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Teaching NeuroImages: Menkes kinky hair syndrome

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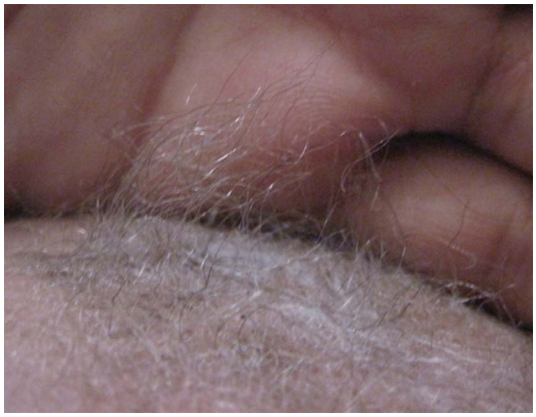
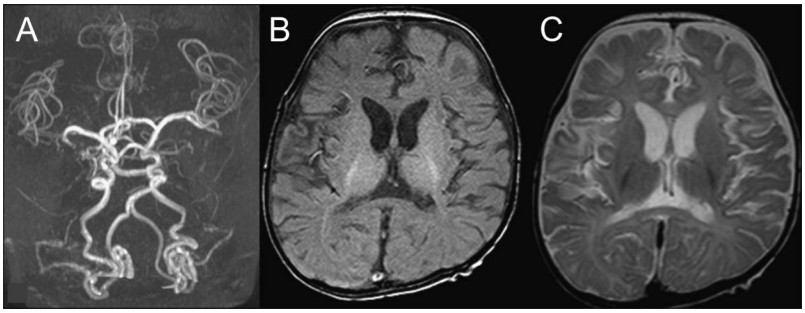
Figure 1 Characteristics of scalp hair in Menkes disease



Hair is sparse, short, thin, fragile, and light-colored, and has a steel-wool appearance.

Figure 2 Imaging



(A) Magnetic resonance angiography reveals highly tortuous arteries of the brain. (B) T1 and (C) T2 MRI sequences demonstrate generalized neuroatrophy and delayed myelination.

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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