is centrally coordinated and historically achieves very high coverage across the country, and early implementation reports indicate that sickle cell screening is reaching nearly all newborns[27].

Before screening was introduced, a considerable proportion of children were diagnosed only after presenting with severe complications, which underlines the importance of achieving complete coverage and reducing diagnostic delay[28].

3. **Standardisation of post-screening referral pathways**

Pilot study data show that newborn screening has strengthened the referral pathway to specialist paediatric haematology services, enabling earlier initiation of penicillin prophylaxis, vaccination and transcranial Doppler surveillance. Prior to the screening programme, pilot data also showed that twenty-seven per cent of children with sickle cell disease were diagnosed during hospitalisation for acute complications, highlighting the previous limitations of case finding[29].

Integration of screening results into long-term follow-up systems is still developing, particularly in ensuring consistent transition from paediatric to adult services and linking screening data with clinical outcomes[30].

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[25] Instituto Nacional de Saúde Doutor Ricardo Jorge (INSA). (2023). Estudo piloto de rastreio neonatal da doença falciforme: Resultados 2021–2023. https://www.insa.min-saude.pt/
[26] European Haematology Association. (2023). Country review: Sickle cell disease care and screening in Southern Europe. https://ehaweb.org/
[27] Programa Nacional de Rastreio Neonatal. (2024). Relatório anual do Programa Nacional de Rastreio Neonatal. Direção-Geral da Saúde, Portugal. https://www.dgs.pt/
[28] Instituto Nacional de Saúde Doutor Ricardo Jorge (INSA). (2023). Estudo piloto de rastreio neonatal da doença falciforme: Resultados 2021–2023. https://www.insa.min-saude.pt/
[29] Instituto Nacional de Saúde Doutor Ricardo Jorge (INSA). (2023). Estudo piloto de rastreio neonatal da doença falciforme: Resultados 2021–2023. https://www.insa.min-saude.pt/
[30] European Haematology Association. (2023). Country review: Sickle cell disease care and screening in Southern Europe. https://ehaweb.org/

V. Coordinated policy and patient partnership

1. Formal national multi-stakeholder SCD coordination platform

Strategic dialogue on sickle cell disease in Portugal is emerging but remains fragmented, with collaboration occurring mainly through paediatric haematology units, the national newborn screening programme and active patient organisations[31]. Portugal participates in European networks such as ERN EuroBloodNet, which supports harmonisation of clinical practice and cross-border collaboration on haemoglobinopathies.

Recent initiatives, including the first international conference on sickle cell disease in Lusophone countries, have strengthened cooperation between clinicians, researchers and patient groups, although these are project-based rather than part of a formal national platform[32][33]. There is currently no dedicated national governance body that brings together clinicians, patients, policymakers and payers to guide sickle cell disease policy or oversee implementation of the newborn screening programme and long-term care pathways.[34]

2. Inclusion of Patient organisations in decision-making

Patient organisations play a prominent role in sickle cell disease advocacy in Portugal, particularly in community outreach, awareness raising and support for families[35][36]. APPDH has been an important voice in campaigning for earlier diagnosis and improved access to specialist care and continues to engage with clinical teams and health authorities[37].

ALUA and DrepaComunidade focus on empowering communities of African origin, promoting health literacy and reducing stigma, and they have contributed to educational initiatives linked to newborn screening[38]. While involvement of these organisations is increasing, their participation in formal decision-making remains limited, and systematic inclusion in health planning processes has not yet been established[39][40].

