ARISE
African Research And Innovative Initiative For Sickle Cell Education

# Newborn screening in SCD:
## a review of current approaches across continents

Prof Jacques Elion,* Dr Wale Atoyebi,** Ms Nkechi Anyanwu***

*Université de Paris, Inserm U1134 Integrative Biology of the Red Cell
National Institute of Blood Transfusion INTS, Paris, France

**Department of Clinical Haematology, Oxford University Hospitals NHS Foundation Trust, Oxford, UK

***Community Services, South East London Sickle Cell & Thalassaemia Centre, Evelina London, UK

Université de Paris

Instituts thématiques

Inserm
Institut national de la santé et de la recherche médicale

NHS
Guy's and St Thomas'
NHS Foundation Trust

NHS
Oxford University Hospitals
NHS Foundation Trust

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# SCD geographic distribution

Healthy carriers ≈ 5% of the world population
300.000 newborns/year (80% in low income countries)

# Newborn Screening and early care

… have dramatically changed life expectancy

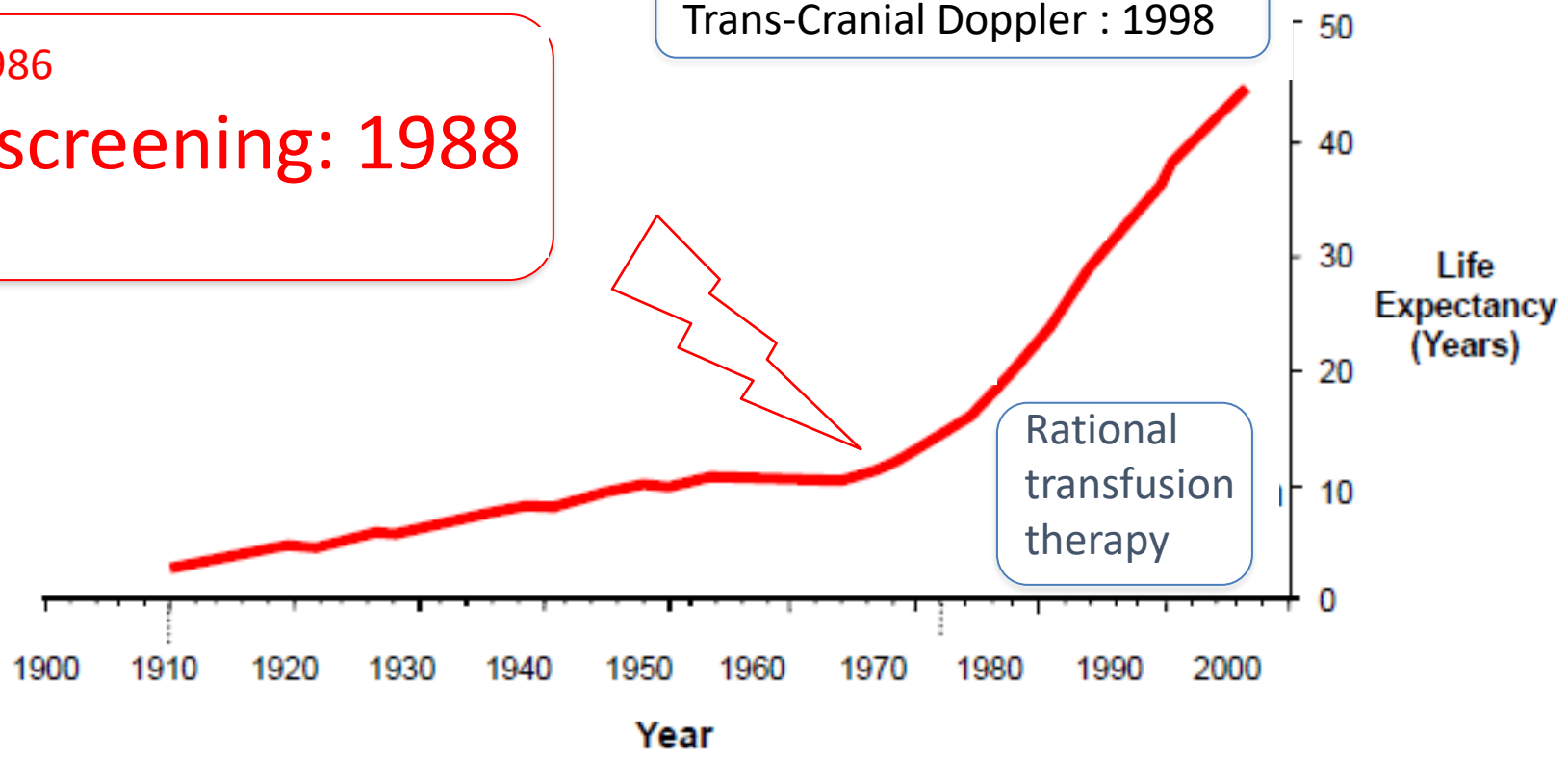
Daily penicillin: 1986

Newborn screening: 1988

Immunization

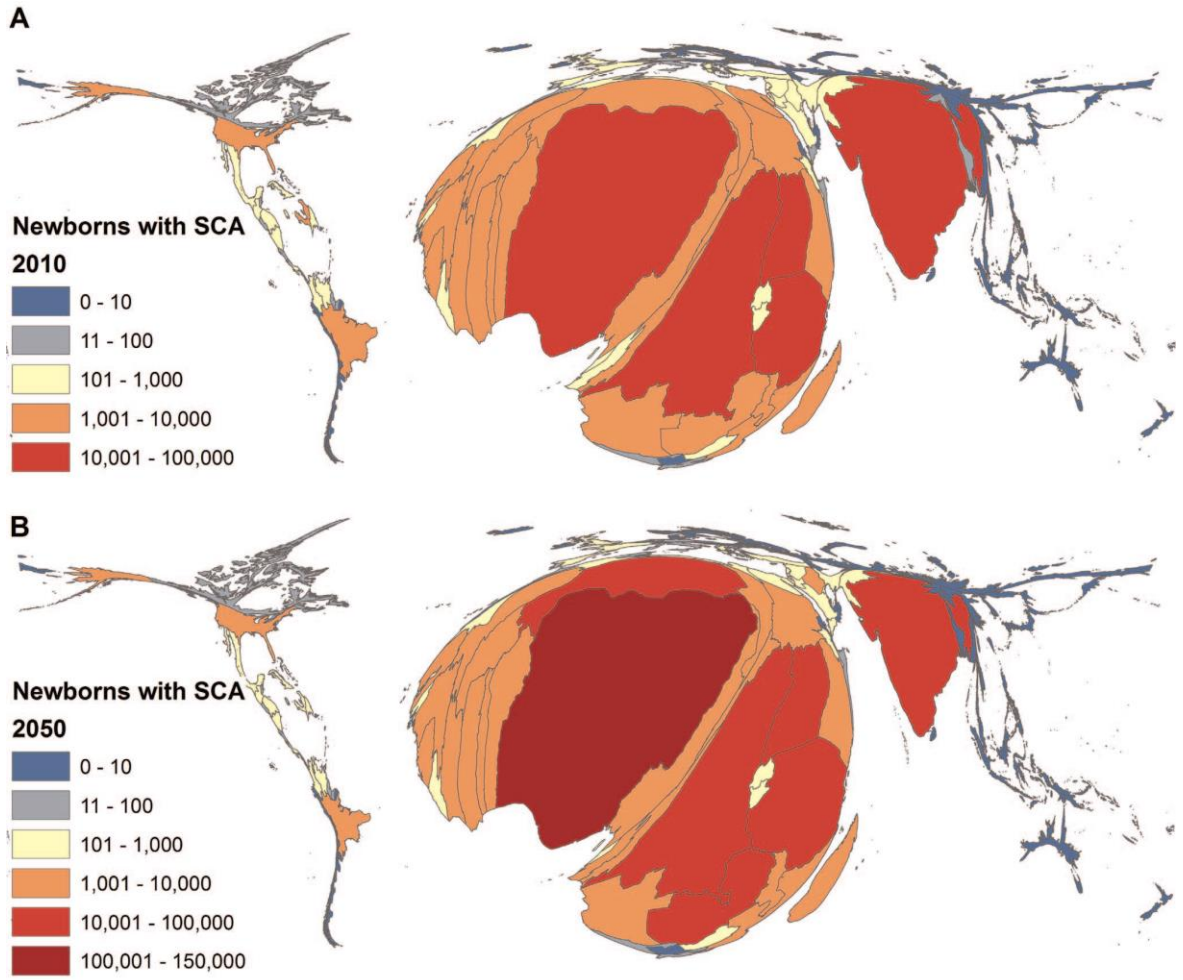
Hydroxycarbamide (HU): 1995

Trans-Cranial Doppler : 1998



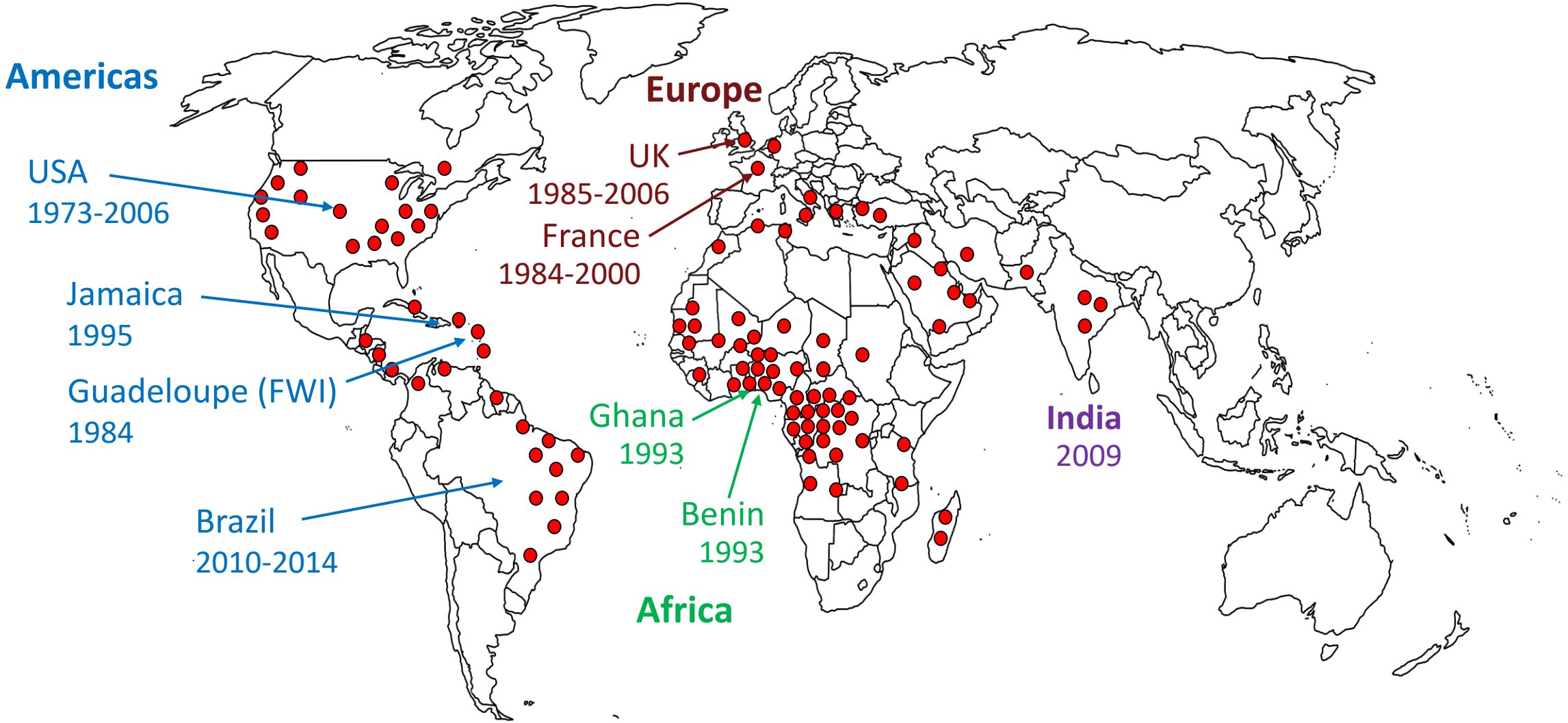
# Geographic disparity of the distribution of newborns affected with SCD

