Opportunities for Genomic Research in SCD

Contribution from Africa

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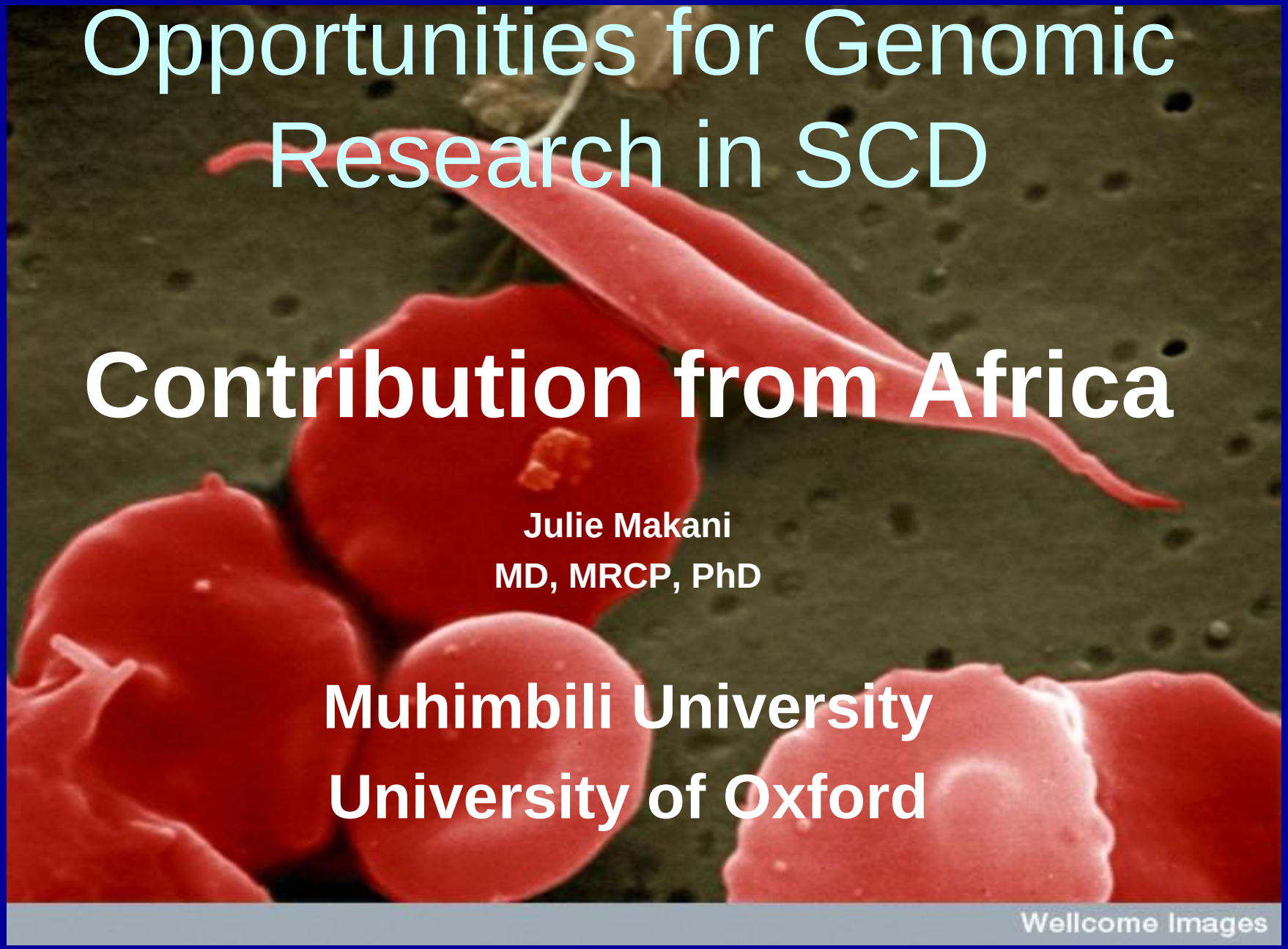
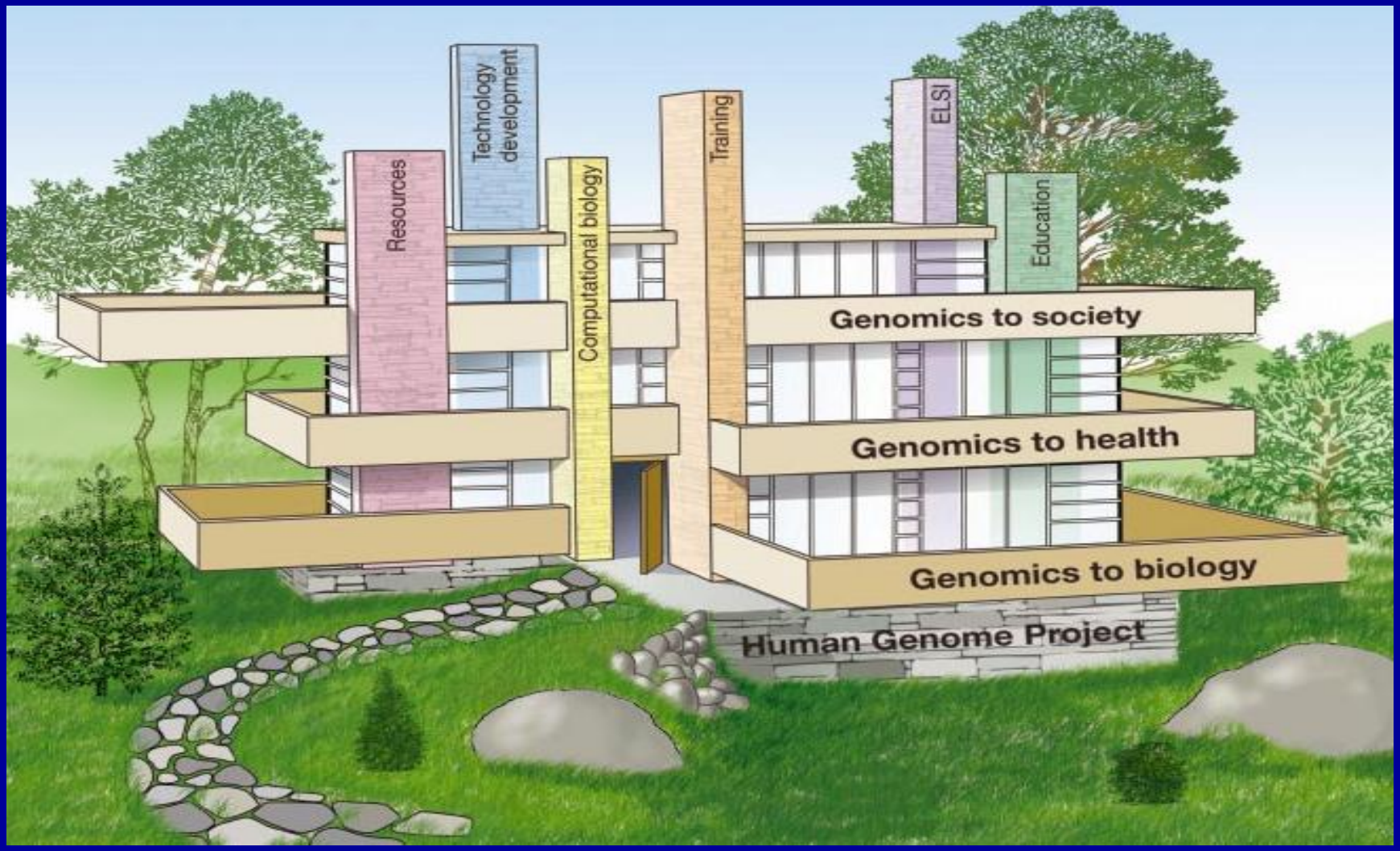


Outline

- 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’

