Neonates with inborn errors of metabolism: spectrum and short-term outcomes at a tertiary care hospital.

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Abstract
We aimed to evaluate the neonates diagnosed as IEM in our neonatal intensive care unit and their outcomes. Among 2994 neonates hospitalized, 51 were diagnosed as IEM (1.7%). Admission complaints were poor feeding, decreased activity, jaundice, seizures, abnormal screening and respiratory problems. Phenylketonuria (11), organic acidemias (8), maple syrup urine disease (5), citrullinemia (5), galactosemia (4), nonketotic hyperglycinemia (4) and tyrosinemia (2) were the most commonly diagnosed IEMs. The follow-up period was 2.5-43 months. Among the 33 neonates followed, 19 had normal development, 9 had developmental delays and 5 had cerebral palsy according to the Guide for Monitoring Child Development. Postnatal age on admission, Apgar score at 5 minutes, being transferred, peritoneal dialysis, cranial ultrasonographic findings, consanguinity and sibling history had significant effects on outcome. Early diagnosis through expanded neonatal screening in countries with high rates of consanguinity, enabling the initiation of early treatment, is essential for achieving low mortality rates and good prognoses.

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