

Newborn screening in Nigeria: will incorporating congenital hypothyroidism with sickle cell disease improve neonatal screening programme?

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Summary. *Introduction:* Nigeria like many African countries has tried to start the newborn screening for congenital hypothyroidism and many failed. Since sickle cell disease is more common in Nigeria, the hypothesis is that incorporating it into a screening programme for congenital hypothyroidism will improve the uptake of the programme by parents and government. *Methods:* Different aspects of newborn screening with difficulties and challenges in running newborn screening were identified and discussed. *Result:* Identifying that for newborn screening to be successful, several key factors have to be put in place including but not limited to organizational structure, system thinking, finance, legislative and political will. A proper recall system for test positives and diagnostic/confirmatory test must be put in place before the programme starts. Since several other screening programmes like sickle cell disease, cervical and breast cancer have run successfully in Nigeria, incorporating one of them into the newborn screening for CH can make the programme succeed as there will be better uptake by the population and the policy makers. *Conclusion:* The difficulty in establishing a newborn screening programme in Nigeria stem from health care financing, organizing the programme from screening through to recall and treatment, and ultimately, prevention of diseases. (www.actabiomedica.it)

Key words: newborn screening, thyroid, sickle cell disease

Introduction

Many African countries, including Nigeria, have tried to do neonatal thyroid screening for congenital hypothyroidism and most have failed (1-3). Only some regions in South Africa have succeeded sparingly to do this continuously, however, South East Asia have succeeded over the last few decades to have newborn screening incorporated into the delivery services offered to all mothers (4). This success is largely due to individual and institutional commitments, advocacy and collaborations (5). For screening to occur, some basic principles and tenets must be met, including but not limited; test must be sensitive and specific, the

condition must be prevalent and also treatable after diagnosis has been made.

In the black America population, while the prevalence of sickle cell trait and disease remains high at 1 in every 400 babies, the CH prevalence is actually less so, at 1 in 10,000 (6, 7). In Nigeria, many parents know more about sickle cell disease than they do about CH, and if they are given the option of having these two conditions diagnosed at birth, they are likely to agree for the process and even pay for them. It is the believe of the author that if Nigeria with a high sickle cell disease burden incorporates the screening into on-going efforts to have congenital hypothyroidism screening, there will be a better yield and uptake of both conditions.

State of Newborn screening in Africa

Newborn screening for any disease in Nigeria is non-existent and the need cannot be overestimated, as many such diseases can lead to long term devastating consequences and loss of economic power and life-style (2). Many developed countries have succeeded in screening their newborns for many metabolic and genetic diseases. In the Middle East and North Africa where there is high rate of consanguineous marriage, it stands to reason that many autosomal recessive disorders will be prevalent (3). This high rate of metabolic diseases was seen in Qatar (8), with 1 in 1327 children born with metabolic disorders. The diseases of interest in most of Sub Saharan Africa are mainly infectious and naturally, most of the resources for health are geared towards eradicating these. The HIV/AIDS scourge ravaged most of Sub Sahara Africa and the most effective means of reducing the incidence and prevalence was the introduction of Maternal to Child transmission of the disease and voluntary testing and counseling. The development of rapid diagnostic testing for HIV and malaria also improved the disease burdens of these conditions, reducing their prevalence and allowing for early treatment to prevent complications. Nutritional screening using weight, and height and other derivatives of these measurements as tools has also helped in reducing the burden and complications of under nutrition and over nutrition. These are done in schools, and well-baby clinics or hospitals just as the child makes contact with the health personnel. The most common noninfectious disease in Nigeria is sickle cell disease with up to 25% of the Nigeria population carrying the sickle cell gene (9). Many churches actually require that the intended couples have a Haemoglobin electrophoresis done and to present these results before they are wedded. Many debate the constitutional rights of such actions but it is done nonetheless. The purpose of this act is to reduce the prevalence of sickle cell disease and anaemia in the country because the resources to manage the conditions are thinning out not to mention the burden of having a child go through acute painful or anaemic crises. All these screening methods evolved over time when many ideas were brought forward and implemented with some of them failing and others succeeding.

