

Menkes Disease



PLAY PICMONIC

X-linked Recessive

X-suit with Recessive-chocolate

Menkes disease is an X-linked recessive disease due to a mutation of the ATP7A gene, which encodes for the Menkes protein, copper-transporting ATPase 1

Defect in ATP7A

Defective ATP-battery with Lucky (7) Slot-machine and (A) Apple

Menkes disease leads to a defective copper-transporting ATPase 1, thereby impairing copper absorption and transport, which eventually leads to defective collagen synthesis.

Defective Copper Transporting ATPase 1

Defective Copper-cop with Transporter and ATP-battery with (1)-Wand

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Defective Collagen

Broken Cola-gem Machine

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Clinical Features

Brittle, Kinky Hair

Kitty with Broken, Kinky Hair

Menkes disease can present with brittle or kinky hair.

Hypopigmentation

Hippo-pig with Hypopigmentation

Menkes disease can lead to brittle hair along with both hypopigmented skin and hair.

Hypotonia

Floppy Hippo-baby

Menkes disease can also lead to hypotonia.



Developmental Delay

Developmental-bus

Menkes disease can lead to developmental delay along with failure to thrive, hypotonia, and epilepsy.

Cerebral Aneurysm

Brain with Bulging-aneurysm

Diagnosis

Genetic Testing

Gene Test-tubes

Menkes disease can be diagnosed via genetic testing checking for the ATP7A defective gene. Other testing can include a skin cell culture, blood tests, and skeletal/skull XR.

Blood Test

Blood Test-tubes

Menkes disease can also be diagnosed with blood tests, which detect lower levels of both copper and ceruloplasmin. Ceruloplasmin is a major copper carrier in the blood.