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[31] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt
[32] ERN EuroBloodNet. (2023). ERN EuroBloodNet: Centres and activities. https://www.eurobloodnet.eu/
[33] Sociedade Portuguesa de Hematologia. (2023). Primeira Conferência Internacional da Drepanocitose nos Países Lusófonos. https://sph.org.pt
[34] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt/
[35] Ibid.
[36] APPDH – Associação Portuguesa de Portadores de Doenças Hematológicas. (2024). Atividades, missão e campanhas. https:/ appdh.org.pt/
[37] Ibid.
[38] DrepaComunidade. (2024). Educação, sensibilização e apoio comunitário sobre a doença falciforme. https://www.drepacomunidade.pt/
[39] Direção-Geral da Saúde. (2024). Plano de Ação para as Doenças Raras 2025–2030. Ministério da Saúde, Portugal. https://www.dgs.pt/
[40] APPDH – Associação Portuguesa de Portadores de Doenças Hematológicas. (2024). Atividades, missão e campanhas. https:/ appdh.org.pt/

Spain
Sickle Cell Disease Barometer

Key Performance Indicator (KPI) Overview

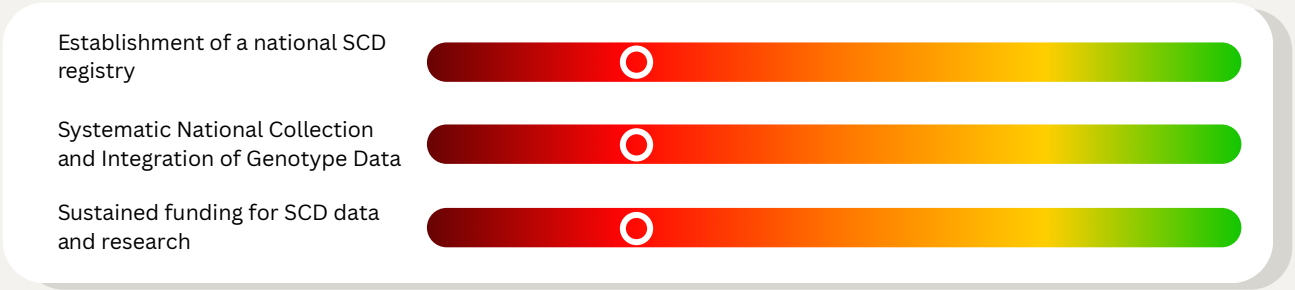
Policy Goal	KPI 1	KPI 2	KPI 3	KPI 4
1. Equitable access to SCD care and treatment	●	●	●	●
2. Strengthened National Workforce Capacity and Preparedness for SCD	●	●		
3. Comprehensive National Data, Registry and Research Infrastructure	●	●	●	
4. Early detection through newborn screening	●	●	●	
5. Coordinated policy and patient partnership	●	●		

Key Performance Indicator (KPI) Overview

1. Equitable access to SCD care and treatment

2. Strengthened National Workforce Capacity and Preparedness for SCD

3. Comprehensive National Data, Registry and Research Infrastructure



4. Early detection through newborn screening

5. Coordinated policy and patient partnership

Policy Goal: 1. Equitable access to SCD care and treatment		
KPI	**Overview**	**Score**
National plan for rare diseases and SCD-specific measures	Spain has a national rare disease strategy and regional plans, but SCD is not specifically prioritised within them, and SCD-related implementation remains uneven across autonomous communities, with inconsistent adult pathways. Spain's 2025–2028 chronicity strategy promotes continuity and care coordination for chronic conditions; although not SCD-specific, it may indirectly support long-term and transition care pathways for SCD.	
Availability and Access to SCD Treatment	Hydroxyurea is available nationally and widely used in paediatric care, but adult uptake varies across regions. Chronic transfusion programmes are available in most tertiary hospitals. Automated red blood cell exchange (RBCx) is widely available via hospital apheresis units (including most hospitals, with only a few small regional sites lacking capacity) and is funded at the hospital level through tenders and direct purchase. Gene therapy has EU approval but is not nationally reimbursed in Spain, with access assessed on a case-by-case basis.	
Access to Automated Red Blood Cell Exchange (aRBCX)	aRBCX is available widely in tertiary hospitals with apheresis units in Madrid, Barcelona and Valencia and other regions and cities. Regional hospitals often lack staffing and infrastructure, which restricts equitable access.	
Access to curative treatment	HSCT is available in specialist transplant centres, but donor availability, referral variability and centre capacity limit access for eligible patients.	
2. Strengthened National Workforce Capacity and Preparedness for SCD		
Integration of SCD into routine clinical training	Paediatric haematologists are well-trained and follow evidence-based guidelines. Adult haematologists, general practitioners and emergency clinicians have variable SCD-specific training, which contributes to regional disparities.	
Emergency care training and protocols for SCD crises	Emergency departments often lack SCD-specific training and consistent crisis protocols. Patients report delays in analgesia and inconsistent acute management.	
3. Comprehensive National Data, Registry and Research Infrastructure		
Establishment of a national SCD registry	Spain maintains a national registry for haemoglobinopathies and rare anaemias, but data are concentrated in a few high-volume centres and adult participation is variable, limiting national visibility of long-term outcomes.	
Systematic National Collection and Integration of Genotype Data	Genotyping data are collected for many patients, but not systematically for all cases, and integration with hospital information systems is incomplete.	
Sustained funding for SCD data and research	Spain lacks a dedicated national funding stream for SCD registries and long-term outcome research. Registry sustainability depends largely on academic and regional support.	

