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Clinical approach to treatable congenital metabolic disorders

Angel Rios

Albany Medical Center, USA

While evaluating a sick newborn infant, the challenges that confronts the physician are to distinguish the more common treatable diseases which can be life threatening from transient phenomena such as transient tachypnea of the newborn. Newborn infants often exhibit nonspecific clinical signs and symptoms that can be encountered in a wide array of disorders that can include sepsis, delay transition, congenital cardiac disease, endocrine disturbances, and inborn errors of metabolism. Because of their rarity, inborn errors of metabolism are not always initially considered and sometimes missed with fatal consequences. Onset of symptoms ranges from a few hours after birth to several weeks of age, depending on the underlying disorder and the initiation of enteral nutrition. The infant might rapidly become tachypneic, lethargic, and develop seizures. Infants with inborn errors of metabolism typically fail to respond to conventional medical therapy. Inborn errors of metabolism are individually rare but if taken collectively can occur as frequently as 1:800 births. Since there is some clinical overlap with more commonly encountered disorders, it becomes increasingly more important to consider inborn errors of metabolism in parallel with common disorders when formulating a differential diagnosis. An example of this mode of thinking is when a physician is presented with a newborn infant with seizures the differential diagnosis usually includes cerebrovascular accidents, infections, hypoxic ischemic encephalopathy without including inborn errors of metabolism such as urea cycle disorders, organic aciduria and maple syrup urine disease. Prioritizing the knowledge and recognition of inborn errors of metabolism that can be treated becomes imperative. The focus of this lecture is to help the physician to expeditiously recognize metabolic disorders and initiate treatment quickly in order to spare the newborn from long-term disabling sequela, brain damage and ultimate death.

riosam@mail.amc.edu