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Karnataka Paediatric Journal

Letter to the Editor

Infantile tremor syndrome masquerading as Menkes kinky hair disease with long-term follow-up

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Received: 12 April 2023
Accepted: 05 June 2023
EPub Ahead of Print: 23 June 2023
Published: 17 August 2023

DOI
10.25259/KPJ_26_2023

Quick Response Code:

Dear Editor,

Infantile tremor syndrome is characterised by anaemia, developmental delay, hair changes and tremors.[1] Menkes disease (MD) is an X-linked recessive disorder due to mutations in the ATP7A gene manifest with developmental delay, seizures, subdural effusion and hair changes (pili torti).[2]

A 12-month boy presented with developmental delay, followed by regression. The child attained neck holding by 6 months, reaching out for objects by 10 months and bisyllables by 1 year. At around 12 months, he developed a fever and subsequently had a regression in all domains. On examination, pallor, hyperpigmented knuckles, hand tremors, weight - 6.59 kg (−5.34 WHO Z), head circumference - 41 cm (−5.64 WHO Z), hypotonia in all limbs with 3/5 power and brisk deep tendon reflexes were noted. Investigations revealed haemoglobin of 5.1 g/dL, a total count of 8200/µL, platelet

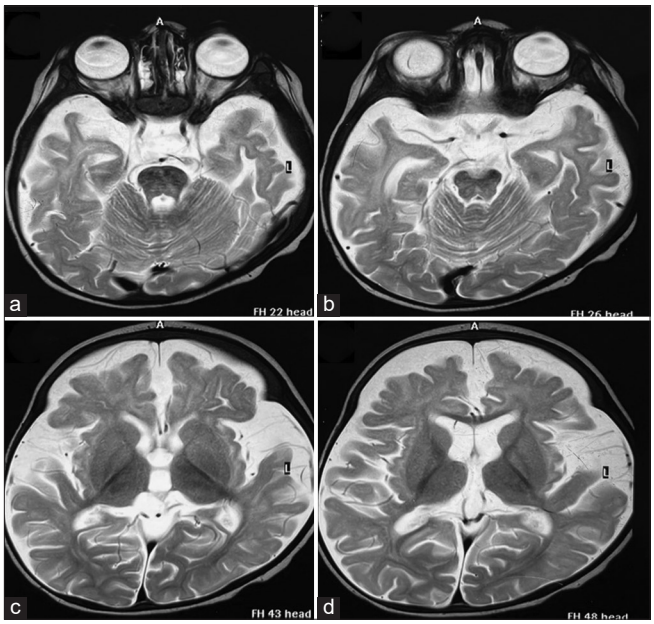

Figure 1: (a-d) Magnetic resonance imaging brain-T2W axial sections at 1 year of age show diffuse cerebral atrophy, widened Sylvian fissure with prominent subdural spaces.

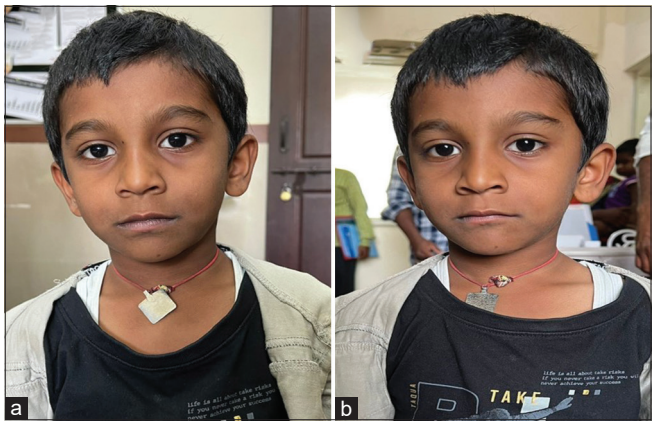

Figure 2: (a and b) Clinical photograph of a child at 6 years of age showing a bright, active and alert child with normal hair and skin.