Public Health

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

Clinical disease

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

Genetic basis

Gene – gene interactions

Gene - environment

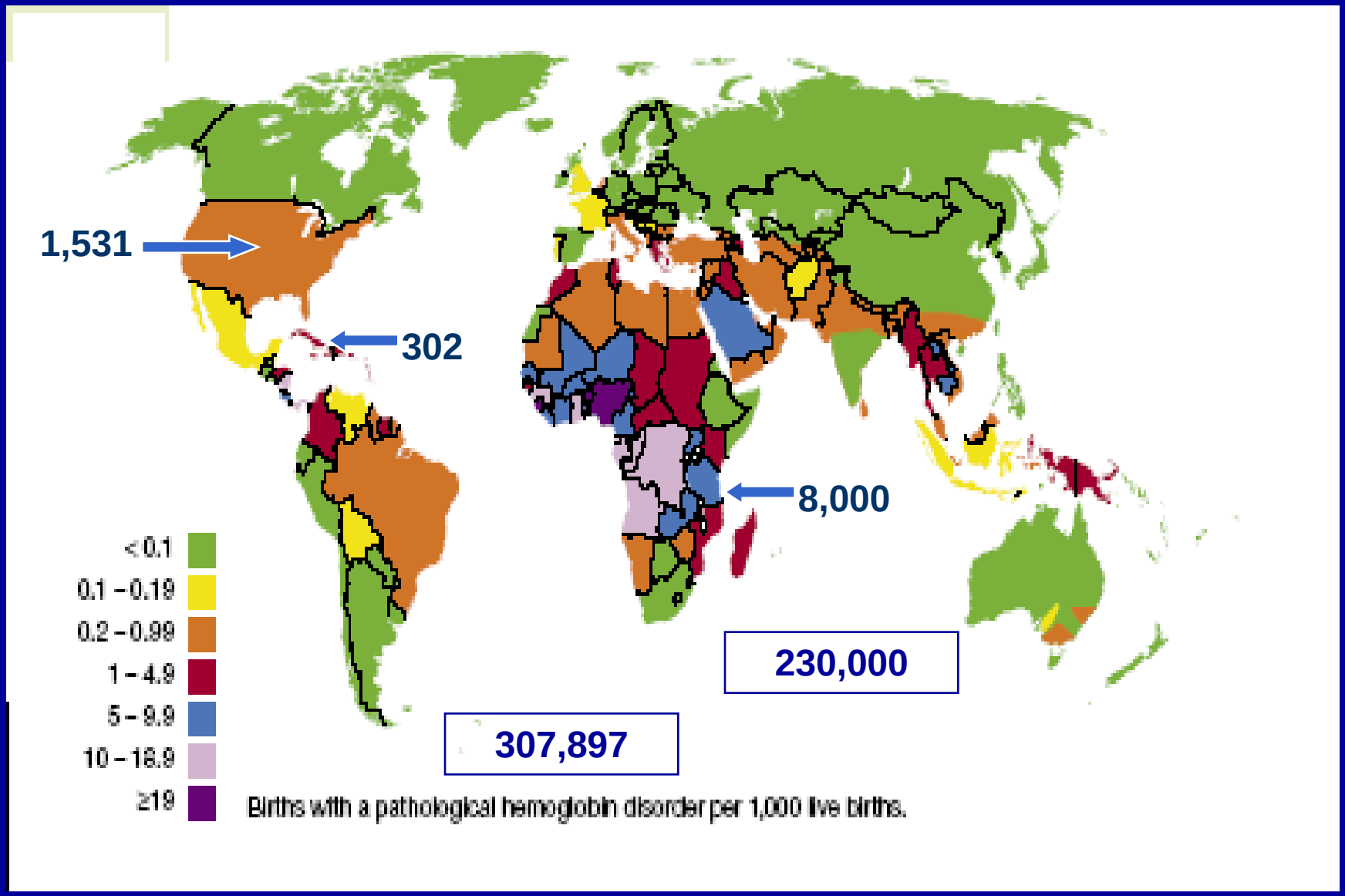
A ‘model disease’ for genetic studies

- 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.

