

INBORN ERRORS OF METABOLISM - II

NEWBORN SCREENING FOR INBORN ERRORS OF METABOLISM IN INDIA - PRESENT STATUS, GAPS AND FUTURE DIRECTIONS

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Abstract: *Newborn screening (NBS) is a proven public health intervention enabling early detection and management of treatable metabolic and genetic disorders. In India, NBS has evolved from isolated pilot projects to structured state-led initiatives like NEEV (Delhi) and Shalabham (Kerala). Despite demonstrated feasibility, nationwide implementation faces challenges of infrastructure, cost and workforce limitations. A tiered, resource-based strategy integrating NBS with existing maternal-child health programs is proposed to ensure equity, sustainability and long-term reduction in preventable morbidity and mortality.*

Keywords: *Newborn screening, Inborn errors of metabolism, India, Public health.*

Points to Remember

- *There is significant experience in India for NBS on DBS of core disorders (CH, CAH, G6PD, Galactosemia, Biotinidase deficiency) primarily through large multicentric, funded projects and some state programs establishing the feasibility and need for a national program.*
- *Ongoing state programs (e.g. NEEV and Shalabham under RBSK) have initiated comprehensive screening which in addition to DBS (disorders tested variable) include externally visible birth defects, hearing and CCHD screening. This strategy seems to be appropriate providing one time approach, easier implementation and avoids running multiple parallel programs thus saving cost.*
- *Experience with expanded NBS using TMS is limited though diagnostic facilities are available both in public and private laboratories. The testing is primarily utilised in high-risk scenarios. Some national pilot programs may be initiated in larger experienced institutions.*
- *FSMPs are now manufactured in India (barring few) at a much lower cost and treatment for small molecule IEMs is supported under the National Rare Disease Policy 2021. There is a need to have more centres with metabolic physicians and a robust referral system.*
- *There is an urgent need to roll out a national NBS program in a phased manner beginning from states with better health statistics. Start with Tier I (essential, universal), expand to Tier II in capable states and aspire for Tier III in future as health financing improves.*
- *A tiered, pragmatic framework aligns with India's public health priorities, economic realities, and infrastructure.*

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