webinar

HEALTH PROFESSIONALS

Thursdays Webinars

Newborn screening for sickle cell disease: what can we learn from the practice in the European Union?

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28 January 2026

ERN | EuroBloodNet

Newborn screening for Sickle Cell Disease

1975

First newborn screening programme for Sickle Cell Disease (SCD) in New York

1986

Prophylaxis with oral penicillin in children with SCD (PROPS)

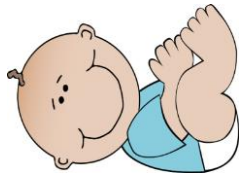
- N Engl J Med . 1986 Jun 19;314(25):1593-9. doi: 10.1056/NEJM198606193142501
- Randomized trial terminated earlier

1987

NIH conference: universal newborn screening for SCD recommended

- Early integration into dedicated care pathway = decrease SCD mortality and morbidity

Newborn screening for Sickle Cell Disease



Birth with SCD

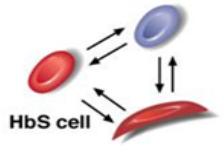
Genetic counselling

Parents education/recommendations

9 months start HU ?

Prevention of infections

2 months and +



Recommendation

(2018 DOI:10.1111/BJH.15600)

Consensus Statement

3. Early diagnosis by NBS, together with anti-pneumococcal penicillin prophylaxis and vaccination, coordinated follow-up and parental education, reduces morbidity and mortality from SCD in childhood.

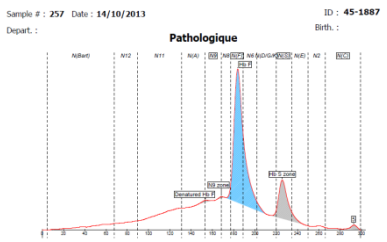
Newborn screening programme for SCD: HOW?

Sampling

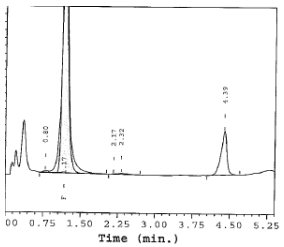
Report result

Integration into a comprehensive care

Analysis/screening



First line



Confirmation

Control new sample

Recommendation *(2018 DOI:10.1111/BJH.15600)*

Consensus Statement

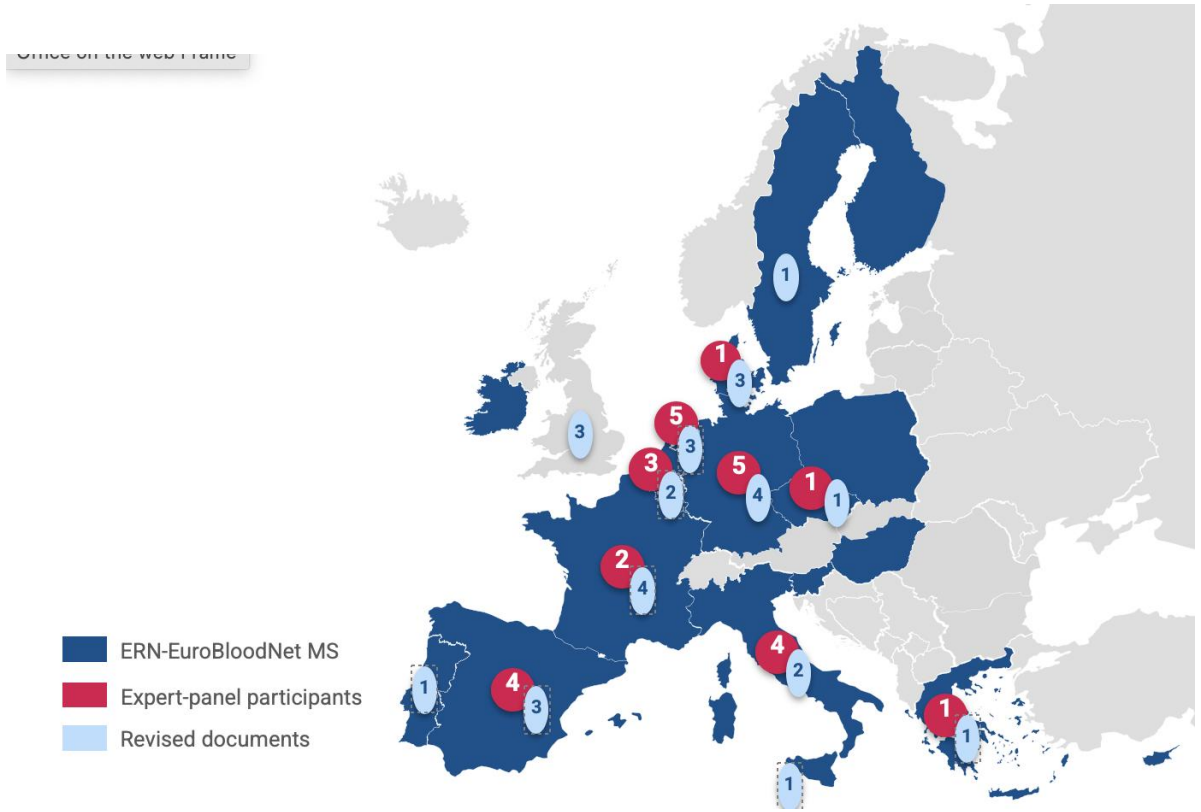
3 8d. All children with SCD should be enrolled in a comprehensive care programme. The programme should ensure equal access to high-level clinical care.

