

Opportunities for Genomic Research in SCD

Contribution from Africa

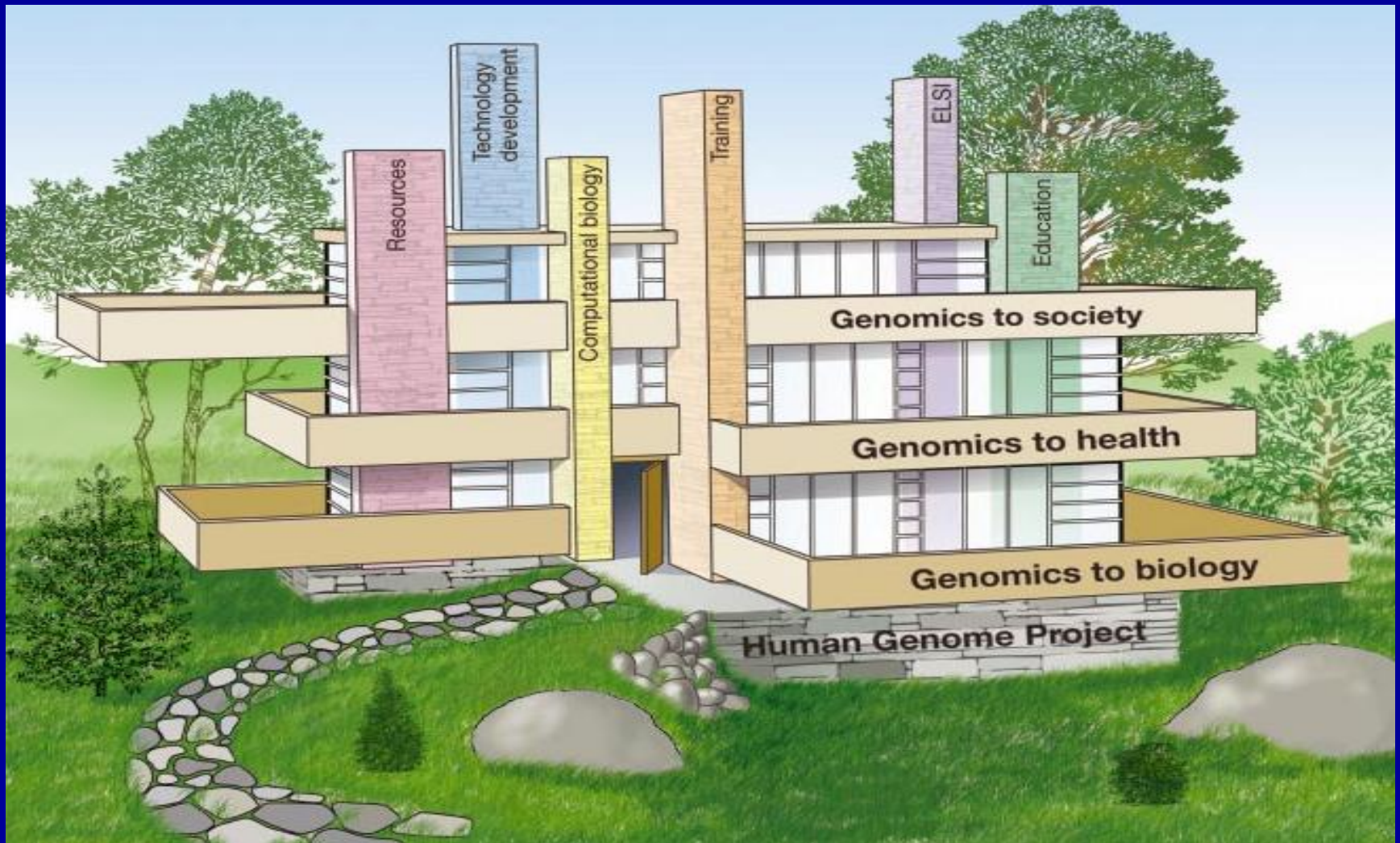
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University of Oxford

Outline

- SCD and the future of genomics
- State of SCD in Africa
- What can Africa contribute?
- What are the challenges that need to be addressed?

The future of genomics



'Genomics to Health'



Genetic basis

Gene – gene interactions

Gene - environment

Clinical disease

- **Multiple mechanisms**
- **Single gene-variable phenotypes**

Public Health

- **Health systems**
- **Socioeconomic**
- **Behavioural factors**

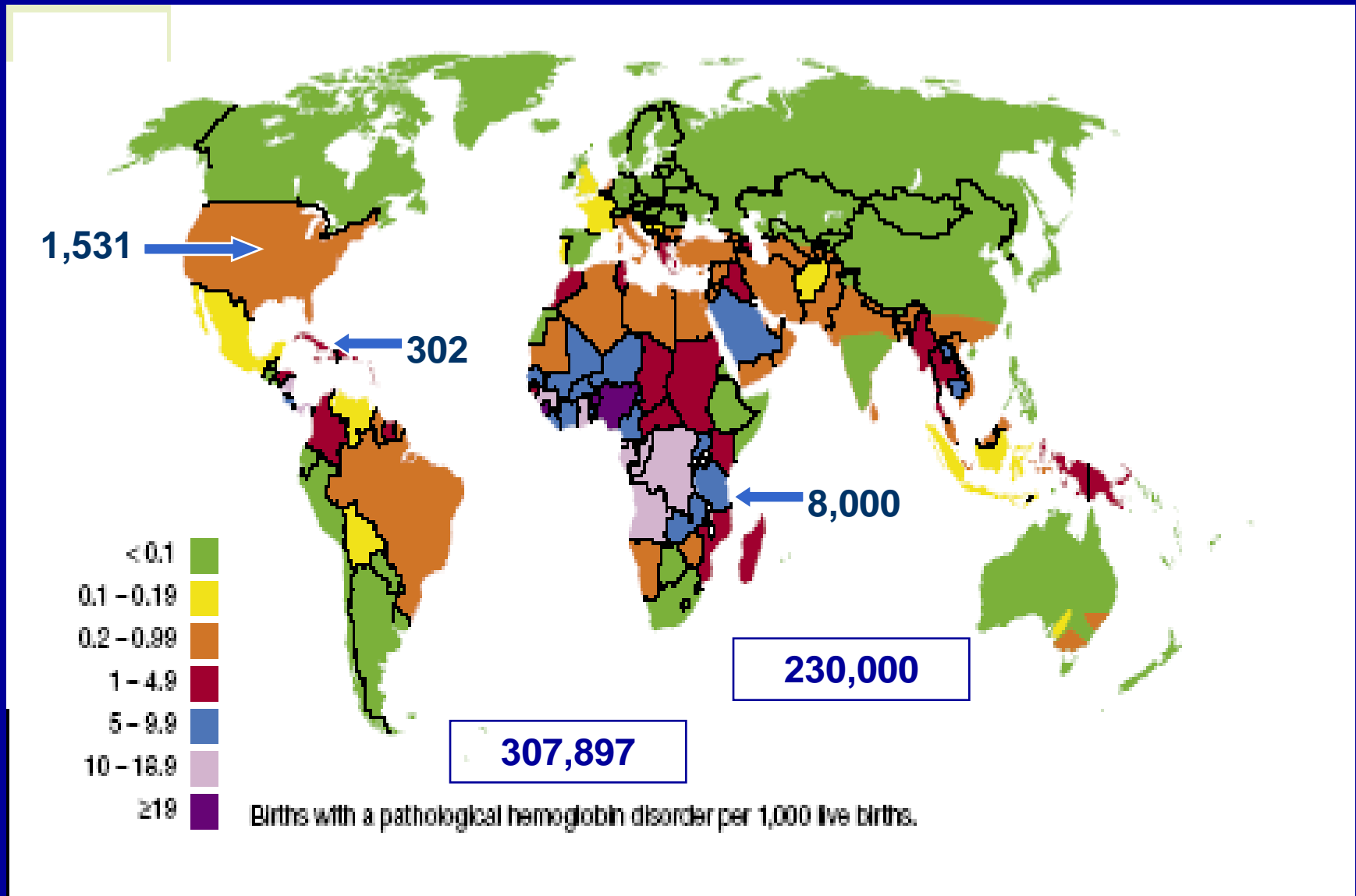
A 'model disease' for genetic studies

- High heritability
 - Coding mutations with large effect .
 - Relatively easy to transfer to cellular and animal models for mechanistic studies
- Unmet medical need
- Limited interventions.

