ERN | EuroBloodNet

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ERN-EuroBloodNet Review Recommendations on sickle cell disease: their application at national level in the European Union, discrepancies and research questions

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European Reference Network | Hematological Diseases (ERN EuroBloodNet)

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Disclosure of Conflict of Interest

"The speaker declares no conflicts of interest inn relation to the subject matter of this presentation ."

EU 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 carrying out an inventory of practice of SCD care within the EU.

The expected results

- Highlighting within EU countries
  - ✓ 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

EU recommendations on Sickle Cell Disease (SCD)

FRAME

- Recommendations available in an EU country + UK (for comparison)
  - Scientific publications (Agency subcontracted by the EC)
  - National documents (Expert panel)
- Last update max10 years ago
- Any language (translated in English

EXPERT PANEL

- 27 experts from 9 MS

DOCUMENTS

- 26 + 3 UK
  - 27 national
  - 20 translated in English

EU recommendations on Sickle Cell Disease (SCD)

TOPICS

1. Diagnosis at birth

2. Prevention and management of complications
    - Standard follow-up
    - ACS
    - VOC
    - Stroke + TCD/MRI
    - Priapism
    - Kidney
    - Splenectomy/Splenic sequestration
    - Cardiomyopathy
    - Pregnancy
    - Pre-surgery
3. Treatments including prevention
    - Vaccination/antibioprophylaxis
    - Analgesia
    - Fever treatment
    - Hydroxyurea
    - Blood transfusion
    - HSCT
4. Transition

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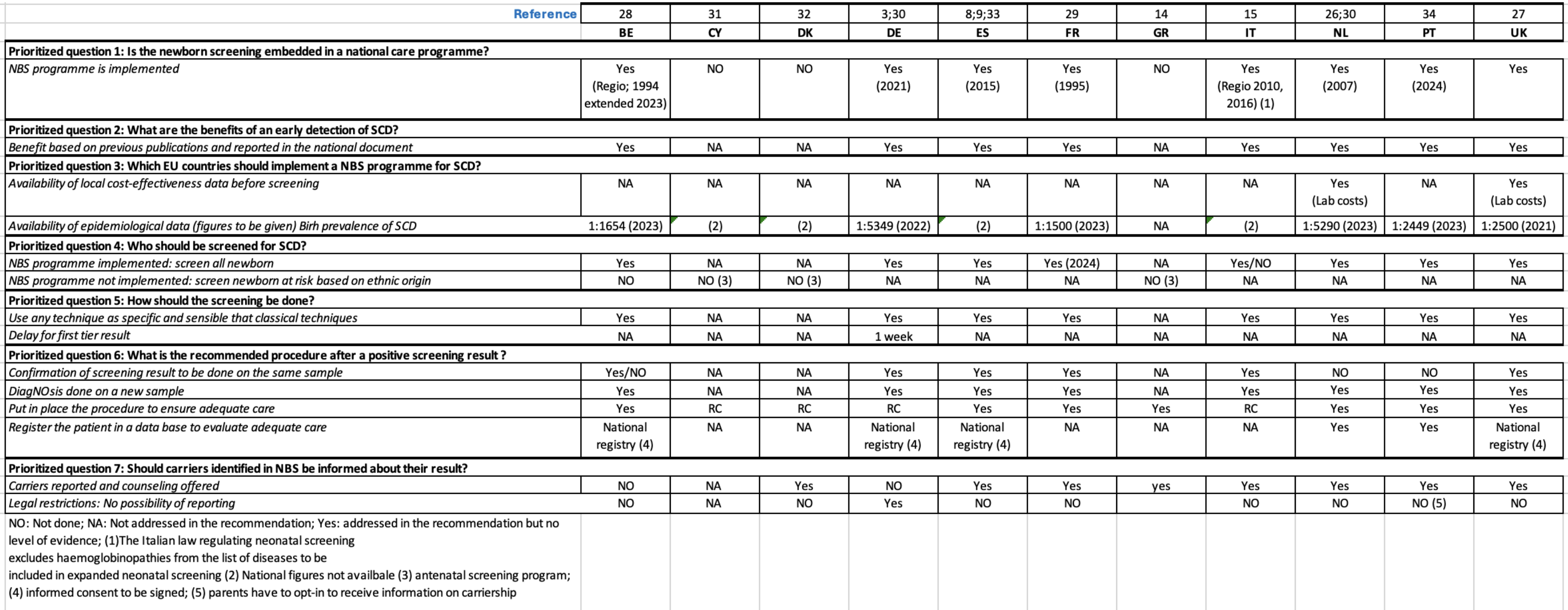
EU recommendations on Sickle Cell Disease (SCD)

