Biomarkers in Friedreich's Ataxia

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SUMMARY

UCLA researchers in the Department of Neurology have identified multiple biomarkers for Friedreich's Ataxia.

BACKGROUND

Friedreich's Ataxia (FRDA) is a progressive neurodegenerative disease that affects the peripheral nerves and the cerebellum. Mutations resulting in loss of function of the iron-binding protein, frataxin are found in inherited forms of the disease. Currently, the disease is diagnosed by assessing symptoms and testing for mutations in the gene encoding, frataxin. There are no biomarkers for monitoring disease progression and effect of therapeutics resulting in long clinical trials with ambiguous results. Furthermore, the disease has a long progression period of 15-20 years making disease progression studies difficult.

INNOVATION

UCLA researchers have identified a set of genes that are biomarkers for the neurodegenerative disease, Friedreich's Ataxia. A total of 21 genes were identified for changes in expression in a small cohort of FRDA patients, heterozygous carriers and healthy control subjects. Testing for changes in their expression in animal and cell culture model systems further validated the genes as biomarkers for FRDA. These genes can be easily tested by PCR based methods providing a novel biomarker for monitoring disease progression and effects of therapies in clinical trials.

APPLICATIONS

- Biomarkers for assessing disease progression in FRDA
- Biomarkers for assessing treatments effects in clinical trials

ADVANTAGES

- Easily tested by cost-effective, PCR-based methods
- Accurate and specific biomarkers
- Multiple genes identified

STATE OF DEVELOPMENT

The biomarkers identified have beenassessed for expression changes in animal model systems. Currently, these biomarkers being tested in a larger cohort of FRDA patients, carriers and healthy subjects.

ADDITIONAL TECHNOLOGIES BY THESE INVENTORS

- A Novel Mouse Model for Friedreich's Ataxia

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INVENTORS

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OTHER INFORMATION

KEYWORDS

Friedrich’s Ataxia, FRDA, biomarker

CATEGORIZED AS

- **Medical**
  - Diagnostics
  - Disease: Central Nervous System

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