Clinical Practice Guidelines & Access to Highly Specialized Care

New CPGs/CDTs produced by the ERN-EuroBloodNet

Sickle Cell Disease

Systematic comparative analysis of national SCD recommendations in Europe



Clinical Practice Guidelines & Access to Highly Specialized Care

New CPGs/CDTs produced by the ERN-EuroBloodNet

Sickle Cell Disease

B. Biemond	The Netherlands
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J. Cermak	Czech Republic
M. Colard	Belgium
A. Collado	Spain
R. Colombatti	Italy
D. Cuzzubo	Italy
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M. Rab	The Netherlands
A. Rijneveld	The Netherlands
G. Russo	Italy
L. Tagliaferri	Germany
E. van Beers	The Netherlands
A. Vanderfaeillie	Belgium

Systematic comparative analysis of national SCD recommendations in Europe

TRANSLATION IN ENGLISH - REPOSITORY

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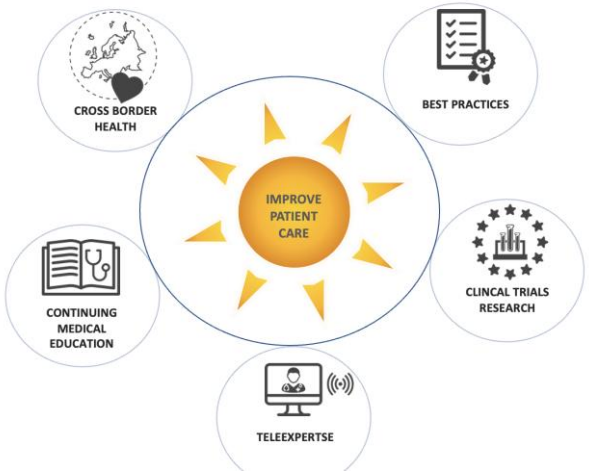
Systematic comparative analysis of national SCD recommendations in Europe

1. Diagnosis at birth
2. Prevention and management of complications
    a. Standard follow-up
    b. ACS
    c. VOC
    d. Stroke + TCD/MRI
    e. Priapism
    f. Kidney
    g. Splenectomy/Splenic sequestration
    h. Cardiomyopathy
    i. Pregnancy
    j. Pre-surgery
3. Treatments including prevention
    a. Vaccination/antibioprophylaxis
    b. Analgesia
    c. Fever treatment
    d. Hydroxyurea
    e. Blood transfusion
    f. HSCT
4. Transition

Systematic comparative analysis of national SCD recommendations

- Improve inequity of care (cross-border care
- Integration of ERNs initiatives into national care



- **Research projects** : harmonized and evidence-based clinical policies
- **Place of registries** (clinical outcome research)

EU (UK) review of recommendations on Sickle Cell Disease (SCD)

The objective of this project is original

Not a question of giving recommendations for the SCD

Rather an inventory of practice within the EU/UK.

The expected results

- Highlighting
  - Aspects not taken into account in a national recommendation
    - ✓ **Suggestion of update**
  - Differences in practice, topics remaining unexplored or insufficiently explored
    - ✓ **Gaps of knowledge (Research topics) identified**

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Systematic comparative analysis of national SCD recommendations in Europe

1. Diagnosis at birth

National recommendations on a website
e.g., Belgium
https://www.depistageneonatal.be/wp-content/uploads/2024/06/guide-de-depistage-des-anomalies-congenitales-vf2024.pdf

National recommendations in a scientific publication
e.g. Portugal
https://doi.org/10.3390/ijns11010010