KPI	Overview	Score
4. Early detection through newborn screening		
National Implementation of SCD Universal Newborn Screening	Spain has universal newborn screening for SCD across all autonomous communities, although implementation and reporting vary regionally. Pilots demonstrated feasibility ahead of national rollout.	
Newborn screening coverage	Screening coverage is very high nationally due to Spain's established newborn screening infrastructure. Detailed regional reporting is still developing.	
Standardisation of post-screening referral pathways	Early diagnosis leads to rapid referral to paediatric haematology. Registry data show strong adherence to penicillin prophylaxis, vaccination and TCD screening.	
5. Coordinated policy and patient partnership		
Formal national multi-stakeholder SCD coordination platform	Clinical societies and specialists contribute to guidelines and awareness, but Spain does not have a dedicated national SCD governance platform bringing all stakeholders together.	
Inclusion of Patient Organisations in decision-making	Spain has active patient organisations that support education and awareness. Formal involvement in national decision-making remains limited and tends to occur through broader rare disease platforms.	

The Hidden Struggle: SCD in Spain

I. Equitable access to SCD care and treatment

1. **National plan for rare diseases and SCD-specific measures**

Spain has a national Rare Diseases Strategy within the National Health System that provides general objectives for prevention, diagnosis, care and research, but it does not contain detailed implementation measures specific to sickle cell disease[1]. Autonomous communities have developed their own rare disease plans, yet these differ in scope and do not consistently address sickle cell disease or ensure equal access to specialist care pathways[2].

In practice, all autonomous communities designate referral hospitals for sickle cell disease care, with access to standard treatments across regions. However, available national data suggest differences in service visibility and activity, which may partly reflect unequal regional participation in registries and reporting rather than true differences in clinical expertise or access.

2. **Availability and Access to SCD Treatment**

Hydroxyurea is authorised and reimbursed in Spain and is widely used in paediatric haematology units that follow national and European guidance[3].

Adult uptake is considerably more variable and depends on the presence of adult haematologists familiar with sickle cell disease management, resulting in differences in therapy use between autonomous communities[4].

Chronic transfusion programmes are available in most tertiary hospitals and are used for stroke prevention and severe disease, although follow-up intensity and coverage vary between adult and paediatric settings[5].

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[1] Ministerio de Sanidad. (2014). Estrategia en Enfermedades Raras del Sistema Nacional de Salud. Actualización 2014.https://www.sanidad.gob.es/organizacion/sns/planCalidadSNS/pdf/EnfermedadesRaras.pdf

[2] EURORDIS. (2009). Rare Diseases Strategy of the Spanish National Health System. https://download2.eurordis.org/documents/pdf/Spain_National_Plan.pdf

[3] Sociedad Española de Hematología y Hemoterapia & SEHOP. (2022). Guía de enfermedad de células falciformes.https://www.sehh.es/guias-y-consensos

[4] Sociedad Española de Hematología y Hemoterapia. (2022). Informe sobre la atención a pacientes con hemoglobinopatías en España. https://www.sehh.es/publicaciones

[5] García Morín, M., et al. (2020). Update of the Spanish registry of haemoglobinopathies in children. Medicina Clínica. https://www.sciencedirect.com/science/article/pii/S0025775320301950

Gene editing therapy for sickle cell disease has received approval from the European Medicines Agency, but it was not approved at the national level in Spain[6].