## estimated evolution 2010-2050

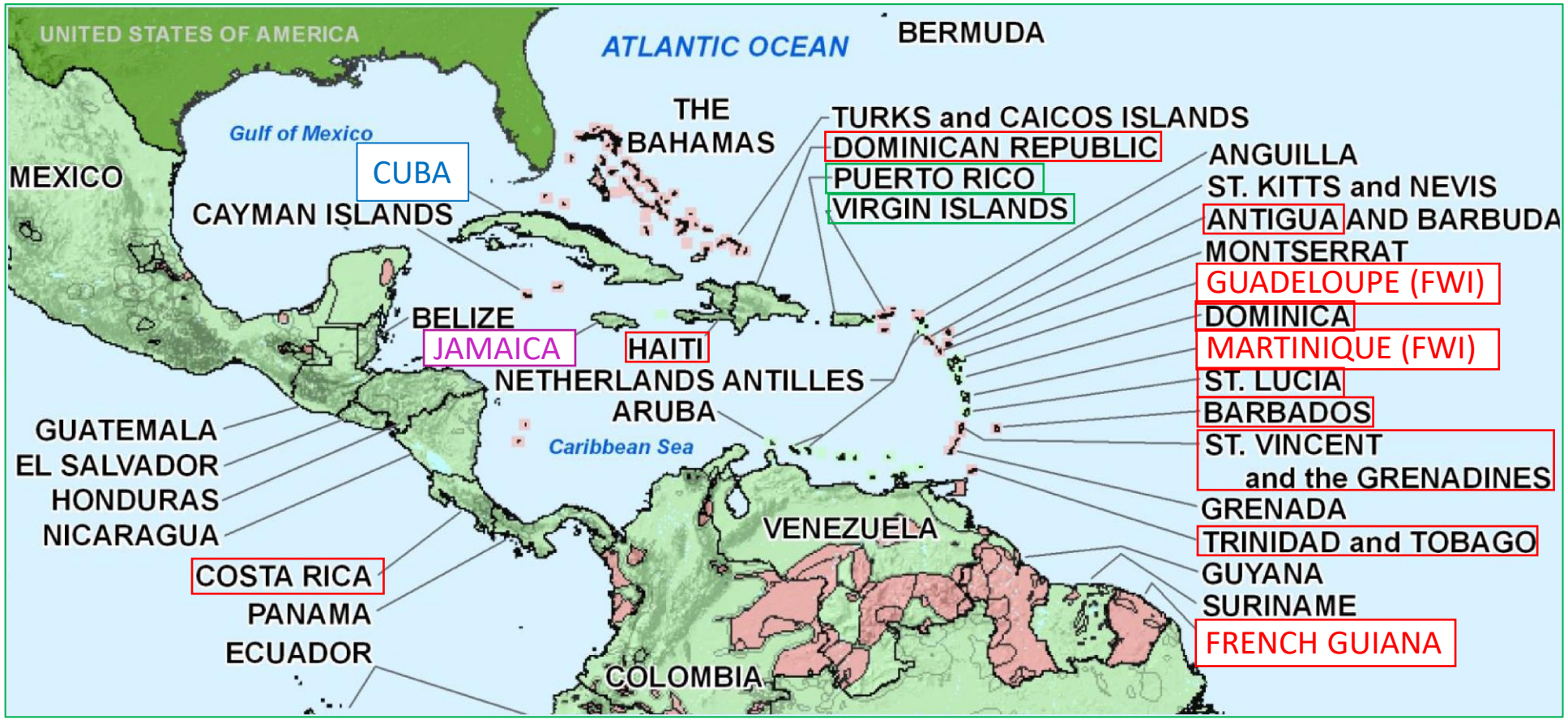



*Courtesy of Fred Piel*

# Chronology of pilot NBS projects



# The Caribbean




CAribbean network of REsearchers on Sickle cell disease a Sickle cell disease and Thalassemia. 2006 - present

https://carest-network.org

# The Caribbean: the case of Cuba

## Sickle Cell Anemia in Cuba: Prevention and Management, 1982–2018

**Beatriz Marcheco-Teruel MD PhD**

*MEDICC Review, October 2019, Vol 21, No 4*


Cuba is the only country is the world using antenatal diagnosis for SCD screening