+ translation in English
+ experts by country in NBS

Clinical Practice Guidelines & Access to Highly Specialized Care

New CPGs/CDTs produced by the ERN-EuroBloodNet

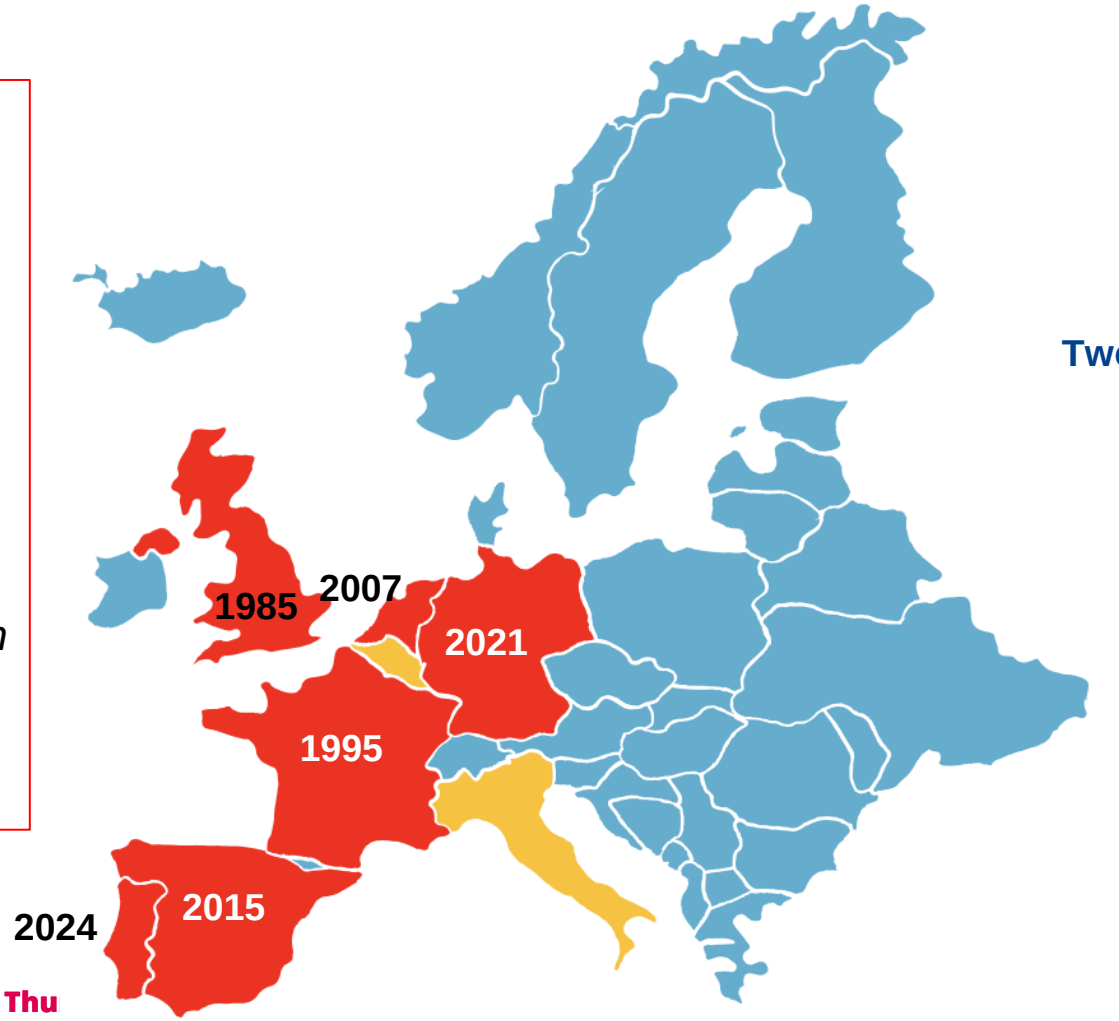
Sickle Cell Disease

	Max level of evidence	REF BE	REF CY
Prioritized question 1: Is the newborn screening embedded in a national care programme?			
Recommendation 1: a newborn screening programme is implemented	C	Yes (Regio; 2023)	No
Prioritized question 2: What are the benefits of an early detection of SCD?			
Recommendation 1: Benefit is based on previous publications and reported in the national document	C	Yes	NA
Prioritized question 3: Which EU countries should implement a NBS programme for SCD?			
Recommendation 1: Newborn screening is implemented at least related to a treshold of birth prevalence	C	No	NA
Recommendation 2: Epidemiological data, as birh prevalence of SCD, are available	C	1:1654 (2023)	57 SCD reported (2021)
Prioritized question 4: Who should be screened for SCD?			
Recommendation 1: if a newborn screening programme is implemented, all newborn are screened	c	Yes	NA
Recommendation 2: if a newborn screening programme is not implemented, at least newborn at risk based on ethnic origin are screened	C	NA	No (1)
Prioritized question 5: How should the screening be done?			
Recommendation 1: Use any technique as specific and sensible that classical techniques	C	Yes	NA
Recommendation 2: Delay for first tier result is reported	C	NA	NA
Prioritized question 6: What is the recommended procedure after a positive screening result ?			
Recommendation 1: Confirmation of screening result is done on the same sample	C	Yes/No	NA
Recommendation 2: the diagnosis is made on a new sample	C	Yes	NA
Recommendation 3: the procedure to ensure adequate care is described	C	Yes	Yes
Recommendation 4: the SCD newborn is registered in a clinical data base to evaluate adequate care	C	Yes (3)	Yes (3)
Prioritized question 7: Should carriers identified in NBS be informed about their result?			
Recommendation 1: Carriers are reported and counseling is offered	C	No	NA

Newborn screening for SCD -National/regional Programmes in Europe

Recommendation
(2018 DOI:10.1111/BJH.15600)

Consensus Statement
4a. The implementation of a national NBS programme for SCD should be informed by a review of national epidemiological data on SCD, but should not be based solely on a threshold birth prevalence. Where not available, these data should be collected.

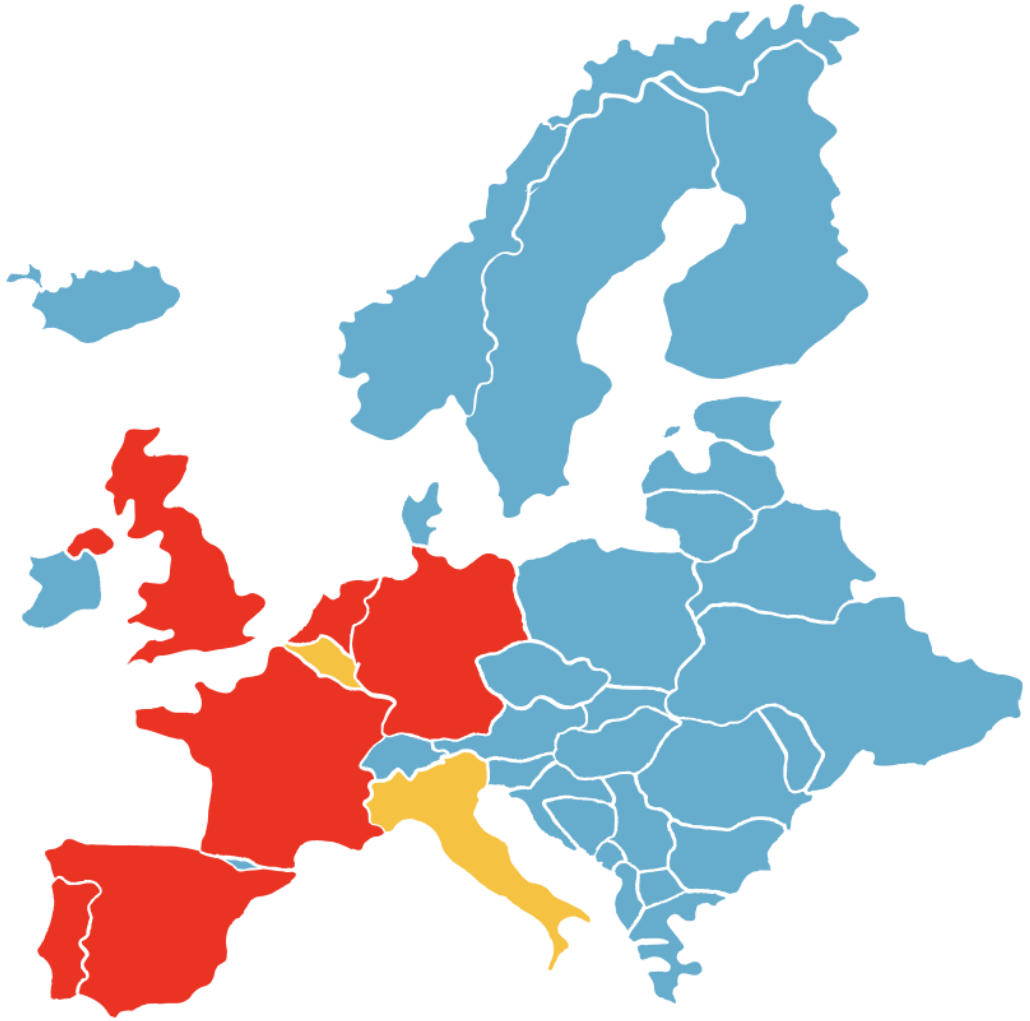


Two programmes integrated SCD after 2018
Germany, Portugal

Newborn screening for SCD – Methods ?