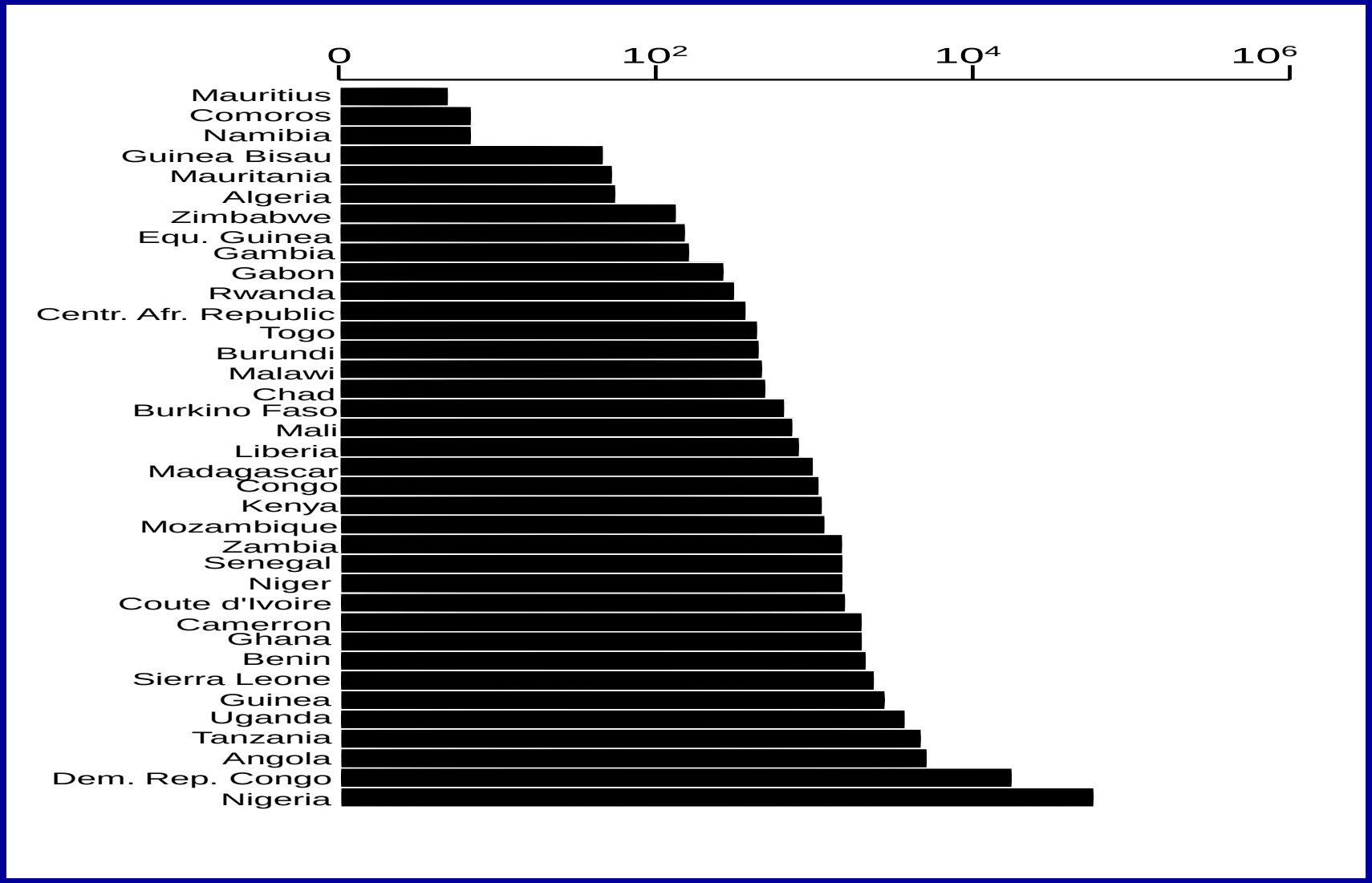
State of SCD in Africa

Global Burden of SCD



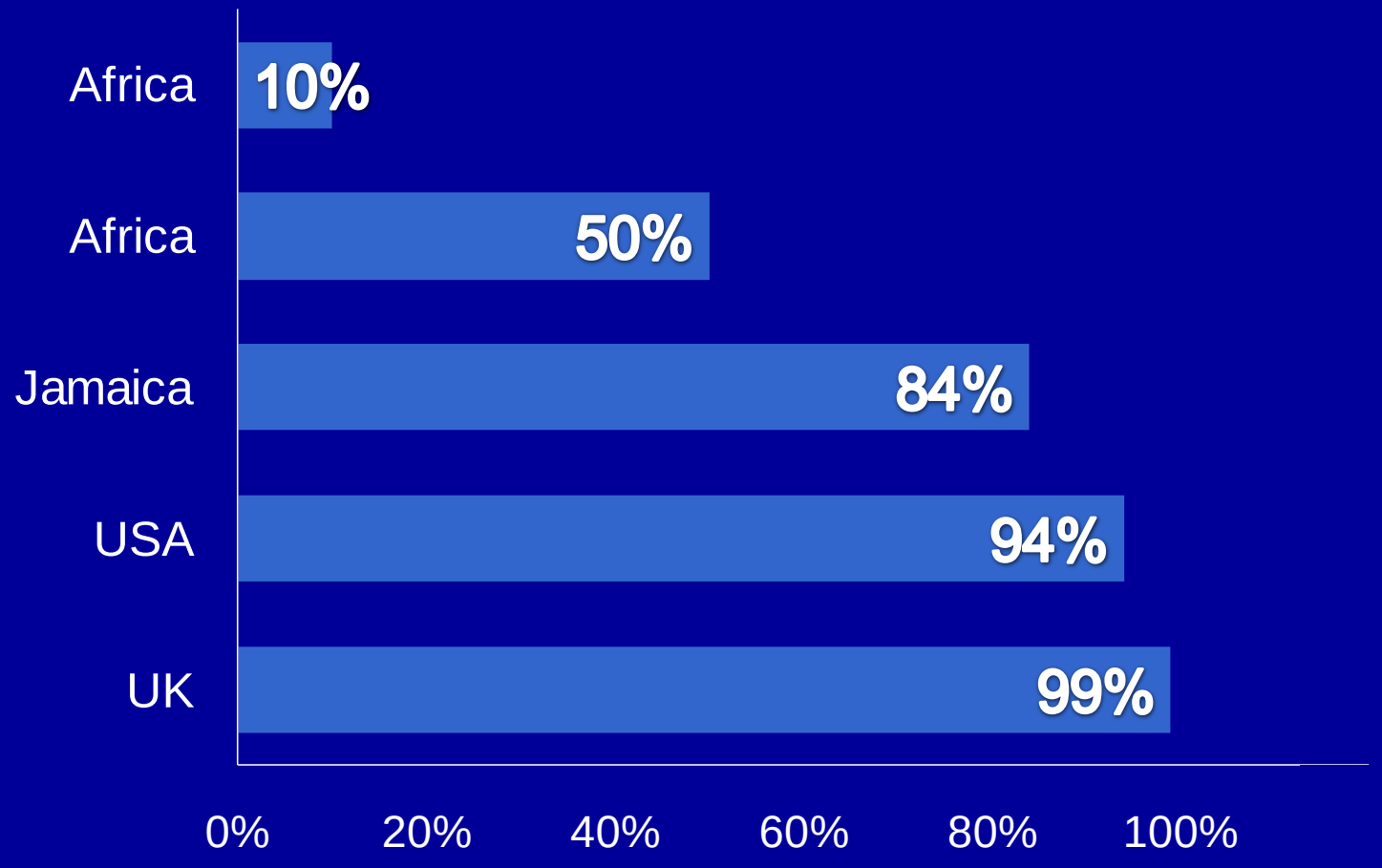
WHO, 1996

Birth estimates



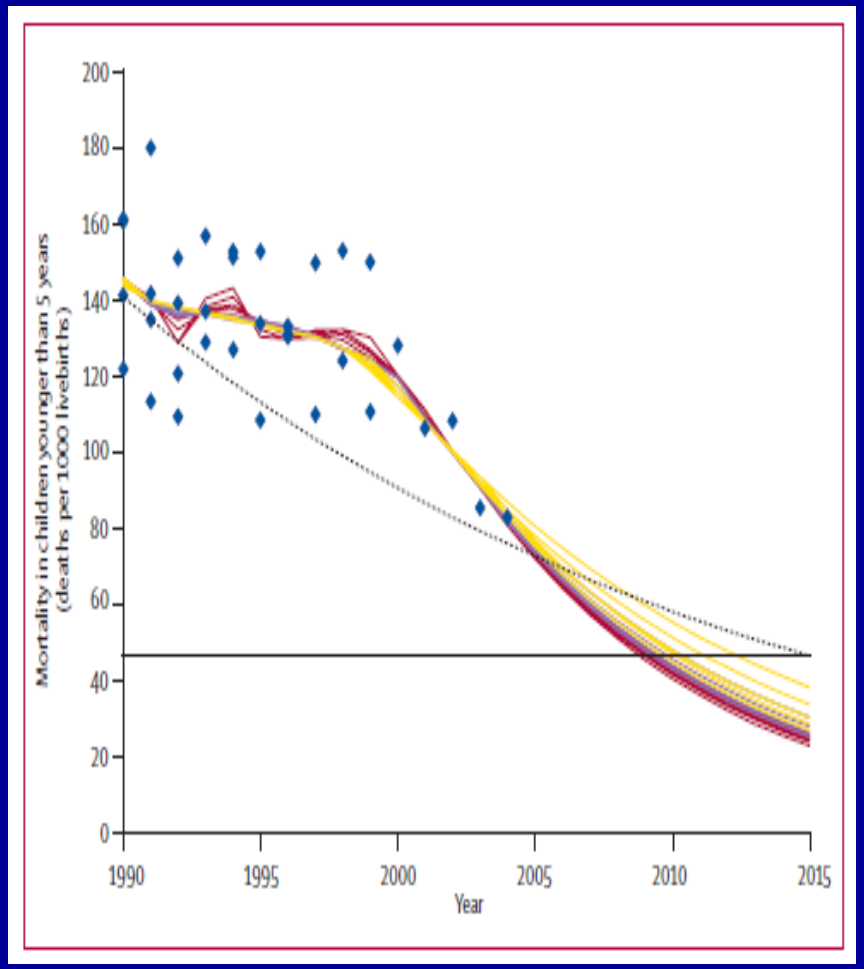
S OA and K OF in Pace, 2007; Modell, 2008

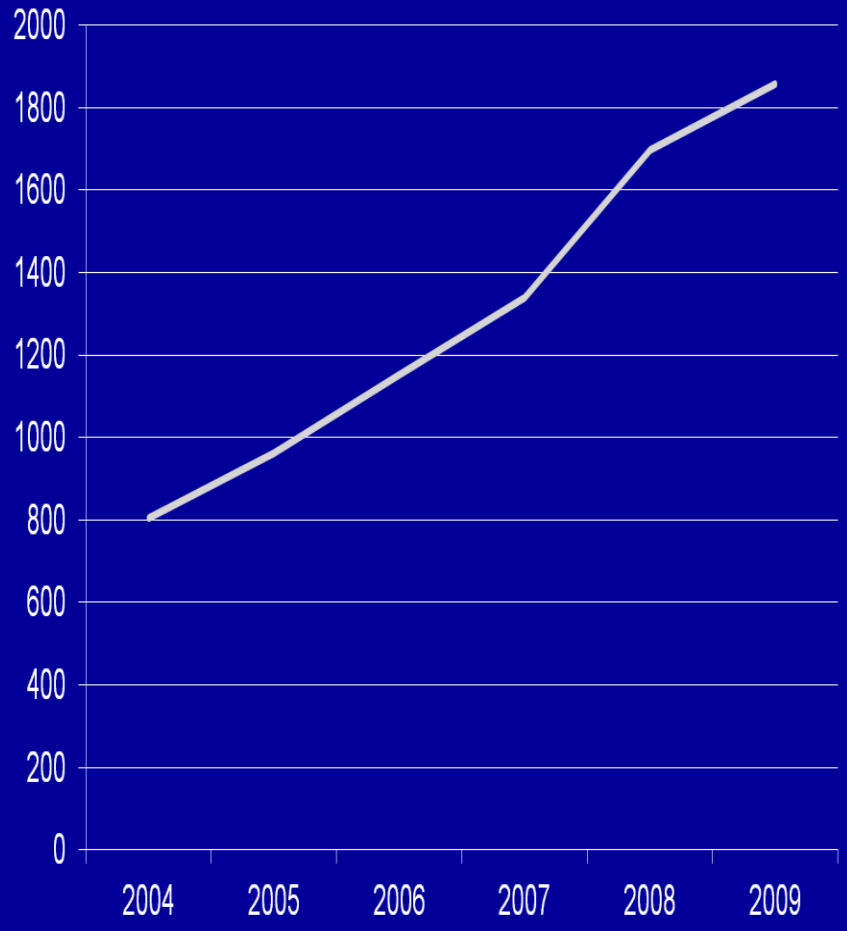
Childhood Survival for SCD



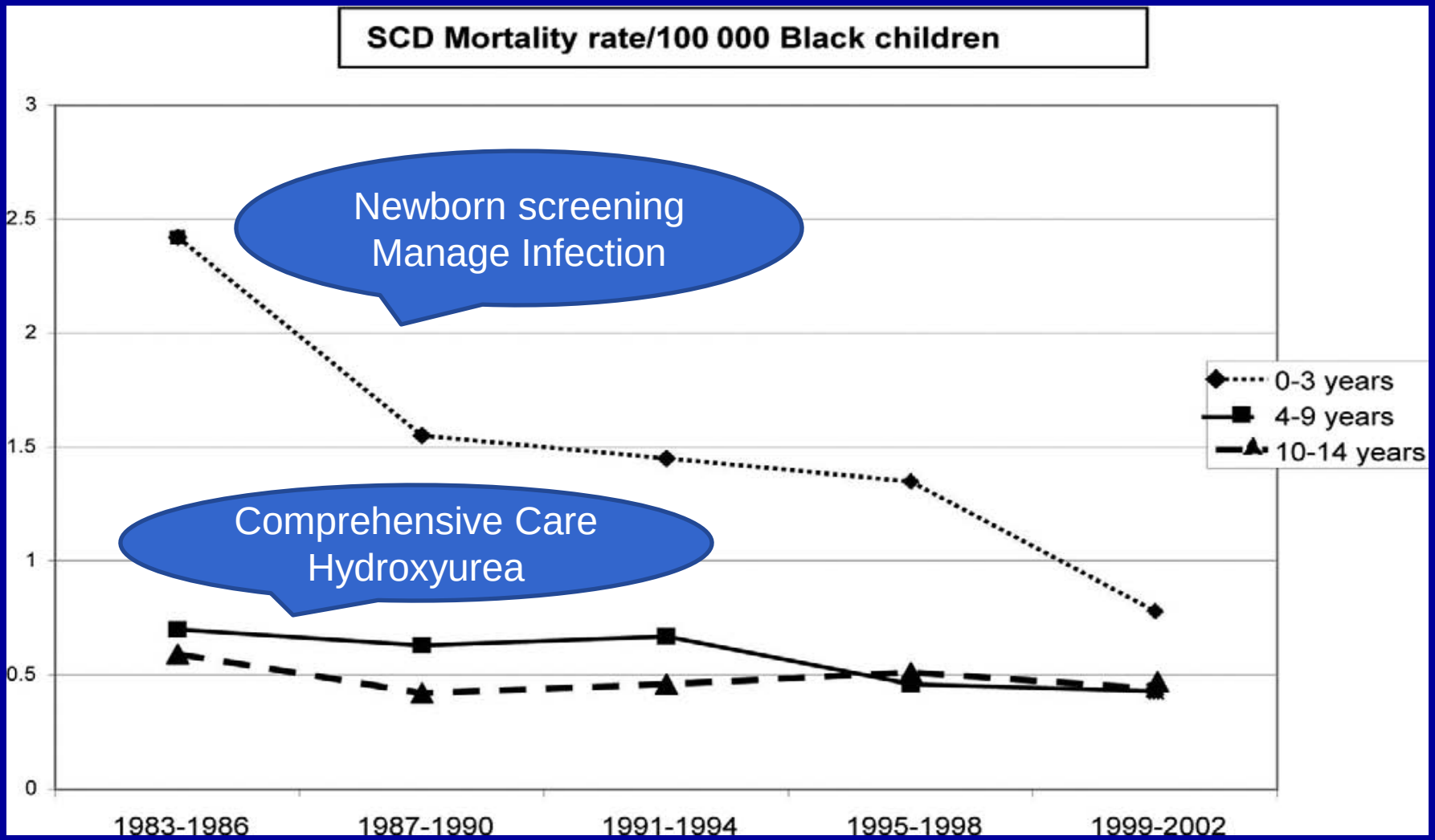
Fleming, 1979; Weatherall, 2006; Wierenga, 2001; Quinn 2004; Telfer, 2007

