Condition: Menkes disease

Inheritance:

X-linked recessive.

Genetic etiology:

Mutation in *ATP7A* gene, encoding copper-transporting ATPase 1.

Frequency:

Approximately 1/100,000 births.

Clinical features:

Affected individuals have impaired copper absorption and consequent reduced activity of copper-containing enzymes. Classically, the disorder presents in infants or young children with neurological regression, seizures, failure to thrive, short, sparse, and kinky hair, with death by three years. A milder form is occipital horn syndrome, due to calcifications in the attachment of the trapezius and sternocleidomastoid muscles to the occipital bone, along with joint laxity and tortuous blood vessels.

Management:

Supportive care.

Genetic counseling:

Based on X-linked recessive inheritance; molecular genetic testing available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.