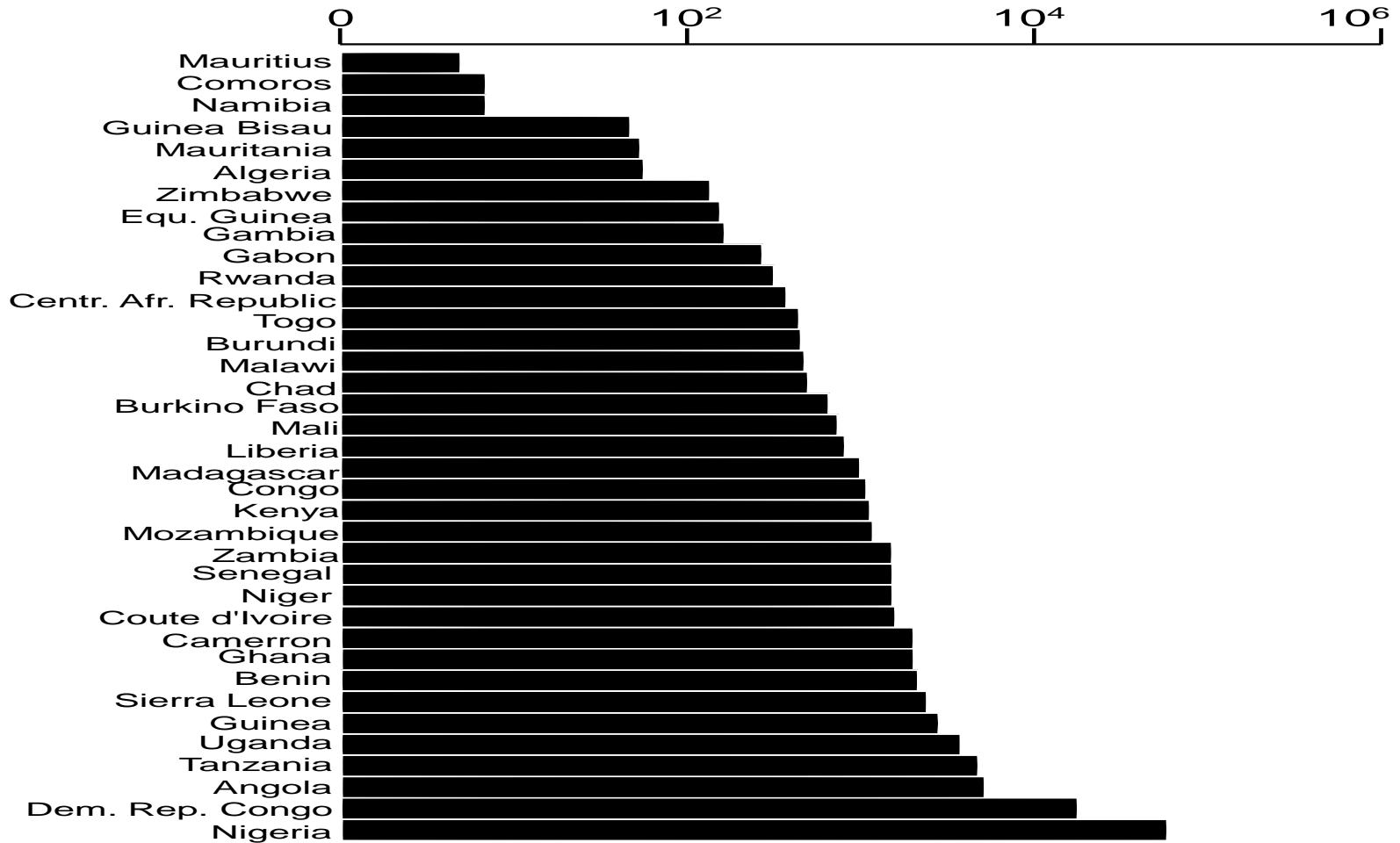
Altshuler *et al*, Science 2008.

State of SCD in Africa

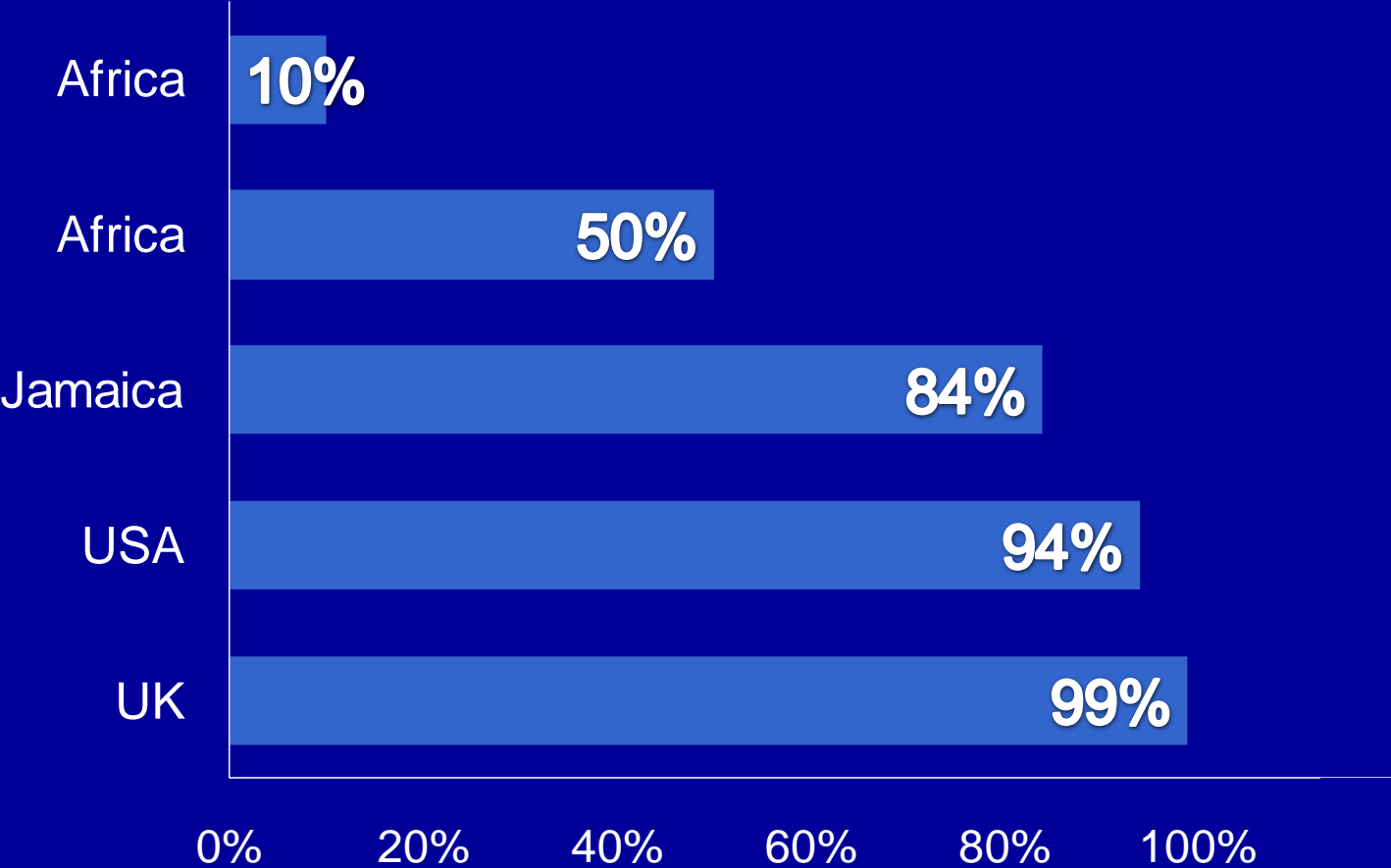
Global Burden of SCD



Birth estimates

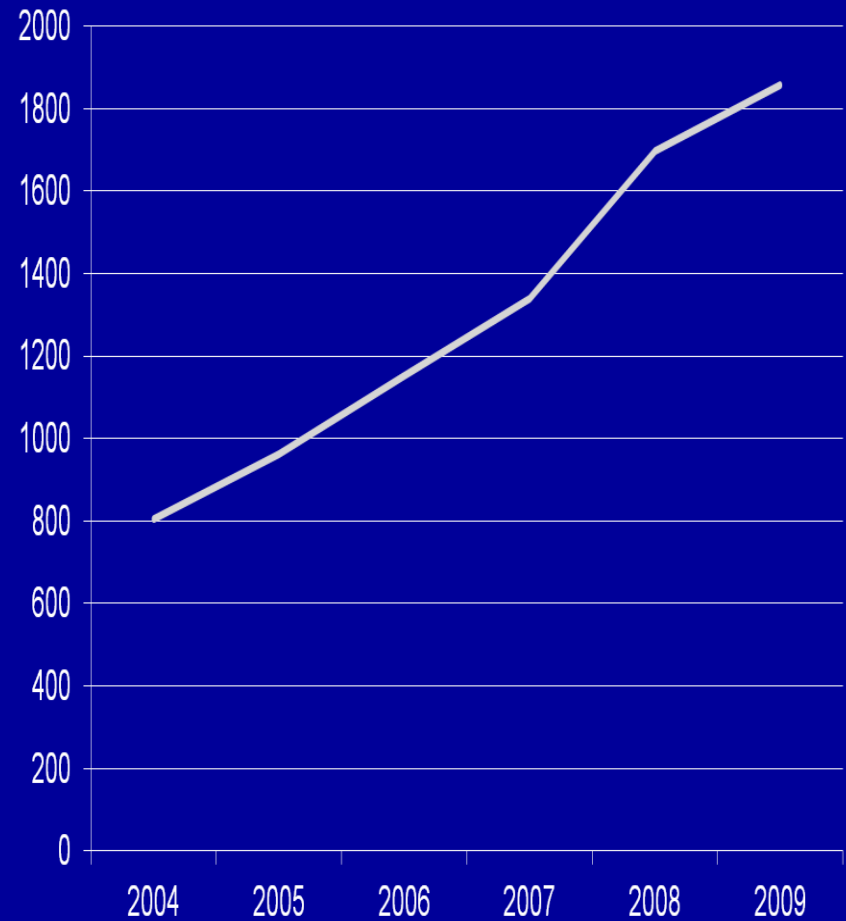
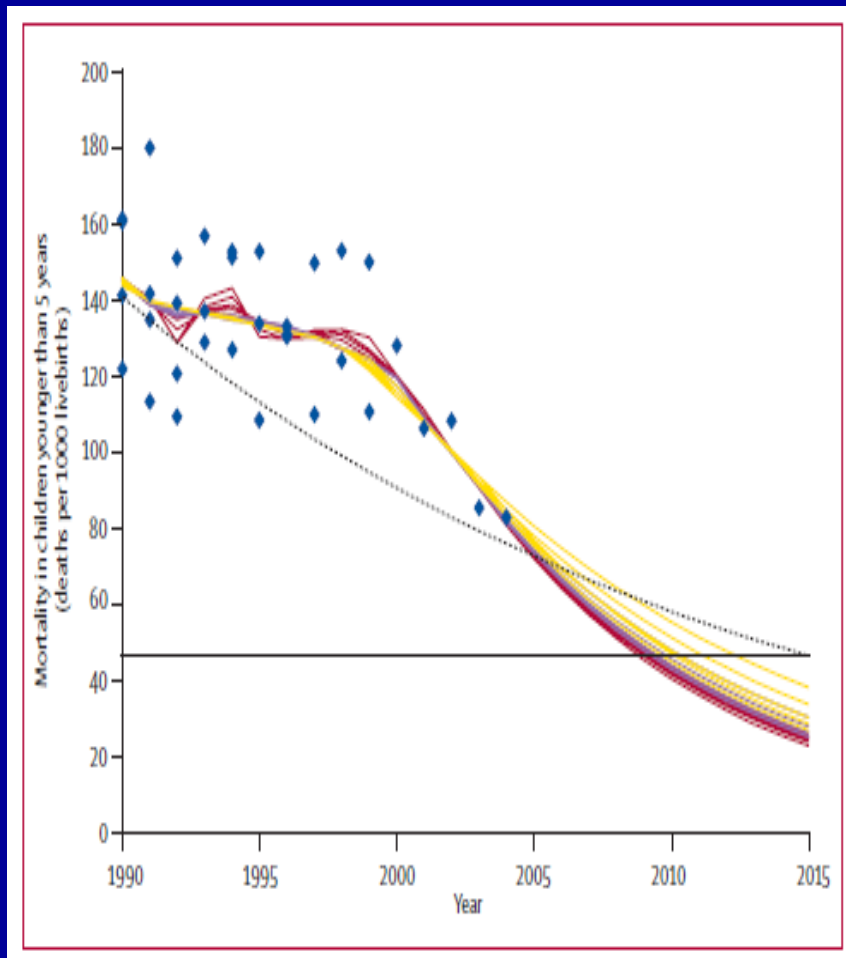


