Studies on Menkes Disease

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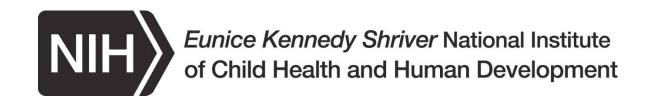
Molecular Medicine Program

Section on Translational Neuroscience

National Institute of Child Health & Human Development

National Institutes of Health, Bethesda, MD





Menkes disease causes premature death



- Death typical by 4 months to 3 years of age
 >90% of untreated patients die by age 3
 51.4% of "late" treated (older than 1 month)
 27.9% of "early" treated (younger than 1 month)
- Currently available treatment:

 Daily copper injections for several years

Early Copper Treatment is Sometimes Very Effective





1998 2013

President Obama with Protocol 09-CH-0059 Subject #34 (Campaign stop in Colorado, September 2012)



ATP7A del exon 3-7

Brain-directed gene therapy with adeno-associated virus: A promising new direction the Kaler Laboratory is pursuing

original article_

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ATP7A Gene Addition to the Choroid Plexus Results in Long-term Rescue of the Lethal Copper Transport Defect in a Menkes Disease Mouse Model

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Molecular Therapy (2012)