Recommendation
(2018 DOI:10.1111/BJH.15600)

Consensus Statement
7a. High performance liquid chromatography (HPLC), capillary electrophoresis (CE), isoelectric focusing (IEF) and tandem mass spectrometry (MS/MS) are appropriate methods for NBS for SCD.
7b. New methods currently being tested should prove to be as specific and sensitive as HPLC and CE before being implemented on a larger scale.

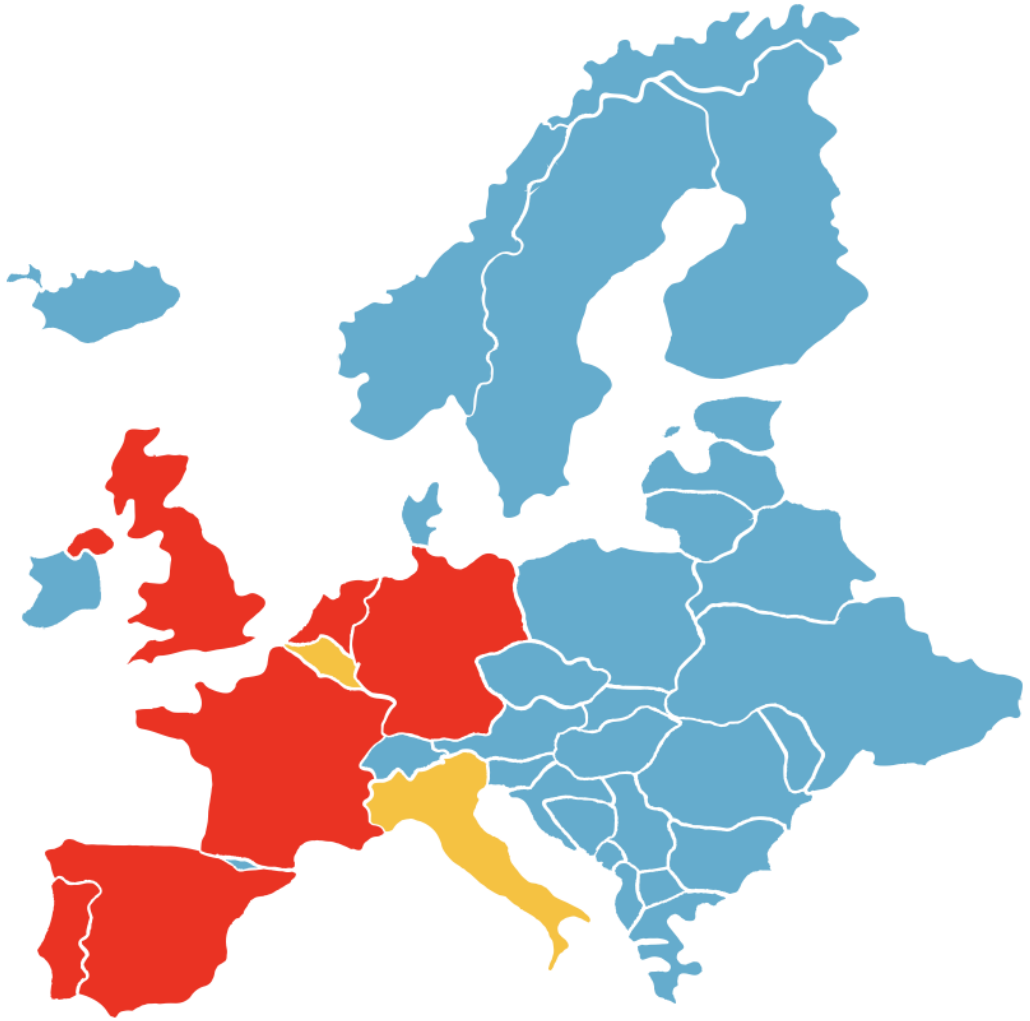


	First tier
Belgium	qPCR MS/MS
England	HPLC CE MS/MS
The Netherlands	HPLC
France	IEF, HPLC, CE, MS/MS
Spain	HPLC

Newborn screening for SCD – Confirmation ?

Recommendation
(2018
DOI:10.1111/BJH.15600)

Consensus Statement
8a. A haemoglobin pattern that is in accordance with any genotype of SCD requires a re-test with a fresh punch from the same sample. If available, a different method from the first one should be used (secondtier screening). If a second alternative method is not available, a re-test with the same method is acceptable.
If the re-test is positive, the newborn should be re-called for confirmatory testing.

