Spectrum of IEMs in Indian NICUs.

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Objective:- India is vast country with a population over one Billion and annual birth of over 28 million babies. However since it is a developing country, there is no nation wide screening program as yet, but pediatricians and neonatologists are now becoming aware of the seriousness of IEMs, especially in the newborn period. We also have a high rate of consanguineous marriages, especially in the southern part of India and some tribal and minority populations, which also increases the risk of autosomal recessively inherited disorders including some of the IEMs. 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 test panel atleast for NICU babies.

Subjects and method: We selected 316 critically ill babies admitted in various NICUs with clinical suspicion of IEM 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 some other specific diagnostic tests were performed in relevant cases e.g. 17 OHP, VLCFA analysis, Transferrin Iso Electric Focusing to rule out CDG, mutation analysis for Cystic Fibrosis. Incidence for various disorders was as follows: -

Galactosemia	22 / 316	6.96 %
Non Ketotic Hyper Glycinemia	19 / 316	6.01 %
Biotinidase Deficiency	11 / 316	3.48 %
Maple Syrup Urine Dis.	11 / 316	3.48 %
OTC Deficiency	8 / 316	2.53 %
Tyrosinemia Type I	8 / 316	2.53 %
Citrullinemia Type I	7 / 316	2.22 %
Propionic Acidemia	6 / 316	1.89 %
Methyl Malonic Acidemia	6 / 316	1.89 %s
Glutaric Aciduria Type II	6 / 316	1.89 %
CAH	5 / 316	1.58 %
Argininemia	4 / 316	1.27 %
Fructose 1,6 Diph-osphatase Def.	4 / 316	1.27 %
CPT II Def. / CACT	4 / 316	1.27 %
Cystic Fibrosis	3 / 316	0.9 %
Suspected Mitochondriopathy	9 / 316	2.85 %

Many newborns were suspected to have Mitochondriopathy due to following clinical and lab. Features. Neurological involvement, acidosis, high lactate in blood, urine and CSF, normal organic acid profile in urine or mildly elevated urinary Orotic acid or presence of high Succinate, Fumarate, Citrate, Aconitate etc. Absence of specific Aminoacidopathy or fatty acid oxidation defect. Since we do not have facilities for further studies in Mitochondriopathy, they were referred to specialized centers for further workup.

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.***

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References: -

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