

Metabolic Disorders in Children: What to Expect in Neonates and Early Childhood

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Inborn errors of metabolism are considered as complicated disorders for many physicians. With an incidence of one in 1500 persons practicing physicians will encounter at least one of these disorders. Advances in technology and knowledge of human genome resulted in significant modification in classification and management of these disorders. It is important for physicians to recognize these disorders in order to know when to order advanced laboratory investigations or referral to subspecialty doctors. Screening tests is designed to identify persons with markers of different diseases. Advances in medical technology tools for investigations such as the use of electrospray ionization tandem mass spectrometry to newborn screening helps in earlier identification of metabolic disorders which in turn reduce death rates caused by them.

In critically ill patients with metabolic diseases aggressive treatment, before definitive diagnosis, is lifesaving. Acidemia is treated with 1 mg of intramuscular vitamin B12. Metabolic acidosis should be treated with sodium bicarbonate. Seizures are treated with usual antiepileptics but in case of rare cases such as pyridoxine dependent epilepsy, they better be treated with oral pyridoxine. Long-term therapies for metabolic diseases include diet control such as protein restriction, prevention of fasting and cofactor supplements. Promising therapies include organ transplantation and enzyme replacement. Treatment through gene therapy is still in research stage, which may be curative. An accurate diagnosis is required for family reassurance, genetic counseling and prenatal screening.

Symptomatic Infants

A previously healthy neonate may begin to show signs of an underlying metabolic disorder within days or weeks postnatal. These signs include tachypnea, lethargy, vomiting, reduced feeding, hypovolemia, and seizures. With disease progression, they develop sleep apnea, stupor, coma, and neurological abnormalities. Increased plasma ammonia, decreased blood glucose, and metabolic acidosis, are suggestive. Besides, any abnormal odor in an infant could be caused by one of the followings; maple syrup urine disease, phenylketonuria, hepatorenal tyrosinemia type 1, or isovaleric acidemia. Through recognition of full pattern of each disorder, physicians may reach a diagnosis and can request diagnostic tests. Chromosomal analysis is not advised in metabolic disorders since most of them are single gene disorders.

Toddlers and Children

Older infants with metabolic disorders are presented also with lethargy, vomiting, FTT, stupor, other neurological findings, skeletal abnormalities, coarse features, organomegaly, eye or skin manifestations. Cognition, language, attention, focus and concentration defects, and other behavioral changes usually present in toddlers and preschoolers with metabolic disorders. Careful family, social and nutritional history taking is essential for better differentiation between primary-genetic and secondary-acquired causes of disorders that presented with developmental delay or FTT.

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