3. **Automated Red Blood Cell Exchange (aRBCX)**

Automated red blood cell exchange is available and used in all regions in Spain, with established apheresis units in almost all hospitals, with the biggest centres in Madrid, Barcelona and Valencia[7].

Regional hospitals without apheresis capability often rely on simple transfusion or must transfer patients to more distant specialist centres, creating regional variation in access to optimal transfusion strategies[8].

4. **Access to curative therapies (HSCT)**

Haematopoietic stem cell transplantation is offered in a limited number of Spanish transplant centres, mainly for paediatric patients with severe disease and a suitable donor[9].

The number of transplants remains low because of donor limitations, centre capacity and transplant-related risk, which restrict access to curative options for eligible individuals[10]. Adult transplantation for sickle cell disease is rare, and there is no national programme that systematically evaluates adult transplant eligibility across autonomous communities, which contributes to unequal access[11].

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[6] European Society for Blood and Marrow Transplantation. (2022). EBMT activity survey 2022.https://www.ebmt.org/ebmt/documents/ebmt-activity-survey
[7] Sociedad Española de Hematología y Hemoterapia. (2022). Informe sobre la atención a pacientes con hemoglobinopatías en España. https://www.sehh.es/publicaciones
[8] García Morín, M., et al. (2020). Update of the Spanish registry of haemoglobinopathies in children. Medicina Clínica. https://www.sciencedirect.com/science/article/pii/S0025775320301950
[9] European Society for Blood and Marrow Transplantation. (2022). EBMT activity survey 2022.https://www.ebmt.org/ebmt/documents/ebmt-activity-survey
[10] Ibid.
[11] Sociedad Española de Hematología y Hemoterapia. (2022). Informe sobre la atención a pacientes con hemoglobinopatías en España. https://www.sehh.es/publicaciones

II. Strengthened National Workforce Capacity and Preparedness for SCD

1. Integration of SCD into routine clinical training

Paediatric haematology expertise in Spain is strong, particularly in tertiary centres where clinicians follow national and European guidance and participate in specialist haemoglobinopathy networks[12].

Participation in national and European registries reinforces structured clinical practice and supports adherence to guideline-recommended screening, prophylaxis and disease-modifying treatment[13].

However, sickle cell disease is not consistently included in the standard training curriculum for internal medicine, family medicine or general haematology, resulting in uneven knowledge among non-specialist clinicians[14]. Studies on rare disease training in Spain show that many healthcare professionals report insufficient preparation for diagnosing and managing rare diseases, which contributes to inconsistent awareness of sickle cell disease outside specialist units[15].

2. Emergency care training and protocols for SCD crises

Emergency departments often lack structured sickle cell disease-specific protocols, and published patient experience reports indicate delayed analgesia and variable recognition of acute complications such as acute chest syndrome[16].

Local training initiatives exist in some tertiary hospitals, but they are not standardised across autonomous communities[17].
National rare disease policy acknowledges the need to strengthen professional training, yet this has not resulted in nationwide emergency care protocols for sickle cell disease[18].

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[12] Ministerio de Sanidad. (2014). Estrategia en Enfermedades Raras del Sistema Nacional de Salud. Actualización 2014. https://www.sanidad.gob.es/organizacion/sns/planCalidadSNS/pdf/EnfermedadesRaras.pdf
[13] ERN EuroBloodNet. (2023). Annual Activity Report. https://eurobloodnet.eu/
[14] Sociedad Española de Hematología y Hemoterapia & SEHOP. (2022). Guía de enfermedad de células falciformes. https://www.sehh.es/guias-y-consensos
[15] Ramalle Gómara, E., et al. (2020). Education and information needs for physicians about rare diseases in Spain. Orphanet Journal of Rare Diseases, 15(22). https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-1327-y
[16] Federación Española de Enfermedades Raras. (2023). Informe sobre experiencias de pacientes con enfermedades raras en servicios de urgencias. https://enfermedades-raras.org/
[17] Ibid.
[18] Ministerio de Sanidad. (2014). Estrategia en Enfermedades Raras del Sistema Nacional de Salud. Actualización 2014. https://www.sanidad.gob.es/organizacion/sns/planCalidadSNS/pdf/EnfermedadesRaras.pdf