Population: 11,000,000
A/S carrier frequency: 1/33
Incidence early 80's: 1/6000

| | |
|---|---|
| 1982-2018 | 4,847,239 pregnant women tested<br>8,180 at-risk couples identifed, 79.2% agreed to an antenatal study<br>20.1% of the tested fetuses had the SS genotype<br>76.2% of the couples decided to interrupt the pregnancy |
| Results | 3-fold reduction in prevalence of SCD in Cuba<br>10-fold reduction of the incidence of infants born with SCD yearly<br>16-year average increase in life expectancy of patients |

# Brazil: nine years to cover the whole country with an exhaustive NBS program

## Population: 211 M

## National NBS Program


| | |
|---|---|
| Phenylketonuria and congenital hypothyroidism | 2006 |
| Sickle cell disease | 2013 |
| Cystic fibrosis | 2013 |
| Congenital adrenal hyperplasia and biotinidase deficiency | 2014 |

all the 27 states are covered

**2010**


**2012**


**June 2014**

# SCD NBS in Brazil

Saúde
Ministério da Saúde

SUS Sistema Único de Saúde

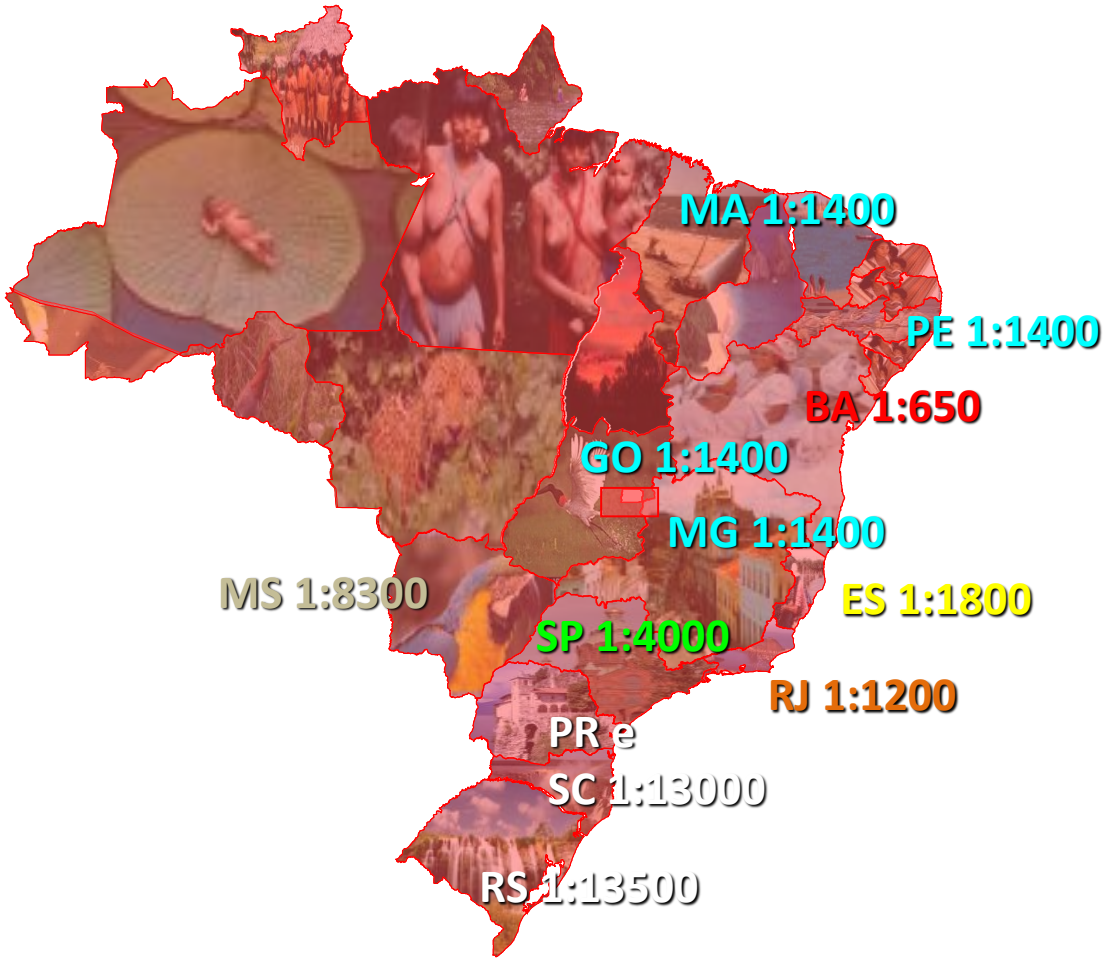
GOVERNO FEDERAL BRASIL PAÍS RICO É PAÍS SEM POBREZA

