

## REVIEW ARTICLE

### NEWBORN SCREENING IN INDIA: WHAT ARE THE CHALLENGES AND PITFALLS?

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#### Abstract

Neonatal screening is the most important preventive public health programme of the 21st century. It is implemented in majority of the developed countries. Most of the developing countries are following suit. In India "it is still in its neonatal stage and yet to evolve into childhood". Currently there is no government funded neonatal screening programme for the masses. It may seem to be an extra financial burden on the country's resources, but when we consider the large population of one billion and a high birth rate, the burden of metabolic disorders with preventable long term morbidity could be very high. According to World Health Organization (WHO), 140 million children are born every year and 5 million die in the first month of life in developing countries. (1,2) Four percent of the population in India are mentally retarded and 5-15% of sick newborns are thought to have a metabolic problem. (1) Thus mass screening will be useful to prevent disability and death by early intervention, follow-up and counselling.

The author describes the challenges & probable pitfalls in the implementation of such a programme in India.

**Keywords:** Newborn screening (NBS), inborn errors of metabolism (IEM)

#### Introduction

The challenges in implementation of the newborn screening programme in India can be discussed under various alphabetical headings A,B,C,D,E,F,G and H for better comprehension.

#### A – Awareness, Attitude, Anxiety

Newborn screening though popularly known as Guthrie test all over the world, after Robert Guthrie [who is considered as father of the newborn screening, credited for having discovered the screening test for phenylketonuria (PKU)] (3) – is not known to many medical professionals including nurses and doctors. The recent survey done in Bangalore (4), a cosmopolitan city in south India, showed 79% obstetricians and 62% of paediatricians were not aware of the Guthrie test. The survey conducted over a period of 12 months included 602 obstetricians, 209 paediatricians, 34 neonatologists, 223 nurses and 210 laymen. Most neonatologists (49%) were sceptical of the benefits and believed that it should only be offered to high-risk babies. They also felt pessimistic about it, as it is not followed up with parallel support like artificial formulas, confirmatory tests, and outcome of the treated children. (2) Ninety-five percent of the nursing staff and 99% of general public were not aware of neonatal screening. Though the awareness among general public was low, there was no eagerness or anxiety among the general population to know about things. Even providing leaflets was not enough to educate them as they feel

it is a "money grabbing exercise" since their doctors didn't mention it. Twenty percent of them got confused with stem cells including some obstetricians. (3)

On the other hand, a study conducted in California showed that improving communication by the state with prenatal care providers and public regarding newborn screening is going to make a difference to the implementation of the programme.(5)

#### B – Budget

Screening programme in India is currently not funded by the federal Government or the State Government. (6,7) Indian Council of Medical Research (ICMR) commissioned a project few years back at four centres in India to look into the feasibility of the programme to be implemented for the whole country. (7,8) This has come after nearly 5 decades of successful screening programme in other developed countries.

The economic boom has brought lots of private players into the arena. Currently there are few major players in this area including one in Delhi, one in Mumbai and two in Bangalore – offering the newborn screening services to the affordable clients for a price ranging from Rs. 2,500 to Rs. 6,000 to cover the screening costs for disorders like congenital hypothyroidism, Glucose 6 phosphatase dehydrogenase (G6PD) deficiency, galactosemia, cystic fibrosis, congenital adrenal hyperplasia (CAH) plus diseases screened by tandem mass spectrophotometry (TMS). This development in some ways is good for the country as the awareness is definitely going to increase, but the disadvantage is the rich get the services and poor are being neglected and the higher infant mortality continues to remain high among this group. (9,10)

This is a classic example where Public Private Partnership (PPP) could work very well for the benefit of the community at large.

#### C – Current Practices, Costing, Counselling, Consent

Current practices in the last few years include many neonatologists doing some screening for the babies born under their care with no national guidelines to follow.

For now the main stay of counselling in India is about the importance of the screening to the parents and about its cost. But once our awareness of the screening increases and if we were to follow the past foot steps of the western experience or the recent experience in Mexico (11) – the focus soon shifts to the counselling services for the paediatricians in explaining the implications of chronic metabolic diseases to the affected families and also the counselling of the parents and families regarding the need for re-test, confirmatory test and treatment in applicable cases.

Major portion of this counselling in countries like ours will be the cost. The cost means – it includes the cost of testing, cost of re-testing, confirmatory tests

and the on-going need for treatment plus the cost of manpower including parental leave of absence to attend doctor's clinic and doctor's consultation fee and in case of affected families, the cost of the future genetic counselling and the appropriate tests. For these – unless the public health sector is well funded and infrastructure is provided by the government, the private sector would just be offering newborn screening and stopping at that juncture, with no ongoing support.

Consent for Guthrie tests has become a major point of discussion in recent days. (12-15) So far in most countries, verbal consent has been obtained for the Guthrie tests. But in recent days the discussion has tended over how long we can save the blood spots (filter paper) and can that be used for future tests? If so do we need further consent or should we have obtained written consent at birth itself for these? Can these blood spots be used for DNA analysis if it is crime investigation, without consent. All these points of discussion have led some countries to try out written consent for the same. In India, currently most hospitals are doing the tests on verbal consent obtained from the parents.