Strategies for improving newborn screening uptake

For new programmes to be established and have public acceptability there has to be education as to the need for the programme from the health care institutions through to the general public that will benefit and the politicians that will make policies to see that there is universal coverage (1, 3-5). The insurance companies, media, government and legislatures have to understand why this kind of programme will benefit the whole country. Understanding that as little as \$5 per infant screening can prevent a loss of intellectual ability in a child who will be dependent on the system could make the legislature enact laws that will make screening mandatory for all children (10). Community leaders who have better understanding of their people they lead should be incorporated ab initio to help mobilise all those to benefit from the programme. Production of flyers, handbills, posters and billboard advertisements should be used to disseminate information about the programme, and collaboration with bodies who have already done relatively similar programmes should be partnered with to help with the planning phase of the programme.

When establishing a newborn screening programme, consideration should be made primarily on the basis of improving the health outcomes of affected individuals, and the making sure the diagnosis is established in the shortest possible time. In this case, the screening should be specific and sensitive so the unaffected newborns are not erroneously labeled as being affected and having to go through a barrage of tests without end (1, 2). A newborn successfully screened and diagnosed will be treated immediately if the symptoms present early, or preventive strategies will be put in place at all levels of prevention to reduce the disease burden.

The model of community health insurance scheme being practiced in some parts of Rivers State, Nigeria under the supervision of Shell Petroleum Development Company should be approached for inclusion of newborn screening into their coverage (11). The success of such inclusion and establishment is likely to spur the nationwide National Health Insurance Scheme to include newborn screening in their health package. This not only reduces the cost of the screening programme, but it also increases uptake of the programme and also

uptake of insurance coverage by the general public. If the public knows that specific diseases can be detected long before symptoms start showing, and they do not have to pay extra from their pocket as the process is already covered in their insurance, there will be dissemination of information and more people will key into the scheme (10, 12).

Developing strict criteria for diseases to be screened is essential in the initial developing and planning stage (13). It makes no sense to include extremely rare and untreatable condition into a screening programme where resources will be expended and outcome will be dismal. The 2 conditions already mentioned in this article (sickle cell anaemia and congenital hypothyroidism) are quite common in Nigeria and should be included in any programme that seeks to screen newborns (1, 2, 6). Other conditions like HIV/AIDS and hearing loss can be included as the programme gets acceptance and structures are well developed as the system progresses. In Lagos state, advocacy for inclusion of hearing assessment for the newborn into the birthing package is ongoing and those with hearing losses are immediately referred to specialist for proper management (14).

The establishment of the structures that will manage the programme from sample collection to recall, diagnosis and treatment will take a long process that must be proper in the first instance (3). Identification of samples and filter papers should be done electronically and a system for archiving put in place. It thus stands to reason that the names on the filter papers with their parents' names and mobile numbers should be legible as was done by Yarhere et al (2). Before samples are sent for laboratory analyses, all details should be entered into a spreadsheet and any sample that is flagged after analysis should have the parents contacted immediately. The laboratory to be used for the screening should be domiciled in Nigeria, with the logistics of retrieving samples from every nook and cranny of the country. The possibility of getting samples to the center must be fully explored making sure samples arrive within 3-4 days of collection. Now, there are many problems and challenges in this aspect and having a system that overcomes these will make the programme succeed.

The cost of screening one newborn is not only in the machine and reagents to be used, but transportation, logistics of collecting samples during or soon after

delivery should be factored in (12, 15). Electrical power should be constant, training, and retraining of support staff must meet contemporary requirements, legal and ethical team must sit regularly to improve the outcomes of the programme, and the recall process will be factored into the cost also (6, 13). Who bears the cost of all of these should be debated before the programme starts. The chances of success of the programme are slim if it is for free to the end users, and the charge should not be prohibitive to prevent continued payment by either the insurance or the end user (15). State governments that are interested in the programme should sign laws into place to make sure the programme is continuous even after their tenures expire. Understanding that the diseases to be screened for are of public health importance, the Ministry of health at Federal, and State levels should take up the responsibility of ensuring the success of the programme.

