The Need for a Paradigm Shift to Non-Communicable Diseases and Newborn Screening of Common Genetic Disorders in Nigeria

TR Kotila, KI Adediran, *PO Olatunji
College of Medicine, University of Ibadan,
*Olabisi Onabanjo University, Ago’woye,
Nigeria
Background

• Malaria is endemic in Nigeria
• Genes that are linked with malaria are therefore prevalent
• There is a well established interdependence between malaria and sickle cell anemia
• Considerable research effort goes into the prevention and eradication of malaria
• Little is done to control sickle cell anemia or other genetic disorders
• There is no program for neonatal screening of genetic disorders in Nigeria
Rationale for Newborn Screening

• There are variations on the reported prevalence rates depending on the study population
• Coinheritance of alpha thalassaemia ameliorates the features of sickle cell anemia but this is not routinely tested for
• The similarities in the clinical presentation of beta thalassaemia and sickle cell anaemia results in misdiagnosis of the former
Laboratory Diagnosis

• Hemoglobin electrophoresis at alkaline pH is sufficient for diagnosis in a symptomatic patient

• This may not be appropriate for a healthy population because of hemoglobins that co-migrate with HbS

• This method is also unsuitable for neonatal diagnosis

• Isoelectric focusing gives better separation than electrophoresis at acidic or alkaline pH
Diagnosis of Haemoglobinopathies in Nigeria

- Electrophoresis at alkaline pH is employed by most hospitals
- HbF and hbA₂ are usually not quantified
- Diagnosis is also not confirmed at the molecular level
Conclusion

• The high prevalence of sickle cell anaemia in Nigeria justifies a need for neonatal screening
• It is also necessary to delineate and determine the prevalence of the different genotypes
• Neonatal screening will not only assist in appreciating the burden of the disease
• It will also help in planning an effective control program
Proposal

• A pilot neonatal screening program
• All levels of health care will be involved
• Tertiary institutions will employ HPLC
• Isoelectric focusing will be used at the primary and secondary health care levels
• A molecular diagnostic centre will be required to resolve difficult cases
Way forward for Nigeria

• Nigeria is a resource-limited country with high disease burden but little intervention.

• SCD and Thalassaemia burden can be transferred to temperate and developed countries through human migration

• Control through neonatal screening requires collaboration between countries advanced in NNS and those developing.

• Through technical assistance in form of training, quality control and sharing equipment required for diagnosis.