Demographic Transition



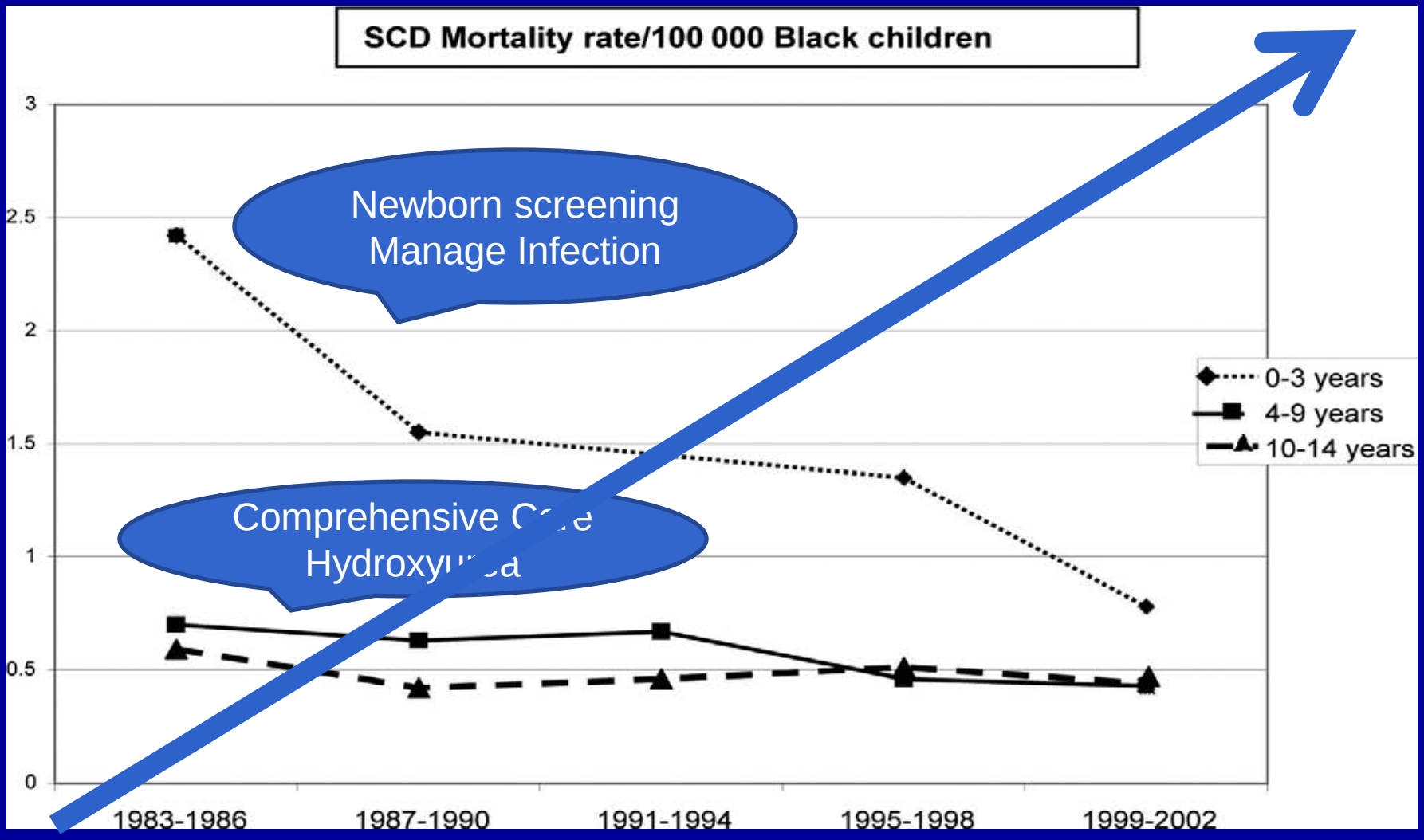


Limited Interventions in SCD



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

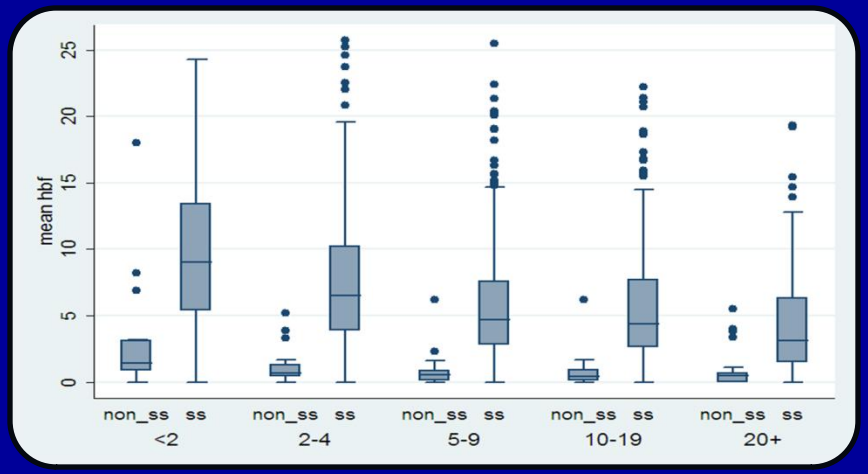
Bioinformatics

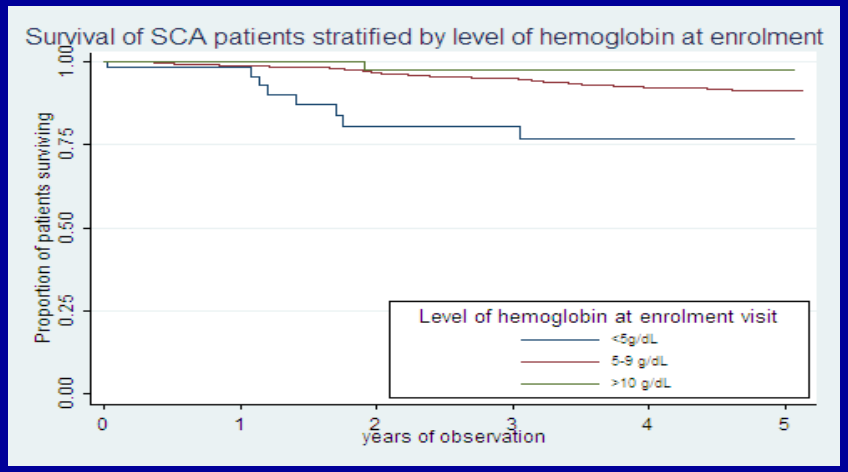
Health benefits

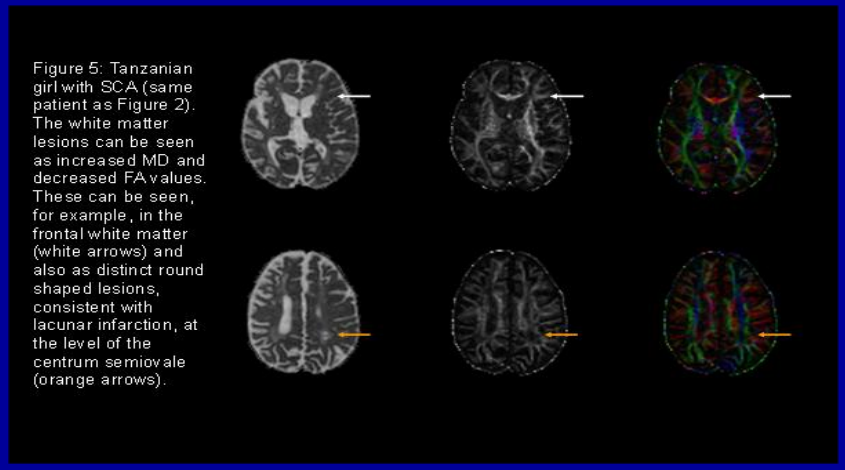
Clinical Studies

Statistical and Epidemiological Analysis

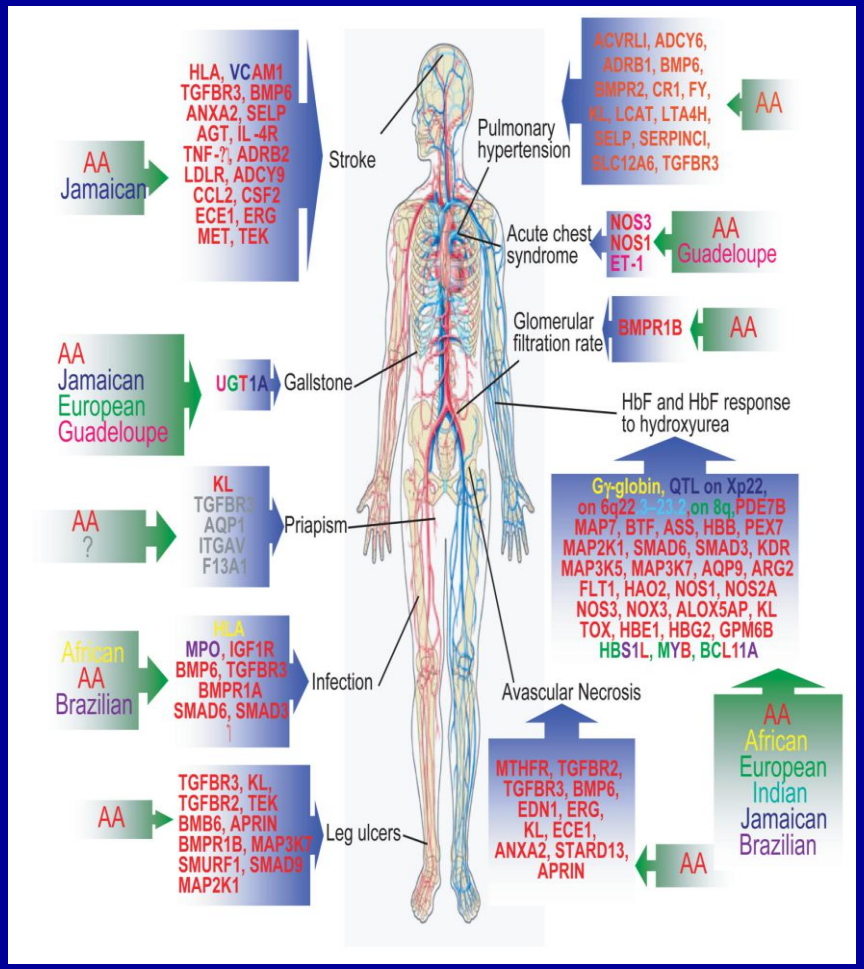
Phenotypic Diversity
HbF, Hb, Stroke, Haemolysis

