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.