Birth rate: - 3,1 million / 2018

Screening coverage: 83%

Newborns with SCD - 2,500 - 3000

Mean incidence 1: 1000

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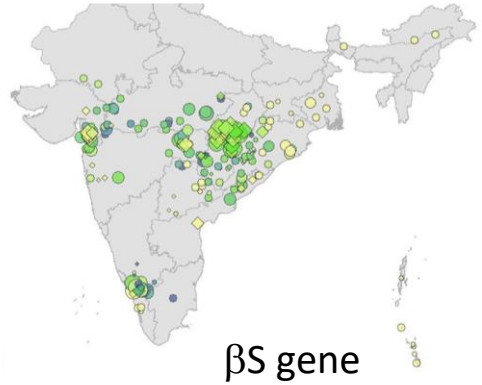
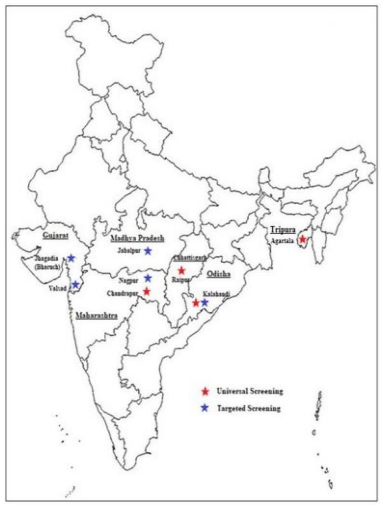
# India

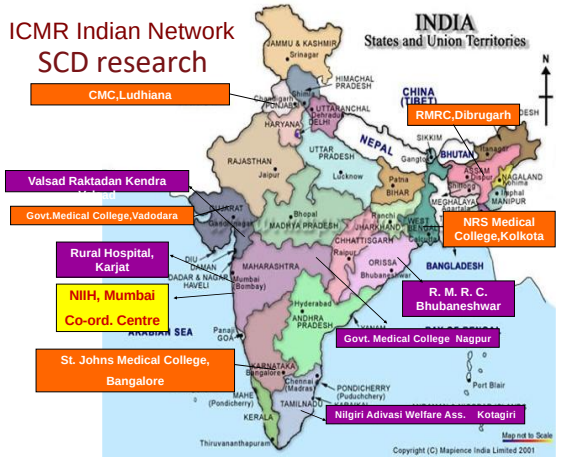
## Population: 1,3 billion



βS gene distribution

SCD Reference Centres




SCA is prevalent in West Central (Gujarat, Maharashtra) and East Central India (Chhattisgarh, Odisha) with a smaller focus in the southern region (Kerala)

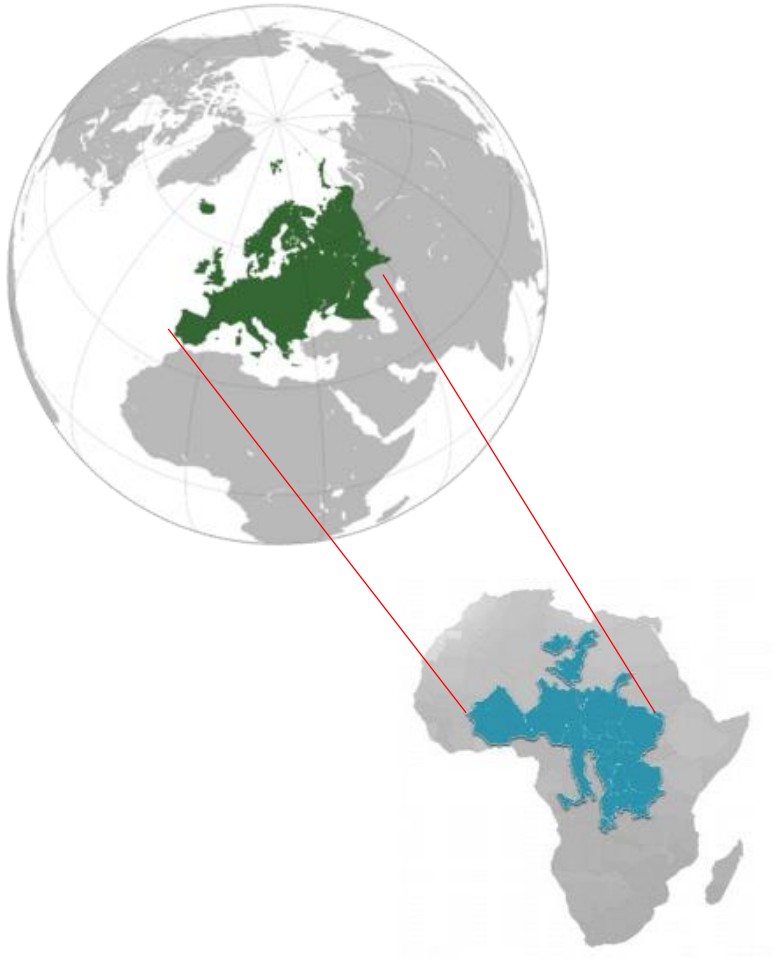
among tribal populations who are considered to be the original inhabitants, scheduled castes and other backward classes (non-tribal populations)

patients often reside in remote rural regions away from the mainstream

carrier frequencies may range from 1 to 35% in these groups

the β thalassemia gene is frequent (icentral India 40% of the SCD patients have sickle-β thalassemia

