INBORN ERRORS OF METABOLISM - I

LABORATORY WORKUP IN SUSPECTED IEM - A STEPWISE APPROACH (BIOCHEMICAL TESTS, METABOLIC PANEL, INTERPRETATION PITFALLS)

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Abstract: *Inborn errors of metabolism are genetic disorders that disrupt normal intermediary metabolism. Early recognition and timely intervention are crucial to prevent severe complications, including disability and death. These conditions often present with variable and non-specific clinical symptoms, making accurate diagnosis challenging. Initial investigations such as blood glucose, ammonia, lactate levels, electrolytes and ketones may provide clues to the underlying metabolic disturbance. The findings of these investigation can guide the selection of more specific, advanced metabolic tests. Diagnostic confirmation typically relies on specialized investigations, including the analysis of acylcarnitines, organic acids, amino acids and specific enzyme assays. This article reviews both the foundational and advanced diagnostic tools, highlighting key biochemical markers used in the identification of Inborn errors of metabolism.*

Keywords: *Inborn errors of metabolism, Laboratory investigations, Acyl carnitines, Organic acids, Amino acids.*

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Points to Remember

- ***Clinical presentation in IEMs can be very helpful in designing the tests in each case and always sharing the clinical details, discussing the possibilities with a metabolic expert can save time and expenses.***
- ***Sample collection should be done before starting any intervention or transfusion. Also, it is important that samples be collected at the right time in correct containers after discussing with the metabolic laboratory.***
- ***Guidelines of proper handling, storage and transport of samples to a metabolic laboratory must be followed.***
- ***Pre- and post- test counseling of parents is of utmost importance***

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