#### **D – Disease burden, Detailed reporting, Dietary advise, Drugs involved.**

India is a single country of multiple cultures and genetic traits. The research and data from one state does not necessarily apply to another region. The incidence of CAH is thought to be higher in Southern India; where as incidence of G6PD is thought to be highest in Punjab and Gujarat. Thalassemia has its highest incidence in Gujarati's. Disorders mimicking methyl malonic acidemia (MMA) is thought to be higher in vegetarians in south India than in north India. These geographical variations and their understandings are very important in planning the health programmes of a country. (7,16-21)

Also the local cultures need to be considered when taking the disease burden into account. South India is considered to be the hub of "consanguineous marriages". Thus, resources need to focus on autosomal recessive disorders and also education of the public regarding its prevention.

Newborn screening reporting requires expertise in its interpretation. Newborn screening even if done and is reported normal may have included just thyroid stimulating hormone (TSH) and 17 (OH) progesterone for hypothyroidism and CAH respectively and may thus be incomplete. We also need to know the method used for screening and the timing of the sample before we can interpret the results. For example, in some hospitals the TSH is done on the cord blood – in which case TSH values upto 40 IU are normal as there is surge of TSH soon after birth, whereas 40 IU after 1 week could indicate a borderline hypothyroidism.

Diseases screened means there should be management protocols for those diseases. Lot of places in India still don't have any special formulas to treat the metabolic disorder that we have started screening. So what is the next step involved? The

people who offer screening should take responsibility for providing the treatment for the affected diseases and their families. Special formulas that are required for diseases like PKU should be allowed to be imported free of duty.

Drug therapy remains the mainstay of treatment of some diseases that we screen. The importance of the drug therapy needs to be understood by pediatricians, families with affected children, people in government and these drugs should be subsidized.

#### **E – Efficiency, Evaluation of detailed diagnosis.**

Efficiency starts from the education of the people involved and the sample collection. Unless sample collection is correct and processed correctly, it can unnecessarily delay the results of the test, causing unnecessary anxiety for the parents as well as chances that the laboratory may ask for a repeat test and then there is a risk of delay in diagnosis. Also if a baby has a positive result this needs to be followed up very quickly to avoid delay in treatment. Certain diseases may require enzymes to be challenged and may have to be sent after adequate milk feeds are started for e.g. urea cycle defects and galactosemia.

The sample collection also need to be on a filter paper which has not been touched by any staff as the sweat of a person can be part of the sample process which can affect the results.

#### **F – Follow up services, Future prospects, Family therapy**

Success of any programme involves adequate follow up. Follow up services involve adequate explanation for the parents involved, financial constraints and implications involved. The parents need to be explained about the future prospects with early diagnosis and the physician involved should allay their fears.

Family therapy is very essential especially in a country like India where majority of the decisions are taken by the grand parents of the child who may have inherent fears about diagnosis of disorders they have never heard of and their fears need to be appropriately allayed.

#### **G – Genetic Counselling & Geneticists**

With the newborn screening, comes the need for genetic counselling and the need for geneticists especially in a country like India, where consanguineous marriages are thought to be higher compared to the rest of the world. Also the need for geneticists is higher once a hereditary diagnosis is made. Qualified geneticists are very few. To increase the number of qualified geneticists, the government need to increase the training posts in medical colleges to cope with the increasing need.

#### **H – Home therapy, Homoeopathy etc.**

The parental belief about the home therapy and alternative medicines like homoeopathy can dictate the success or failure of the programme. Once a disease is diagnosed, many parents in India cannot imagine some diseases need dietary restrictions or life long

supplementation – which automatically lead them with the social pressures to seek alternative therapies. For example, parents of a child with congenital hypothyroidism may start seeking some dietary modifications to “cure” hypothyroidism rather than giving thyroxine for lifelong or may turn to alternative medicine treatments like homoeopathy. This can be reduced significantly by appropriate counselling.

### I –Indian Scenario.

The situation currently in India is that there is no newborn screening in government hospitals. There are few autonomous institutions screening for congenital hypothyroidism. Most private hospitals in India have started screening for congenital hypothyroidism by measuring TSH either at birth or at 2 days of age at discharge. (22,23) Some hospitals are also doing tests for G6PD screening by measuring the activity of the enzyme.(24) Few hospitals are screening for CAH by measuring 17-hydroxyprogesterone (25), but not following the nomograms for the results issued by International Society of Newborn Screening (ISNS) (26) which is probably leading to too many false positive results. Very few hospitals in the country are offering the screening by TMS for inborn errors of metabolism as a routine.

### Conclusion

It is important to educate not only the public but also the medical and nursing faculty regarding newborn screening. The Organisations like ISNS, WAPM (World Association of Perinatal Medicine) and March of Dimes should step in and help implement newborn screening programme in developing countries to save lives as an urgent measure. (10)

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