FTIR and INAD

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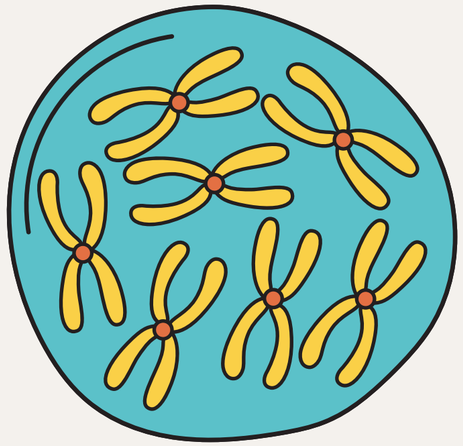
INAD AND DIAGNOSIS

- INAD is a rare genetic disorder with symptoms appearing before age three, including muscle tone issues, abnormal movements, vision impairments, and cognitive challenges.
- Diagnosis traditionally relied on clinical observations, electrophysiological assessments, imaging studies, and skin biopsies, often requiring multiple biopsies for confirmation.
- Before advanced sequencing technologies, diagnosis was prolonged and costly.
- Decreasing costs of gene sequencing and targeted gene panel testing, along with increased physician awareness, have shortened diagnostic timelines significantly.
- Despite advancements, challenges in diagnosing INAD persist due to its rarity and complex symptomatology, necessitating ongoing research and collaboration among medical professionals.

PROBLEM



The diagnosis of INAD is a time-consuming and costly process. An alternative, more effective method should be found.

METHOD

Fourier-transform infrared (FTIR) spectroscopy is a technique used to display absorption bands of molecules in biological samples, enabling simultaneous monitoring of various molecules. It has been extensively utilized for decades in diagnosing diseases like cancer, diabetes, and neurodegeneration, alongside chemometric analysis methods. FTIR's sensitivity to biochemical changes during disease progression or treatment shows potential in identifying new biomarkers and monitoring treatment response beyond cancer screening. Various biological samples, including blood and tissue, have been investigated using FTIR spectroscopy for disease diagnosis due to their accessibility and low cost. Despite promising results in diseases like Alzheimer's, Parkinson's, and cancer, there have been no studies conducted on Infantile Neuroaxonal Dystrophy (INAD) using FTIR spectroscopy.

The FTIR spectroscopy method can be started to be used as a new diagnostic method for INAD by working with blood and cerebrospinal fluid. This way, an early and easily accessible diagnostic method can be found.

CHALLENGES

Firstly, INAD's complex nature may yield biochemical signatures not fully understood, complicating interpretation of FTIR spectra. Secondly, INAD's rarity limits available data for comparison, hindering accurate analysis. Additionally, the variability in symptom manifestation and progression makes identifying specific biomarkers challenging. Optimizing FTIR protocols tailored to INAD diagnosis requires extensive research and validation. Thus, while FTIR spectroscopy holds promise for disease diagnosis, addressing these challenges is crucial for its effective application to INAD diagnosis.

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