

## 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](#)

Menkes disease is an X-linked recessive disease that occurs due to a mutation of the ATP7A gene, which encodes for the Menkes protein, copper-transporting ATPase 1. This now defective ATPase 1 leads to impaired copper absorption and transport. This impairment thereby reduces the activity of copper-dependent enzymes, such as tyrosinase and lysyl oxidase. Thus, the reduced activity of copper-dependent enzymes leads to defective fibril synthesis and collagen crosslinking.

### 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](#)

Menkes disease is associated with an increased risk of cerebral aneurysm, which involves an abnormal dilation of the cerebral artery or vein, especially around the circle of Willis. The berry (saccular) is the most common type.

## 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.