Indications for Newborn Screening

A case for newborn screening in South Africa

A personal view from Prof Chris Vorster - Chemical pathologist and Head of the Centre for Human Metabolomics at North West University.

The incidence of inherited metabolic diseases in South Africa

There is a perception amongst many South African clinicians that inherited metabolic diseases are exceedingly rare in our country. These perceptions are based on a lack of recognition of these conditions when they present and on misleading earlier research. Hitzeroth and co-workers could only find one case of Phenylketonuria and one case of Tyrosinemia amongst 60 000 screened cases and concluded that metabolic diseases in South Africa are too rare to justify screening[1].

It is not clear from the article which diseases were included in the screening panel. On the other hand Van der Watt et al predicted an incidence of 1 in 5000 newborns for type I glutaric acidemia in a selected South African black population through estimation of the carrier frequency of a commonly occurring mutation.[2] Prof LJ Mienie has headed the metabolic laboratory on the Potchefstroom Campus of the North West University for the past 30 years and has received more than 50 000 requests for the metabolic workup of patients. In his experience the most frequently occurring metabolic diseases that are also included in the newborn screening panel are isovaleric acidemia, propionic acidemia, galactosemia, vitamin B responsive methylmalonic acidemia, maple syrup urine disease and glutaric acidemia type I. Several other conditions, most notably peroxisomal biogenesis disorders, are also common but does not currently form part of the screening panel.

Based on the experience obtained from the screening program it appears as if Biotinidase deficiency may also be quite common. The March of Dimes global report on birth defects found that genetic and congenital disorders cut across all nations with little regard for ethnic background and socioeconomic status. All in all there is little evidence to suggest that South Africa is spared from metabolic diseases.

Economic considerations of a newborn screening program

One of the common constraints to the implementation of a newborn screening program is the perception such a program will come a huge expense and will divert funds from competing health priorities. The reality is that ample evidence exists to illustrate that the economic benefit of screening offsets the costs and that it is economically far better to screen and initiate early treatment than to diagnose late and deal with the associated morbidity and mortality.[3],[4],[5],[6],[7]

While most health care funders will agree that preventative health initiatives will lower the health care expenses of an individual over his or her lifetime there seems to be less clarity on how preventative programs are prioritised. In South Africa screening for HIV, hypercholesterolemia, breast cancer and prostate cancer is fairly common practise and is uncertain why these conditions were prioritized before newborn screening when at least some evidence indicates the opposite.[7]

A more realistic economic barrier is that the initial cost of screening is incurred straight away while the benefits are only realised over an extended period. In the light of this most developing countries have opted for an incremental approach to implementation through pilot programs. In addition, a country must have reached a certain level of economic prosperity to make the implementation of a screening program feasible.

Human rights and social responsibility

Given the vulnerability of the newborn, newborn screening should be regarded as a human right and a social responsibility. The South African Bill of Rights in chapter 2 Section 28 (c) supports this view when it states that "A child has the right to basic nutrition, shelter, basic health care services and social services". In addition to this United Nations Convention on the Rights of the Child (Art. 24[1] & Art. 24[2], which was signed by South Africa in 1993, requires signatories to "recognise the right of the child to the enjoyment of the highest attainable standard of health" and states that parties are to take appropriate measures to "diminish infant and child

mortality and to ensure the provision of the necessary medical assistance and health care to all children with emphasis on the development of primary health care."

Alignment with national priorities and political drive

South Africa was one of 189 nations who have adopted the United Nations Millennium Declaration in 2000. The declaration calls for people across the globe to be freed from extreme poverty and deprivation. The declaration was turned into 8 Millennium Development Goals, of which, Millennium Development Goal 4 calls for the reduction of child mortality by two thirds over the period from 1990 to 2015. This was translated into several targets in South Africa including the reduction of the infant mortality rate from 54 per thousand in 2001 to 18 in 2015. In the 2010 South African Country Progress Report on the Millennium Development Goals, it is stated that the country is unlikely to achieve this target. This is despite the good progress that has been made in as far as infectious diseases, malnutrition and access to health care is concerned. One explanation for this lack of progress is the lack of a clear strategy to deal with genetic and congenital conditions and in fact clear evidence exists that indicates the growing contribution of these conditions when the infant mortality rate reaches 50 per 1000.[8]

It is noteworthy that consideration of genetic and congenital conditions was absent in the Strategic Plan for a Campaign on Accelerated Reduction of Maternal and Child Mortality in Africa (CARMMA) and based on available data it is to be expected that these ever-increasing efforts will produce fewer and fewer results when genetic and congenital conditions are not considered.

Legal and ethical considerations

A number of clinicians in South Africa have had malpractice lawsuits filed against them for failing to diagnose a metabolic condition before irreversible complications sets in. In these cases, it must be kept in mind that the inability to diagnose these conditions clinically is exactly the reason why screening programs were started in the United States and other parts of the world in the 1960's. Over the past 5 decades more and more conditions were added to screening panels based on the criteria a timeous clinical diagnosis is simply not possible while effective interventions can be instigated when screening is available.

Conclusion

There is an overwhelming case for newborn screening in South Africa. For many of the diseases in the newborn screening profile simple and cost-effective treatments are available and it seems completely unethical to deny the children of South Africa such interventions especially given the arguments that are put forward in this document. The technology and knowhow to perform newborn screening is available while the logistical infrastructure already exists in many instances. The issue cannot be ignored any longer…

References

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