Childhood Survival for SCD

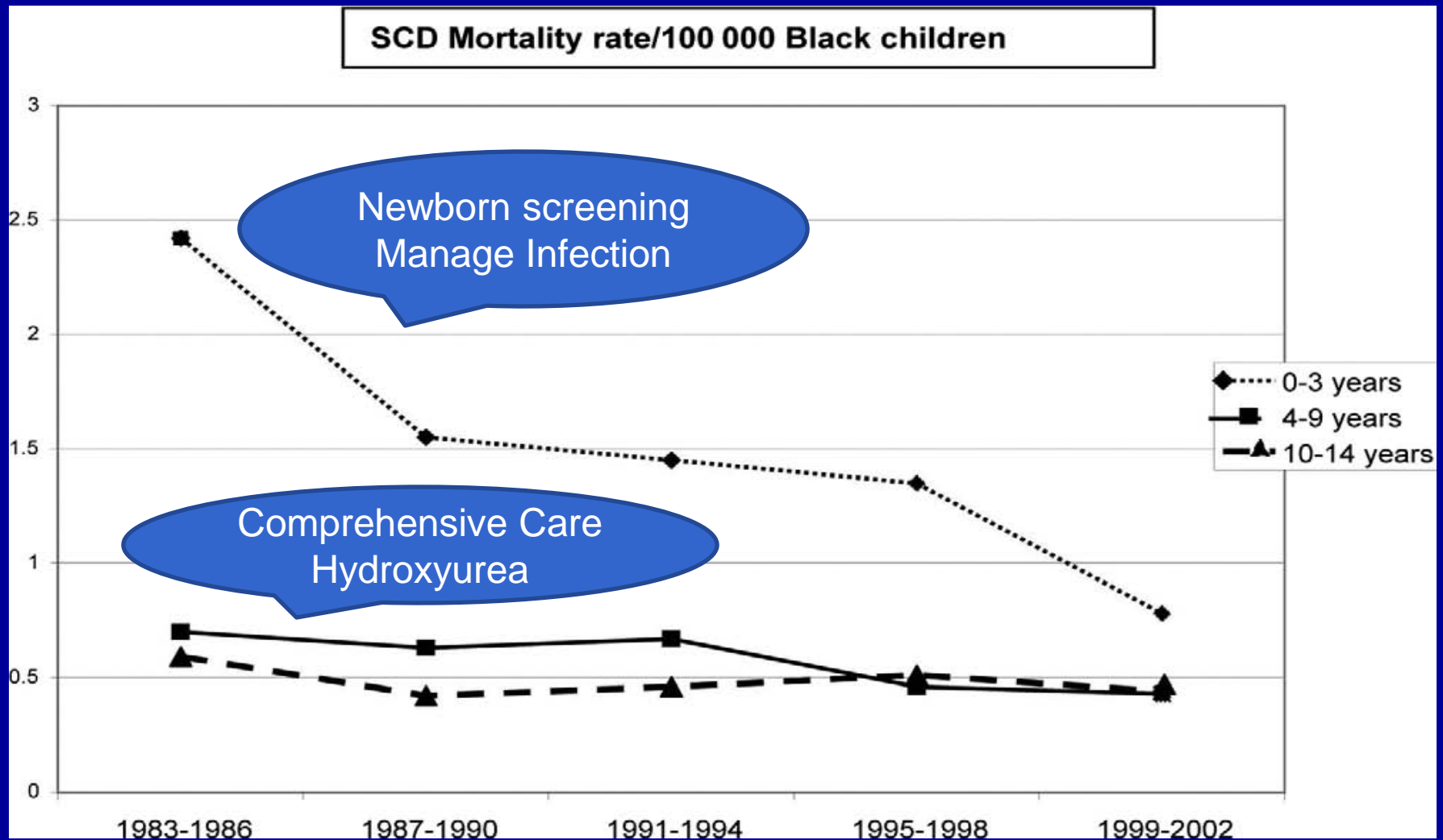


Fleming, 1979; Weatherall, 2006; Wierenga, 2001; Quinn 2004; Telfer, 2007

Demographic Transition

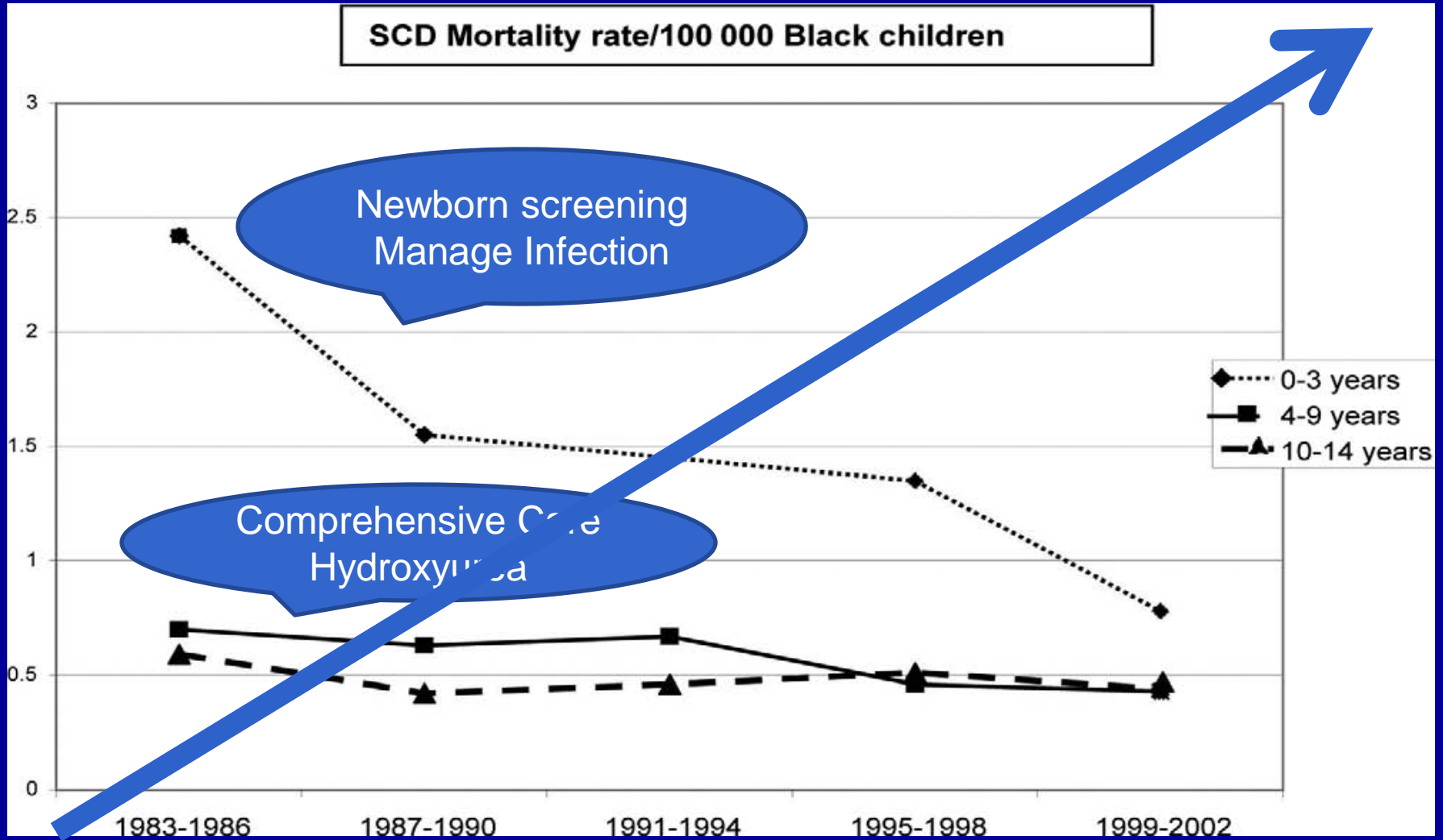


Limited Interventions in SCD



Trend in Mortality in SCD in the USA

Interventions in SCD in Africa



Trend in Mortality in SCD in the USA

What can Africa contribute?