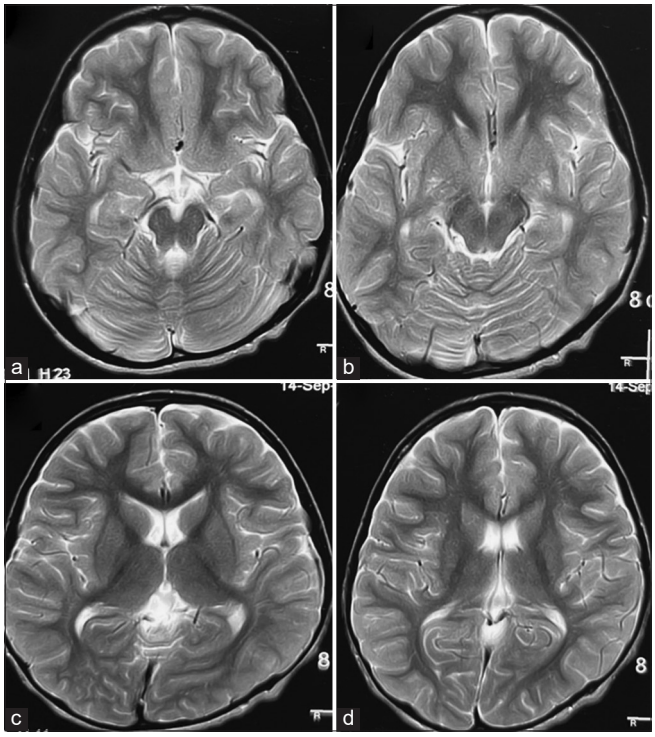

Figure 3: (a-d) Magnetic resonance imaging of the brain-T2W axial sections at 6 years of age shows a complete reversal of cerebral atrophy.

of 1.62 L/μL, mean corpuscular volume of 103 fL, low Vitamin B12 of 85 (normal: 156–672 pg/mL), normal serum copper of 18 (17.50 ± 4.10 μmol/L) and ceruloplasmin of 30 (31.5 ± 8.0 mg/dL) levels. Magnetic resonance imaging (MRI) brain showed diffuse cerebral atrophy, widened Sylvian fissure with prominent subdural spaces [Figure 1a-d]. Magnetic resonance angiography showed a tortuous left middle cerebral artery. The hair examination was normal. The child was treated with injectable Vitamin B12 and showed improvement.

During the last follow-up at 6 years, he has normal motor and cognitive development, a weight of 14 kg (−2.04 WHO Z), a head circumference of 47 cm (−2.50 WHO Z), mild spasticity, and brisk deep tendon reflexes. The hair and skin changes had resolved [Figures 2a and b] with the reversal of MRI changes [Figure 3a-d].

We considered MD as a child who had a developmental delay with regression, hair changes and MRI changes; however, the hair examination was normal. The neuroimaging feature of tortuous intracranial vessels forms a typical diagnostic feature and is a handle in MD.[3]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Gowda VK, Kolli V, Benakappa A, Srinivasan VM, Shivappa SK, Benakappa N. Case series of infantile tremor syndrome in tertiary care pediatric center from Southern India. J Trop Pediatr 2018;64:284-8.
2. Danks DM, Campbell PE, Stevens BJ, Mayne V, Cartwright E. Menkes's kinky hair syndrome. An inherited defect in copper absorption with widespread effects. Pediatrics 1972;50:188-201.
3. Manara R, D'Agata L, Rocco MC, Cusmai R, Freri E, Pinelli L, *et al*. Neuroimaging changes in menkes disease, Part 1. AJNR Am J Neuroradiol 2017;38:1850-7.

How to cite this article: Gowda VK, Challa VS, Srinivasan VM. Infantile tremor syndrome masquerading as Menkes kinky hair disease with long-term follow-up. Karnataka Paediatr J 2023;38:57-8.