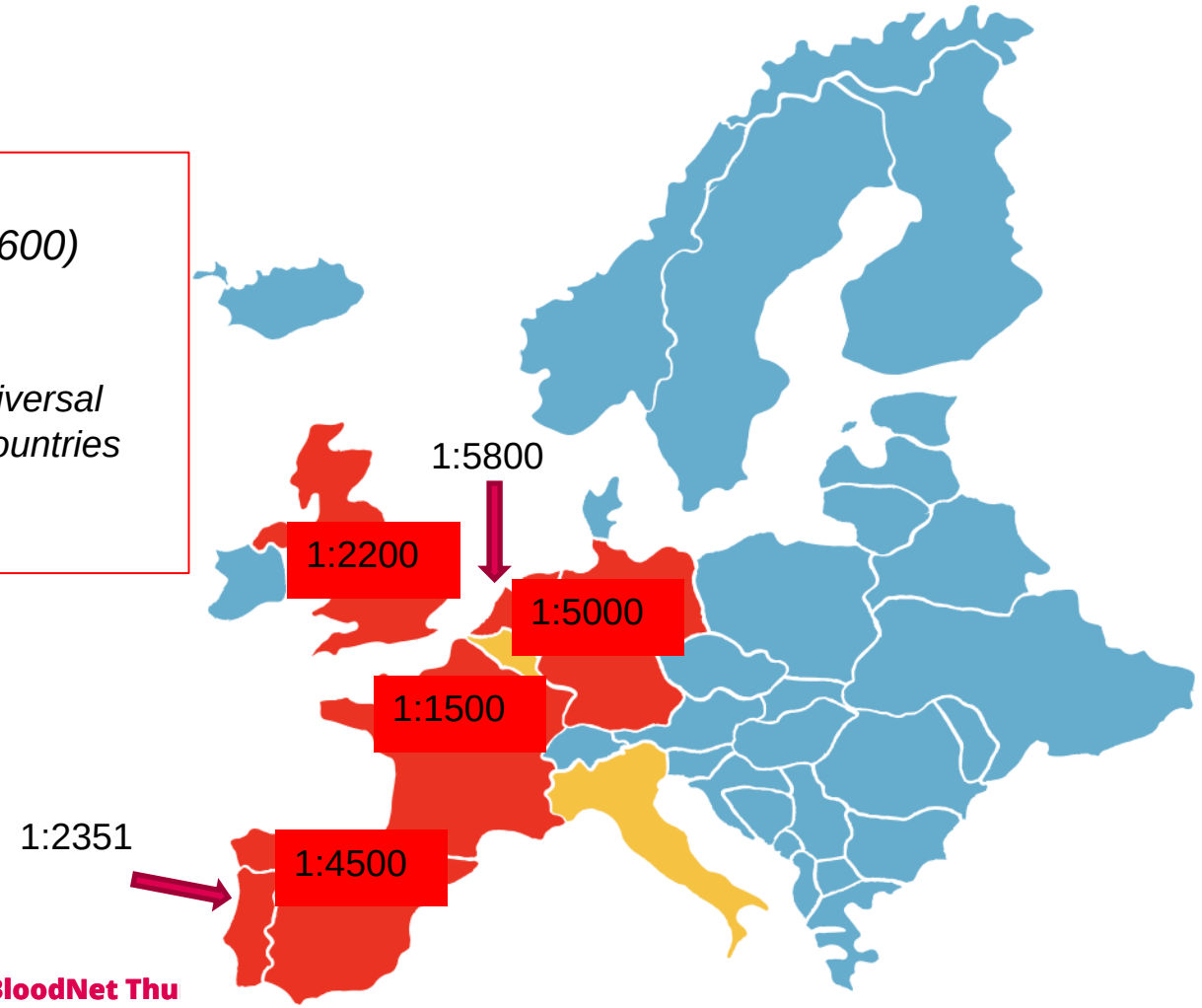


	Confirmation *(same sample)*
Belgium	MS/MS qPCR
England	IEF HPLC HPLC
The Netherlands	-
France	CE or HPLC
Spain	CE

Newborn screening for SCD - Data on birth prevalence

Recommendation
(2018 DOI:10.1111/BJH.15600)

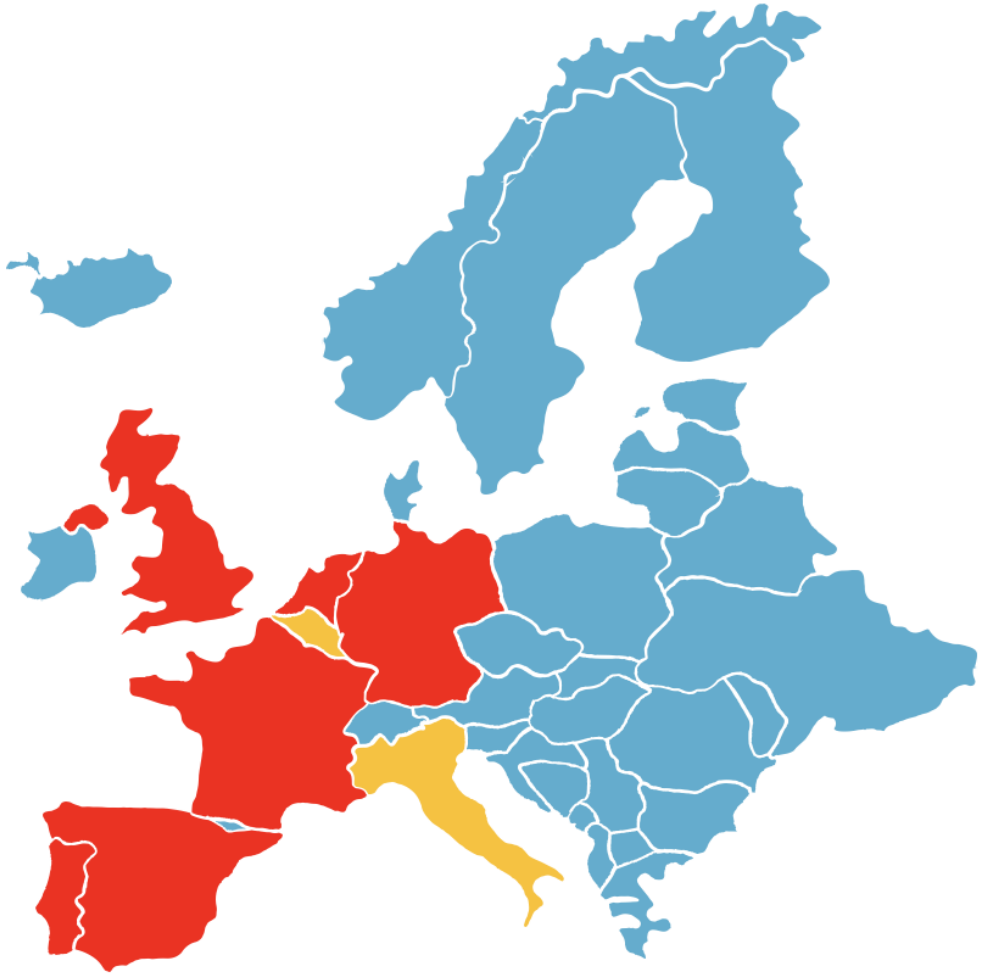
Consensus Statement
5a. The panel recommends universal NBS screening for SCD in all countries participating in the conference.
(13 Member States).



Newborn screening for SCD - Targeted (population at risk) ?

