

Stitch in time saves nine

BY ARTI DAS

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PANJIM: A stitch in time indeed saves nine literally, as the six-month-old Neo Gen Labs at the Goa Medical College (GMC) and hospital launched to test metabolic and genetic disorders in newborns, has demonstrated.

Of the 7000-odd infants tested by the lab, nine were found with the G6PD deficiency, a condition which if not detected, can destroy the baby's blood. These nine lives were saved as the condition, once detected early can be remedied with mere dietary dos and don'ts

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and by avoiding certain types of medication.

GMC in partnership with NeoGen Labs, set up the laboratory using Tandem Mass Spectrometry (MS/MS) technology licensed from PerkinElmer Genetics, Bridgeville, USA in June last year.

This newborn screening programme helps identify the problem in advance. Most importantly, it can help children who may otherwise develop mental retardation in the course of their lives due to undetected disorders.

In India, such disorders cumulatively occur in nearly 3 out of every 200 babies.

According to Dr Mimi Silveira, assistant professor at GMC's Paediatrics depart-

The baby screening laboratory, a public-private partnership initiative at the GMC, has scored high. Of 7,000-odd babies screened so far, around 42 have been detected with some deficiency or the other including nine with blood-damaging G6PD deficiency, greatly helping save lives.



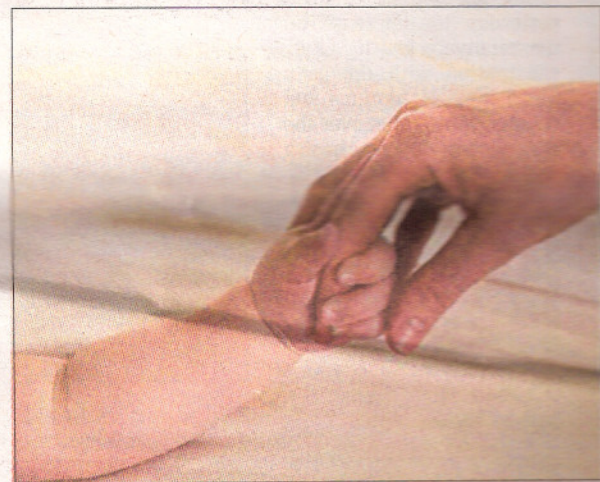
ment 2,415 babies born at the GMC and 4,722 from other hospitals, including Asilo at Mapusa, Hospicio at Margao and some of the Primary Health Centres in Goa, have been tested thus far. Of these, 12 from GMC and 30 from the other hospitals were detected positive for defects.

"For this test we just send blood samples on a

special filter paper to the lab situated at Bangalore and get the results in one or two days. But, in case the test is positive, then they inform us on the phone itself," Dr Silveira said.

The test screens babies for over 40 metabolic disorders.

Dr Silveira said nine babies were detected for G6PD deficiency.



"If this deficiency is not detected in advance, it can result in destroying blood of the baby. Avoiding certain medicines and some dietary changes can rectify it. Thus, we have already given a list of medicines to these cases and their diet will follow soon as now they are quite young and still on breast feeding."

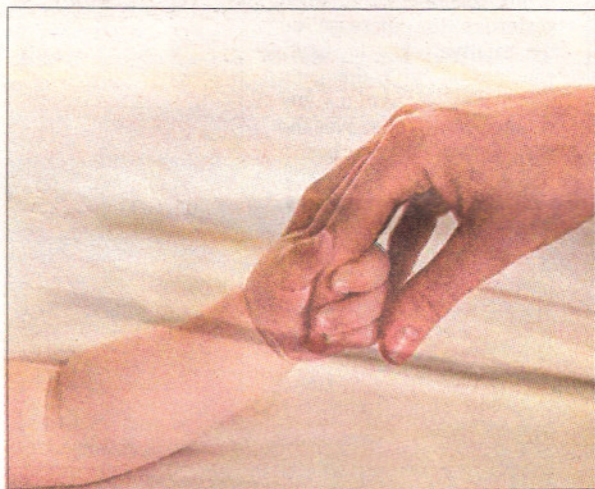
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"We have got four cases of thyroid deficiency. The detection of this deficiency is important as thyroid plays a crucial role in the development of the brain, especially during the first four years of the baby's life. If not treated, then it can lead to mental retardation. For those babies who've tested positive, we've al-

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Another deficiency that is quite common among the tested babies is the thyroid

deficiency.

"We have got four cases of thyroid deficiency. The detection of this deficiency is important as thyroid plays a crucial role in the development of the brain, especially during the first four years of the baby's life. If not treated, then it can lead to mental retardation. For those babies who've tested positive, we've already started treatment, Dr Maria said.

However not all babies were lucky though.

"Out of these positive babies, eight babies have died. It is because sometimes the disorder is not curable and babies don't survive," she said.

These disorders come to light only after testing and are not detected during the pregnancy level.

"The sonography conducted during pregnancy detects only anatomic defects and doesn't pick up enzyme deficiency," Dr Silveira informed.

These tests are restricted not only to babies born in government hospitals but also those born in private hospitals can avail the facility. However for this, parents would have to pay around Rs 4000, otherwise it's free at the GMC and other hospitals. "Some of the parents come on their own, or are recommended by their doctor."

She opined that these tests are a boon for a baby, as early detection helps in giving proper treatment and helping it to lead almost a normal life.

About metabolic disorders

- It is a medical disorder which affects the production of energy within the individual's human cells.

- Most metabolic disorders are genetic, though a few are acquired as a result of diet, toxins, infections, etc.

- Genetic metabolic disorders are also known as inborn errors of metabolism and are caused by genetic defects that result in missing or improperly constructed enzymes necessary.