RARE fNIRS: A Novel Approach to Identifying Brain Biomarkers in Rare Genetic Disorders

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Abstract (This version 328 words):

We are exploring the use of functional near-infrared spectroscopy (fNIRS) to identify brain biomarkers associated with rare genetic disorders that involve brain dysfunction. Our research aims to enhance early diagnosis and intervention for these disorders by implementing clinical protocols at St. Jude Children's Research Hospital, where participants complete age-related cognitive tasks and provide detailed medical and lifestyle information through pre-experiment questionnaires. This approach aims to capture brain activity across different stages of the cognitive task, allowing for a more comprehensive understanding of how these rare conditions affect brain function.

To ensure robust data analysis, advanced signal processing techniques are employed to correlate brain activity with the disease history and dietary factors, further enhancing the diagnostic value of the data. Preliminary results indicate significant differences in brain activation patterns in the prefrontal cortex (PFC) between individuals with rare disorders and control subjects.

We have developed the **RARE fNIRS** paradigm, an acronym that encapsulates key aspects of our methodology:

R – Resting state and response: Measuring both spontaneous and task-related brain activity to capture a comprehensive picture of brain function.

A – Anatomical targeting: Ensuring accurate probe placement in disease-relevant cortical regions.

R – Reproducibility: Standardizing protocols to ensure reliable and comparable results across different studies.

E – Ethical considerations: Prioritizing patient consent, minimizing burden, and ensuring accessibility for rare disease populations.

f – Functional connectivity: Evaluating disruptions in neural networks that may be specific to the disease.

N – Neurovascular coupling: Monitoring hemodynamic responses that reflect underlying neural activity.

I – Individualized approach: Adapting protocols to the specific capabilities of each patient and the nature of the disease.

R – Robust signal processing: Applying methods that mitigate motion artifacts and other systemic confounds to ensure data integrity.

S – Standardized data collection: Following best practices for data collection to ensure comparability across research sites.

Through this innovative approach, we aim to support the rare disease community by providing earlier, more accurate diagnostic tools, ultimately improving the quality of life for individuals affected by these rare and complex disorders.