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

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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 hbA_2 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.