Better Late than Never: The Newborn Screening Programme in Pakistan

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Physicians worldwide are familiar with the concept of newborn screening (NBS), but this idea seems not to be discussed as much in Pakistan. This might be because of the fact that healthcare delivery system is still curative here, rather than preventive; even though it seems to be an emerging preventive health care system in some areas. Newborn screening was first introduced by Dr. Robert Guthrie (MD, PHD) in 1962, around 5 decades ago. It initially tested for phenylketonuria (PKU) using a spot of blood on a filter card, and more tests for different diseases have been added subsequently over the years. The list of tests for various disorders differs from country to country, but generally the screening programme is seen as a preventive public health measure when it comes to the diagnosis of conditions such as phenylketonuria (PKU), congenital hypothyroidism (CH), cystic fibrosis (CF) and sickle cell disease (SCD) etc. Conditions such as amino-acidopathies, free fatty-acid oxidation defects and many more which can be detected using tandem mass spectrometry (TMS) have also been added to various screening programmes in the last decade. Generally, a blood test is performed when the baby is 24 to 48 hours of age. Timing is important because certain conditions may go undetected if blood sample is drawn before 24 hours of age.

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It is not too hard to see the positive implications of such programmes; children who have such conditions detected early can be offered replacement therapy (RT) and nutritional therapy (NT) which may lead to normal lives. In most developed countries, the screening programme is running as a public health activity aimed for early identification and detection of such congenital conditions and helps in reduction of morbidity and mortality through timely intervention. On the other hand, in developing countries such as Pakistan, NBS is not yet a priority or has just started to emerge as one. Centres such as Aga Khan University Hospital (AKUH) and a few other hospitals do perform screening for congenital hypothyroidism[1]. Congenital hypothyroidism is one of the most common preventable diseases that has an incidence of 2% in Pakistan, which is around 3 to 4 times more than in the West[2-4]. NBS covers a wide range of diseases, with some states in the United States covering upto 34 conditions and disorders. However, if the diagnosis is delayed, babies end up requiring frequent hospitalisations; usually in intensive care units (ICUs) and high dependency units (HDUs). These patients may become a burden on the healthcare system due to the need of high financial support. A study carried out in Faisalabad, Pakistan showed that only 13% cases of hypothyroidism were detected before the age of 3 months in children, whereas 42% of the cases were diagnosed at around 1-5 years of age[3]. This delay in diagnosis could seriously affect the prognosis of the patient. Most of the disorders carry serious clinical consequences in the affected neonates and young infants. Most of the metabolic and endocrine disorders can lead to growth retarda-

tion, developmental delays, learning disabilities, coma and even death. Newborn screening is a state provided service, however, for its successful implementation, a proper infrastructure should be present. Ethics, evidence and cost of the programme is very important prior to forming any policy regarding it. A successful NBS system is now generally considered to include six essential elements: (i) education (of health professionals, parents, the general public and politicians); (ii) screening (proper timing and specimen collection, transport, laboratory testing and reporting); (iii) early follow-up (including abnormal test notification, tracking and confirmatory testing); (iv) diagnosis (through clinical and biochemical evaluation); (v) management (including counselling, treatment monitoring and long-term follow-up); and (vi) evaluation (through system-wide quality assurance and outcome monitoring)[5]. Creating an infrastructure that will sustain the NBS system is necessarily the focus of programme development as screening is implemented. To ensure high level screening quality, all system components must be part of a system-wide quality assurance using measurable indicators for each component[6]. The American College of Medical Genetics along with a commission from Maternal and Child Health Bureau of Health Resources and Administration recommended a uniform panel of conditions for inclusion in state newborn screening programs in 2006. These included clinical characteristics (e.g. incidence, burden of disease if not treated and phenotype in the newborn); analytical characteristics of the screening test (e.g., availability, features of the platform), diagnosis, treatment and management of the condition in both acute and chronic forms (including the availability of health professionals experienced in diagnosis, treatment, and management). Since there is not yet a screening programme in Pakistan, it is important that the data being generated are available at a national level for review so that plans and strategies can be devised to use resources and finances efficiently. Some people claim that the way healthcare is provided will change significantly after the introduction of these screening tests[7]. Tandem mass spectrometry (MS/MS), which is able to detect many metabolic variants, expanded the capacity of the screening program in many countries. Many countries, however, chose to report only a limited number of disorders detectable by MS/MS[8].

Conclusion

Newborn screening programme (NBS) has already been started more than a half century earlier in the developed world, but has not yet started or implemented in developing countries for example Pakistan, India, Bangladesh, Nepal, Bhutan and even gulf countries except Saudi Arabia, Qatar, Kuwait and UAE, where consanguinity is very high. It is time to start spreading awareness and starting a motivational programme for healthcare providers and the community and also consider starting a national new-born screening programme in the high risk areas of the country. Early detection and intervention of many preventable metabolic, haematological and endocrine disorders can prevent disability, morbidity and mortality.

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