

## ORIGINAL ARTICLE

# Selective Screening For Inborn Errors Of Metabolism In Children: Single Centre Experience From Karnataka

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### ABSTRACT

A large number of inborn errors of metabolism (IEM) in children remain undetected in India due to lack of investigative facilities and economic restraints. We screened 50 children presenting with neurological and metabolic problems at a tertiary level teaching hospital in Karnataka for inborn errors of metabolism using a standard protocol. There was male preponderance of cases (75%). The commonest clinical presentation was convulsions (30%) followed by metabolic acidosis (15%). 15% of the cases showed history of sibling deaths. We have come across four interesting cases in the course of our study - Phenylketonuria, Methyl malonic aciduria, Mucopolysaccharidosis and Branched Chain aminoaciduria, which will be presented in detail in our paper.

**Key words:** Inborn errors of metabolism, Phenylketonuria, Urine screening tests for IEM, Mucopolysaccharidosis, Methylmalonic aciduria.

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