III. Comprehensive National Data, Registry and Research Infrastructure

1. **Establishment of a national SCD registry**

Spain maintains a national registry for haemoglobinopathies and rare anaemias that includes sickle cell disease cases from both paediatric and adult centres.
Paediatric participation in the registry is strong, with recent analyses showing robust data collection on diagnosis, treatment patterns and clinical outcomes. Registry data are currently concentrated in a small number of high-volume centres, notably Gregorio Marañón, Vall d'Hebron and Sant Joan de Déu hospitals, resulting in Madrid and Catalonia accounting for the largest share of registered patients. Adult participation and contribution from other hospitals remain more variable, which may partly reflect incomplete registry participation and limit the national visibility of long-term complications and transition outcomes[19].

2. **Systematic National Collection and Integration of Genotype Data**

The national registry captures clinical and genetic information for many patients, although genotyping is not yet fully systematic across all registered individuals[20]. Incomplete integration of registry data with hospital information systems limits the ability to conduct large-scale genotype-phenotype studies that could support risk stratification and eligibility assessment for advanced therapies[21].

3. **Sustained funding for SCD data and research**

Registry activities in Spain rely largely on academic centres, professional societies and project-based funding, since there is no dedicated national mechanism for long-term financing of sickle cell disease registry infrastructure[22].
Analyses of rare disease funding in Spain highlight the need for more stable support for data collection systems and for sustained monitoring of clinical outcomes[23].

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[19] Marco Sánchez, J. M., Bardón Cancho, E. J., Benéitez, D., et al. (2024). Haemoglobinopathies and other rare anemias in Spain: Ten years of a nationwide registry (REHem AR). Annals of Hematology.
https://link.springer.com/article/10.1007/s00277-024-05899-3
[20] Marco Sánchez, J. M., Bardón Cancho, E. J., Benéitez, D., et al. (2024). Haemoglobinopathies and other rare anemias in Spain: Ten years of a nationwide registry (REHem AR). Annals of Hematology.
https://link.springer.com/article/10.1007/s00277-024-05899-3
[21] Ibid.
[22] Zozaya, N., et al. (2023). Strategic discussion on funding and access to therapies targeting rare diseases in Spain: An expert consensus paper. Orphanet Journal of Rare Diseases.
https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02803-4
[23] Ibid.

IV. Early detection through newborn screening

1. National Implementation of SCD Universal Newborn Screening

Spain has progressively implemented newborn screening for haemoglobinopathies across autonomous communities and now includes sickle cell disease within the national newborn screening panel[24]. Pilot programmes in regions such as Madrid and Catalonia demonstrated the feasibility of screening and highlighted the importance of detecting sickle cell disease early in life[25]. National evaluations indicate that all regions now provide universal newborn screening for haemoglobinopathies, positioning Spain in line with international recommendations[26].

2. Newborn screening coverage

Coverage for newborn screening in Spain is very high due to the established screening infrastructure, although detailed performance indicators for sickle cell disease are still being developed and standardised at the national level[27].

Regional studies show that the incidence of sickle cell disease varies considerably between autonomous communities and confirm that screening enables the timely identification of affected infants[28].

3. Standardisation of post-screening referral pathways

Referral pathways from screening laboratories to paediatric haematology centres are well-defined and function effectively, ensuring the timely initiation of prophylaxis and specialist follow-up[29]. Paediatric registry data indicate high adherence to recommended penicillin prophylaxis, vaccination schedules and transcranial Doppler screening in children diagnosed through newborn screening[30].

