

Studies on Menkes Disease

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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)