

# Webinars

for patients

Sickle Cell Disease

Topic on Focus

**EuroBloodNet** 

## Neonatal screening

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European  
Reference  
Network

for rare or low prevalence  
complex diseases

Network  
Hematological  
Diseases (ERN EuroBloodNet)



**Nothing to declare.**



- **30-35min presentation (30 slides max) + 15 min Q&A session**
- **Microphones will be muted by host to avoid back noise**
- **Please, stop your video to improve internet connexion**
- **Send your questions during the presentation through the chat**



**After listening to this webinar on sickle cell disease you will be able to :**

- 1. Explain what means screening**
- 2. Understand the purpose and benefit of neonatal screening**
- 3. Understand for whom, how and where the screening is offered**



# WHAT MEANS SCREENING?

# What is screening ?



- To offer (not mandatory) a group of people not apparently affected but who are at risk of a condition (example: sickle cell disease) to be tested



Positive screening

Offer more tests to make the diagnosis of the condition

Comprehensive care



- **WHO 1968** — Principles and practice of screening for disease – *JMG Wilson & G. Jungner*

## – 10 criteria

1. The disease is a serious public health problem
2. The natural history of the disease is known
3. The disease must be detectable at a pre-symptomatic stage
4. A treatment of proven efficacy is applicable to patients
5. Appropriate means of diagnosis and treatment of patients are available
6. An effective screening test exists
7. The screening test is acceptable to the population
8. Screening must be accompanied by a precise therapeutic protocol (What criteria to benefit from the treatment?)
9. The economic cost-benefit ratio must be assessed
10. The sustainability of the program must be ensured and evaluated



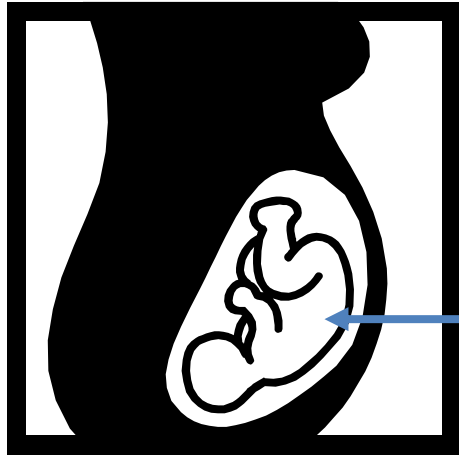
# THE PURPOSE AND BENEFIT OF NEONATAL SCREENING FOR SICKLE CELL DISEASE



# Why a neonatal screening for sickle cell disease?



- 1. Sickle cell disease is an inherited **serious blood illness**.



