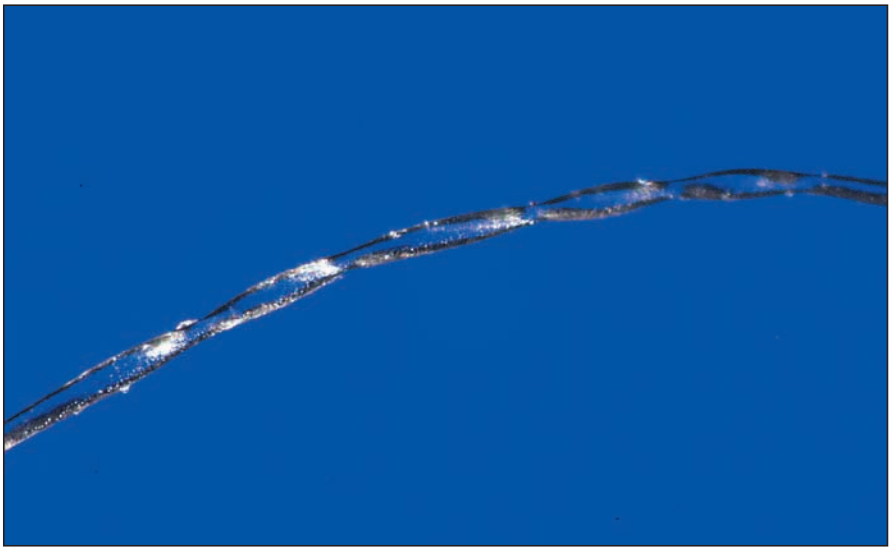
Menkes' syndrome is an X-linked recessive genetic disorder of copper metabolism first described in 1962.[1,2] It is a rare condition (incidence rate of 1 in 298 000 live births).[3] Mutations in the *ATP7A* gene, which encodes a copper membrane transporter protein,[4] lead to deficiencies in intestinal absorption of copper and in intracellular processing of copper in the central nervous system and connective tissues, where enzymes requiring copper as a cofactor no longer function properly.

Clinical manifestations usually begin within the first 1–2 months of life and include failure to thrive due to feeding difficulties, twisted hair (pili torti, also described as "coarse" or "steely" hair), skin laxity, joint hypermobility, cerebral vessel tortuosity, bladder diverticula, osteoporotic bones with flared metaphyses (there may also be wormian bones) and progressive cerebral atrophy (leading to mental retardation, seizures, hypotonia and optic atrophy).[5] The condition is suspected in infants with low serum copper and ceruloplasmin levels and is confirmed by means of copper uptake studies on fibroblast cultures. Menkes' syndrome is characterized by progressive deterioration, and death usually occurs by 3 years of age.

Oral copper treatment is not effective, but subcutaneous injections of copper histidine appear to provide encouraging results in preventing neurodegeneration and prolonging survival,[6] especially if started within 1 month of life. Copper treatment unfortunately does not prevent the connective tissue complications associated with the syndrome, and many patients subsequently die of these complications.



Courtesy: Erik Swartz

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[Permission was obtained from the boy's family to not conceal his face.]

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