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[24] Vives Corrons, J. L. (2025). Insights into hemoglobinopathies in Spain. eJHaem. https://doi.org/10.1002/jha2.70093

[25] Sánchez Villalobos, M., et al. (2023). A newborn screening programme for sickle cell disease in Murcia, Spain. International Journal of Neonatal Screening, 9(1). https://www.mdpi.com/2409-515X/9/1/7

[26] Colomé, C., et al. (2020). Newborn screening for metabolic disorders in Spain and worldwide. Anales de Pediatría, 93(1), 60.e1–60.e9. https://www.analesdepediatria.org/en-newborn-screening-for-metabolic-disorders-articulo-S2341287920301096

[27] Ibid.

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National technical guidance on haemoglobinopathy screening provides instructions for confirmatory testing, family counselling and referral, although full integration of newborn screening data with the national sickle cell disease registry remains incomplete[31].

V. Coordinated policy and patient partnership

1. **Formal national multi-stakeholder SCD coordination platform**

Strategic dialogue on sickle cell disease in Spain is emerging but remains fragmented, with activities spread across rare disease and haemoglobinopathy structures rather than coordinated through a single national platform focused on sickle cell disease[32].

The Spanish Society of Haematology and Haemotherapy and paediatric haematology networks provide expert guidance on haemoglobinopathies and have contributed to clinical recommendations and policy debates on newborn screening and access to innovative therapies[33].

Spanish centres participate in ERN EuroBloodNet and RADeep, which support harmonisation of care pathways, development of clinical recommendations and collaboration on registries at the European level[34][35].

However, Spain does not yet have a dedicated national sickle cell disease council or formal governance body that regularly brings together clinicians, patients, payers and policymakers around a structured sickle cell disease agenda[36].

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2. Inclusion of Patient organisations in decision-making

Patient organisations play an increasingly important role in sickle cell disease in Spain, including the Asociación Española de Enfermedad Falciforme, which provides information, peer support and advocacy for people with sickle cell disease and their families[37].

Federation-level platforms such as the Federación Española de Enfermedades Raras highlight the need for stronger and more systematic participation of rare disease patient organisations in health decision-making.[38].

Despite this, involvement of sickle cell disease patient groups in formal national processes, such as guideline development and the assessment of new therapies, remains limited and often occurs indirectly through broader rare disease coalitions[39][40]. Recent analyses of patient participation in Spain indicate progress but also underline that mechanisms for involving patient representatives in strategic planning and health technology assessment are still uneven between regions and institutions[41].

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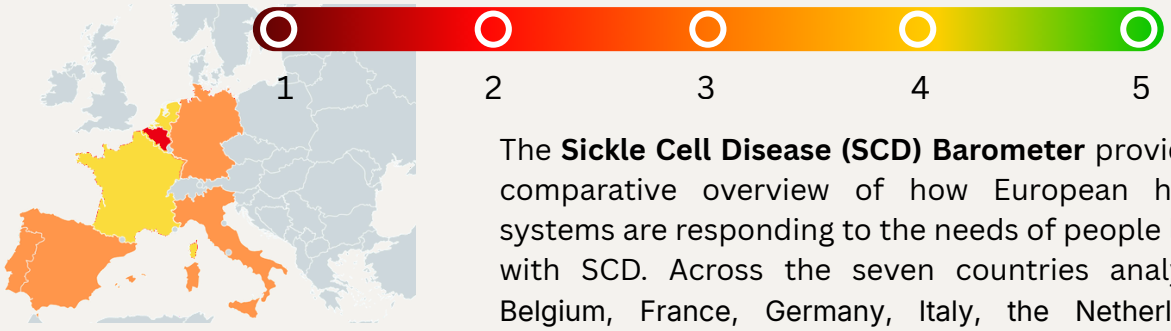
Conclusion



The **Sickle Cell Disease (SCD) Barometer** provides a comparative overview of how European health systems are responding to the needs of people living with SCD. Across the seven countries analysed, Belgium, France, Germany, Italy, the Netherlands, Portugal, and Spain, **the findings reveal increasing awareness of the disease but also persistent structural gaps in screening, care organisation, data collection and policy prioritisation.**

Although national health systems differ in governance and demographic context, several common trends emerge. In most countries, **SCD is addressed within broader rare disease frameworks rather than through dedicated national strategies.** While these frameworks provide an important policy foundation, they often lack targeted measures addressing the specific clinical and long-term care needs associated with SCD. As a result, policy attention to the disease remains uneven and fragmented across countries.