Partnering with organisations that have done screening before, like family Health International 360, World Health Organisation, United Nation International Children's Emergency Funds etc will improve the outcome of the programme as these bodies will give technical support and advice on how to proceed and where not to thread or to cautiously thread (7, 13). These partners that are well informed and experienced in managing these programmes can also serve as part of the external quality assurance team to ensure system continuity and audit. They will establish guiding principles and criteria for subsequent inclusion of other conditions into the screening panel as needs arise. It must be noted that the usual screening for diseases are voluntary, like the VCT and cervical cancer screening, which individuals usually pay for knowing the implications of not getting tested on time. The case is different in this instance because the end users, newborns, cannot give consent or assent so they do not have a say in the decision on whether or not to be screened.

Methods used for screening congenital hypothyroidism and sickle cell disease

For Congenital hypothyroidism screening, the samples are usually analysed for TSH, with levels greater than 50uIU/ml indicative of hypothyroidism, as

done in Europe and other Asian countries that screen (12, 13). In the USA however, samples are analysed for TSH and T4 immediately to ascertain whether the CH is primary or secondary. In the African and Nigeria model, authors advocate a primary TSH analysis in the first instance since a high level tells that the pituitary gland is functional and the thyroid is likely unresponsive (1, 2). A lower than normal TSH reveals a dysfunctional pituitary gland but does not tell if the thyroid gland is anatomically intact and functional. Now both models have their advantages but cost in the African and Nigerian model should make a primary TSH screening more feasible. The machines used are usually sensitive for TSH up to 0.005 μ IU/ml.

For Sickle cell disease, High performance liquid chromatography is the most feasible method in the newborn period (6, 15). Specimens with HPLC profiles consistent with SCD, sickle cell trait, β -thalassaemia, or variant haemoglobins other than HbS can subsequently be analyzed by Capillary Electrophoresis as a confirmatory method. Other screening methods for HB genotype include Isoelectric focusing, cellulose acetate electrophoresis, Citrate ager electrophoresis, capillary zone electrophoresis and molecular methods.

The challenge of funding and overcoming this special barrier

For any programme to succeed, there has to be funding, and for a programme that the short and long term benefits are not monetary, getting government or organisations to fund them will be difficult (10, 15, 16). Now Nigeria and Africa face more peculiar problems as they are more into curative than preventive medicine and fighting infectious diseases with little devotion to metabolic or other systemic conditions. In countries where preventive medicine takes centre stage, the citizens rarely fall ill and many of their health care professionals devote their time to more innovative technologies and science. Asking the WHO or UNICEF to fund screening programmes will be over bearing because they are more concerned with infectious disease prevention and thus concentrate on vaccination as the main stay of prevention in these countries. Individual governments, at state and federal levels need to take

up this challenge and increase budgetary allocations to primary health care and preventive medicine to allow for this type of screening. If health insurance were patronized and not shrouded in cesspool of corruption, many individuals will buy these insurance schemes and the insurance will cover for the screening of their newborns. Already, there is a sickle cell foundation in Nigeria that drives many charitable programmes geared at improving the care of these children and advocating for continued support. This organisation may be willing to get more financial aids to start screening all newborns for sickle cell disease and getting a system of recall for those who are positive. Foundational grants are a veritable source of funding for such programmes and this option should be explored.

Conclusion

The difficulty in establishing a newborn screening programme in Nigeria stem from lack of financing, organizing the programme from screening through to recall and treatment, and ultimately, prevention of diseases. Political will to improve the primary health care system and facilities where many of these children will be delivered and eventually cared for should be made priority so that actions can be backed by laws that govern the whole process. The cost effectiveness of this programme eventually translates to cost saving knowing the interventions that are available once there is early diagnosis and limitation of disability. The children diagnosed and treated early become less of a burden to the healthcare system and the society and ultimately economically productive and self-sufficient.

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