Example - Newborn screening – Tables support for writing



Reference	28	31	32	3;30	8;9;33	29	14	15	26;30	34	27
	BE	**CY**	**DK**	**DE**	**ES**	**FR**	**GR**	**IT**	**NL**	**PT**	**UK**
Prioritized question 1: Is the newborn screening embedded in a national care programme?											
NBS programme is implemented	Yes (Regio; 1994 extended 2023)	NO	NO	Yes (2021)	Yes (2015)	Yes (1995)	NO	Yes (Regio 2010, 2016) (1)	Yes (2007)	Yes (2024)	Yes
Prioritized question 2: What are the benefits of an early detection of SCD?											
Benefit based on previous publications and reported in the national document	Yes	NA	NA	Yes	Yes	Yes	NA	Yes	Yes	Yes	Yes
Prioritized question 3: Which EU countries should implement a NBS programme for SCD?											
Availability of local cost-effectiveness data before screening	NA	NA	NA	NA	NA	NA	NA	NA	Yes (Lab costs)	NA	Yes (Lab costs)
Availability of epidemiological data (figures to be given) Birh prevalence of SCD	1:1654 (2023)	(2)	(2)	1:5349 (2022)	(2)	1:1500 (2023)	NA	(2)	1:5290 (2023)	1:2449 (2023)	1:2500 (2021)
Prioritized question 4: Who should be screened for SCD?											
NBS programme implemented: screen all newborn	Yes	NA	NA	Yes	Yes	Yes (2024)	NA	Yes/NO	Yes	Yes	Yes
NBS programme not implemented: screen newborn at risk based on ethnic origin	NO	NO (3)	NO (3)	NA	NA	NA	NO (3)	NA	NA	NA	NA
Prioritized question 5: How should the screening be done?											
Use any technique as specific and sensible that classical techniques	Yes	NA	NA	Yes	Yes	Yes	NA	Yes	Yes	Yes	Yes
Delay for first tier result	NA	NA	NA	1 week	NA	NA	NA	NA	NA	NA	NA
Prioritized question 6: What is the recommended procedure after a positive screening result ?											
Confirmation of screening result to be done on the same sample	Yes/NO	NA	NA	Yes	Yes	Yes	NA	Yes	NO	NO	Yes
DiagNOsis done on a new sample	Yes	NA	NA	Yes	Yes	Yes	NA	Yes	Yes	Yes	Yes
Put in place the procedure to ensure adequate care	Yes	RC	RC	RC	Yes	Yes	Yes	RC	Yes	Yes	Yes
Register the patient in a data base to evaluate adequate care	National registry (4)	NA	NA	National registry (4)	National registry (4)	NA	NA	NA	Yes	Yes	National registry (4)
Prioritized question 7: Should carriers identified in NBS be informed about their result?											
Carriers reported and counseling offered	NO	NA	Yes	NO	Yes	Yes	yes	Yes	Yes	Yes	Yes
Legal restrictions: No possibility of reporting	NO	NA	NO	Yes	NO	NO		NO	NO	NO (5)	NO
NO: Not done; NA: Not addressed in the recommendation; Yes: addressed in the recommendation but no level of evidence; (1)The Italian law regulating neonatal screening excludes haemoglobinopathies from the list of diseases to be included in expanded neonatal screening (2) National figures not availbale (3) antenatal screening program; (4) informed consent to be signed; (5) parents have to opt-in to receive information on carriership											

Writing ongoing

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THANK YOU!

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