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Menkes Disease: Report of Two Cases

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Abstract:

Introduction: Menkes disease is a rare X-linked recessive disorder of copper metabolism. It is characterized by progressive cerebral degeneration with psychomotor deterioration, hypothermia, seizures and characteristic facial appearance with hair abnormalities.

Case Presentation: We report on two cases of classical Menkes disease with typical history, (progressive psychomotor deterioration and seizures}, clinical manifestations (cherubic appearance, with brittle, scattered and hypopigmented scalp hairs), and progression. Light microscopic examination of the hair demonstrated the pili torti pattern. The low serum copper content and ceruloplasmin confirmed the diagnosis.

Conclusion: Menkes disease is an under-diagnosed entity, being familiar with its manifestation and maintaining high index of suspicion are necessary for early diagnosis.

Keywords:

Menkes disease ، Copper metabolism ، Pili torti ، Cerebral degeneration

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