## Open Peer Review on Qeios

## **Menkes Disease**

National Cancer Institute

## Source

National Cancer Institute. <u>Menkes Disease</u>, NCI Thesaurus. Code C75486.

An X-linked disorder caused by mutations in the AT P7A gene resulting in the abnormal transport and metabolism of copper. It affects primarily male infants. It is characterized by hypotonia, seizures, failure to thrive, and peculiar colorless or steel-colored brittle hair.