Fetal hemoglobin

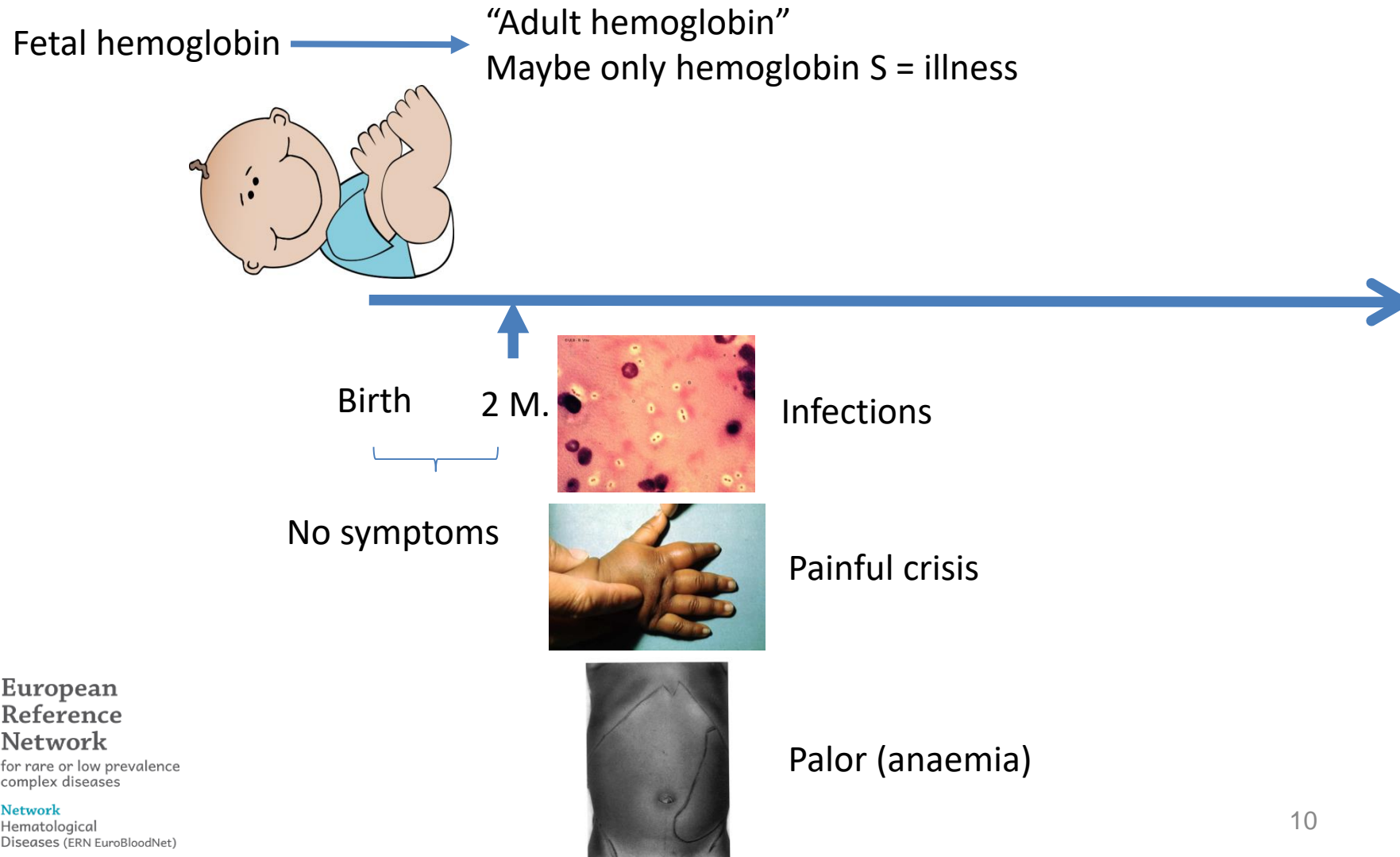


“Adult hemoglobin”  
Maybe only hemoglobin S = illness





- 2. Natural history is known
- 3. Pre-symptomatic phase for screening

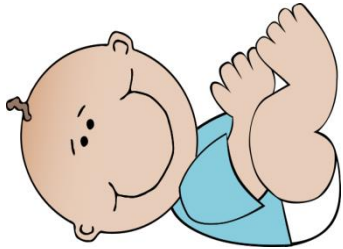


# Why a neonatal screening for sickle cell disease?



- DECREASE OF COMPLICATIONS : 4. and 5. early preventive measures and improvements in comprehensive care

*ENERCA guidelines  
Am J Hematol. 2011 Jan;86(1):72-5  
M. De Montalembert et al.*



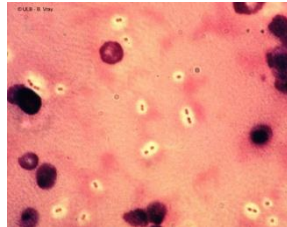
2 M.

