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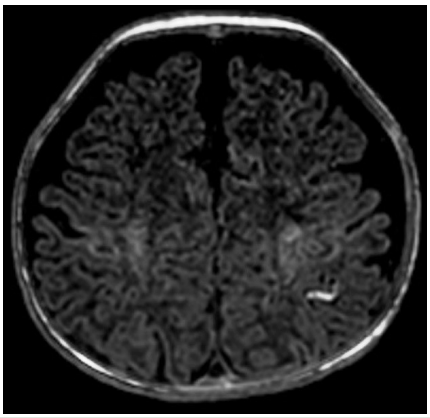

Figure 1. Brain MRI (T1W) revealed mildly delayed myelination, benign subarachnoid space enlargement, and a vascular anomaly in the left parietal lobe.

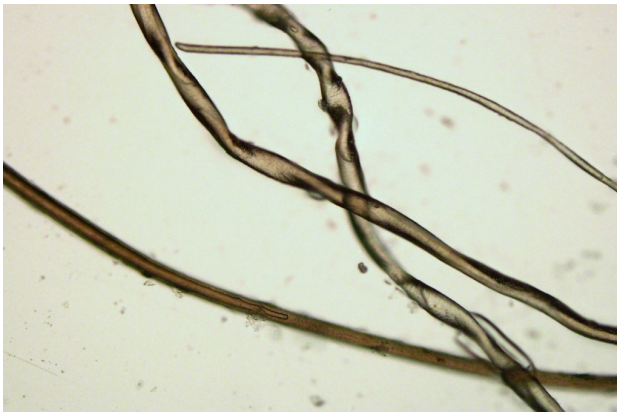

Figure 2. Microscopic examination of the hair showed *pili torti,* hair shaft thinning, and decreased pigmentation.

The patient was a 2-month-old male infant with seizure, hypotonia, poor feeding, and vomiting. He had fair complexion and hypopigmented sparse woolly hair. Brain magnetic resonance imaging (MRI) revealed a mild delay in myelination and benign subarachnoid space enlargement with 6.5 mm craniocortical and 10.5 mm interhemispheric diameter. It also exhibited cortical hyperintensity in T1-weighted sequences (T1W) in the left partial lobe that could be due to vascular anomaly (Figure 1). Laboratory tests were unremarkable except for low serum ceruloplasmin (100 mg/L, normal range 200-600 mg/L). Hair microscopy revealed *pili torti*, hair shaft thinning, and decreased pigmentation (Figure 2).

What is your diagnosis?
See the next page for your diagnosis.

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Photoclinic Diagnosis

Menkes Disease

Menkes disease (MD) is characterized by copper deficiency in contrast with Wilson's disease. MD is a rare X-linked genetic disorder caused by ATP7A mutations with a frequency of 0.8–2/100 000 live male births. Its pathogenesis is explained by the defective activity of several essential copper-containing metalloenzymes like cytochrome c oxidase, lysyl oxidase, superoxide dismutase, dopamine β hydroxylase, ascorbic acid oxidase, and tyrosinase.[1,2]

Classical clinical manifestations of MD that begin from the neonatal period include hypothermia, hypoglycemia, poor feeding, impaired weight gain, and less often hemorrhagic diathesis like cephalohematoma. Later, at the age of 2–3 months, MD presents with seizure, failure to thrive, and developmental delay. The most striking finding is colorless and friable hair. Microscopic examination of the hair commonly exhibits *pili torti* that is the hair shaft flattening and twisting 180 degrees on its axis.[1,3,4]

Neuroimaging studies frequently show impaired myelination, cerebral atrophy, regions of low density within the cortex, tortuous and enlarged intracranial vessels, and subdural hematoma. MD diagnosis is suggested by typical presentations like seizure, developmental delay, and unusual hair. Finally, it is diagnosed by reduced levels of serum ceruloplasmin, and copper. However, serum copper and ceruloplasmin are within normal limits during the first 6 weeks of life and cannot be utilized diagnostically very early. Abnormal catecholamine metabolites (due to dopamine β hydroxylase deficiency) can be utilized as a sensitive and specific diagnostic test and may contribute to earlier diagnosis of MD.[5]

Copper replacement therapy with daily subcutaneous injection of copper histidine is the treatment of MD and might have a relatively satisfactory outcome if initiated in the early days of life. Nevertheless, the overall prognosis is poor and death happens in the first 3 years of life.[1,6]

Authors' Contribution

MA: Involved in patient clinical management and drafting the manuscript. MS and DG: Involved in pathology interpretation, image preparation and drafting the manuscript.

Conflict of Interest Disclosures

The authors have no conflicts of interest.

Ethical Statement

Informed consent was obtained from the patient's parents.

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