8. Screening for Inborn Error of Metabolism in Critically ill Newborn

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Objective: We sought to determine the incidence of various genetic and metabolic disorders in critically ill newborns in NICU setup and to determine the feasibility of comprehensive newborn screening for early identification of these disorders.

Methods: Total 286 critically ill newborns admitted over last 4.5 yrs, in NICUs were studied in detail for genetic and metabolic disorders.

Results: - The commonest IEM detected in our patients were Galactosemia (18), Biotinidase deficiency (8), Partial Biotinidase deficiency (24), MSUD (8), Tyrosinemia (1), Phenylketonuria (0), Homocysteinemia (1), CAH (4), Hypothyroidism (1), Propionic Acidemia (4), Methyl Malonic Acidemia (2), Iso Valeric Acidemia (2), Fructose, I,6 diphosphatase deficiency (1), Ketothiolase deficiency (1), CPT1 def. (1), CPT II def. (4), MCAD Def. (1), LCHAD Def. (1), Severe Carnitine def. (9), HMG CoA Lyase def. (1), Respiratory chain defect (8), GA Type II (5), Cystic fibrosis (3), G6 PD Def. (2).

Conclusions: - Comprehensive metabolic screening for 30+ disorders will detect almost 68.13% of all IEMs in these babies. We hereby strongly recommend comprehensive screening of all NICU babies.