# Europe



50 countries
Population 750 million

USA
pop. 320 million

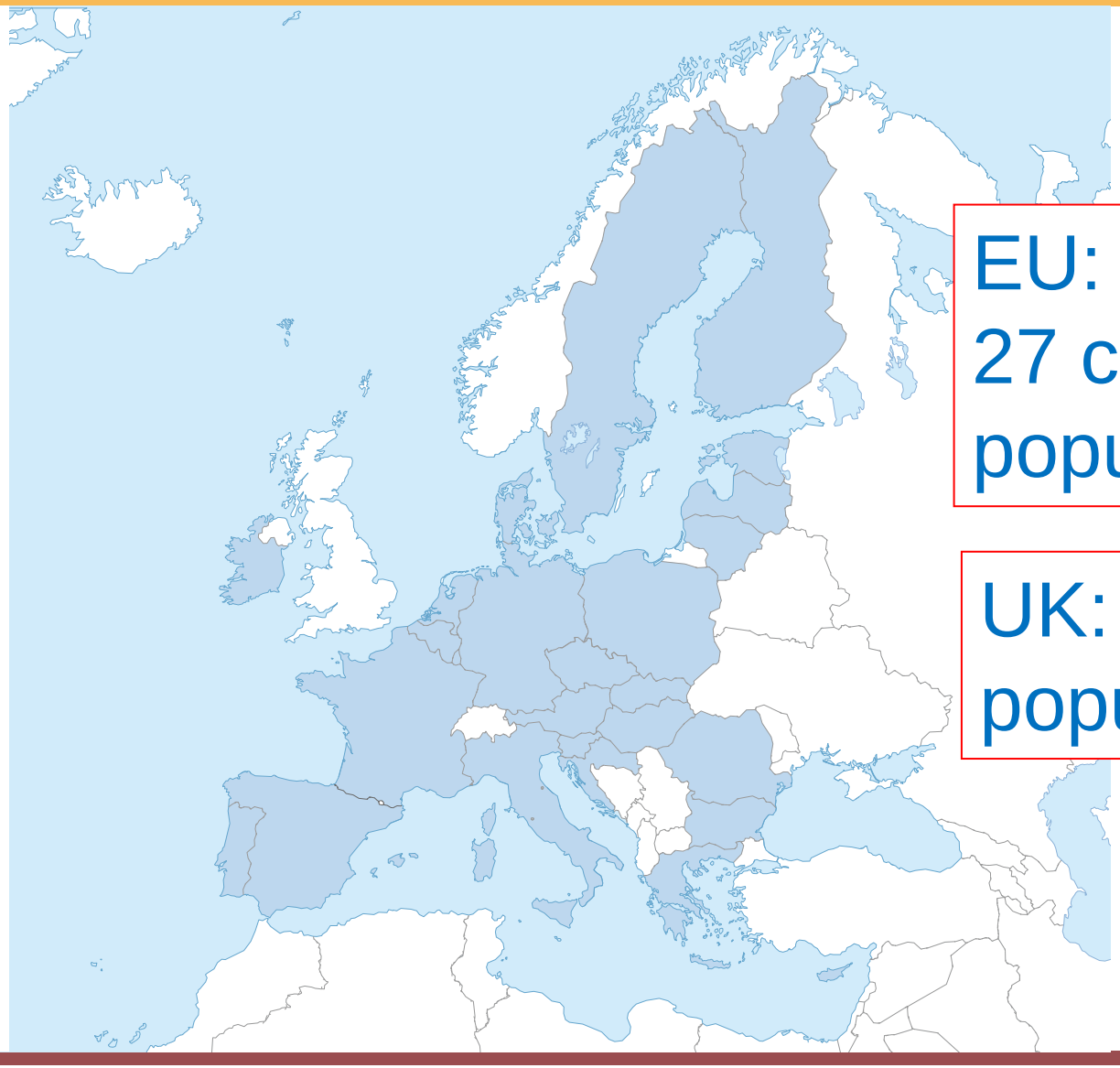
India
pop. 1.3 billion

Africa

50 countries

population 1.2 billion

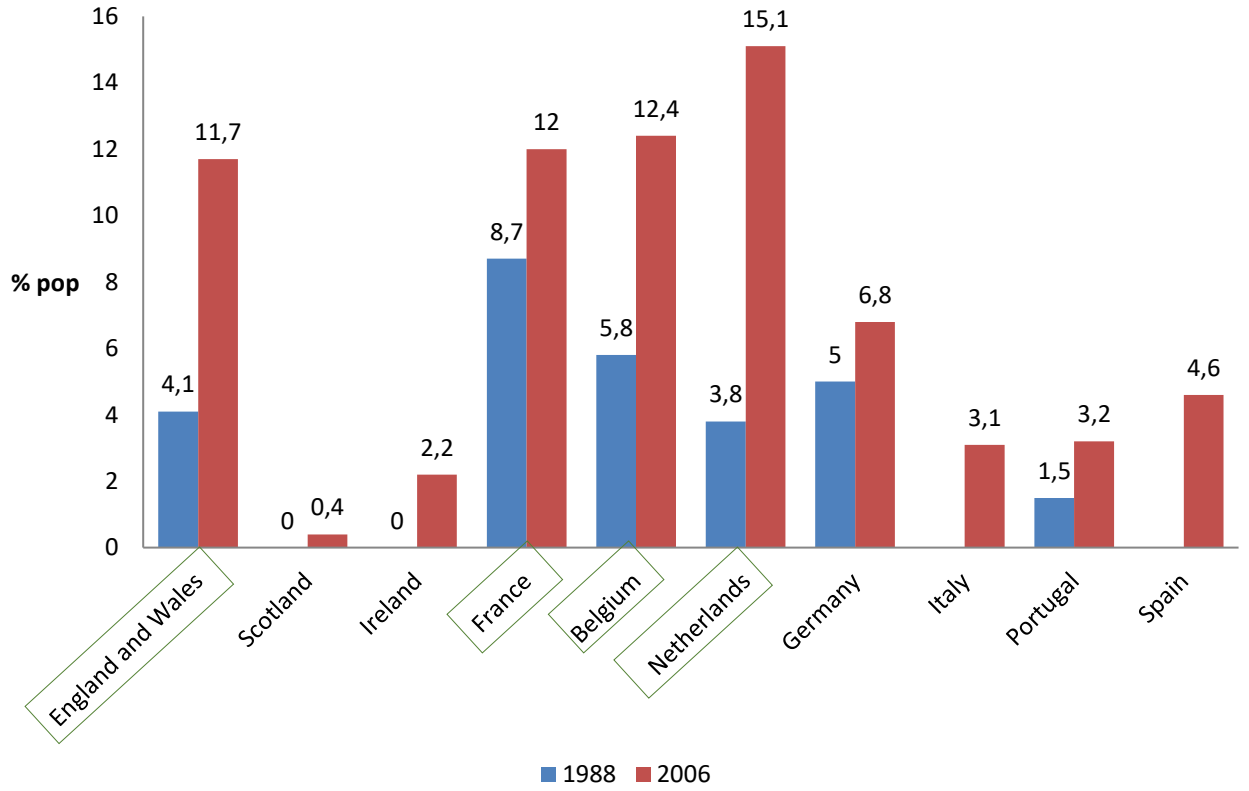
