Dried Blood Spot Compound Movement for ADA2 Deficiency

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Abstract

Adenosine deaminase 2 (ADA2) deficiency is a rare autosomal recessive disorder associated with systemic infammation and vasculopathy. Timely diagnosis is crucial for initiating appropriate treatment and improving patient outcomes. In this study, we investigate the utility of dried blood spot (DBS) compound movement analysis as a symptomatic device for ADA2 deficiency. By assessing the movement patterns of specific compounds in DBS samples collected from patients suspected of ADA2 deficiency, we aim to establish a non-invasive and cost-effective screening method. Preliminary results demonstrate promising correlations between compound movement characteristics and ADA2 deficiency, suggesting the potential of DBS compound movement analysis as a diagnostic tool for this rare disorder. Further validation studies are warranted to confirm these findings and optimize the diagnostic accuracy of this approach.

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References

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