2 Y.

## VACCINATIONS/ANTIBIOTHERAPY

*Davies et al.  
Health Technol Assess.  
2000;4:1*

*Gaston et al.  
NEJM 1986;314:1593-*



*Transcranial doppler = prevention*

## GENETIC COUNSELLING RECOMMENDATIONS PARENTS' EDUCATION



# Why a neonatal screening for sickle cell disease?



- Parents and children education = BENEFIT

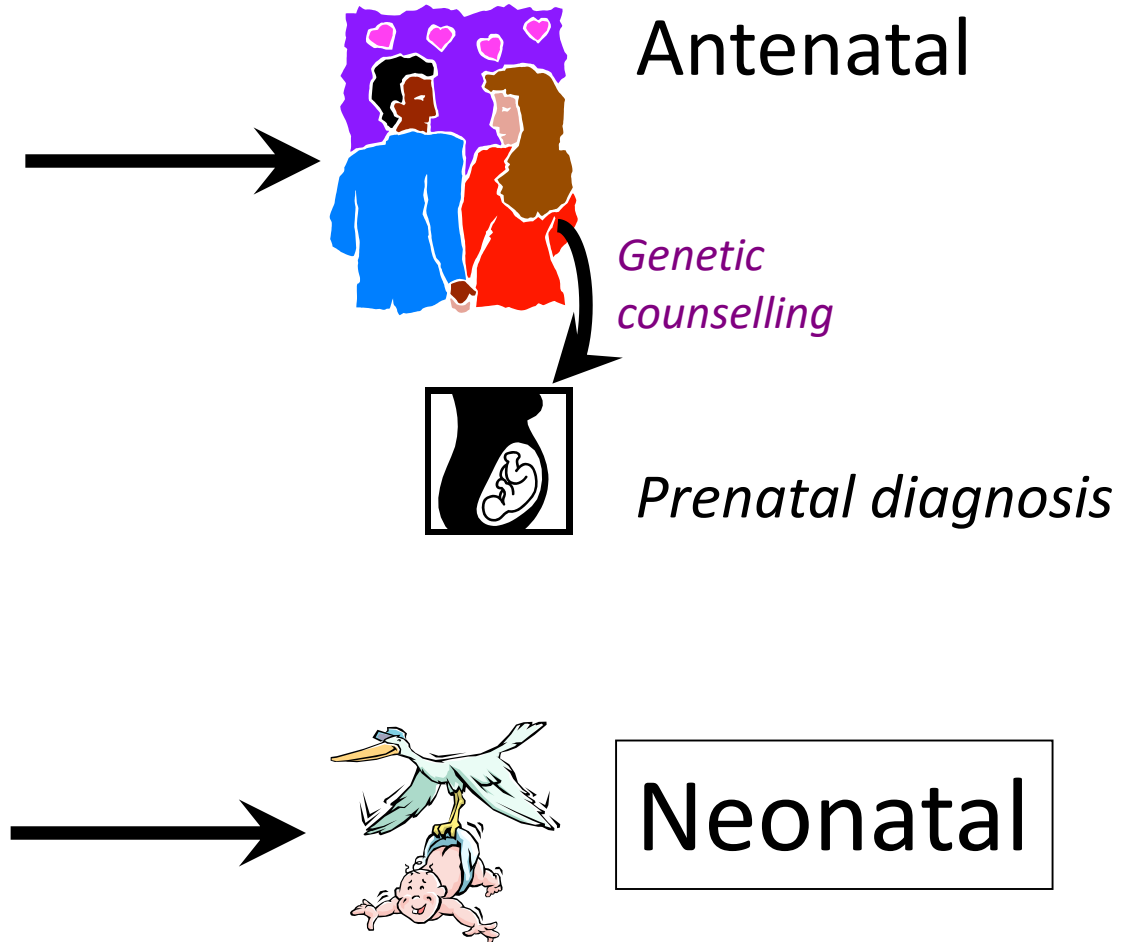
Example for painful crises

Don't forget some tips to avoid painful crises

- Cover up when its cold**  
Avoid extreme heat
- Don't give germs a chance  
Wash yourself well
- Oxygen yourself Go for walks, taking care to cover yourself
- If you play sports, avoid sports that are too intense
- Beware of high altitudes
- Avoid sitting too long, your blood needs to circulate
- Drink regularly throughout the day
- Go regularly to the doctor to follow your evolution Warn him at the slightest problem (fever...) or at the slightest question