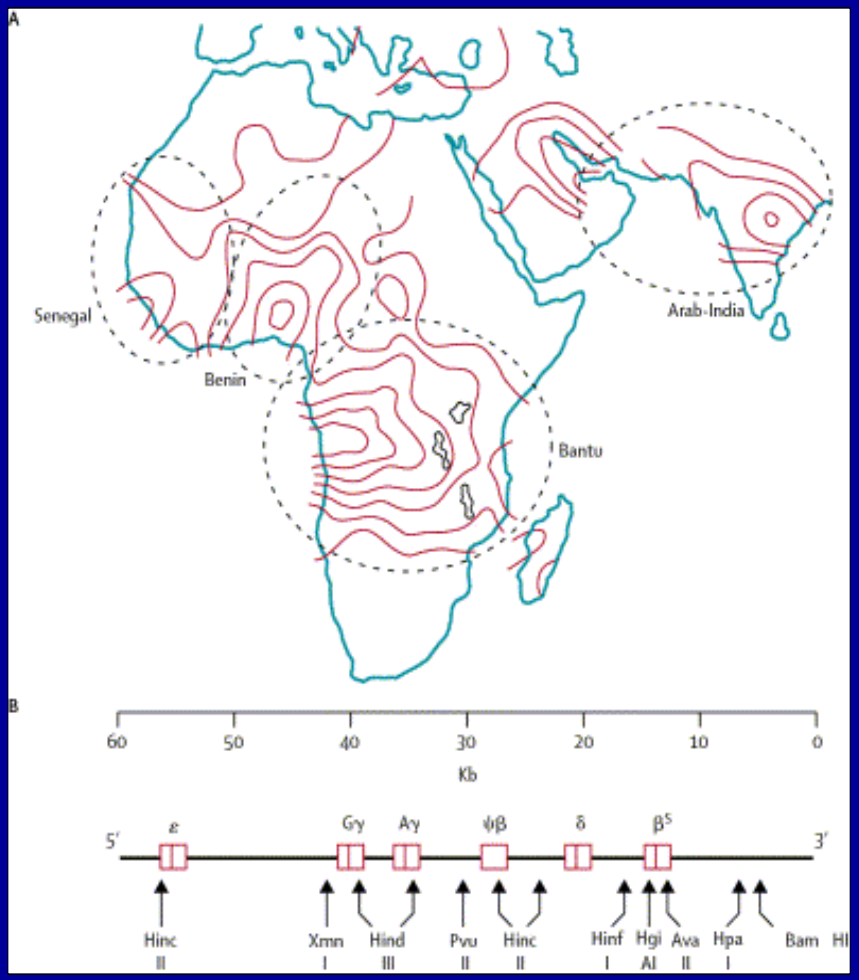






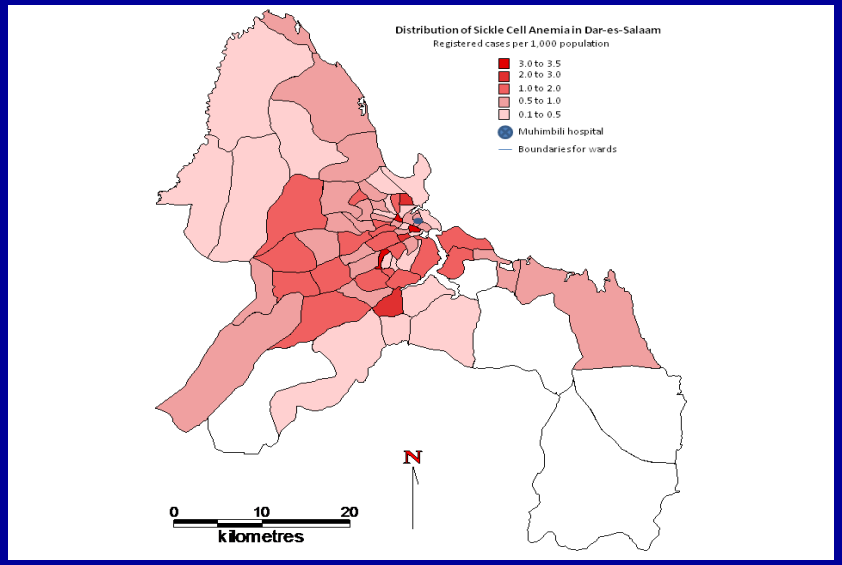
Genetic diversity

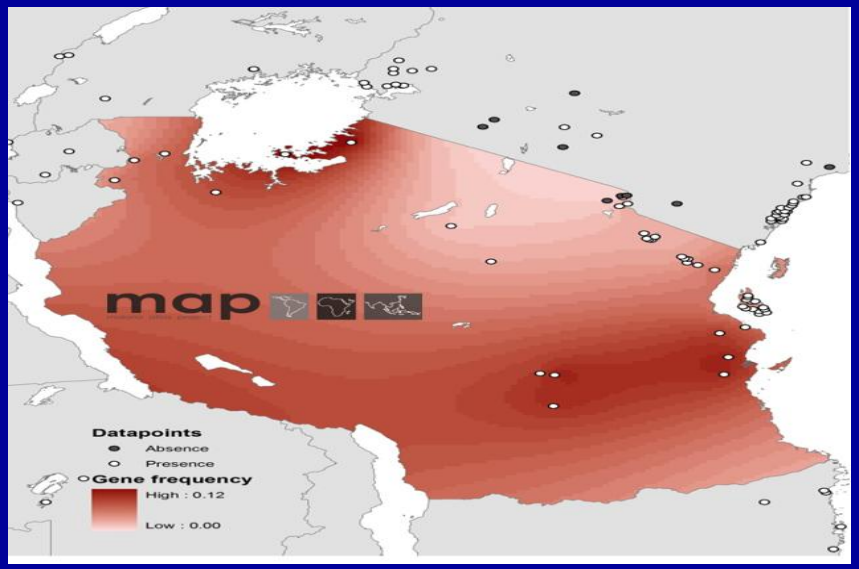


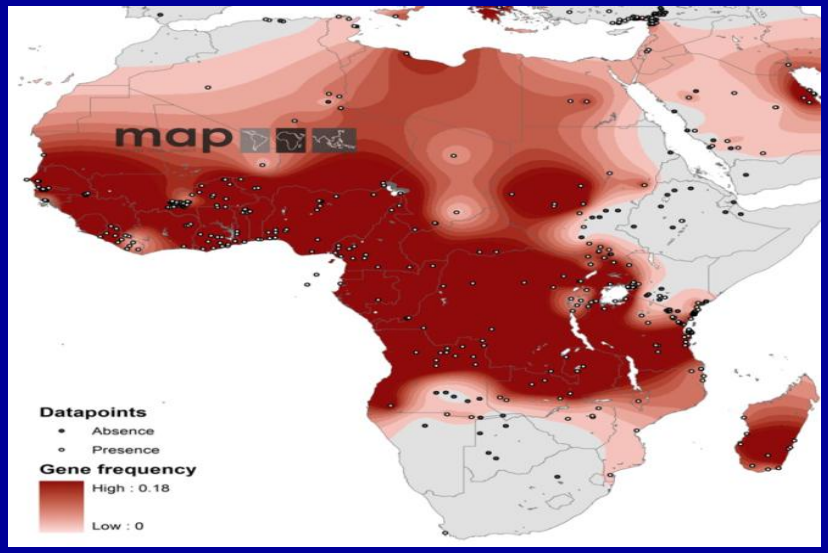


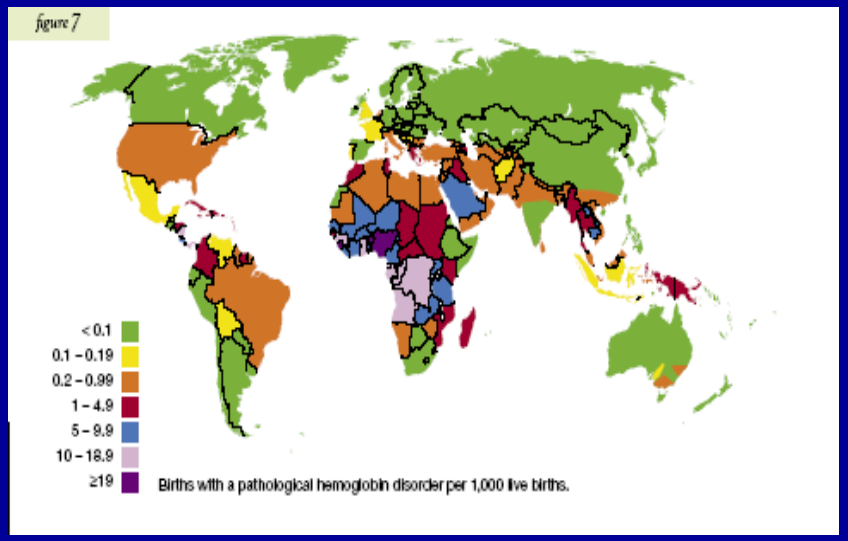
Environmental diversity

Population diversity



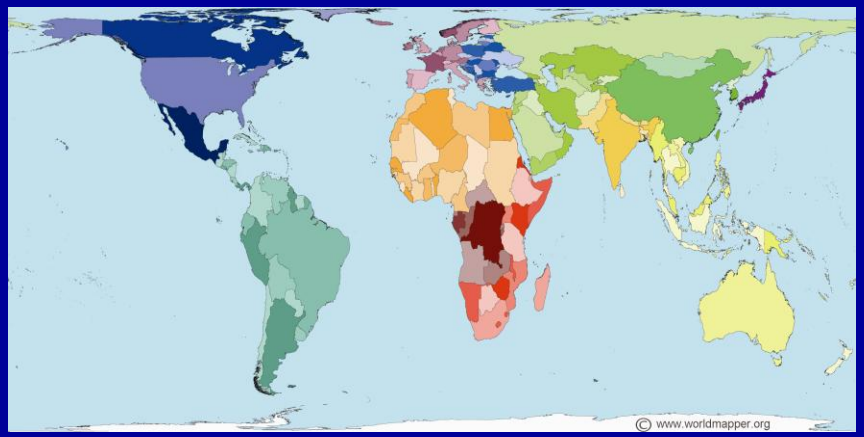


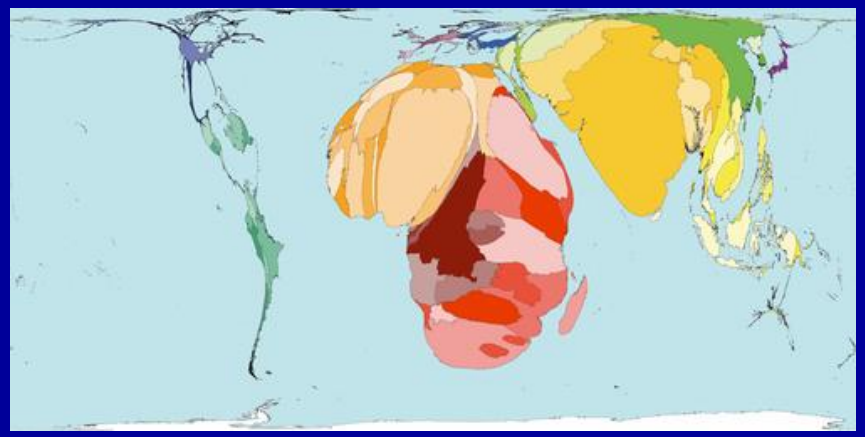


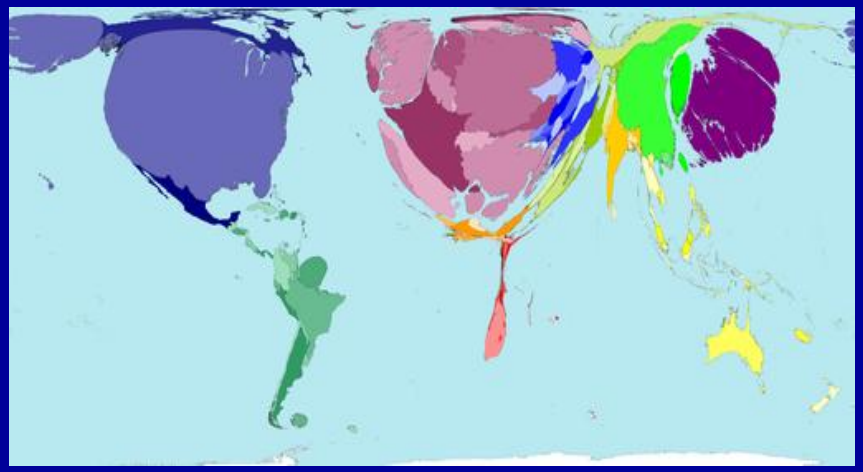


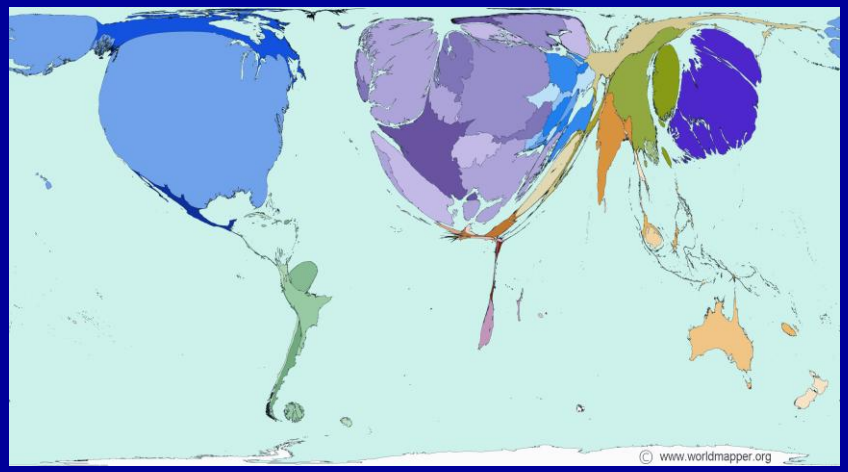
What are the challenges?