Genomic Research

Genotyping

Laboratory capacity

Phenotyping

Hb, Haemolysis, HbF, Stroke

Bioinformatics

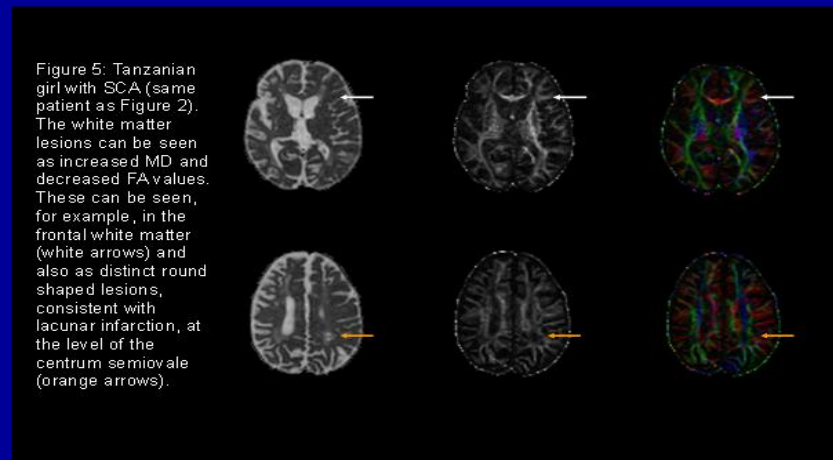
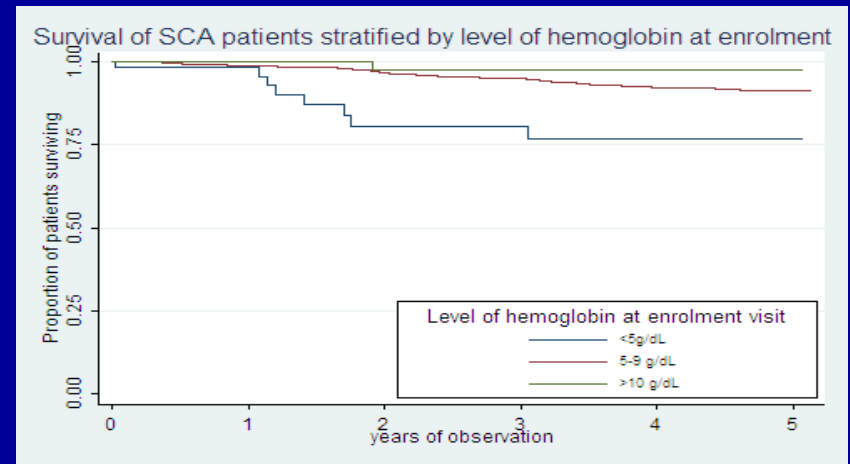
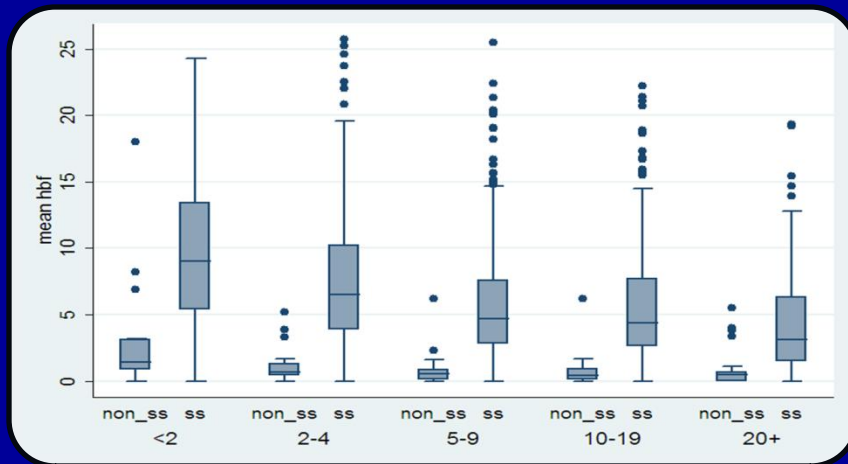
**Health
benefits**

Clinical Studies

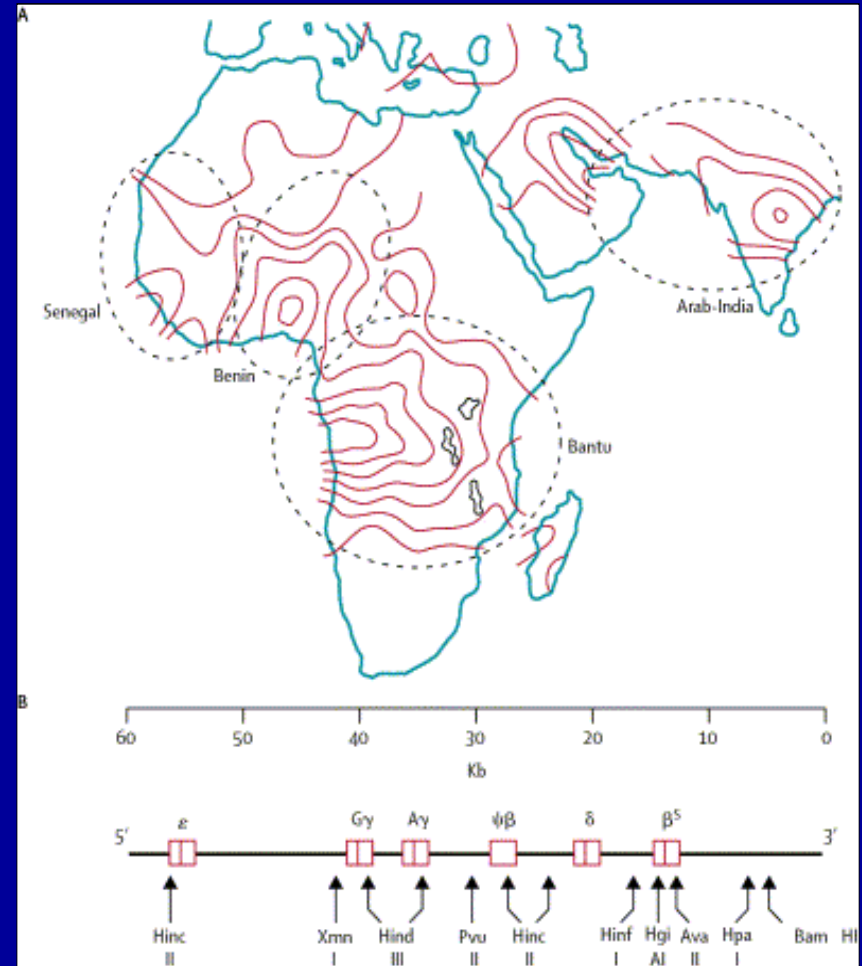
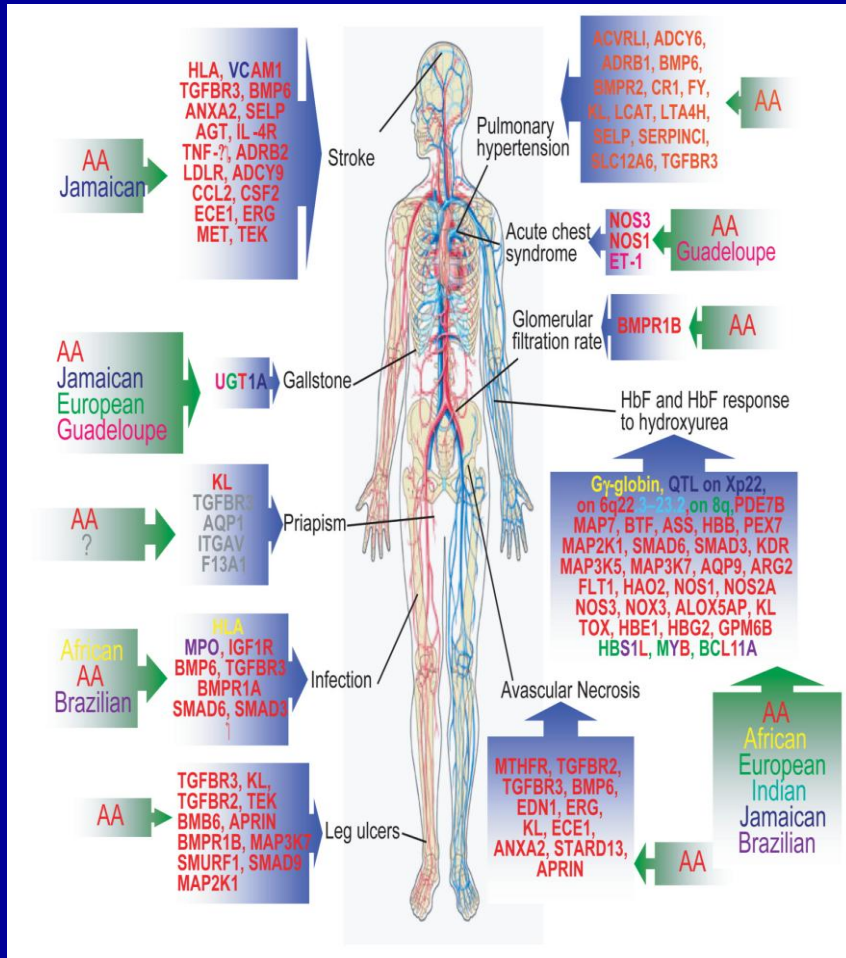
Statistical and Epidemiological Analysis