# European Union / UK



EU:
27 countries
population 447 millions

UK:
population 65 millions

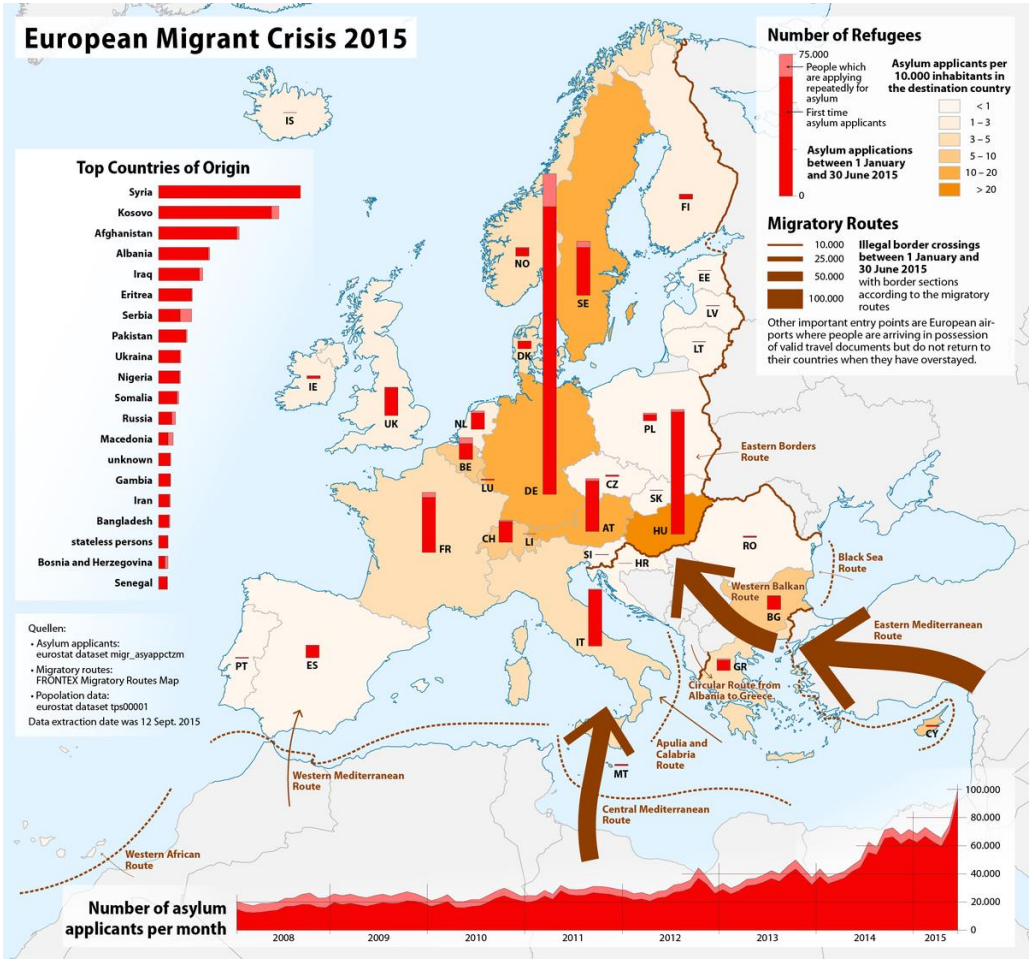
# Europe: Trends in populations at-risk for SCD