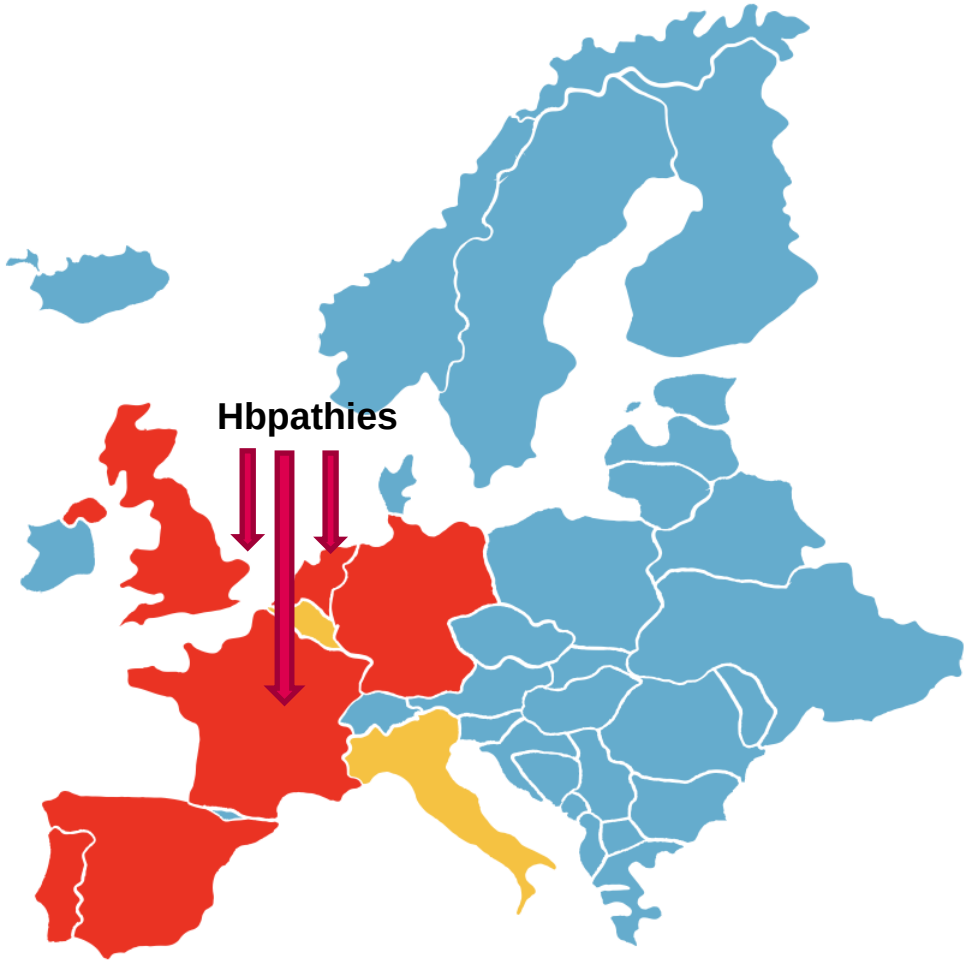
Recommendation
(2018
DOI:10.1111/BJH.15600)

Consensus Statement
5b. Targeted screening based on ethnic origins is not recommended because of the higher risk of failure to identify an affected newborn.



France moved to universal screening in November 2024

Newborn screening for SCD – extended to haemoglobinopathies?



Recommendations
(2018 DOI:10.1111/BJH.15600)

Consensus Statement

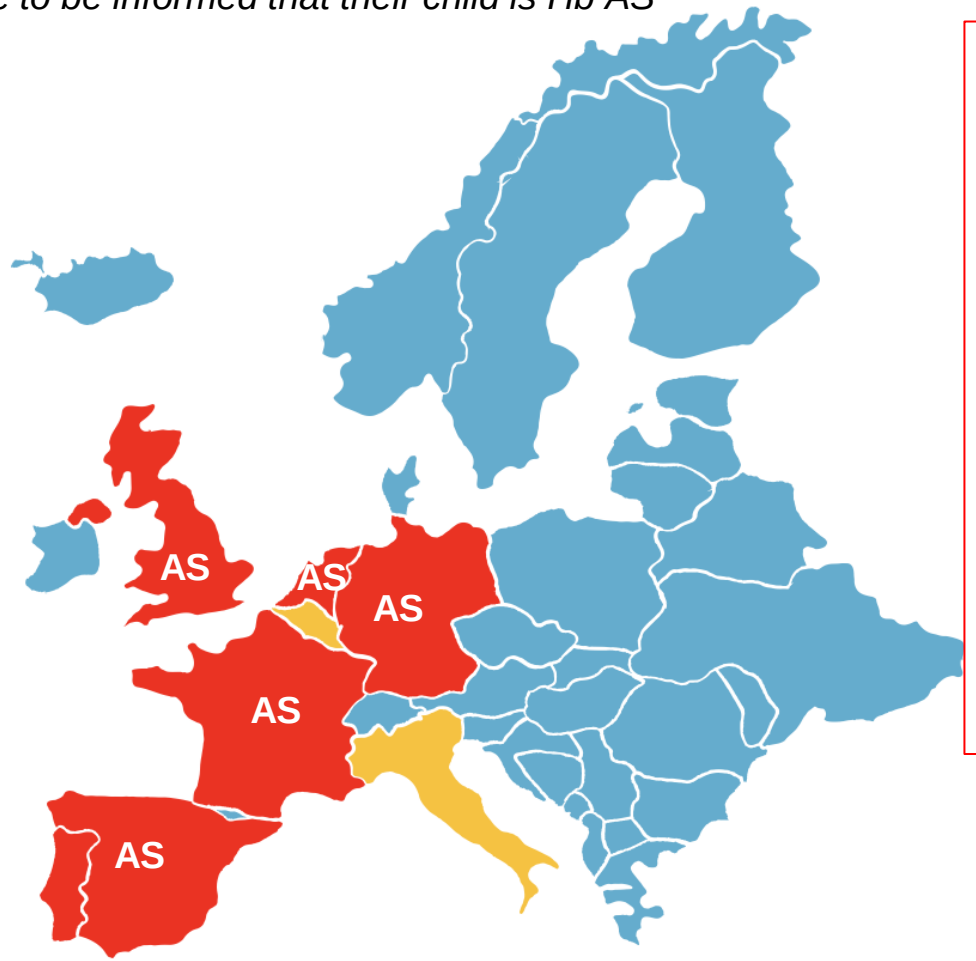
2a. The target disease of a NBS programme for haemoglobinopathies is SCD, including all genotypes.

