Diagnosis and Prognosis of Fabry Disease by Detecting Neuronal Apoptosis Inhibitor Protein (NAIP) Expression

Published date: Feb. 1, 2012

Technology description

Summary

Fabry disease is a severe metabolic disorder that affects the vascular system of multiple tissues and organs. An estimated 1 in 40,000 individuals inherit this rare disease, and suffer from various complications including stroke, renal failure, and cardiac arrest. At present, molecular markers that directly measure cellular dysfunction to not exist, thus, prognosis for Fabry disease therapy can not be assessed.
Available for licensing and commercial development is a rapid diagnostic assay to identify individuals with Fabry disease and an effective mechanism of evaluating enzyme replacement therapy. It provides a quick, inexpensive device for determining expression patterns of the neuronal apoptosis inhibitor protein (NAIP). Peripheral blood white cells of Fabry disease patients are analyzed for elevated levels of the marker NAIP, which is over-expressed in patients suffering from acute strokes. These elevated levels have been found in children with Fabry disease and point to the need for preventive therapies. Additionally, this test can be routinely utilized for evaluation of specific and non-specific therapies that aid in minimizing the complications associated with Fabry disease.
Market:
Individuals genetically susceptible to Fabry disease.

Application area

Rapid diagnostic test to identify person at risk for Fabry disease.
Reliable diagnostic test to identify subject response to Fabry disease therapy.

Institution

NIH - National Institutes of Health

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