5. Spectrum of IEMs in Indian NICUs.

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Objective:- Ours is one of the major referral center for diagnosis and treatment of IEM in India. Aim was to determine the spectrum of common IEMs presenting in the first 90 days of life as critical illness, so as to develop recommendations for newborn screening tests atleast in NICU.

Subjects and method: We selected 316 critically ill babies admitted in various NICUs with clinical suspicion of IMD from a period 2000-2006.

Results: - All these 316 babies were subjected to a standard battery of tests to detect IEM – Plasma Ammonia, Lactate, Blood Sugar level, ABG, Anion Gap, Urine Ketones, MRST, urine Orotic Acid, HPLC Aminoacids of plasma and if required CSF, Urine GC-MS for Organic acids and MS/MS for Carnitine/ Acyl Carnitine and Aminoacid profile. In addition to these specific diagnostic tests were performed in relevant cases. Incidence for various disorders was: Galactosemia 22(6.96%), NKHG 19 (6.01%), Biotinidase deficiency 11 (3.48%), MSUD 11 (3.79%), OTC deficiency 8 (2.53%), Tyrosinemia Type I 8 (2.53%), Citrullinemia 7 (2.22%), Propionic Acidemia 6 (1.89%), Methyl Malonic Acidemia 6 (1.89%), GA Type II 6 (1.89%), CAH 5 (1.58%), Argininemia 4 (1.27%), Fructose 1,6 Diphosphatase Def. 4 (1.27%), CPT II Def. 4 (1.27%), Cystic Fibrosis 3 (0.95 %) and suspected Mito-chondriopathies 9 (2.85%).

Conclusion: - It was evident that there are atleast 15 common IEMs [with incidence > 1%] in NICU babies. Screening for Galactosemia, Biotinidase deficiency, MSUD and Urea Cycle defects should be recommended for all NICU babies in India.