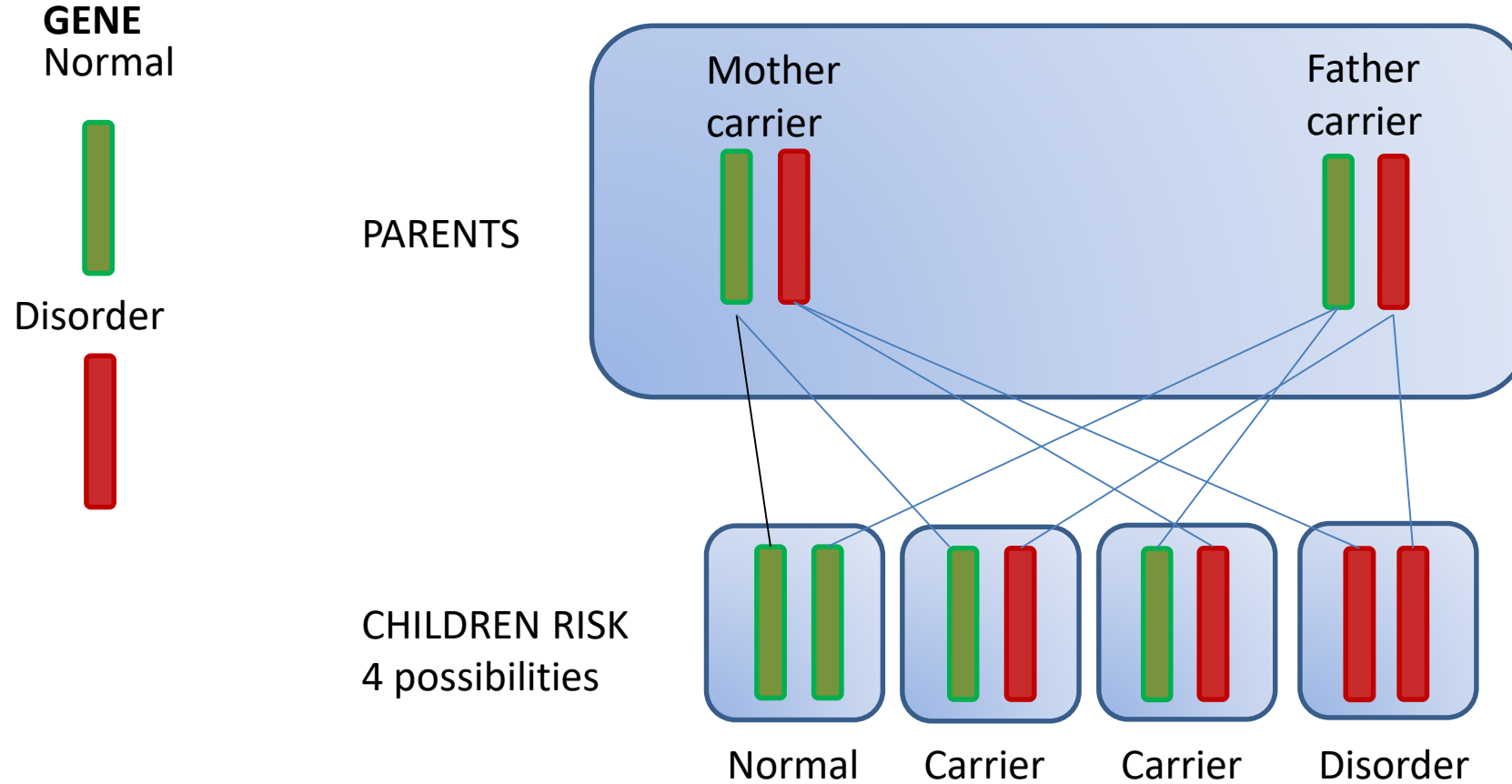
# FOR WHOM AND HOW NEONATAL SCREENING FOR SICKLE CELL DISEASE IS PERFORMED?



# Why a neonatal screening for sickle cell disease?



Both parents must at least be carrier of the disorder gene



*It affects both males and females*

**6. Can be detected at birth and the test is effective**

**8. Screening must be accompanied by a precise therapeutic protocol (What criteria to benefit from the treatment?)**

# Why a neonatal screening for sickle cell disease?



- Sickle cell disease: different “forms” and clinical expression >> 8 . Screening must be accompanied by a **precise therapeutic protocol** (What criteria to benefit from the treatment?)







The screening is performed in view to find babies at higher chance of sickle cell disease, a serious blood illness.

