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A NOVEL METHOD OF MULTI-OMICS ANALYSIS IN METHYLMALONIC ACIDEMIA

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Methylmalonic acidemia (MMA) is a rare genetic metabolic disorder characterized by elevated levels of MMA in the body [1]. In recent years, with the rapid advancement of multi-omics technologies, an increasing number of studies have employed multi-omics analysis methods to investigate the pathogenesis of MMA, identify diagnostic biomarkers, and explore potential therapeutic targets.

Keywords: methylmalonic acidemia, multi-omics analysis, transcriptomics, proteomics, metabolomics.

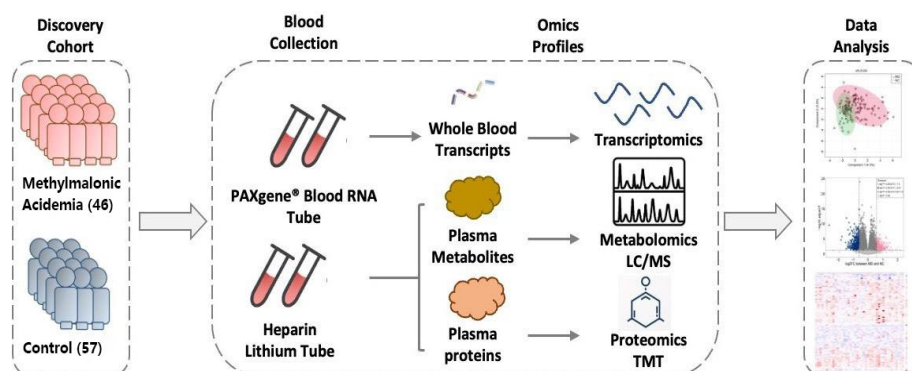


Fig. 1 - Overview of MMA cohort for omics analysis. Study design.

Integrating data from multiple omics levels has deepened our understanding of MMA. Transcriptomic studies have revealed dysregulated gene expression patterns, highlighting potential molecular pathways involved in MMA development. Proteomic investigations have identified disease-specific protein signatures and networks, shedding light on the molecular mechanisms of MMA. Metabolomic profiling has facilitated the detection of altered metabolic pathways and potential biomarkers for diagnosis and monitoring. The integration of multi-omics datasets has been invaluable in MMA research, enabling a comprehensive analysis of the intricate molecular networks driving the disorder. This approach has the potential to identify novel therapeutic targets and personalized treatment strategies [fig 1].

We firstly introduce the disease characteristics and pathogenesis of MMA, followed by a detailed discussion of various multi-omics analysis methods in MMA research. By integrating data from different omics levels, we can gain a comprehensive understanding of the pathophysiological processes underlying MMA, identify molecular markers associated with the disease, and provide novel insights and approaches for the diagnosis and treatment of MMA.

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