

Q&A

Testing times

DR ROHIT CARIAPPA, AN EXPERT IN THE FIELD OF NEWBORN SCREENING WAS IN CHENNAI RECENTLY. HE TALKS TO ET M+ ABOUT HIS WORK AND ITS IMPORTANCE TO THE WIDER COMMUNITY

sangeetha shinde tee

What is newborn screening?

Screening newborns for genetic and metabolic disorders (also referred to as Inborn Errors of Metabolism or IEMs) involves taking a blood spot by a simple heel prick from the baby within 24 hours after the birth of the baby. The sample is then sent to a centralized laboratory by courier and the results are communicated to the doctor within 48 hours if the screening test is positive.

What is the potential risk of not conducting such a test?

Newborns that suffer from IEMs are usually born healthy, with no visible sign of illness. If these IEMs are not detected at birth, the baby will develop symptoms later, when it is too late for treatment to have any positive outcome. If affected babies are not diagnosed very early on they will suffer mental retardation, physical disabilities and even death. A common experience for babies born with an IEM in India, is that they are not screened at birth and spend several months, even years, suffering numerous medical tests to find out what is wrong with them. **What are the medical resources available for those detected with a problem?**

The results are always accompanied by material containing information on the disorder, the treatment, diets, IEM specialists and genetic counsellors in India, which allows the doctor to begin treatment immediately. Most of these disorders can be avoided through a strict diet regimen.

What brought you to this particular field?

I gave up a high paying job in the US to come back to India and offer these services here because I believe it is a need in our country and I want to be able to do something about it. I have good friends whose family went through the horrors of this problem with their child, and it inspired me to do something about it. In most developed countries, Newborn Screening is mandatory. The health and economic benefits of detecting and treating a disorder have proven to outweigh the costs of screening.

What's your advice to parents?

I believe expectant parents should ask their pediatrician about Newborn Screening. They owe it to themselves and their child.