Distribution of Resources

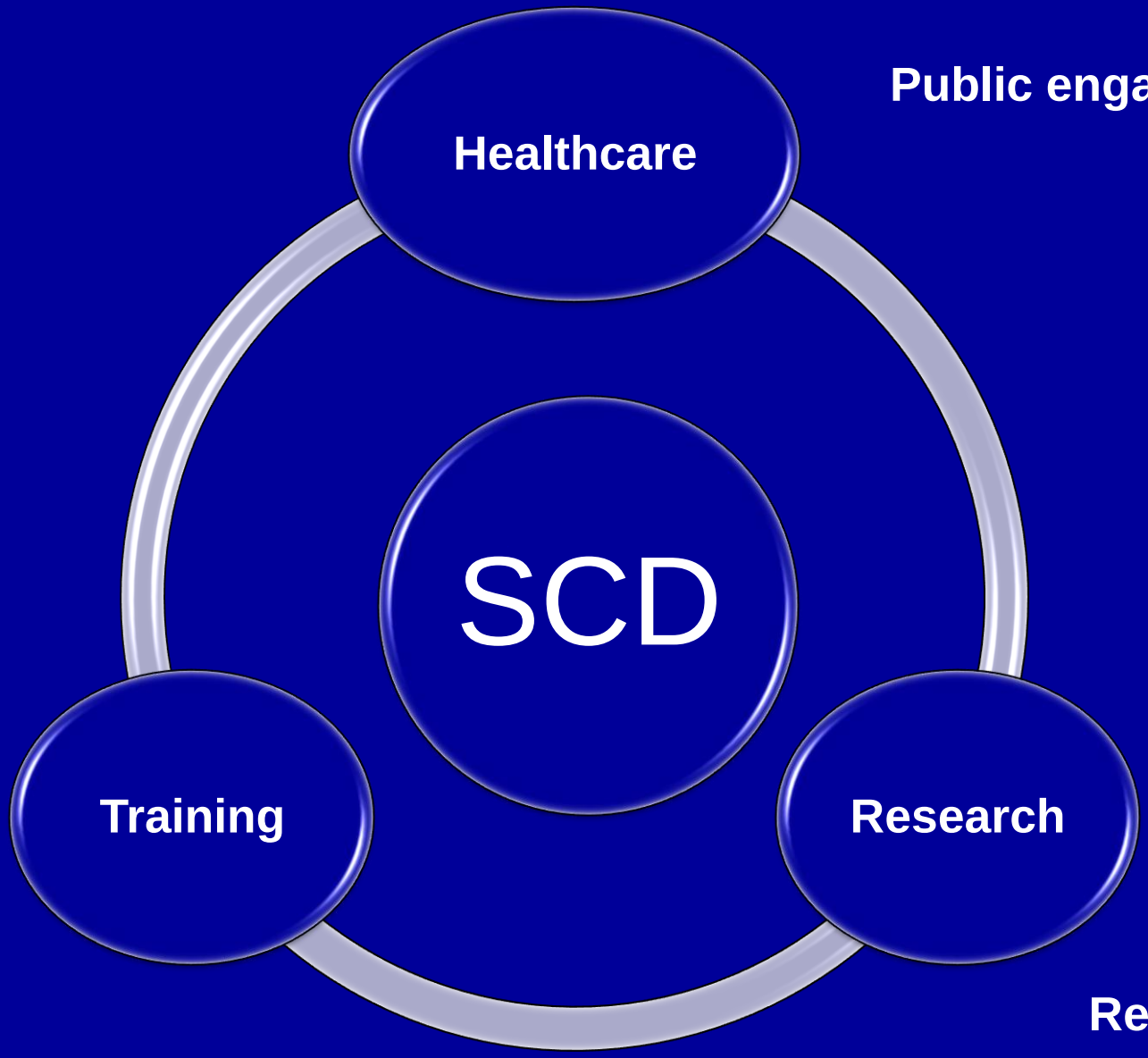








The Strategy



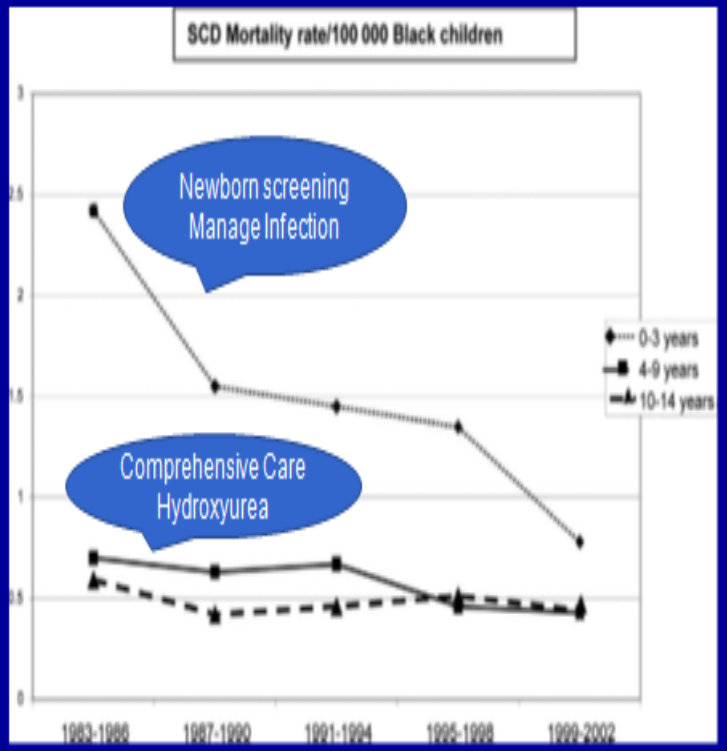
Public engagement

Funding

Resources

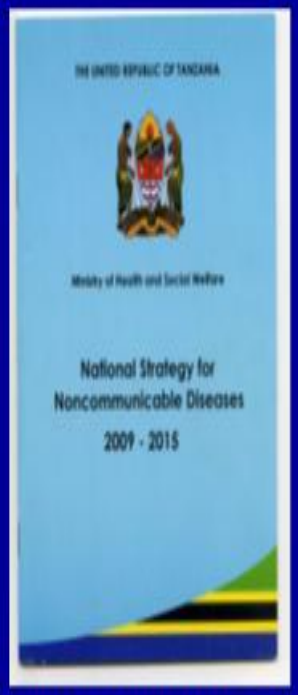
Healthcare and Policy

Healthcare - Interventions in SCD



Trend in Mortality in SCD in the USA

Healthcare - Policy

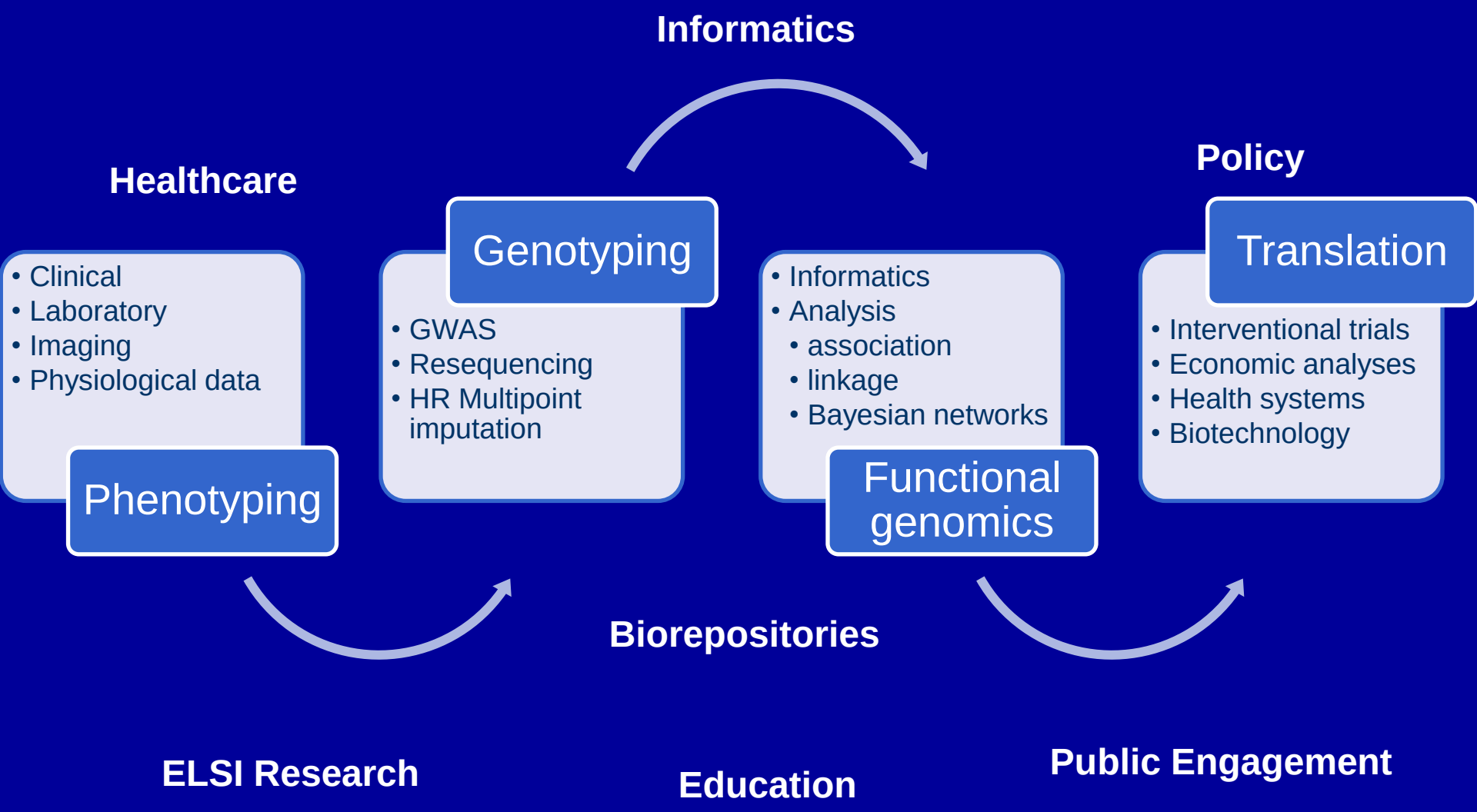


National Strategy for NCD - 2009

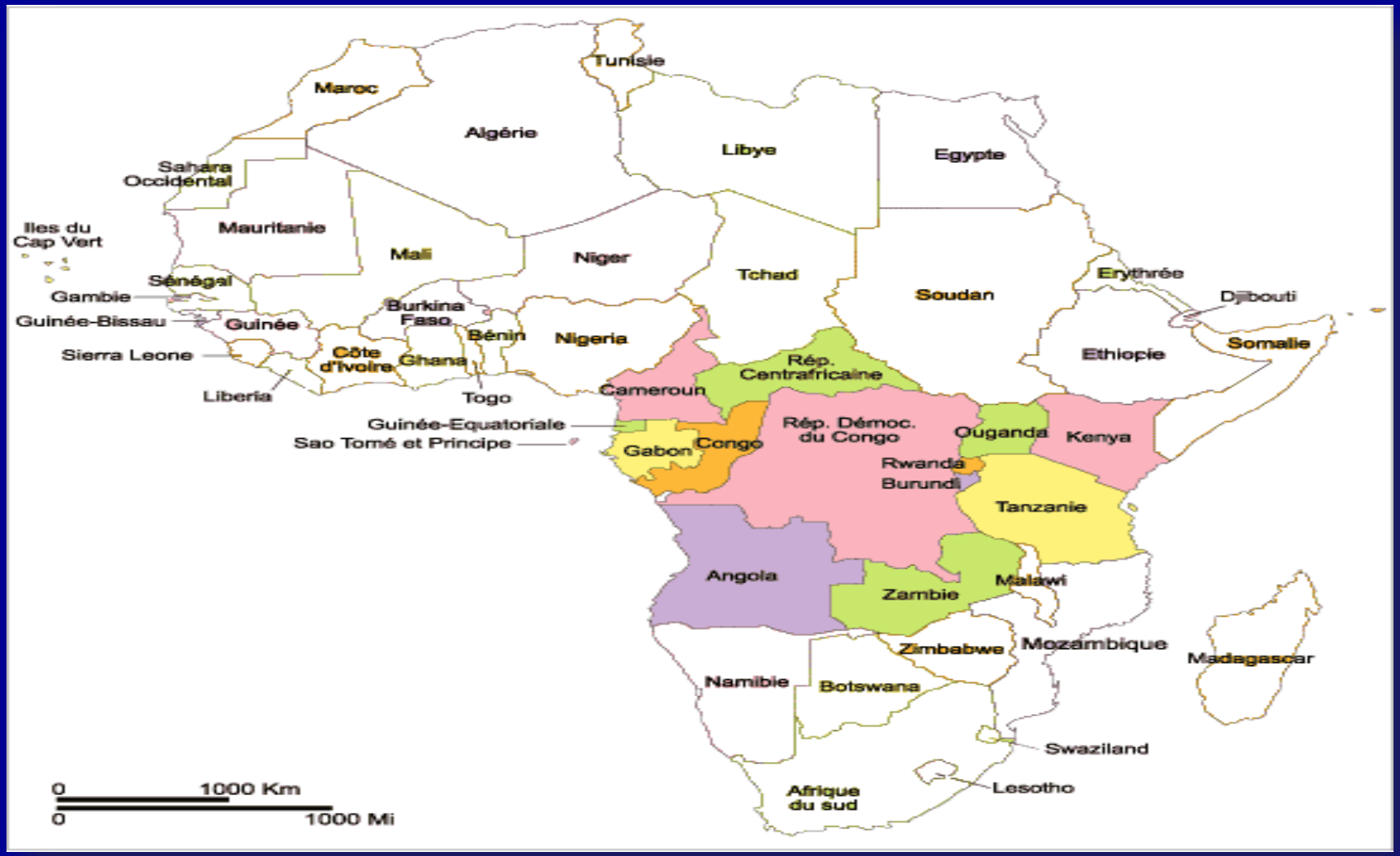
Training

- 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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Egypte
Sahara Occidental
Iles du Cap Vert
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Mali