### Who is at risk?

All populations where malaria (mosquito-borne infectious disease) is or was endemic



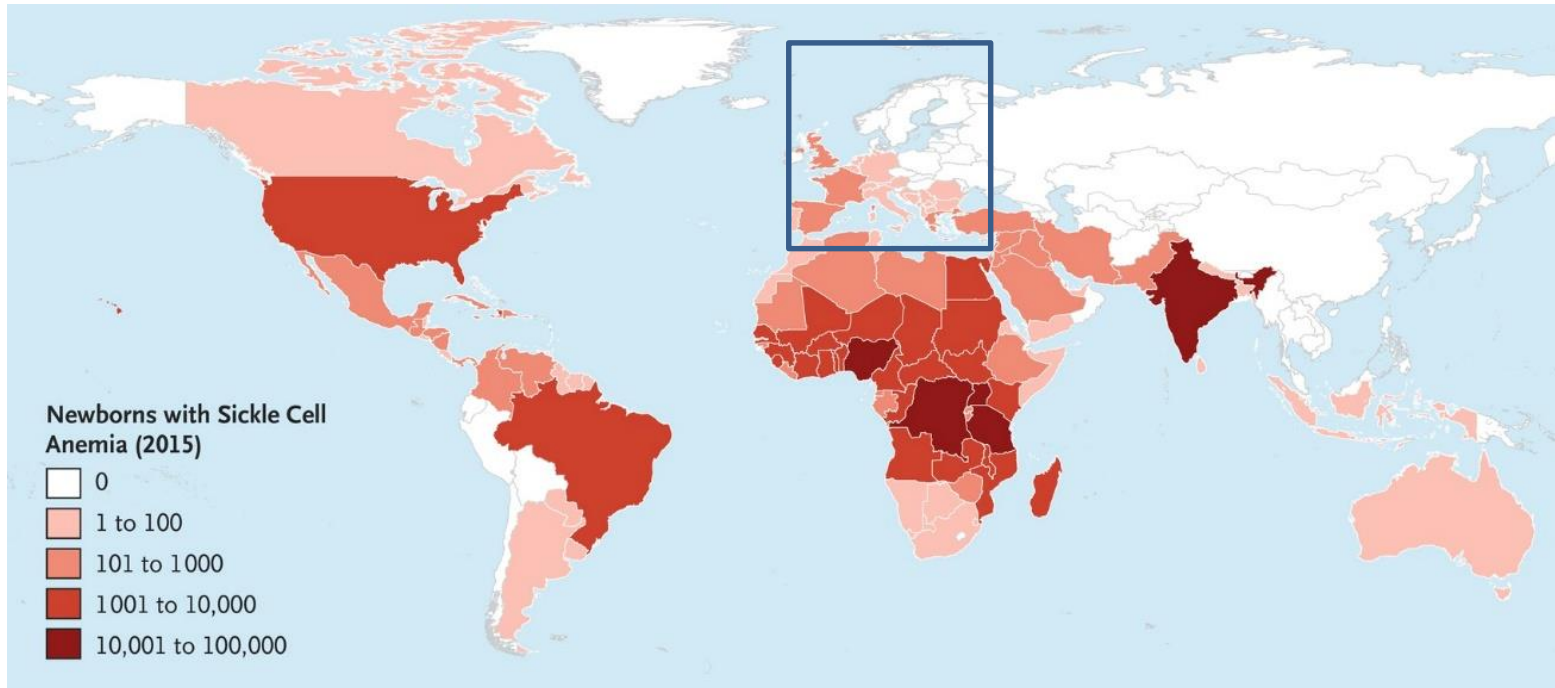
### IMPACT OF IMMIGRATION and ANCESTRY

#### Why this condition?

Because people with a sickle haemoglobin trait (carrier the condition) are more resistant to malaria infection



- 9. The economic cost-benefit ratio must be assessed



*Piel et al., 2017*



# HOW AND WHERE NEONATAL SCREENING FOR SICKLE CELL DISEASE IS PERFORMED?



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Reference  
Network**

for rare or low prevalence  
complex diseases

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Sickle Cell Disease

Topic on Focus

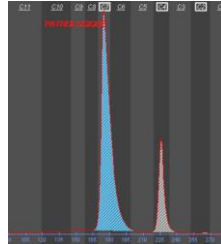
**EuroBloodNet**





- 6. An **effective screening** test exists
- 7. The screening test is **acceptable to the population**

