

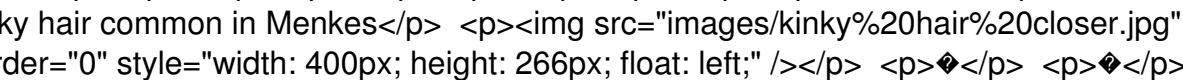
What is Menkes Disease?

Written by Menkes Australia

Saturday, 20 August 2011 10:11 - Last Updated Wednesday, 28 December 2011 09:13

What is Menkes Disease? Menkes Disease is a rare and fatal genetic disease affecting copper transportation around the body, depriving the brain and other tissues of this essential mineral. It is also known as Menkes Kinky Hair Syndrome or Menkes Syndrome.

Who is affected? Menkes Disease usually only affects male infants. Females can carry the disease (70% of cases are inherited). Menkes Disease can occur spontaneously (30% of cases). Menkes Disease is found in all ethnic groups. Menkes Disease occurs in 1 in 100,000 to 1 in 250,000 live births (meaning approximately 1-2 babies will be born in Australia each year with Menkes Disease).

<p>What are some symptoms of Menkes Disease?</p> <p>Children with Menkes Disease usually exhibit some, if not most of the following characteristics:</p> <ul style="list-style-type: none">- short, sparse, kinky hair which is often colourless.- failure to gain weight and grow at the expected rate (failure to thrive)- weak muscle tone (hypotonia)- sagging facial features- seizures- developmental delays (both physically and intellectually)- pudgy, rosy cheeks- low body temperature- irritability- pale, pigmented skin colour- frequent urinary tract infections (UTI) caused by bladder diverticuli.- respiratory difficulties- weak/brittle bones	<p>Below: Example of short, sparse, kinky hair common in Menkes</p> 
---	---

What treatments are available?

Presently there is no cure for Menkes Disease, however copper treatment (either in the form of copper histidine, copper chloride, copper sulfate or copper acetate) are available for boys with Menkes Disease. Please refer to the [Copper Supplementation](index.php?option=com_content&view=article&id=53&Itemid=34 "Copper Supplementation") section for more information.

Other treatments for specific symptoms are also available. Please also refer to the [Other Treatments](index.php?option=com_content&view=article&id=47&Itemid=53 "Other Treatments") section for more information.

How is Menkes Diagnosed? Diagnosis of Menkes Disease is often hard due to the rare nature of the disease and also the fact that affected newborn boys usually appear healthy at birth with few, if not no symptoms of Menkes Disease for the first 2-3 months. However, some babies born with Menkes Disease will be born with unusual kinky hair which should warrant further investigation. There are several different tests that can be performed to confirm Menkes Disease however most commonly blood tests will show low copper and ceruoplasmin levels and a microscopic examination of hair will show the twisted or kinky quality of Menkes Disease.

What genes are involved in Menkes Disease? A mutation of the ATP7A gene causes Menkes Disease. This mutation can occur anywhere along the ATP7A gene and can be any of the following- missense mutations, splice site mutations, nonsense mutations, insertion mutations or deletion mutations.