Approach to IEM in Indian NICU

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Introduction: - IEM is quite common in sick newborns and correct diagnosis is very important for proper treatment and prognostication. Unfortunately in India, we don't have any authentic data on incidence of IEM in general population or CINB. Because of this there is no standard protocol or SOP in NICUs. In India, there is no nation wide newborn screening. Also there is no insurance or 3rd party payment for newborn babies. Most of the expenses for sick newborn are to be born by the parents. Per capita income of average Indian person is approx. 200 US $ and per day cost for NICU admission is about 100 US $. Therefore most of the NICUs try to adopt seemingly economical approaches, which are based on personal assumptions, whims and fancies rather than scientific facts and data.

Objective:- Main aim of the project was to identify the a protocol which will be cost effective and yet comprehensive. Many different NICUs use different approaches, e.g. some NICUs use either TMS or GC-MS alone, whereas some use both TMS + GC-MS as the starting point for workup of a sick newborn suspected to have IEM. Since ours is a referral center, we use rather detailed approach and each and every newborn referred for suspected IEM undergoes a battery of tests which are all undertaken at the same time.

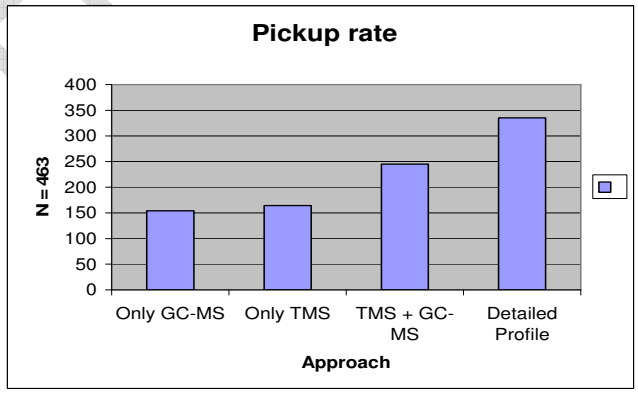
Subjects and method: - Of the 601[Yr 1999 – 2007] referred critically ill babies for evaluation of IEM; we could complete analysis in 463 children. We followed a standard protocol which we labeled as CINB profile for our center. This included - Urine metabolic screening tests, Blood Ammonia, Lactate, Blood Sugar Levels, Blood gases with Sr. electrolytes, GALT and Galactose, Biotinidase enzyme assay,17 OHP,TMS for Carnitine / Acyl Carnitine profile, GC-MS of urine for Organic acids, HPLC - Amino acids, Orotic Acids, Purine & Pyrimidines. Some of the babies also required CDG screening and VLCFA analysis. This type of comprehensive approach allowed us to pick up many more as compared to isolated approaches. We also analyzed the data depending upon the hypothetical isolated approach in the same set of subjects. It appears that had we used only TMS or only GC-MS we would have picked up less than half the cases which we picked up by comprehensive approach. Even a combined TMS + GC-MS seem less effective than comprehensive profile.

Results: -

	Only GC-MS	*Only TMS*	*TMS & GC – MS*	*CINB profile*
N=463	154	164	245	335
	33.26 %	35.42 %	52.91 %	72.35 %

The data given below shows some of the common defects identified by CINB Profile in our cohort of 463 babies. It appears that we have almost all kinds of IEMs in India. This is probably due to high incidence of 2nd and 3rd degree consanguineous marriages, especially in southern part of India and some communities like Muslim, Parsis and some tribal communities in interiors of India

	N=463
Galactosemia	**25**
NKHG	**21**
MSUD	**16**
Severe Biotinidase deficiency	**15**
Respiratory Chain Defect	**15**
Severe Carnitine def.	**14**
Citrullinemia	**13**
Methyl Malonic Acidemia	**12**
Propionic Acidemia	**11**
OTC Deficiency	**8**
GA Type II	**8**
Tyrosinemia Type I	**7**
CPT II / CACT Def.	**6**
Cong. Adrenal Hyperplasia	**5**
Argininemia	**4**
Fructose 1,6 Diphosphatase Def.	**4**
Cystic Fibrosis	**4**
G 6 PD Def.	**3**
Iso Valeric Acidemia	**3**
Peroxisomal Disorder	**2**
Pyridoxin dependant Seizures	**2**
Suspected Mitochondriopathy	**37**



Discussion: - The cost of detailed CINB profile comes to about 350 US $ and for TMS(50 US $), GC-MS(100 US $), TMS + GC-MS (150 US $). However even if baby stays for an additional 5 days for want of further investigations etc., there is an additional expense of approx. 500 US $ which defeats the basic cost saving approach adopted by single test protocols. Moreover one can never come know what really s/he missed till someone else finds that out, which is more problematic especially from the point of Consumer Protection Act applicable to all Indian hospitals, labs and clinics.

Conclusion: - Detailed symptomatologic approach along with a clinical geneticist's opinion is far better than other approaches for sick neonates. This way we will pickup almost all (~100 %) cases.

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