A method of screening and monitoring glycogen storage diseases with a single biomarker

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Unmet Need

Glycogen storage disease type I (GSD I) is an autosomal recessive inherited disorder of carbohydrate metabolism associated with life threatening fasting hypoglycemia, and significant morbidity and mortality. Currently, the standard of care therapy is nutritional intervention. This requires frequent scheduled meals or snacks and uncooked cornstarch in-between meals to maintain normoglycemia throughout the day and night. Meals and snacks must limit fructose, sucrose and galactose and include complex carbohydrates to prevent excursions in glucose levels due to the effect of an insulin response. Monitoring the metabolic status of patients with GSDI is essential to prevent complications. Monitoring the amount of carbohydrate intake in patients with GSDI is also of prime importance given too much or too little is hazardous, leading to increased glycogen and fat storage in the liver and kidneys or hypoglycemia and late complications respectively. However, there is no single biomarker for monitoring the metabolic status and diet control in patients with GSD I. Current clinical monitoring depends on daily frequent blood glucose checks throughout the day and night using a glucometer or continuous glucose monitor, as well as lactate, uric acid and triglyceride serial evaluations during clinic visits. Additionally, GSD Ib requires the monitoring of white blood cell, absolute neutrophil counts, and inflammatory markers. There is a need to improve patient care for those with GSD I.

Technology

Duke inventors have reported a method of screening and monitoring glycogen storage diseases. This is intended to be used as a test on patients' biological samples to assess therapeutic and dietary interventions. Specifically, the inventors have discovered that levels of 1,5-anhydroglucitol (1,5-AG) serve as a biomarker in GSD1 (a, b) and other types of glycogen storage disorders. This has been validated using global biochemical profiling data from patients with GSD Ia, GSD IIIa, and liver GSD IX.

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Links

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Other Applications

This can also be used as a diagnostic test of GSD I, which may be especially useful for filling a need in newborn screening. There is currently no marker available for newborn screening of GSD I (a,b).

Advantages

- Could improve patient care for those with GSD I, and other GSDs
- Simplifies monitoring by using a single biomarker
- Has been validated using global biochemical profiling data from patients