Phenotypic Diversity

HbF, Hb, Stroke, Haemolysis



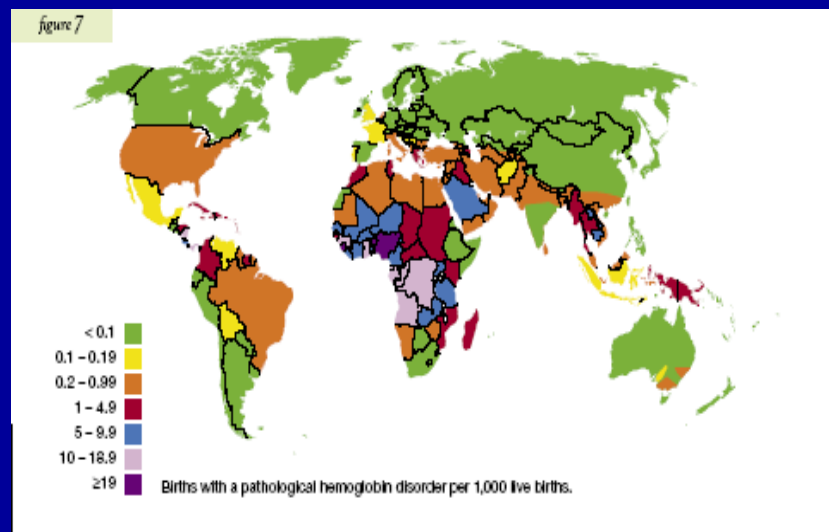
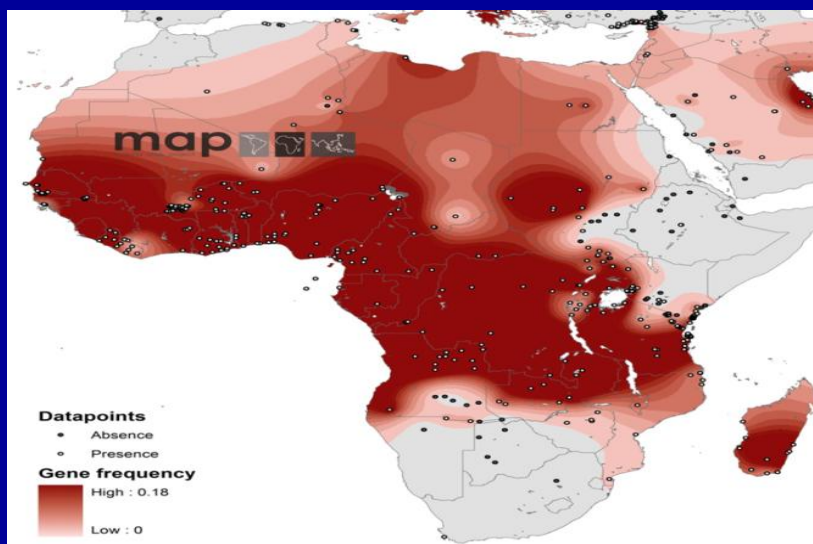
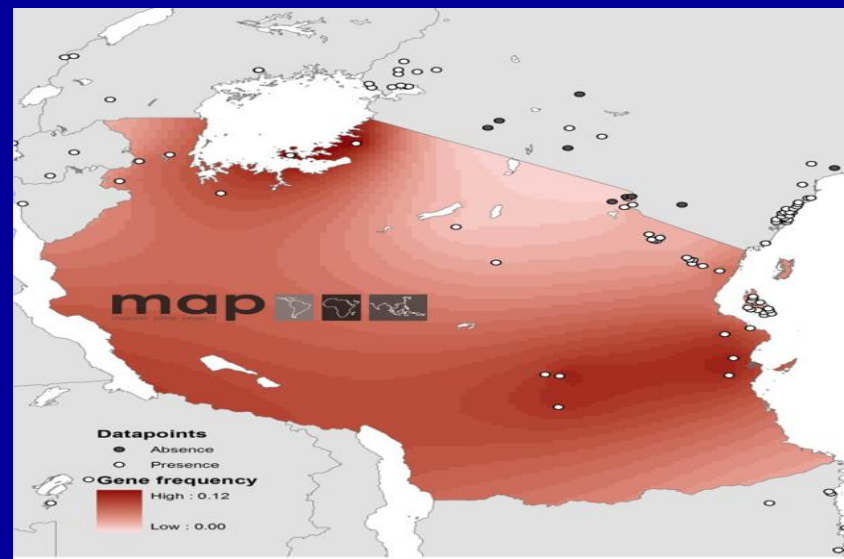
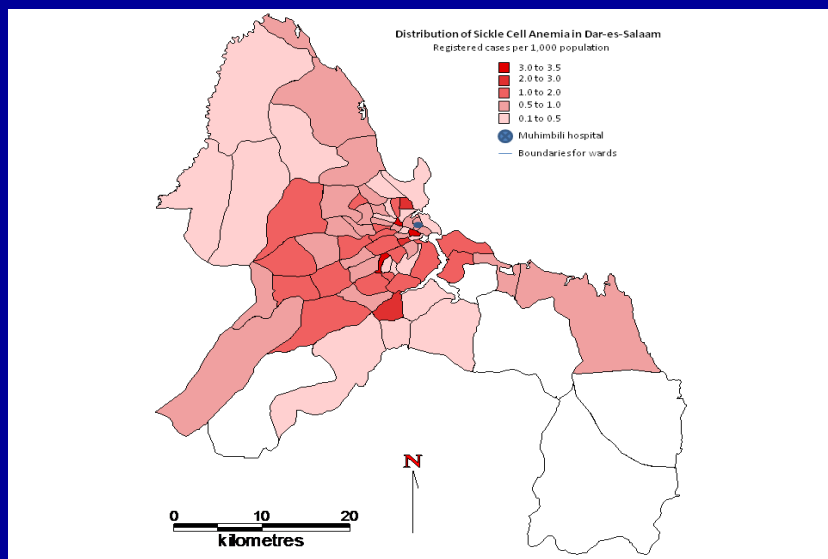
Genetic diversity



Environmental diversity

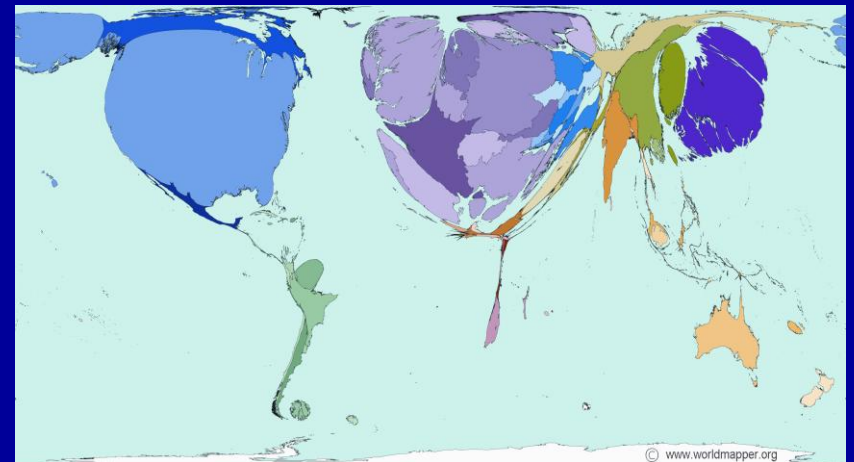
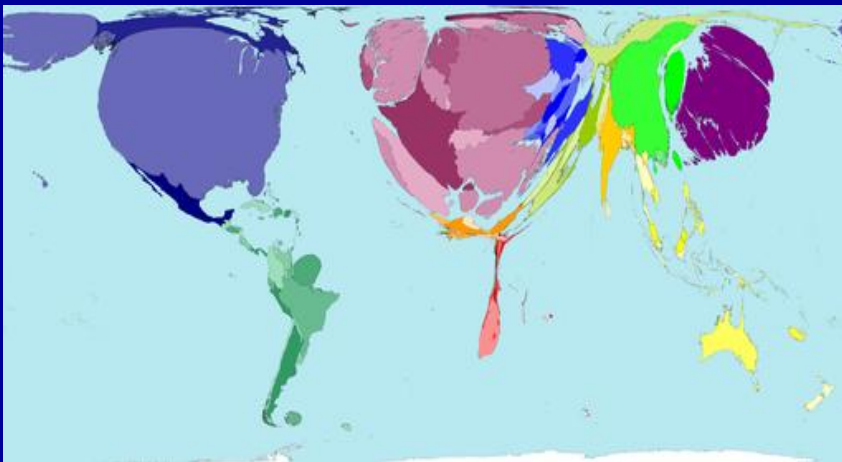
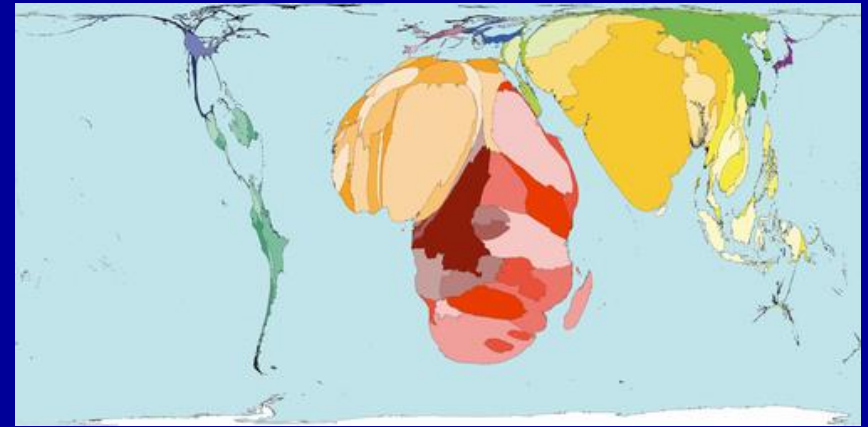
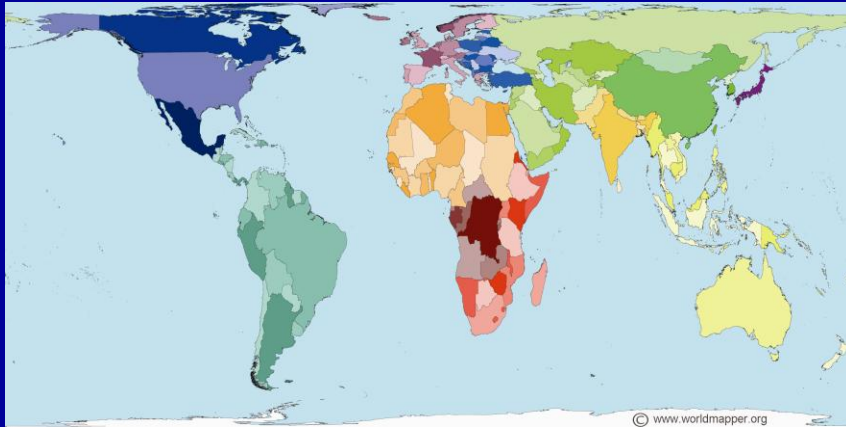


