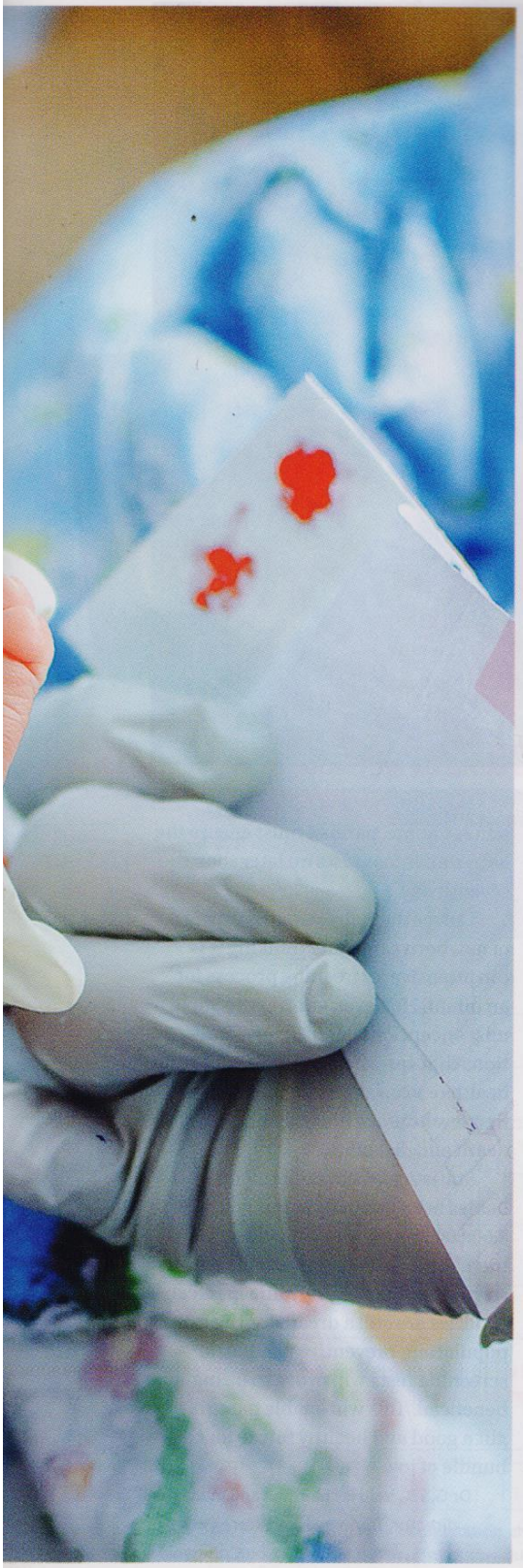

Spot 'em early

Over 45 disorders can be detected through blood tests done within 72 hours of a baby's birth

BY DR G.S.K. VELU

About 27 million babies are born in India every year. But despite the overwhelmingly high birth rate, infant care services in our country are grossly inadequate. Globally, September is observed as the Newborn Screening Awareness Month. There is need for awareness in India about this essential screening.

Studies conducted across the world indicate that 1 in every 2,000 babies has a genetic disorder. Based on the most recent statistics, 3.1 million newborn deaths occurred in 2010 and a quarter to half of them within the first 24 hours after birth.

Newborn screening (NBS) allows us to evaluate the health of infants immediately after birth, even before symptoms of diseases show. It helps in identifying several metabolic and genetically acquired diseases.

Diagnostic tests performed within 72 hours of birth can help identify an infant's possible risk of conditions such as mental retardation, severe anaemia and milk intolerance. This will help in timely medical intervention that can save many infants from disabilities.

How it is done

In a typical newborn screening test, about five drops of blood drawn from the infant's heel through a simple prick is placed on a dry filter paper and tested for various diseases in a lab. For premature babies, a test is first conducted during the first 72 hours and another once the baby is full-term.

This procedure is adopted to detect over 45 disorders. The blood collected is screened to detect abnormalities, which may be a sign of a treatable medical condition. A hearing test is conducted using special equipment, usually when the baby is asleep.

Need for screening

Babies who do not have a family history of disabilities and disorders may still have critical medical conditions. Therefore, it is advisable for parents to mandatorily get newborn screening done within hours of the birth of their babies. It is a preventive paediatric screening service to assess the possibility of genetic disorders in the area of 'Inborn Errors of Metabolism'.

A baby with genetic disorders may not show symptoms immediately after birth. However, without timely treatment the baby usually develops serious problems later in life. Newborn screening helps diagnose these problems early and initiate treatment.

Treatment advice

If the abnormalities or diseases are detected within two to three days of the child's birth, future problems can be averted. Once the disorder is detected, further treatment is suggested, along with advice on the baby's diet.

Although treatments for some conditions are more complicated, it is still helpful to know about the condition early. A baby with sickle cell disease is at risk for harmful infections. Identifying this problem in the baby at the right time would mean that the baby can receive a daily dose of penicillin to help avoid infections and prevent serious problems.

Today, newborn screening has evolved from a simple blood or urine test to a more comprehensive and complex screening system capable of detecting various conditions.

Government initiative

In many countries, hospitals have made these tests mandatory but, unfortunately, in India, there is no government-funded neonatal screening centre. Therefore, it always becomes an extra financial burden for the parents who wish to perform these tests.

Another drawback in our country is the limited awareness about newborn screening. Only a few hospitals in metros like Delhi, Mumbai and Bengaluru perform these tests. On the other hand, Government of Kerala and some of the Central government hospitals have already initiated the active newborn screening programme. Newborn screening should be made mandatory in hospitals. When implementing new programmes, a thorough analysis is needed of the impact on public health and cost effectiveness.

While in developed countries newborns are screened for 50 to 70 disorders in their universal newborn screening programmes using immunoassay and mass spectrometry platforms, many developing countries have adopted a basic programme that screens for at least five to eight parameters using immunoassay platforms. Screening for at least three to five core parameters—congenital hypothyroidism, congenital adrenal hyperplasia, phenylketonuria, galactosemia and G6PD disorders—should be made compulsory for every baby born in India as part of the National Child Health Programme.

If the child is born in a hospital that offers newborn screening, the staff will take care of everything from taking blood samples, to sending them for tests. And the results will be reported back to the hospital. If the birth happens at home, parents must ensure the baby undergoes tests no later than the seventh day.

Currently, many parents opt out of newborn screening thinking it is too intensive a screening process for an infant. However, contrary to such misconceptions, identifying conditions that can affect a child's long-term health or even survival in the critical first few hours of life will help prevent death and disability.

Statistically, out of the 27 million babies born annually in our country, 3,60,800 infants are detected with metabolic disorders. Given the current scenario, where high-profile medical facilities are still a dream for the rural population, preventive paediatric screening tests will prove to be highly beneficial. This will enable parents to gift a good and healthy future for their bundle of joy.

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