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Abstract Title : Newborn screening for sickle cell disease in a hospital setting in luanda, Angola: Local implementing learning from an international consortium

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Abstract Body

Early diagnosis of Sickle Cell Disease (SCD) is critical to reducing mortality and morbidity in affected children; however, it remains largely unavailable in Sub-Saharan Africa, where disease prevalence is highest. This a severe monogenic recessive disorder has an estimated mortality rate of 50–90% by age five if left undiagnosed. The Lancet Haematology Commission strongly recommends that all children worldwide be screened for SCD by 2025.

The aim of this work is to share the results from implementing newborn screening for Sickle Cell Disease at Hospital Materno-Infantil Dr. Manuel Pedro Anzacot de Menezes, Angola.

From June 2023 to December 2024, all children born or vaccinated at Hospital Materno-Infantil Dr. Manuel Pedro Anzacot de Menezes, Angola, who had parental consent, were screened for SCD. Blood was collected by heel prick test, and hemoglobin electrophoresis was performed by Isoelectric Focusing (IEF) in the equipment Migele (acquired with the support of Revvity and Arise project). All SS results were confirmed by PCR-RFLP, and other hemoglobin variants were sequenced.

A total of 13256 samples were collected and analysed by IEF. Results indicate a prevalence of 1,4% SS (183) and 20,2% of carriers (2681). Other hemoglobin variants were identified in 44 children, and included E, C and B-thalassemia, and other variants in alpha genes. Of those children diagnosed with SCD a total of 167 (92%) parents were contacted and only 131 (72%) children were commenced periodic medical follow-up consultations and prophylactic treatments (penicillin, multivitaminics and vacinations);. Three of these children are already deceased. Despite receiving the diagnosis, a total of thirty-six parents (20%) refused treatment or follow-up.

These results align with existing estimates of the high prevalence of Sickle Cell Disease in Angola and underscore the critical importance of newborn screening programs to reduce the substantial under-five mortality and morbidity associated with the disease. Furthermore, the notably high refusal rate for attending medical consultations highlights the urgent need to complement screening efforts with robust investments in community health education and literacy, ensuring families understand the importance of follow-up care and treatment.

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Keywords: Sickle Cell Trait, Clinical Research, Diseases, Newborn Screening, Study Population, Epidemiology, Molecular Biology, Hemoglobinopathies, Technology and Procedures, Sickle Cell Disease, Research, Human, Biological Processes, Real-World Evidence

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