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.

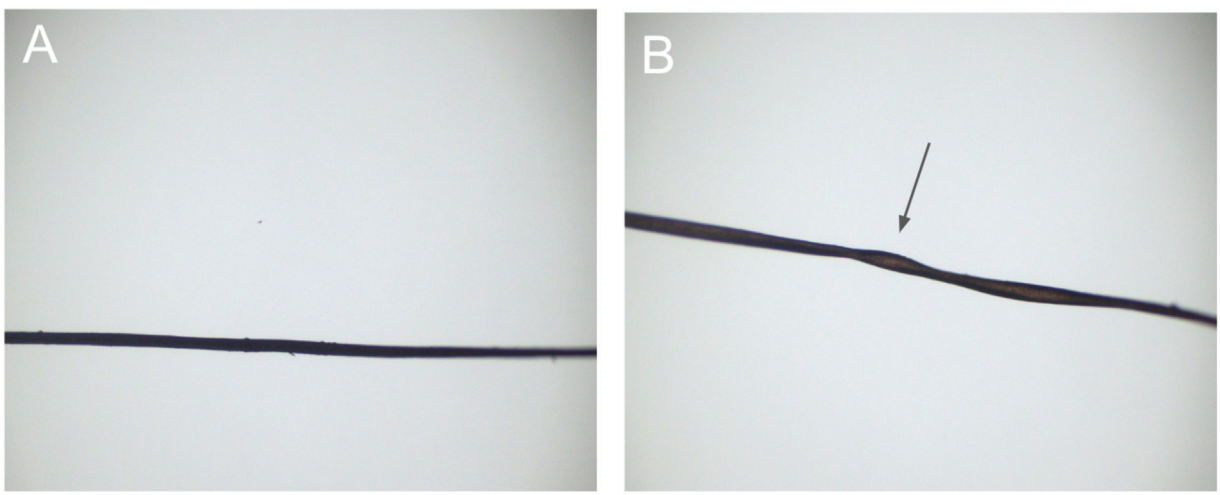

Figure 1 (**A**) Normal hair at 3 months of age; and (**B**) hair strand at 9 months of age with classic *pili torti* seen in Menkes disease.

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Conflict of Interest

The authors have no conflict of interest to declare.

References

1 Tümer Z, Møller LB. Menkes disease. Eur J Hum Genet 2010;18 (05):511–518. Doi: 10.1038/ejhg.2009.187

2 Vairo FPE, Chwal BC, Perini S, Ferreira MAP, de Freitas Lopes AC, Saute JAM. A systematic review and evidence-based guideline for diagnosis and treatment of Menkes disease. Mol Genet Metab 2019;126(01):6–13. Doi: 10.1016/j.ymgme.2018.12.005

3 Kodama H, Murata Y, Kobayashi M. Clinical manifestations and treatment of Menkes disease and its variants. Pediatr Int 1999;41 (04):423–429. Doi: 10.1046/j.1442-200x.1999.01095.x