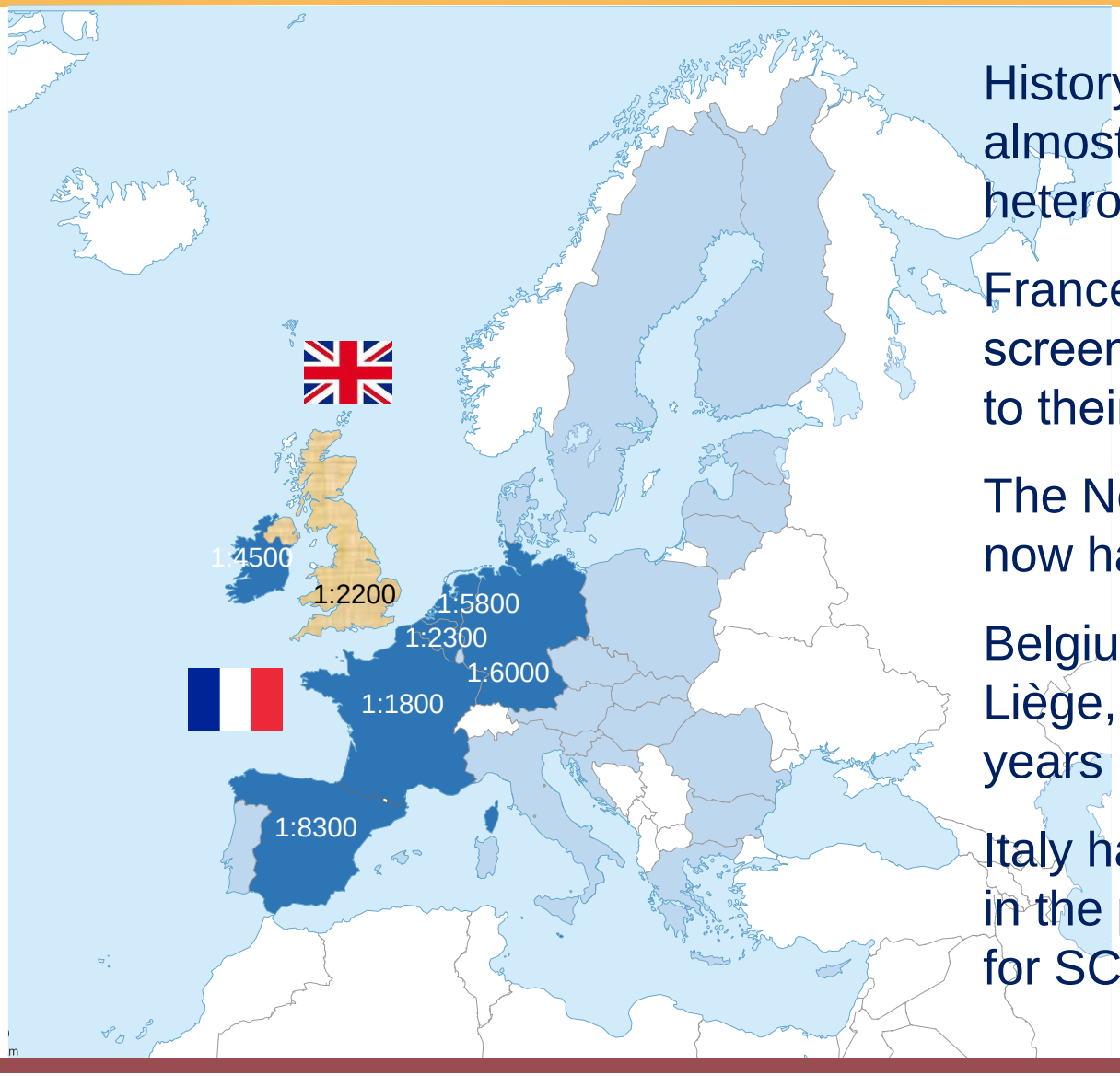

*Adapted from Modell et al, Scan J Clin Lab Invest 2007; 67: 39-70*

# Europe: Contemporary migrant crisis

- dramatic change of migration patterns from the Middle East and Africa
- between 2010 and 2017, nearly 1 million asylum claims from sub-Saharan Africa



# SCD NBS in Europe




History of NBS for SCD in Europe goes back almost 40 years, still the situation is highly heterogenous from one country to another

France and the UK were the first to introduce SCD screening in the 1980's and to extend its coverage to their entire national territories in the 2000's

The Netherlands, Spain, Malta and Germany also now have national programs

Belgium screens in the regions of Brussels and Liège, Ireland has been running a pilot for many years

Italy has completed several pilot studies but is still in the preparatory phase of national NBS programs for SCD

# Reference

International Journal of
*Neonatal Screening*

## Special Issue "Newborn Screening for Sickle Cell Disease and other Haemoglobinopathies"

Stephan Lobitz, Jacques Elion, Raffaella Colombatti and Elena Cela, Eds.

https://www.mdpi.com/journal/IJNS/special_issues/hemoglobinopathies
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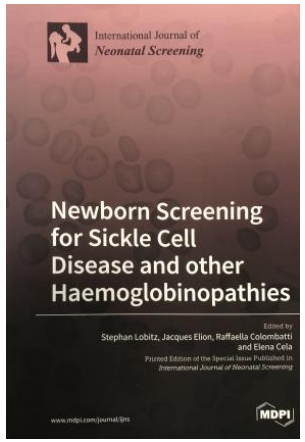

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