Menkes Syndrome
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**History**
3 month old male with seizures, apnea and respiratory failure. Born prematurely at 29 weeks.

**Diagnosis**
Menkes Syndrome

**Additional Clinical**
Hair is fuzzy and palpably dry. Ears are large with elongated ear lobules. Nonmeasurable copper level.

**Discussion**
Menkes disease is a broad range of conditions that affects males (X-linked). One third of the time infants represent a de novo occurrence rather than an inherited condition. The condition occurs because of an inability to transport copper across the intestinal mucosa. First presenting signs and symptoms occur at 2-3 months of age. Findings include low muscle tone, osteopenia, seizures, intracranial hemorrhage, failure to thrive and developmental regression. Additional characteristics may include coarse facial features, coarse hair, temperature instability and hypoglycemia in the neonatal period. This condition results in death, usually by 3 years of age. The inevitable progression to childhood death is preceded by neurological regression. Early treatment consists of injections of copper histidine to replace the non-absorbed copper but copper therapy has not been definitely proven to prevent neurologic damage. It is though the opportunity to prevent CNS damage is lost if therapy is not initiated by about 10 days of age. Therapy may not be effective in some individuals. Fractures in Menkes may follow minor trauma, and metaphyseal fractures with spurring may also occur that appear very similar to those seen in nonaccidental trauma and have erroneously led to that diagnosis.

**Findings**
CR – Moderate metaphyseal deformities of the proximal humeri and constochondral junctions.
MR – Marked decreased T1 and increased T2 white matter signal with loss of the cerebral cortical ribbon in the bilateral temporal regions and thinning of the cortical ribbon in the periorlinal regions.

**Reference**

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