Population diversity

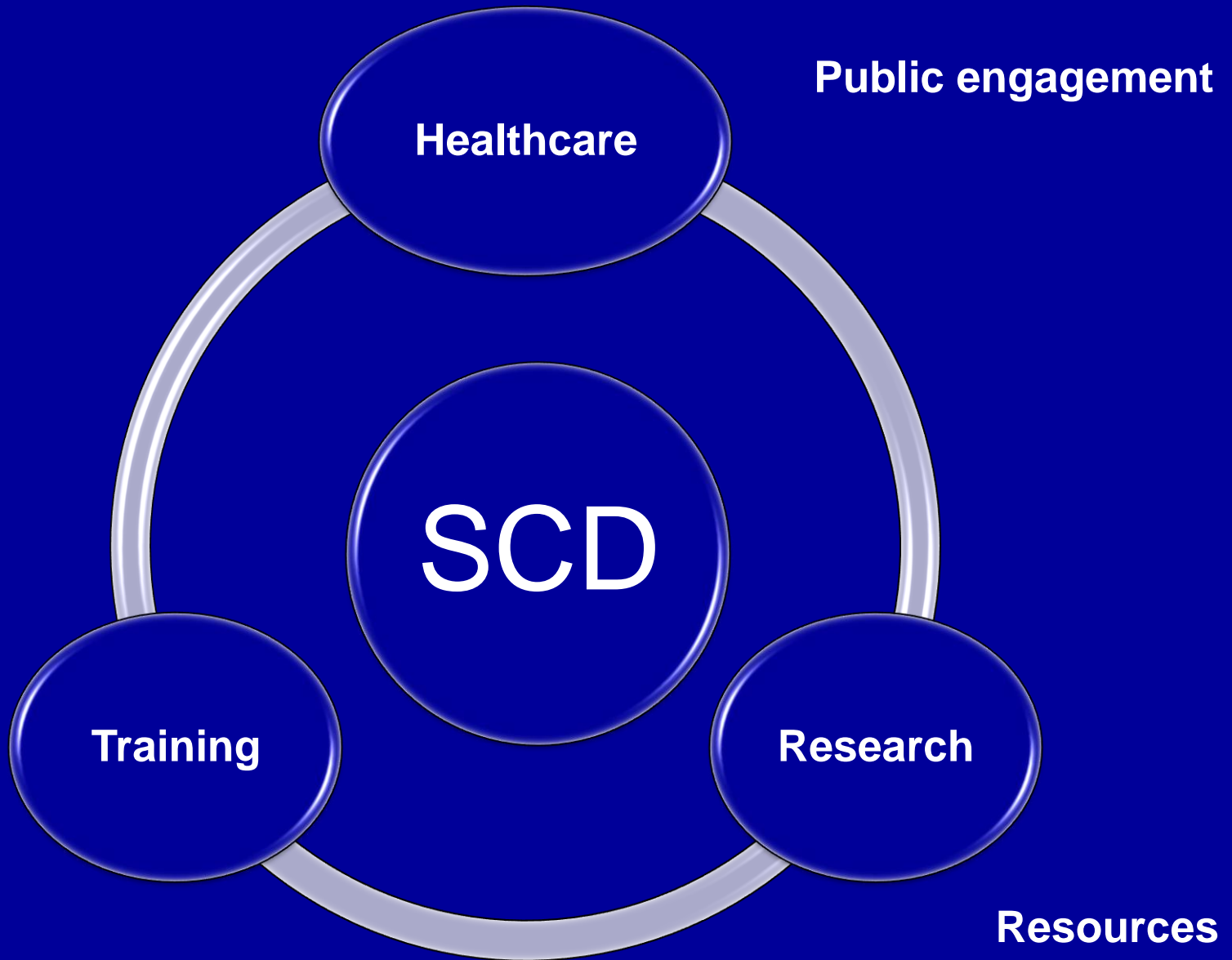


What are the challenges?

