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A prick in time....

Ten-day-old Jagriti slept all the time, "but all infants sleep", insisted elders in the family. She had no interest in feeding, and this was scaring Puja Shah, 28-year-old, mother. She decided to visit her paediatrician and a heel prick done for inborn errors of metabolism (IEM) screening confirmed that Jagriti was suffering from congenital hypothyroidism. Treating hypothyroidism was of the medicine that protected the newborn from mental retardation. "I'm happy that I was skeptical", sighs relieved Pooja

Dr. Raghuram Mallaiah, head of the neonatology department at Fortis La Femme hospital, New Delhi says, "Congenital hypothyroidism is the most picked up condition by an IEM screening. Almost one in 1500 babies screened suffer from the disorder."

Genes went awry

According to Indian Journal of Paediatrics, annually 24 million babies are born, of which around 21,000 babies are affected by IEMs accounting for one in every 1000 in the country. "Imagine the plight of parents if their child is one in 1000?" All parents are willing to walk an extra mile to protect their children, and Dr. Mallaiah strongly recommends this test to all parents who can afford it. Newborn screening test costs Rs. 3000-4000 but it screens the infant against 50 metabolic disorders. "And, mind you in most of the instances the newborn appears normal and both parents are healthy", warns Dr. Mallaiah.

IEM disorders are a group of genetic conditions that prevent the baby from processing food properly. This leads to an accumulation of toxic chemicals in the blood harmful to the baby's normal development. If the condition remains undetected and untreated at birth it can cause mental retardation in the baby. By the time symptoms manifest it is too late for treatment to be effective. IEM screens newborns for 45-50 disorders and if tested positive in quite a few instances the line of treatment is inexpensive involving dietary modifications--adding trace vitamin or hormone.

India watch

Thomas Mookken, founder of Bangalore based NeoGen--newborn screening lab for IEM had a personal brush with IEM. His son Ayden Bhagwati tested positive for IEM in a newborn screening test in a private US hospital. Immediately baby Ayden and his mother, as she was breastfeeding were put on avoidance therapy. Young Ayden has to refrain from foods containing sulphur and sulpha based drugs, mehandi and naphthalene balls all his life, but Mookken considers himself lucky. Awakened by the incident he opened up a company called NeoGen Labs, using the latest and world's best technology, solely dedicated to IEM screening.

Phenylketonuria (PKU)--the most frequently picked up condition in IEM screening, says Dr. Mallaiah. Left untreated, this condition can cause problems with brain development, leading to progressive mental retardation and seizures. However, PKU is one of the few genetic diseases that can be controlled by diet.

"Other conditions are--galactosemia (a condition in which absence of an enzyme in liver does not allow metabolism of glucose) and albinism (a condition in which the body does not make melanin pigment, essential for skin colour), informs Dr. Rajiv Chabra, paediatrician and consultant neonatal intensivist at Gurgaon based Artemis hospital. "Children belonging to Parsi and Gujrati communities are at extra risk for G6PD deficiency. This deficiency can normally cause children haemolytic anaemia, usually after exposure to certain medications, foods, or even infections, says Mookken. Another genetic disorder found commonly in the Indian population is Maple Syrup Urine Disease. In this condition the body is unable to process certain amino acids, and the infants affected with this condition have characteristic sweet-smelling urine. "The affected child will be dogged down by conditions such as poor feeding, vomiting, lethargy and developmental delay", informs Mookken.

In India, as of now routine screening of babies born for IEM is not done with the exception of the State of Goa where it will be mandatory to screen all babies for IEMs, who are born at government hospitals. Dr. Chabra says, "IEM screening is recommended if the newborn has jaundice, unusual odour in urine, dysmorphic body structure and the family has a history of unexplained neonatal deaths." "However it is also a fact that 99% of IEMs are detected in families with no history of metabolic disorders and when the baby appears completely normal", says Rohit Cariappa, chief scientist NeoGen.

Plan for the baby

Newborn screening can provide clue for unexplained deaths of baby. Once the metabolism error is decoded it is a great help for parents while planning their next baby, because there is 1:4 probability that the second child could test positive for IEM too.

IEM screening is a complicated procedure and many hospitals have outsourced it to Bangalore based NeoGen Labs. Dr. Lal Path Lab also has the facility for IEM screening

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My Nutrition Guide



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