NEW OPPORTUNITIES FOR HEMOGLOBINOPATHIES SCREENING IN NIGERIA

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The aim of the study is to evaluate effectiveness of new method of newborn screening (NS) for the most common hemoglobinopathy (sickle cell disease, SCD).

Materials and methods: we performed an analysis of the study conducted by Chukwuemeka Odumegwu Ojukwu University, Akwa, Nigeria, 2017. The screening of SCD was done by determining the genotype through the use of isoelectric focusing eletrophoresis machine. In children who were found to have HbF and HbS, they were sent to pediatric sickle cell unit for follow up and management. The total number of new births within the four years amounted to 4961 of which 2410 were males and 2551 were females.

Results: the results of this study revealed the following of the 4961 newborn children within the age range of 0-30 days, 75.2% had the hemoglobin AA genotype, 24.3% had the hemoglobin AS genotype (carriers), 0.32% of them had the hemoglobin SS genotype and 0.08% had hemoglobin AC genotype which is a variant of the AA genotype; having lysine substitute in place of glutamic acid. Patients with SCD and non-sickling disorders, as well as carriers of abnormal hemoglobin variants, can be detected by this NS method. Choosing primary and secondary target diseases influences false negative and false positive results of the overall NS program and, therefore, are important to the evaluation of the screening process. NS for hemoglobinopathies has sensitivity and specificity close to 100% for SCD with regards to the different genetic forms defined as target diseases; however, it loses sensitivity and specificity when non-sickling disorders are included. Pre-analytic factors like prematurity and blood transfusions prior to sampling influence the sensitivity.

Conclusion: early detection is key to improving the quality of life of people living with SCD especially in childhood. The availability of this NS for SCD for hospitals in Nigeria will help a great deal in proving early diagnosis as well proper follow up, prevention of exacerbating factors and management of associated events. As life-threatening events can occur in children with SCD from three months of age onward, early diagnosis is desirable to establish preventive measures. Patients with thalassemia and other major hemoglobin disorders do not benefit in the same way from early diagnosis. However, these disorders have important implications for family health, and the burden of disease is very high in several regions of the world, thus demanding preventive action. In this context, many NS programs make use of the possibility to detect carriers of hemoglobin variants as a by-product to provide families with reproductive knowledge and informed decision-making regarding hemoglobinopathies.