Teaching NeuroImages: Menkes kinky hair syndrome

Six-month-old twins, born to a consanguineous couple, presented with hypotonia, hypothermia, seizures, and developmental delay. The babies were fair complexioned; scalp hairs were sparse, light-colored, and fragile (figure 1), with regularly spaced twists (pili torti) and nodes (trichorrhexis-nodosa). MRI brain revealed generalized atrophy with tortuous arteries (figure 2). Low plasma copper and ceruloplasmin levels confirmed Menkes syndrome.

Menkes syndrome is an infantile X-linked recessive neurodegenerative disorder caused by missense mutations in the \( ATP7A \) gene (copper transport gene on chromosome Xq21.1), which causes copper deficiency. Reduced activity of copper-dependent enzymes leads to abnormalities in connective tissue, blood vessels, and hair.1 X-rays might reveal widening of metaphysis with spurring.1,2 Early treatment with parenteral copper histidine might stop the progressive neurodegeneration.1,2

**AUTHOR CONTRIBUTIONS**
Dr. Seshadri studied the case, collected the information, and compiled the manuscript. Drs. Bindu and Gupta reviewed the manuscript.
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REFERENCES
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