

# Imaging features that allow for the recognition of Menkes disease

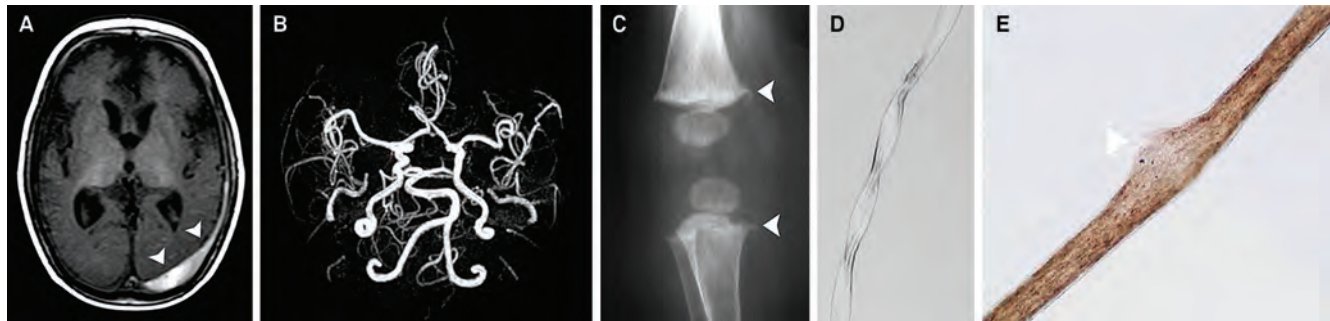
Características de imagem que permitem reconhecer a doença de Menkes

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Menkes disease (MD) is an infantile-onset, X-linked recessive neurodegenerative disorder (ATP7A gene on chromosome Xq21.1) in which copper transport and connective tissue arrangement is disturbed<sup>1</sup>. Variable MD phenotypes are strictly related to abnormal collagen and elastin formation that are more due to the lysyl-oxidase deficiency, which results in skin and hair

alterations associated with tortuous and elongated intracranial vessels, in addition to bladder diverticula and bony abnormalities<sup>1,2,3</sup>.

When imaging suggests MD, laboratory results are used to confirm the diagnosis (Figure). Genetic counseling and early treatment with daily copper injections is suggested to improve outcomes for this rare disease<sup>2,3,4</sup>.



**Figure.** A 9-month-old boy presented with hypotonia, seizures, developmental delay, failure to thrive and hypopigmented skin and hair (copper=99.8 ug/dL (183-152 ug/dL) and ceruloplasmin=4 mg/dL (20-60 mg/dL)). Axial T1WI (A) showed brain atrophy associated to left parietal subdural hemorrhage (arrowhead). Kinking and coiling of the intracranial vessels were depicted on MR angiography (B). Plain radiograph (frontal view) of the left lower limb (C) showed femoral and tibial metaphyseal spurs (arrowhead). In scalp hairs, regularly spaced twists compatible with pili torti (D) and nodes of trichorrhexis-nodosa (E) were also observed (arrowhead).

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