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Malnutrition Turned Into Menkes Disease: A Case Report

Medical Science

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*** Dr. Archan Sil**	MD Pediatrics, RMO cum Clinical Tutor , Department of Paediatric Medicine, Burdwan Medical College, District-Bardhaman, State-West Bengal, India, PIN-713104 * corresponding author
Dr. Kaustav Nayek	MD Pediatrics, Professor, Department of Paediatric Medicine, Burdwan Medical College, District-Bardhaman, State-West Bengal, India, PIN- 713104

ABSTRACT

We report a six month old male infant who was referred to our institute as a case of malnutrition. The child had recurrent episodes of convulsion, developmental delay and hypotonia. He had history of hospital admission twice before due to pneumonia and convulsion. The child had cherubic appearance with brittle, sparse, hypopigmented scalp hairs. Microscopic examination of those hairs showed pili torti (twisted pattern). MR angiography revealed tortuous cerebral blood vessels (hairpin bending). Finally, low serum copper level helped to reach the diagnosis of Menkes disease. The child was treated symptomatically.

Introduction

Menkes disease, also known as kinky hair disease is a rare X-linked recessive disorder of copper metabolism due to mutation of p ATPase 7gene. (1,2) This is a progressive neurodegenerative disease characterised by cerebral atrophy, gross psychomotor retardation, intractable seizures, generalised hypotonia. (2) Phenotypic appearance is remarkable with chubby cheeks and thin, brittle, hypopigmented hair.(3) Microscopic examination of hair reveals pili torti (twisted hair), monilethrix(varying diameter of hair shaft) and trichorrhexis nodosa (fracture of hair shaft at regular intervals). (4) Prognosis is poor. Death occurs mostly by 3-4 years of age. We report a case of a Menkes disease which was previously misdiagnosed as a case of protein energy malnutrition at multiple peripheral centres.

Case Report

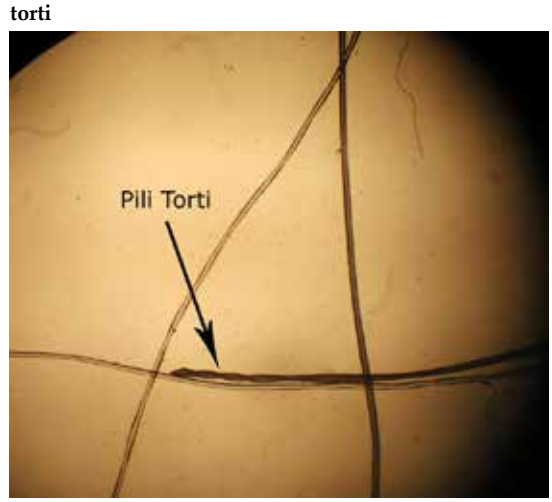
A six month old male infant was referred to our hospital as a case of protein energy malnutrition with developmental delay. The child was being treated as a case of malnutrition in the peripheral hospitals for months. On admission the child had recurrent episodes of convulsion, which were not controlled by injection phenytoin and phenobarbitone. So, the child was shifted to pediatric intensive care unit and ultimately those episodes of seizure were controlled by injection levetiracetam. The child was also admitted twice before in other hospitals. Once with episode of pneumonia and the second time with convulsion. Mother also stated that the child had delayed developmental milestones in comparison to other children of same age and sex. The boy was born at term to non-consanguineous parents. At birth, his head circumference and body weight were 34 cm and 3 kg respectively. There was history of death of one sibling at the age of 18 months.

On physical examination the child had fair complexion with chubby cheeks. The scalp hairs were light coloured, thin, brittle, woolly and sparse (Image 1). On anthropometry, weight of the child was 4.2 kg (severe undernutrition), length-60cm and head circumference-39cm. The child also had mild pallor. Neurological examination revealed generalized hypotonia. He had no eye contact and poor head control.

Image 1: showing Physical cherubic appearance with fair complexion and thin, sparse, hypopigmented hair

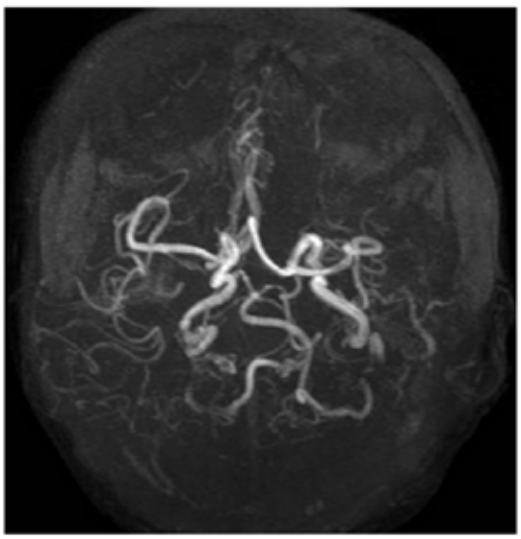
Investigations showed mild anaemia (Hb-9 gm/dl). Sepsis work up and CSF examination were normal. Skeletal survey of the long bones showed osteopenia. MRI study of the brain revealed prominent bilateral extra-axial CSF spaces. Light microscopic examination of the scalp hair showed classical sign of pili torti (twisted hair shaft) (Image 2). With high suspicion of Menkes disease serum copper level was determined and it was found to be 15 microgram/dl(ref-70-155), which confirmed our diagnosis.

Image 2: microscopic examination of hair showing pili torti



Furthermore, MR angiography revealed tortuosity of the cerebral blood vessels with hairpin bending (Image-3). Electroencephalogram showed gross, generalised polyspike waves. Due to non-availability of copper histidine the child was treated symptomatically with anticonvulsant drugs.

Image 3: MR angiography showing tortuous cerebral blood vessels



Discussion

Menkes disease (MD) was first described by Menkes as a syndrome in the year 1962.(5) Later, it was associated with copper metabolism by Drank et al. (6) The disease is caused by defect in the intracellular copper transport protein due to mutation in the p type ATPase7A gene. (7) Clinical picture is due to deficiency of key copper dependent enzymes. Management is mainly supportive with a trial of copper histidine therapy.(8)

The infants are usually asymptomatic for first 2-3 months of age. Developmental delay is the first clue which is generally detected at 3-4 months of age. This picture is followed by hypotonia, refractory seizures, failure to thrive and features of cerebral degeneration.(1) Our case presented to us at the age of 6 months. History and clinical features in our case were suggestive of classical Menkes disease.

The typical cherubic appearance with sparse, brittle, hypopigmented scalp hair is similar to reported cases in the past. One case was reported from the same institution in the year 2008.(9) The investigation finding of low serum copper, ceruloplasmin, pili torti on microscopic examination of hair and MR angiography showing hair-pin bending of cerebral blood vessels clinched our diagnosis. Due to limited facility genetic study could not be done.

The first reported case of MD with EEG changes showed multiple spike and wave activity.(10) In our case EEG also showed gross, generalised polyspike waves. We treated the infant with anticonvulsants for refractory seizures. We could not start copper histidine due to non-availability. Genetic counselling and prenatal diagnosis are helpful in prevention of this lethal disease.

The uniqueness of this case was that it was misdiagnosed as malnutrition on multiple occasions. This makes MD as one of the under-diagnosed disease entity. Prompt recognition of typical clinical features and high index of suspicion make early diagnosis possible.

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