Sampling at birth or a few days after → → Analysis in the laboratory → → Transmission of the result → → Control of the result with other tests





Sampling at birth or  
a few days after



Analysis  
in the laboratory



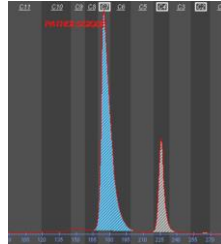
**Transmission  
of the result**



Control  
of the result  
with more tests



**Genetic counselling**



The baby is a carrier of a haemoglobin disorder  
(no risk of illness)

- For the child: future family
- For the parents:  
Already both being tested?  
Risk of having a child with the disease?



Sampling at birth or  
a few days after



Analysis  
in the laboratory



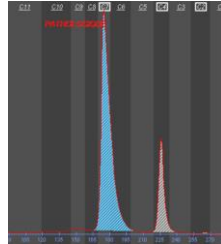
**Transmission  
of the result**



Control  
of the result  
with more tests



**Comprehensive  
Care management**



The baby has a positive test





It is included in a national (regional) programme for neonatal screening: rare metabolic and inherited diseases like sickle cell disease

- Targeted to the population at risk (France)
- Universal (all babies are screened) (UK, The Netherlands, Belgium, Spain, ...)

*Lobitz et al 2019*

Table I. Newborn screening programmes for sickle cell disease in Europe.

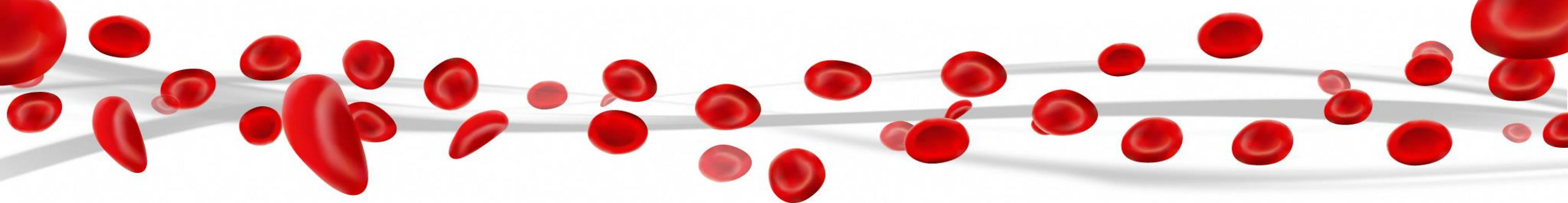
Country	Level	Coverage	Reference
Belgium	Regional (Brussels)	Universal	Gulbis <i>et al</i> (2009)
Belgium	Regional (Liège)	Universal	Gulbis <i>et al</i> (2009)
France	National	Targeted in metropolitan France and universal in overseas territories	Bardakdjian-Michau <i>et al</i> (2009) Saint-Martin <i>et al</i> (2013), Thuret <i>et al</i> (2010)
Netherlands	National	Universal	Bouva <i>et al</i> (2010)
Spain	National	Universal	Manu Pereira and Corrons (2009)
United Kingdom (England, Scotland, Wales, Northern Ireland)	National	Universal	Ryan <i>et al</i> (2010) Streetly (2000, 2005) Streetly <i>et al</i> (2008, 2010, 2018)

Please note: The UK has a linked antenatal and neonatal screening programme for haemoglobinopathies. Cyprus and Turkey have antenatal programmes only (Angastiniotis & Hadjiminias, 1981; Kolnagou & Kontoghiorghes, 2009; Canatan, 2014; Kountouris *et al*, 2016).



1. Screening means to test a group of individuals for a severe disease while a treatment is available; if positive, further tests are done to give the final diagnosis
2. Making the diagnosis of sickle cell disease early in life is important; it makes possible to prevent serious events
3. The frequency of sickle cell disease varies greatly from one European country to another: see migration of people from European or non European countries where malaria infection is or was endemic
4. Screening for the disease is simple, effective and not dangerous; it is implemented in several European countries where it has been proven to be cost-effective





## Discussion