

Emergence of kinky hair in Menkes disease

Desenvolvimento de kinky hair na doença de Menkes

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A three-month-old male presented with developmental delay, rib fractures, and seizures. He had normal-looking hair.¹ A magnetic resonance imaging (MRI) scan of the brain was normal. Pathogenic *ATP7A* mutation was identified, which confirmed Menkes Disease. Only at 9 months of age did he develop patches of kinky hair.

Parenteral copper histidine supplementation can modify Menkes disease progression if initiated within days after birth.^{2,3} The clinical diagnosis relies on phenotypic presentation, especially sparse and lusterless scalp

hair typically apparent by 2 to 3 months of age; however, our patient demonstrated that this feature may not appear until much later in life. Therefore, hair abnormalities should not be relied upon to initiate genetic testing (► **Figure 1**).

Authors' Contributions

HAO, MAM: conceptualization, data curation, formal analysis, writing of the original draft, and writing – review and editing.

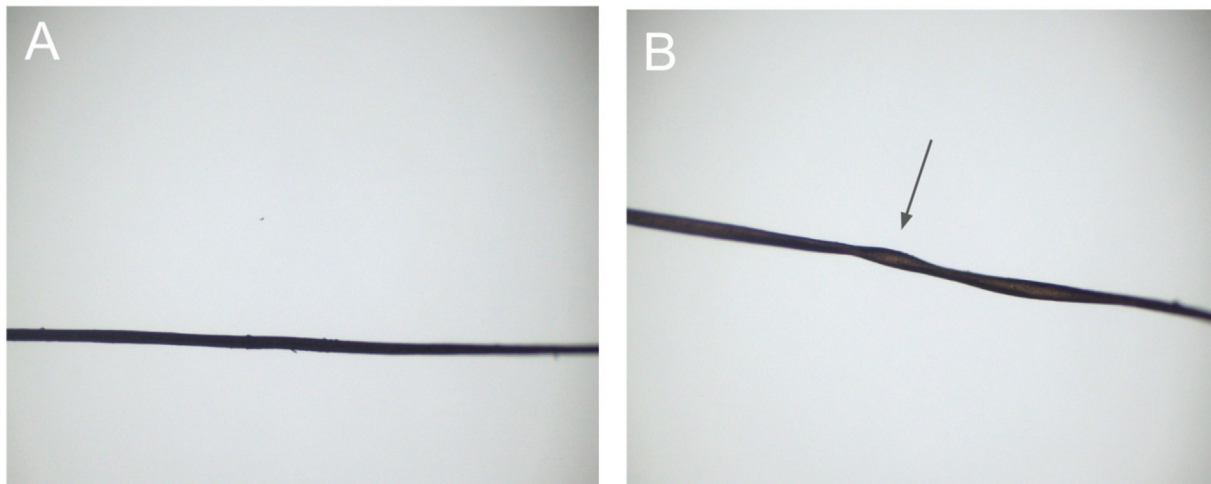


Figure 1 (A) Normal hair at 3 months of age; and (B) hair strand at 9 months of age with classic *pili torti* seen in Menkes disease.

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Conflict of Interest

The authors have no conflict of interest to declare.

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