Significant variation also exists in early detection. **Early diagnosis is critical to improving outcomes and preventing severe complications, yet newborn screening for SCD is not implemented uniformly.** Some countries have established national screening programmes, while others rely on targeted or regional approaches. These differences can affect the timing of diagnosis and the ability of patients to access specialised care early in life.

The organisation of care pathways presents another major challenge. SCD requires coordinated, multidisciplinary care over the course of a patient's lifetime. **Across the countries assessed, specialised expertise is often concentrated in a limited number of centres, and referral pathways are not always clearly structured**. This can lead to fragmented care and unequal access to specialised services, particularly for patients living outside major urban areas.

The Barometer also highlights persistent gaps in data and disease monitoring. Comprehensive information on SCD prevalence, patient outcomes and healthcare utilisation remains limited in several countries. While some registries and hospital-based datasets exist**, data collection is often fragmented and not systematically integrated at national level.** Strengthening data systems would significantly improve the ability of policymakers and healthcare planners to understand the burden of the disease and develop evidence-based responses.

At the same time, advances in SCD treatment are rapidly transforming the therapeutic landscape**. Emerging disease-modifying therapies and potential curative approaches offer new opportunities to improve patient outcomes.** However, integrating these innovations into national health systems will require appropriate clinical expertise, specialised treatment infrastructure and sustainable reimbursement mechanisms.

Overall, the Barometer shows that **countries are at different stages in the development of comprehensive SCD care frameworks.** While progress has been made in several areas, important opportunities remain to strengthen early detection, **improve care coordination, enhance data collection and ensure that health systems are prepared to deliver emerging therapeutic innovations**. Addressing these challenges will be essential to improving health outcomes and quality of life for people living with SCD across Europe.

Recommendations to the EU

The following recommendations outline key policy actions that can support these objectives and help address the systemic gaps identified across the Barometer countries.

Improve Access to Treatment to Address Health Inequities

Introduce an EU-wide action plan for rare diseases, prioritising health equity, and featuring a dedicated section to enhance the diagnosis, treatment, and care for SCD patients. The action plan should aim at ensuring equitable access to resources and treatment, engaging communities, educating stakeholders, and advancing research efforts. This would address increasing demands from EU institutions and civil society.

Invest in Training and Skill Development

Allocate funding to train healthcare providers in SCD treatments, enhancing awareness and minimizing biases. The underfunded healthcare staffing requires increased investment, especially under EU4Health, Digital Europe, and Horizon Europe initiatives. Prioritizing this matter through proactive funding opportunities would incentivize essential training on the different existing SCD treatments.

Encourage an EU-Wide SCD Registry

Create a robust EU-wide SCD registry for efficient patient tracking, policymaking, and genotyping data collection. This registry would enable the systematic and wide collection of genotyping data, offering valuable insights into the genetic diversity of SCD across the European population. This initiative aligns with the goals of the European Health Data Space (EHDS), aiming to establish a secure infrastructure for sharing healthcare data across the European Union.

Newborn screening applied to EU 27 Member States

Early detection is crucial, and screening programs are the first essential step. However, the current decentralized approach in the EU leads to significant disparities among Member States, affecting individuals' access to timely diagnosis and treatment based on their location. Advocating for universal newborn screening, as successfully practised in France, the Netherlands, Portugal and Spain, ensures equitable outcomes. To enhance effectiveness, screenings should be accompanied by referrals and follow-ups to support patients and their families, addressing barriers to care.

Strategic dialogue with relevant stakeholders

Collaboration with relevant stakeholders like ERN-EuroBloodNet is crucial to address SCD challenges. The EU should forge partnerships and open dialogue with healthcare professionals, patient organizations, research institutions, Member States, and industry for shared expertise, research support, advocacy, and a coordinated approach to enhance SCD management.

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