



SCREENING OF MENKES DISEASE IN NEWBORNS THAT ARE LIKELY TO BENEFIT FROM COPPER TREATMENT

SUMMARY

The Eunice Kennedy Shriver National Institute of Child Health and Human Development's (NICHD) Molecular Medicine Program is seeking statements of capability or interest from parties interested in collaborative research to further develop, or evaluate on a large-scale, population-based newborn screening for Menkes disease (also known as kinky hair disease).

REFERENCE NUMBER

E-186-2008

PRODUCT TYPE

- Diagnostics

KEYWORDS

- Diagnostics
- Newborn screening
- Menkes Disease
- Mass Spectroscopy
- Diagnostic Assays
- Neurological Diseases

COLLABORATION OPPORTUNITY

This invention is available for licensing and co-development.

CONTACT

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DESCRIPTION OF TECHNOLOGY

The Eunice Kennedy Shriver National Institute of Child Health and Human Development's (NICHD) Molecular Medicine Program is seeking statements of capability or interest from parties interested in collaborative research to further develop, or evaluate on a large-scale, population-based newborn screening for Menkes disease (also known as kinky hair disease).

Menkes disease is a fatal neurodegenerative infancy disorder caused by diverse mutations in a copper-



transport gene, ATP7A. Affected infants appear healthy at birth but show distinctive neurochemical abnormalities detectable in blood. They develop normally for 6 to 8 weeks but subsequently develop hypotonia, seizures, and failure to thrive and death by 3 years of age is typical unless early diagnosis and treatment occurs. The current treatment, daily copper injections, has been shown to improve the outcome in Menkes disease if commenced within days after birth; however, early detection is difficult because clinical abnormalities in affected newborns are absent or subtle and newborn screening for this disorder is not currently available.

There is a need for improved methods for early detection of infants with Menkes disease in order to improve outcomes. This technology describes biochemical and molecular methods to identify patients likely to benefit from early copper treatment.

R&D Status: Pre-clinical screening of human samples

Role of Collaborator:

- Collaborate with the NICHD to validate the biochemical test on a tandem mass spectroscopy platform consistent with standard methods for newborn screening

IP Status:

- PCT Application No. PCT/US2008/078966 filed 06 Oct 2008

POTENTIAL COMMERCIAL APPLICATIONS

- Newborn screening and genetic tests to identify patients who would benefit from copper treatment

COMPETITIVE ADVANTAGES

- Ability to more reliably detect Menkes disease-specific neurochemical and/or molecular biomarkers among the general population of newborn infants.

DEVELOPMENT STAGE

- Discovery (Lead Identification)

PATENT STATUS

- **Not Patented:** Research Tool--This invention will not be patented

THERAPEUTIC AREA

- Cancer/Neoplasm