2b. Beta thalassaemia, whilst not a formal target disease of a NBS programme for haemoglobinopathies, should also be reported.

Newborn screening for SCD report heterozygosity for Hb S?

Examples

- *The Netherlands, parents can refuse to be informed that their child is Hb AS*
- *Germany, it is not authorized by law*



- ***Recommendation s****(2018 DOI:10.1111/BJH.15600)*

Consensus statement

*6. SCD is a genetic condition. The knowledge of the carrier state in the family provides opportunities for prevention of affected births. The carrier status (all mutations that might cause SCD) should be reported and counselling offered to carriers. The panel acknowledges that there is virtually no other evidence for this recommendation than solely "**expert opinion**" and encourages future research on this question. Any national decision-making process should take this into account.*

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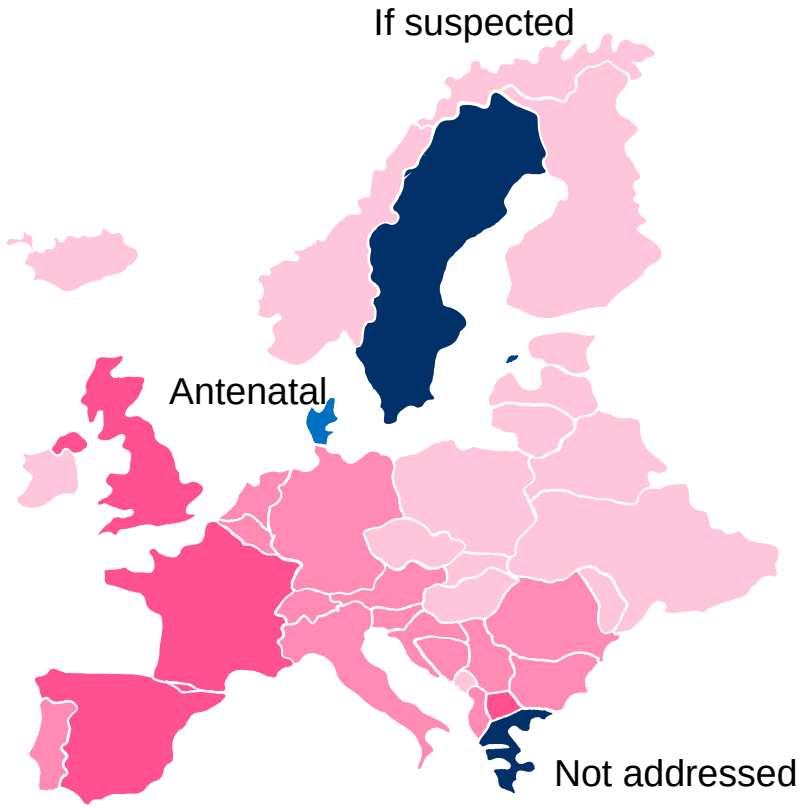
No national newborn screening programme for SCD – screening/diagnosis at birth recommendation ?



- ***Recommendation s****(2018 DOI:10.1111/BJH.15600)*

Consensus statement

5c. In countries where national NBS screening for SCD is not implemented, an interim policy should be agreed for testing at-risk newborns on a case-by-case basis according to family origins

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Newborn screening for SCD – National registries?

Recommendations

(2018 DOI:10.1111/BJH.15600)

Consensus Statements

1a. In Europe the burden of Sickle Cell Disease (SCD)
has increased and will continue to increase.
1b. It is desirable that all European patients with SCD
are enrolled onto registries, with standardized data
collection and coordinated follow-up.

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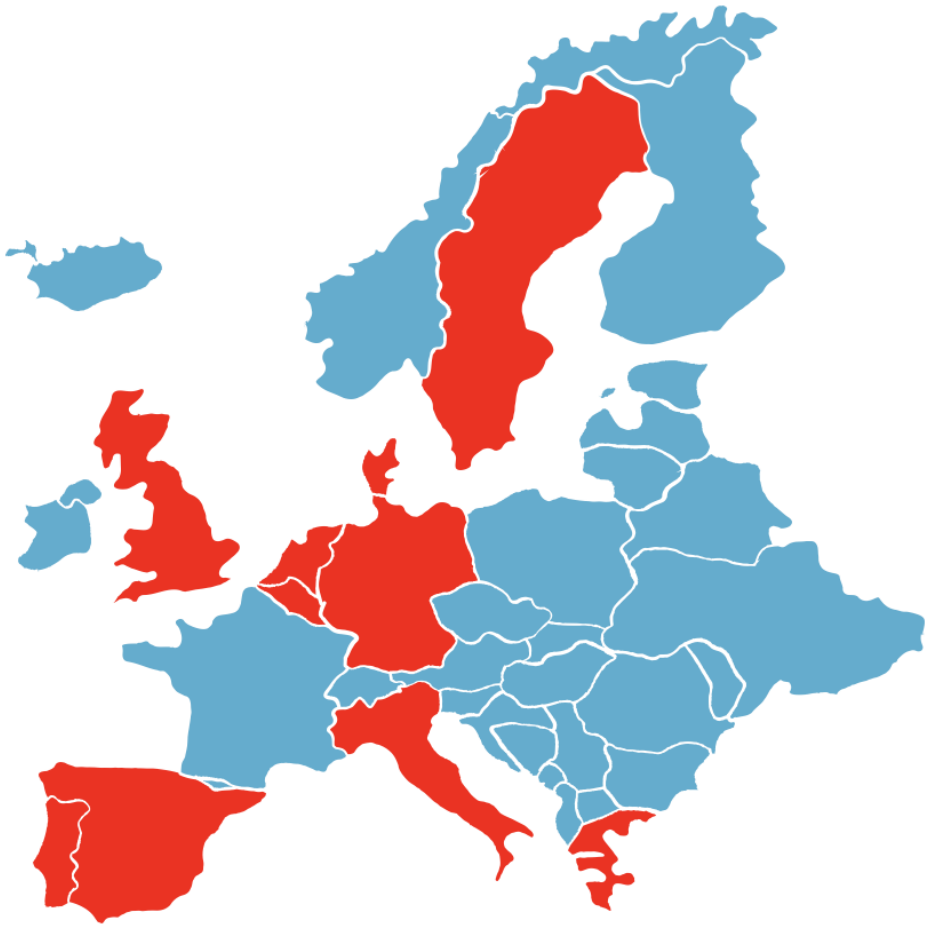
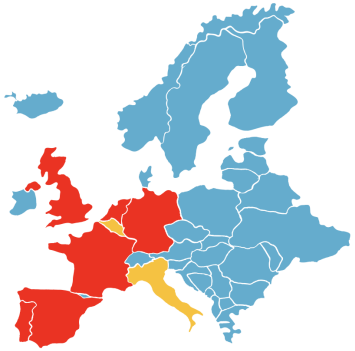


