

FSSAI launches special initiative 'Diet4life' for Inborn errors of metabolism

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New Delhi: Inborn errors of Metabolism (IEM) is a life-threatening medical condition, in which the patient is unable to metabolize certain nutrients and thus suffers from cognitive and physical disabilities for life. IEM patients have special dietary needs, and without special diets, children born with IEM would often not survive infancy. IEM is estimated to affect over 30,000 – 40,000 children in India but in the absence of adequate screening facilities for this disorder, 30,000 diagnosed cases most likely represent only the tip of the iceberg.

To address the challenges faced by IEM patients, Food Safety and Standards Authority of India (FSSAI), Ministry of Health and Family Welfare launched the 'Diet4life' initiative in partnership with various stakeholders comprising of IDA (Indian Dietetic Association), ISIAM (Indian Society for Inborn Errors of Metabolism), MERD (Metabolic Errors and Rare Diseases organization of India), IAP (Indian Academy of Paediatrics) NNF (National Neonatology Forum), ICMR (Indian Council of Medical Research) ISPGHAN (Indian Society of Paediatric Gastroenterology, Hepatology and Nutrition) and Infant and Young Child Nutrition Council of India (IYNCI). Diet4Life initiative is a collaborating effort that aims to provide a holistic service for IEM patients, with facilities of diagnosis, treatment and management of IEM.



What is Inborn Errors of Metabolism?

Inborn errors of metabolism are genetic disorders in which the body cannot convert food (carbohydrates, proteins and fats) into energy and other necessary elements like Aminoacids and sterols. These disorders are usually caused by gene defects that make a particular enzyme defective or deficiencies of the substances that activate the enzymes, or faulty transport of compounds. Enzymes are proteins that help in converting food in to energy, which is required to do work. If the food is not broken down properly it may produce toxins that cause medical problems including growth, problems in the newborn such as poor feeding, vomiting, diarrhea, dehydration, temperature instability, seizures; and altered level of consciousness.

How does it manifest in children and what are the symptoms?

Metabolic Errors present itself at different stages of a child's life, depending upon its severity. Inborn errors of metabolism (IEM) are sometimes referred to as "silent killers" because they can strike healthy-appearing full-term infants without warning. The signs of IEM present at birth, can be subtle, difficult to detect or easy to mistake for other more common neonatal pathologies. It requires a high index of suspicion to include an IEM in the differential diagnosis of an initially healthy full-term baby who begins to display hypoglycemia or poor feeding. temperature instability.

Neonate –

Symptoms for inborn errors of metabolism of substrate and intermediary metabolism develop once a significant amount of toxic metabolites accumulate following the initiation of feeding and may include the following: poor feeding vomiting, diarrhea, and/or dehydration; temperature instability; tachypnea; apnea; bradycardia; poor perfusion; irritability; involuntary movement; posturing; abnormal tone; seizures; and altered level of

consciousness and May be associated with an increased risk of sepsis.

Infants –

Recurrent episodes of vomiting, ataxia, seizures, lethargy, coma, fulminant hepato-encephalopathy or a combination. Dysmorphic or coarse features, skeletal abnormalities and abnormalities of the hair or skin. Poor feeding, failure to thrive. Developmental delay, occasionally with loss of milestones.

Children, adolescents, and adults –

Common findings include mild to profound mental retardation, autism, learning disorders, behavioral disturbances, hallucinations, delirium, aggressiveness, agitation, anxiety, panic attacks, seizures, dizziness, ataxia, exercise intolerance, muscle weakness.

How do we diagnose IEM?

Popularly known as NBS, newborn screening is a medical procedure where a newborn baby is screened within 72 hours of birth for any disorders that might affect the baby's normal functions. NBS is the only way to diagnose IEM. Doctors suggest it to parents if they suspect anything amiss in the baby. The idea is to perform the tests and detect diseases if any at the earliest stage possible so that the necessary treatment can be provided at the very starting of the disease. In many countries, hospitals have made these tests mandatory.

For patients with suspected or known Inborn errors of metabolism, successful emergency treatment depends on prompt institution of therapy aimed at metabolic stabilization. Asymptomatic neonates with newborn screening results coming positive for an inborn error of metabolism may require emergency evaluation including confirmatory testing and as appropriate initiation of disease-specific management.

If experienced pediatrician /metabolic diseases experts are involved, diagnosis can be made through a combination of clinical observations, laboratory evaluations, cerebral imaging and muscle biopsies

How does special formula/foods play a role?

Many inborn errors of metabolism often require diet changes with the type and extent of the changes dependant on the specific metabolic disorder. The particular enzyme absence or inactivity for each inborn error of metabolism dictates which components are restricted and which are supplemented. Registered dieticians and physicians can help an individual assess the diet changes needed for each disease. The goals of nutrition therapy are to correct the metabolic imbalance and promote growth and development by providing adequate nutrition, while also restricting (or supplementing) one or more nutrients or dietary components. Additional goals in some disorders include reducing the risk of brain damage, other organ damage, episodes of metabolic crisis and coma and even death. These restrictions and supplementations are specific for each disorder and they may include the restriction of total fats, simple sugars or total carbohydrates and proteins.

These special diets are carefully designed and manufactured with highest quality standards at few select plants across the world by companies like Danone, Nestle, Abbott and Mead Johnson. There are different types of diets for different metabolic disorders.