Distribution of Resources



The Strategy



Public engagement

Healthcare

SCD

Training

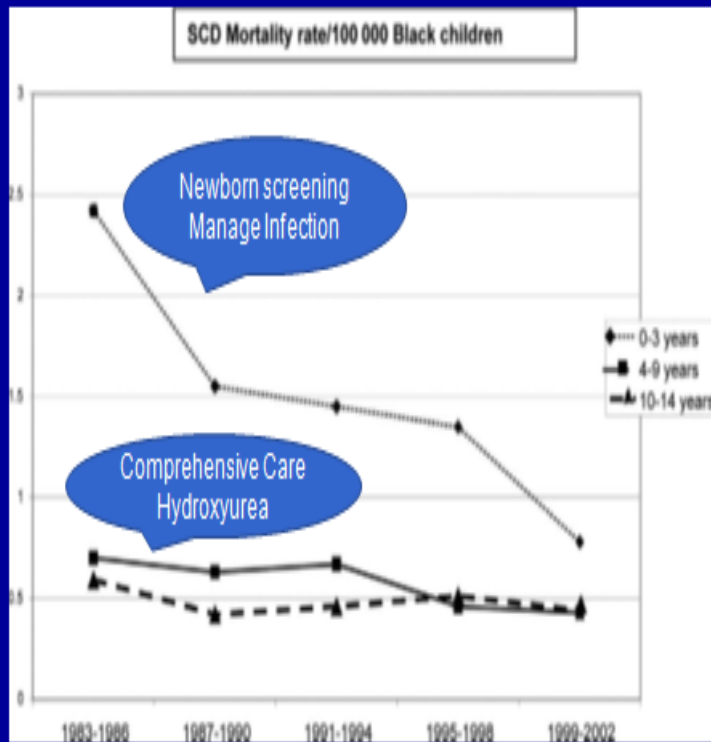
Research

Funding

Resources

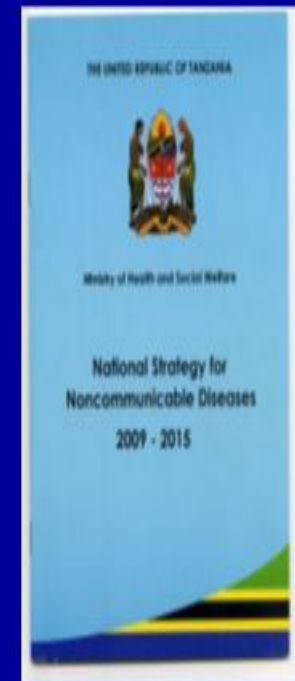
Healthcare and Policy

Healthcare - Interventions in SCD



Trend in Mortality in SCD in the USA

Healthcare - Policy

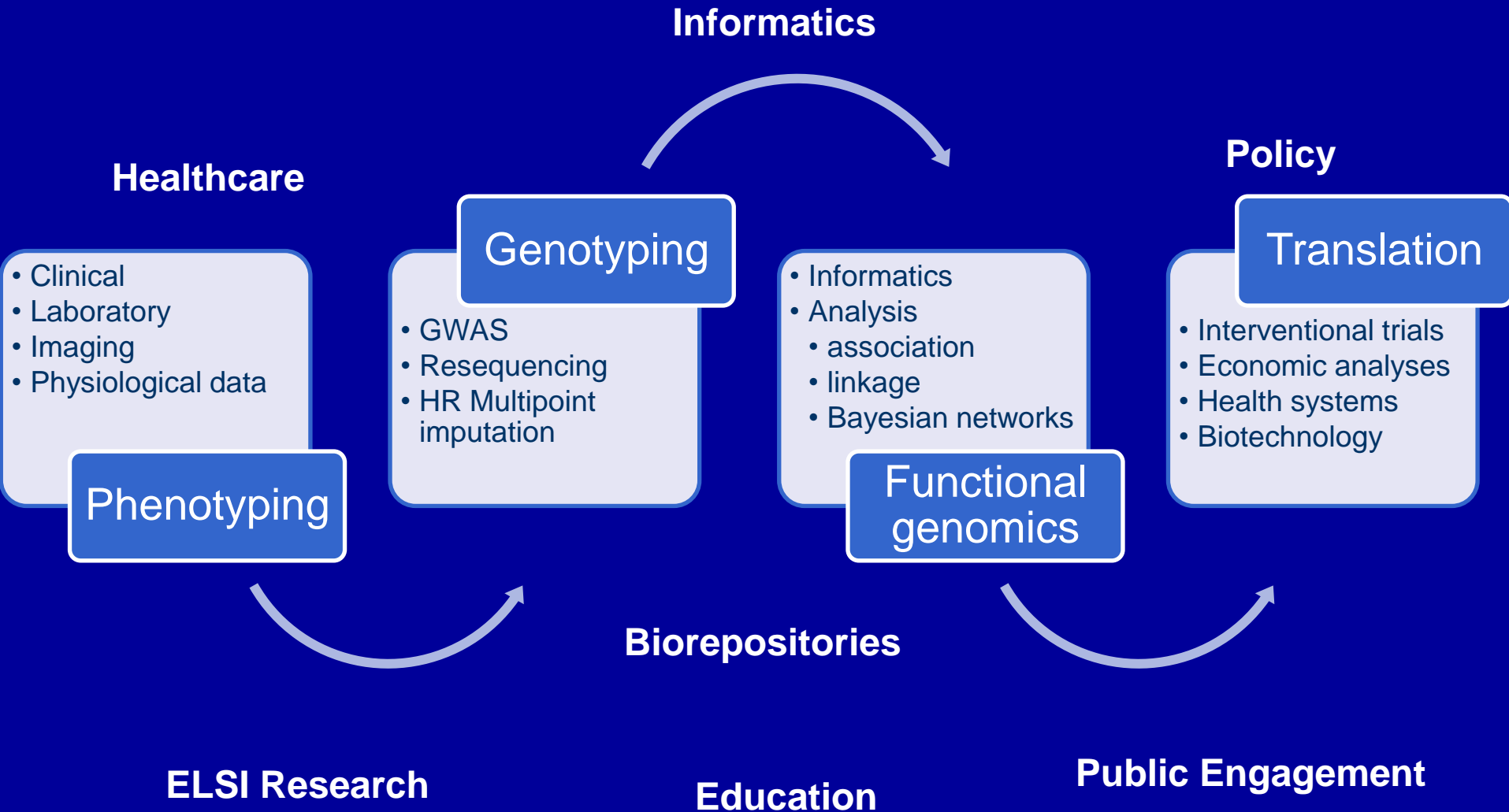


National Strategy for NCD - 2009

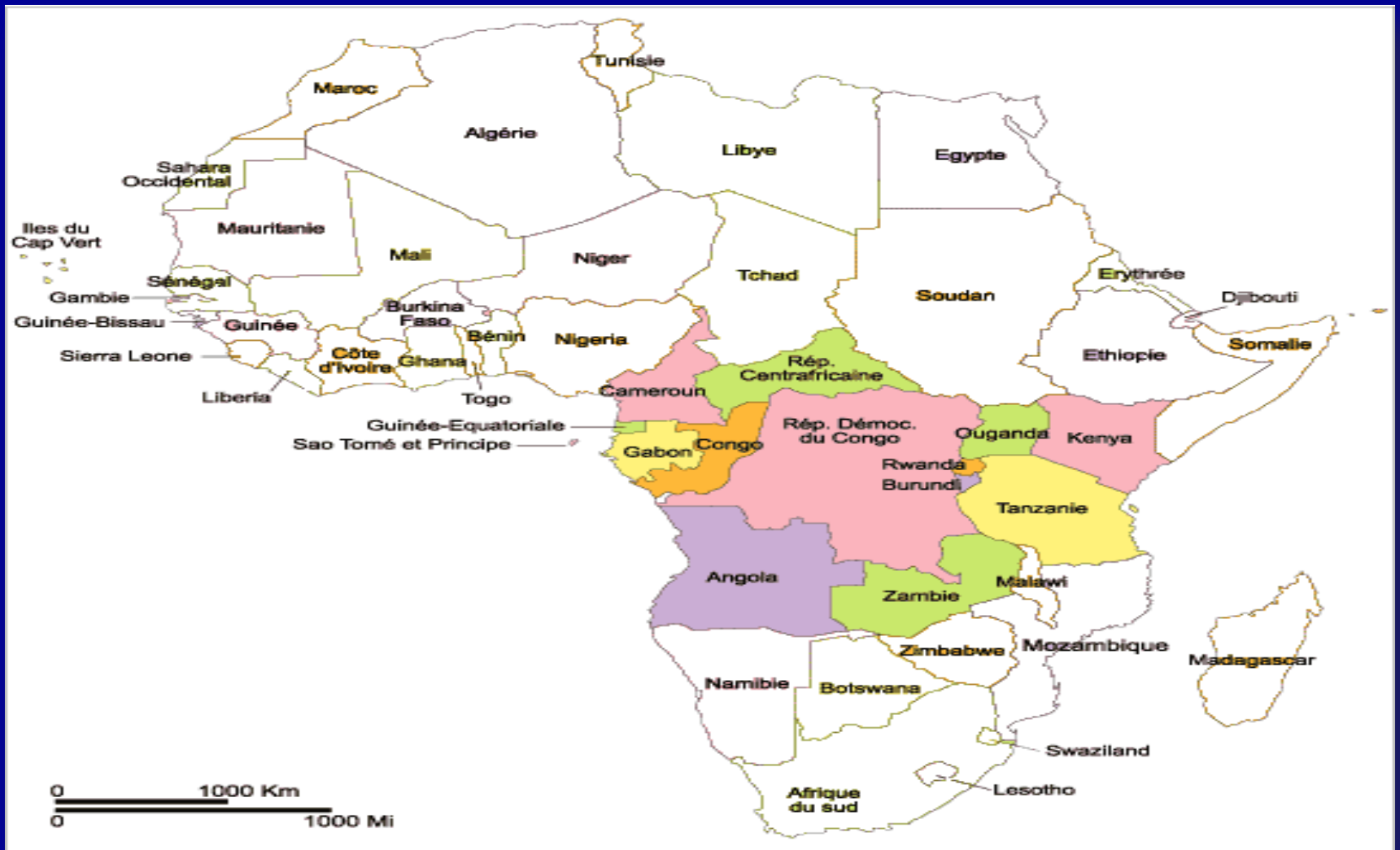
Training

- Strengthen training programmes in universities
 - Avoid short courses, workshops – parallel system
- All cadres of health care workers at different levels of health care
- Areas of training
 - Clinical – undergraduate, postgraduate and CME
 - Four Haematologists qualified in last 2 years.
 - Scientific Training
 - Support – Vocational and technical training

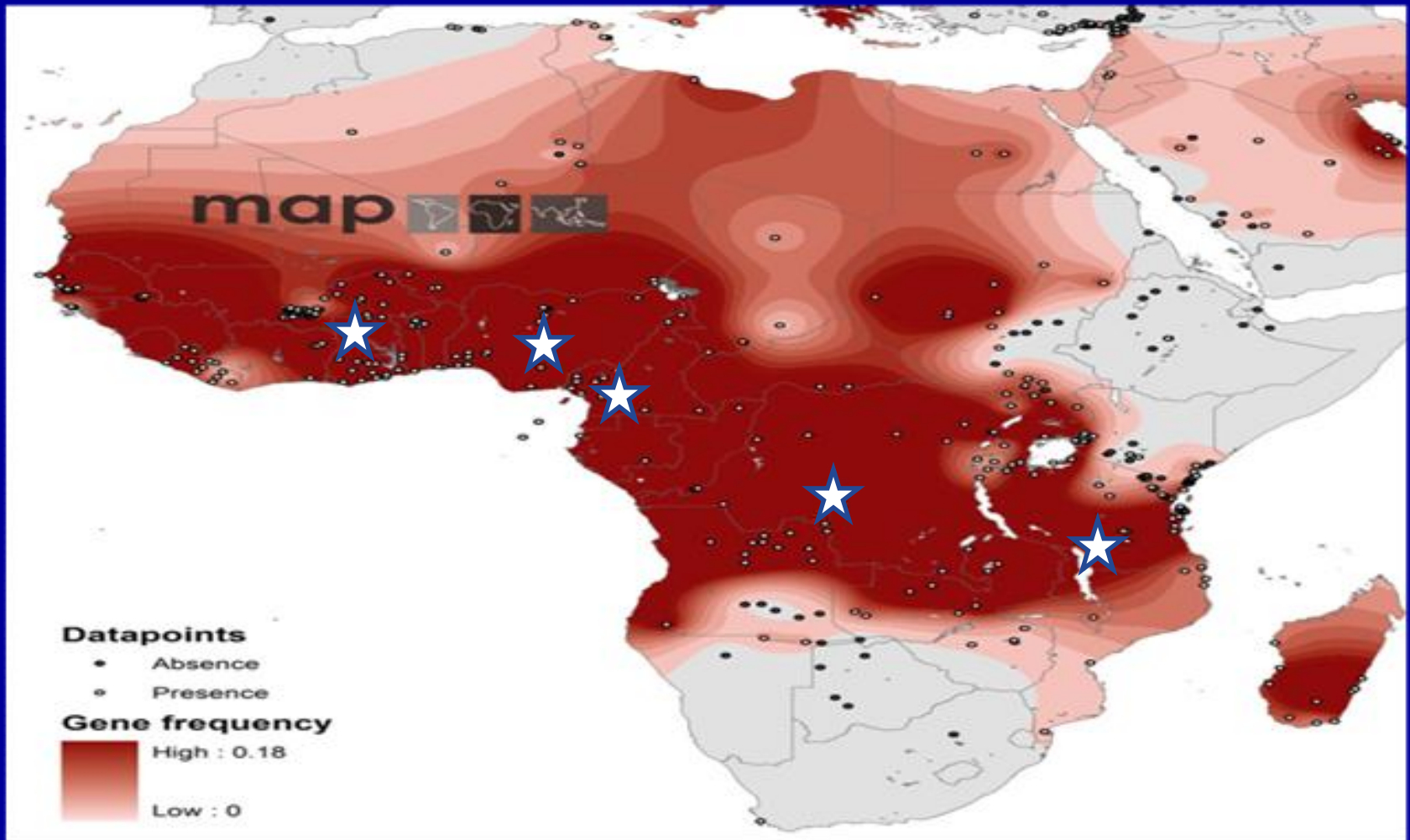
Strategic areas for genetic studies in Africa