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Djibouti
Sénégal
Gambie
Guinée-Bissau
Guinée
Burkina Faso
Bénin
Nigeria
Sierra Leone
Côte d'Ivoire
Ghana
Ethiopie
Somalie
Liberia
Togo
Cameroun
Rép. Centrafricaine
Guinée-Equatoriale
Sao Tomé et Principe
Gabon
Congo
Rép. Démoc. du Congo
Ouganda
Kenya
Rwanda
Burundi
Tanzanie
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Zimbabwe
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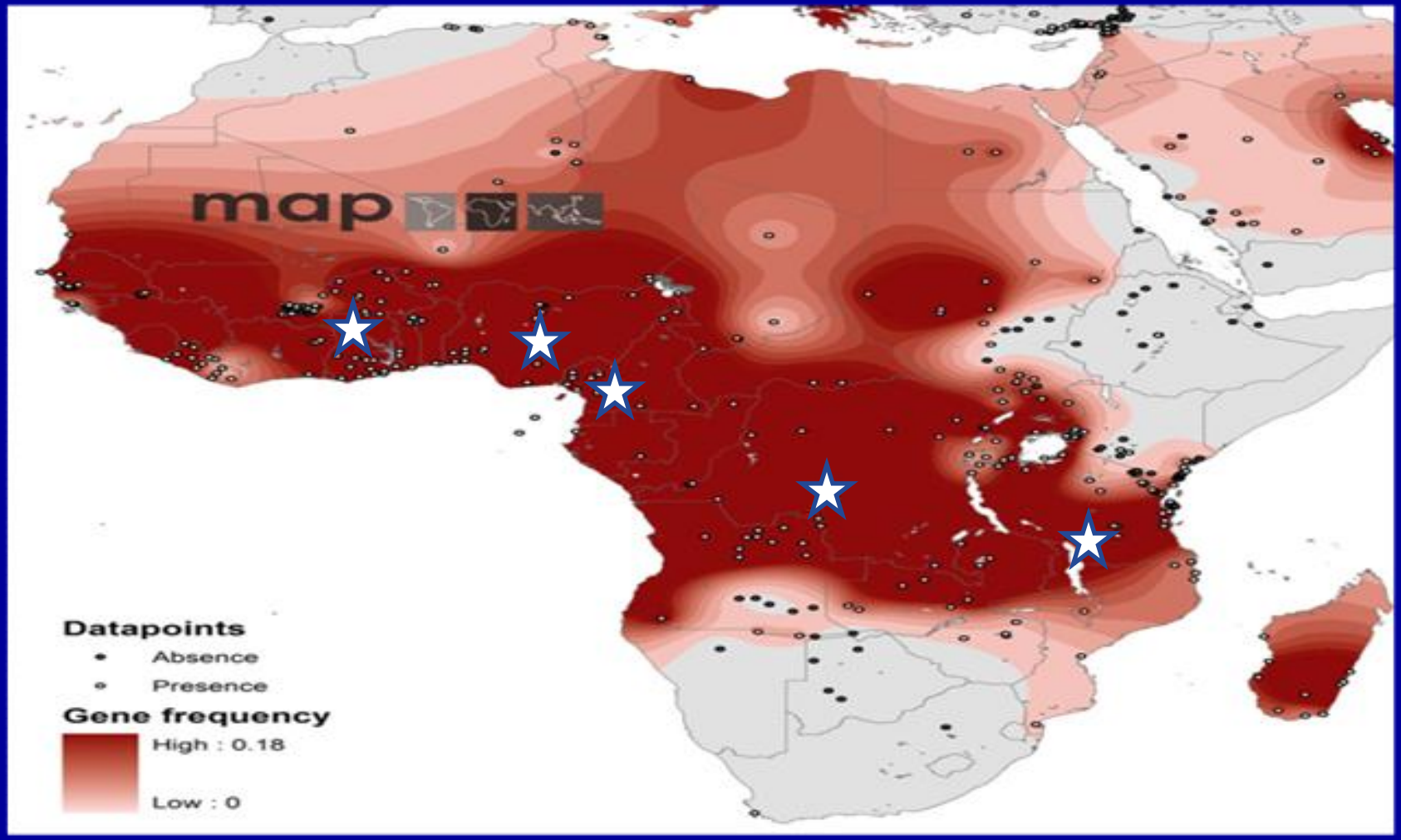
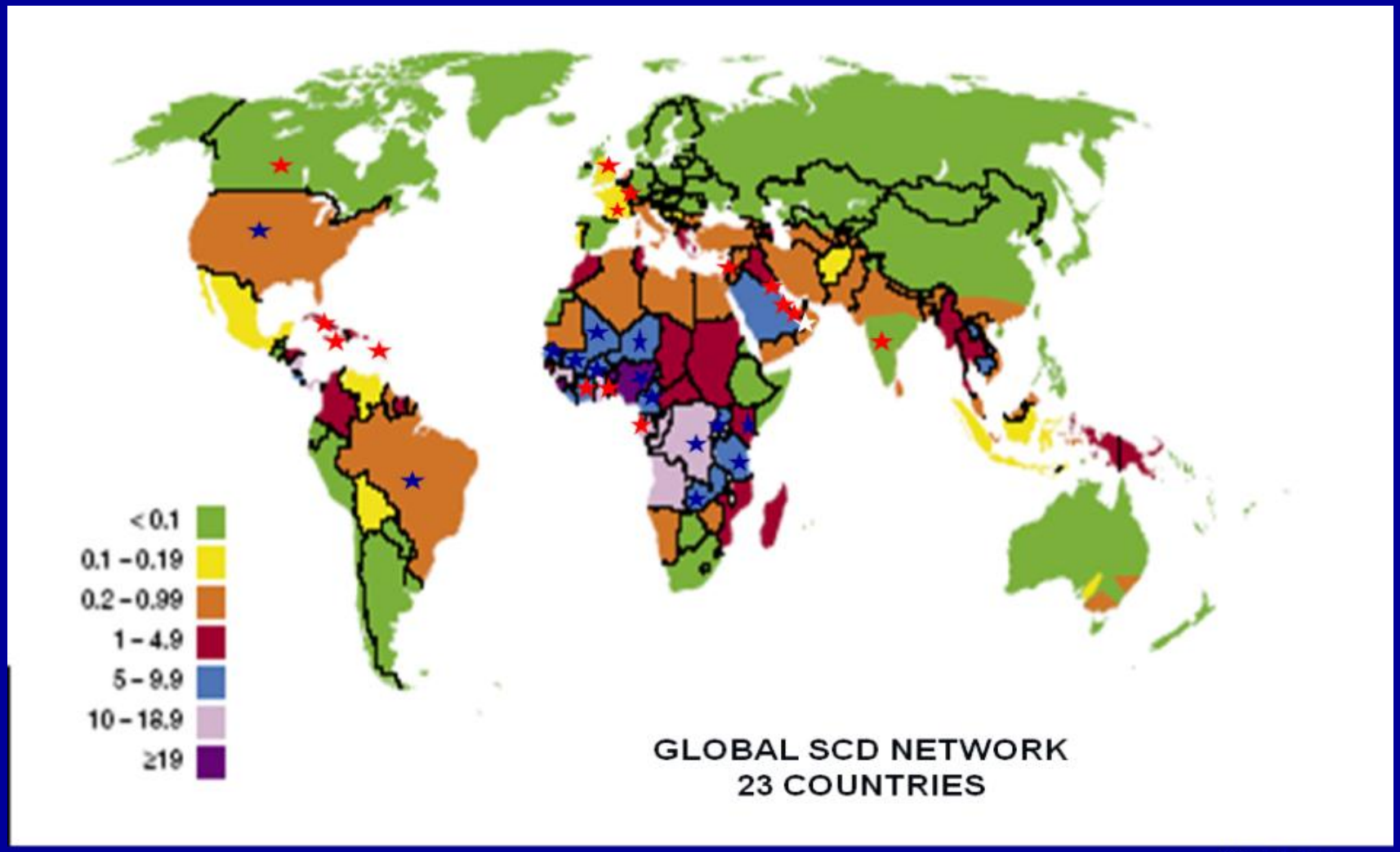
Sickle CHARTA

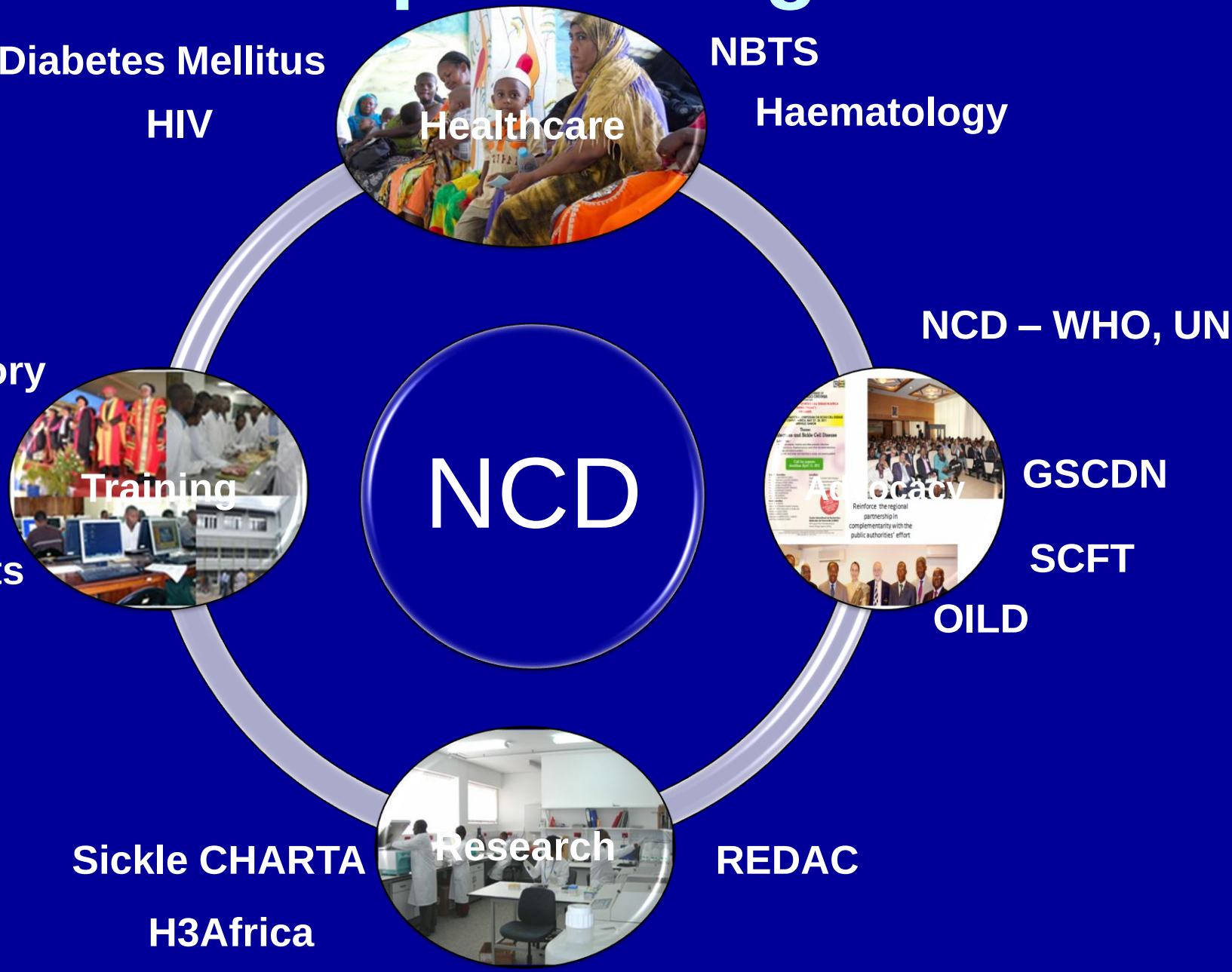


Global Sickle Cell Disease Network



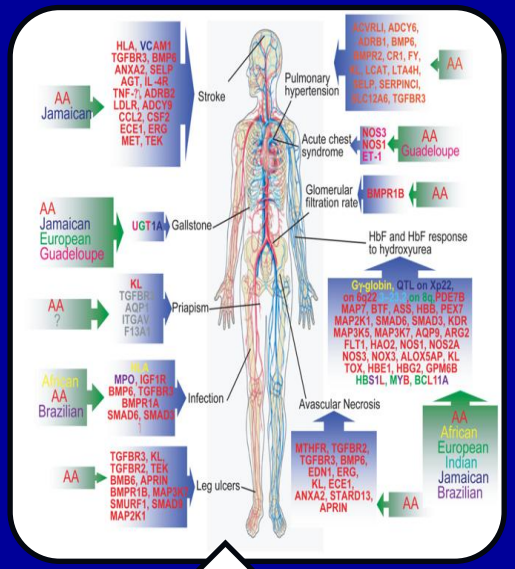
WHO, 1996,

Partnerships & Integration



Genes – Disease - Health

Social and Cultural factors



Environmental factors

Acknowledgements

All patients and staff at Muhimbili

Haematology, Paediatrics & Medicine

MUHAS/MNH

- S Cox, B Lowe, A Komba, D Soka, E Meda, S Rwezaula, H Mwamtemi, P Swai, M Lyimo, A Makubi, C Kindole, E Nyange
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Collaborators

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- F Kirkham
- D Kwiatkowski, J Barrett, I Barrosso
- D Roberts, B Davis, A Qureshi, T Marlowe, J Glanville, P Mwasandube, R Lingwood
- SL Thein, S Menzel

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