

Identification of Subjects Likely to Benefit from Copper Treatment

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Technology description

Menkes disease is an infantile onset X-linked recessive neurodegenerative disorder caused by deficiency or dysfunction of a copper-transporting ATPase, ATP7A. The clinical and pathologic features of this condition reflect decreased activities of enzymes that require copper as a cofactor, including dopamine-beta-hydrolase, cytochrome c oxidase and lysyl oxidase. Recent studies indicate that ATP7A normally responds to N-methyl-D-aspartate receptor activation in the brain, and an impaired response probably contributes to the neuropathology of Menkes disease. Affected infants appear healthy at birth and develop normally for 6 to 8 weeks. Subsequently, hypotonia, seizures and failure to thrive occur and death by 3 years of age is typical. Occipital horn syndrome (OHS) is also caused by mutations in the copper transporting ATPase ATP7A, although its symptoms are milder than Menkes syndrome, including occipital horns and lax skin and joints.

Treatment with daily copper injections may improve the outcome in Menkes disease if commenced within days after birth; however, newborn screening for this disorder is not available and early detection is difficult because clinical abnormalities in affected newborns are absent or subtle. Moreover, the usual biochemical markers (low serum copper and ceruloplasmin) are unreliable predictors in the neonatal period, since levels in healthy newborns are low and overlap with those in infants with Menkes disease. Although molecular diagnosis is available, its use is complicated by the diversity of mutation types and the large size of ATP7A (about 140kb). Thus, there is a need for improved methods for early detection of infants with Menkes disease or OHS in order to improve outcomes.

This technology relates to methods of identifying individuals who may benefit from treatment with copper, particularly those having Menkes disease or Occipital Horn Syndrome.

Institution

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