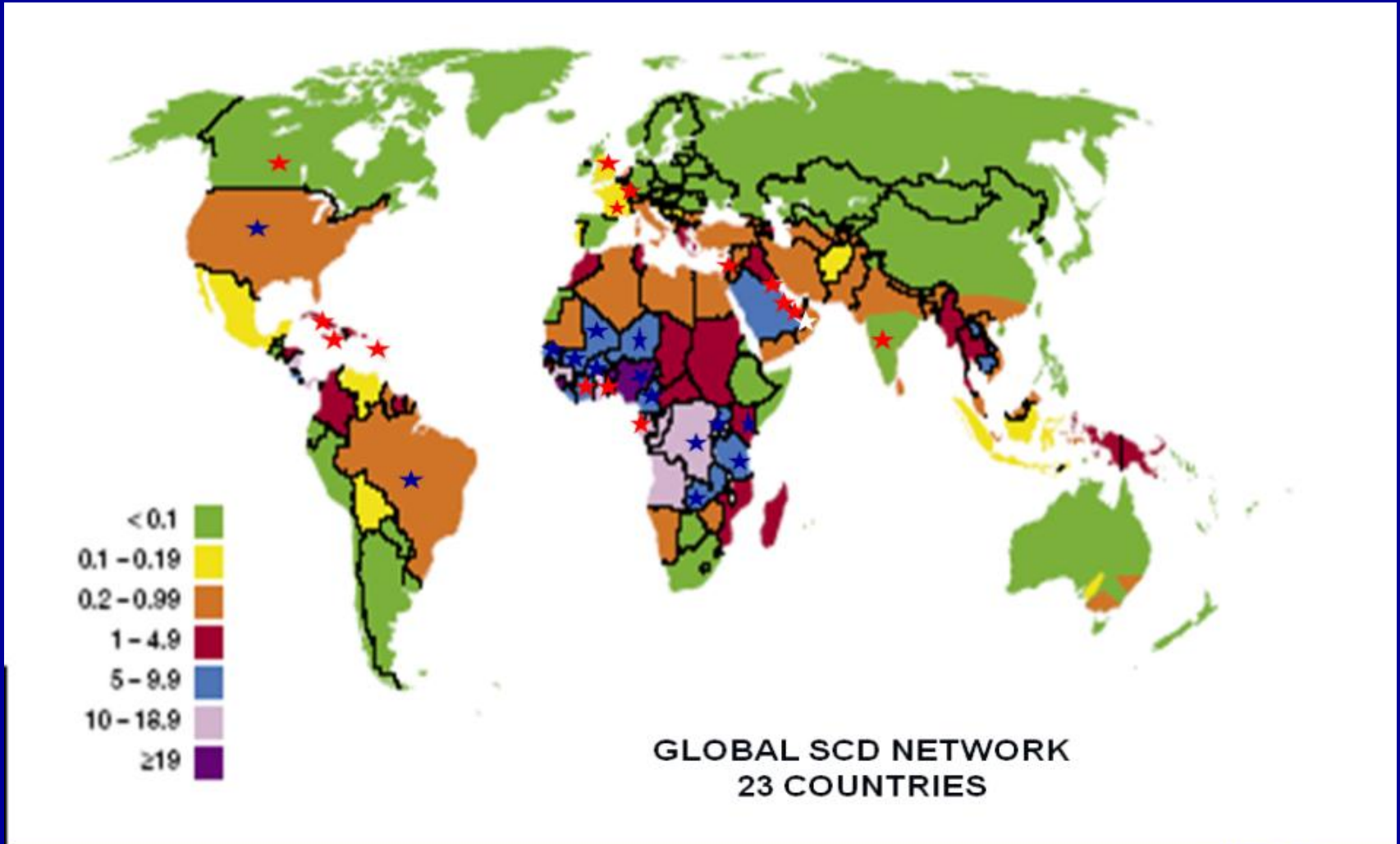
REDAC



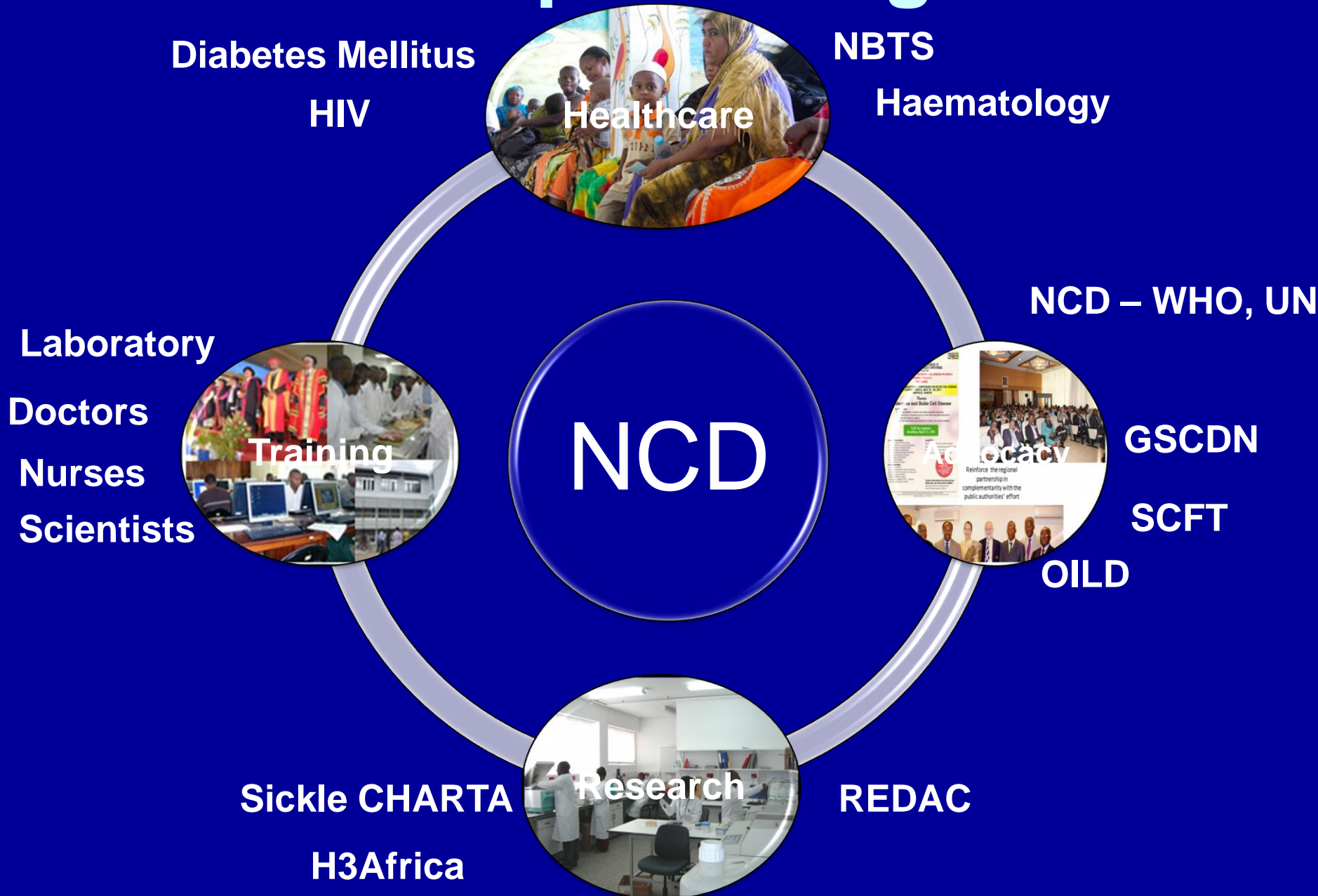
Sickle CHARTA



Global Sickle Cell Disease Network

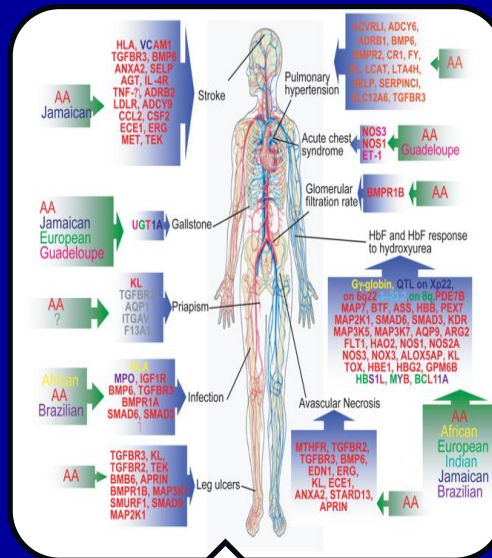


Partnerships & Integration



Genes – Disease - Health

Social and Cultural factors



Environmental factors

Acknowledgements

All patients and staff at Muhimbili

Haematology, Paediatrics & Medicine

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Collaborators

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- F Kirkham
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