Newborn screening

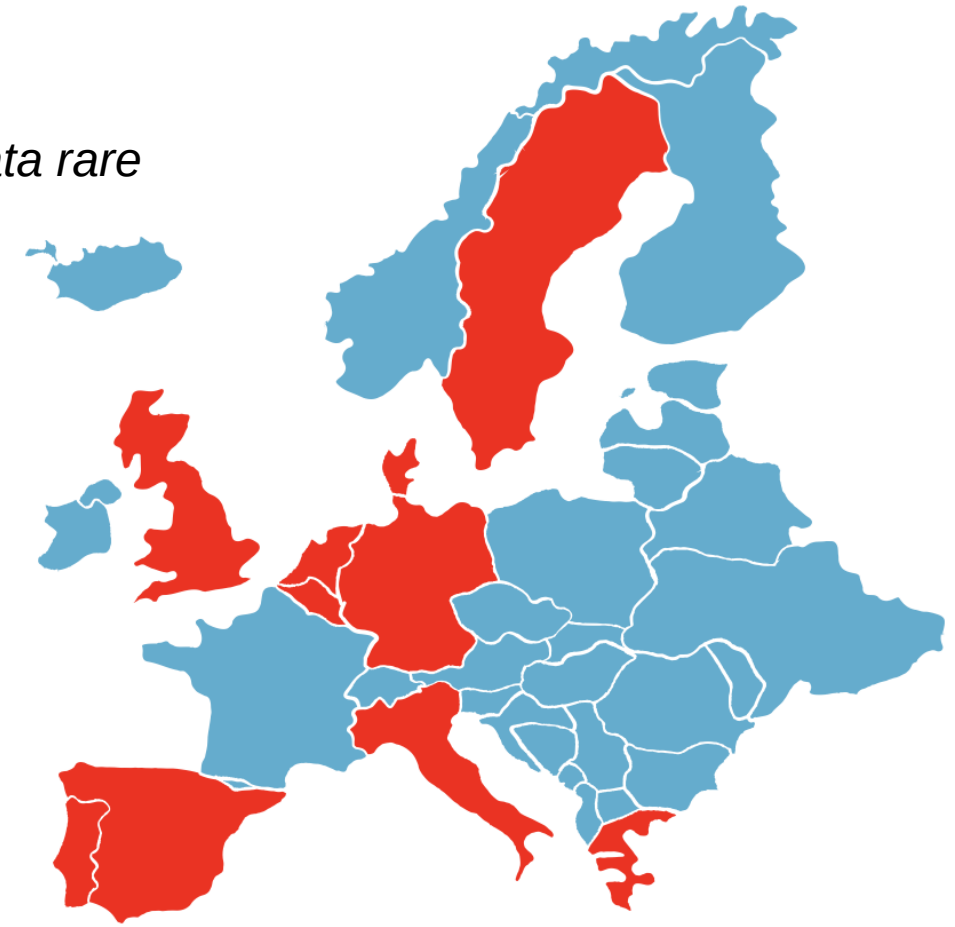


Newborn screening for SCD – National registries?

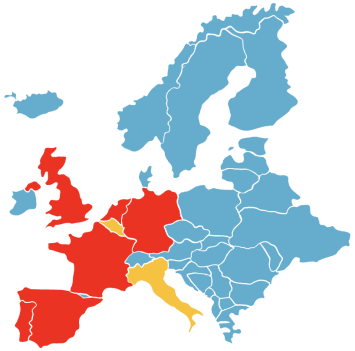
European Commission: ENROL registry (data rare diseases in haematology

RADeep: registry on rare anaemias

https://eurobloodnet.eu



Newborn screening



Newborn screening for SCD – Follow-up

Arch Dis Child. 2017 Nov 5. pii: archdischild-2017-313213. doi: 10.1136/archdischild-2017-313213. [Epub ahead of print]

Evaluation of newborn sickle cell screening programme in England: 2010-2016. *Streetly A1,2, Sisodia R1, Dick M3, Latinovic R4, Hounsell K1, Dormandy E1.*

DESIGN:
Cohort of resident infants with sickle cell disease (SCD) born between 1 September 2010 and 31 August 2015 and followed until August 2016.

CONCLUSION:
The SCD screening programme is effective at detecting affected infants. Enrolment into specialist care is timely but below the programme standards. Mortality is reducing but adherence to antibiotic prophylaxis remains important for IPD serotypes not in the current vaccine schedule.

THE CLOSING MESSAGE

Newborn screening for sickle cell disease: what can we learn from the practice in the European Union?

Pan-European recommendations in 2018

- Germany and Portugal implemented NBS after 2018; extension in Belgium
- NBS programmes are now all universal
- Methods for screening are evolving

- Lack of recommendation if no universal NBS
- Lack of national registries
- Still debate about reporting of heterozygosity for Hb S
- Few data on the follow-up

THANK YOU

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