INBORN ERRORS OF METABOLISM - I

RECOGNISING TREATABLE INBORN ERRORS OF METABOLISM IN CHILDREN WITH DEVELOPMENTAL DELAY, AUTISM, EPILEPSY, MOVEMENT DISORDER OR CEREBRAL PALSY MIMICS

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Abstract: *Inborn errors of metabolism may present with neurological dysfunctions like developmental delay, autism, epilepsy and movement disorders. Due to overlapping clinical features and non-specific systemic features, these conditions may be missed leading to delayed diagnosis, treatment and poor outcomes. Advances in expanded newborn screening methodology, biochemical diagnostics and genomic sequencing help in precise diagnosis of these treatable inborn errors of metabolism. Many inborn errors of metabolism can be treated with dietary modifications, cofactor supplementation, enzyme replacement, organ transplantation and supportive measures. Early appropriate treatment results in better developmental outcomes. This review proposes a practical diagnostic approach to identify metabolic red flags and provides an evidence-based summary of therapeutic strategies.*

Keywords: *Treatable inborn errors of metabolism, Developmental delay, Autism, Epilepsy, Movement disorders, Cerebral palsy mimic.*

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Points to Remember

- ***Treatable inborn errors of metabolism often masquerade as common neurodevelopmental disorders, leading to missed diagnostic opportunities.***
- ***Clinical red flags-such as developmental regression, episodic encephalopathy, refractory seizures or multisystem involvement-should prompt metabolic evaluation.***
- ***A tiered biochemical workup with genetic testing provides a cost-effective strategy in most clinical settings.***
- ***Early treatments like dietary therapy, cofactor supplementation and targeted metabolic treatments can improve outcomes.***

Note : Readers are requested to refer to standard text books for abbreviation - expansion (of metabolic disorders).

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