

Case Report

“ Menkes kinky hair disease – A rare cause of focal seizures”

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Abstract

Background: Menke’s disease presents with focal seizures, abnormal facies and characteristic microscopic hair texture. **Case characteristics:** Four month old male presented with focal seizures, fair complexion and sparse silvery convoluted hairs with developmental delay. **Observation:** S. ceruloplasmin was low, metaphysical widening in long bones on X ray and pili Torti on microscopic examination of hairs. **Conclusion:** Menke’s disease is rare genetic disorder can present as focal seizures since birth and typical clinical presentation, needs high index of suspicion

Introduction

Menkes disease is a rare, lethal, X-linked recessive, multisystem disorder caused by ATP7A mutations. It has been rarely reported in India. one needs to suspect Menkes disease in a case of refractory early onset seizures especially in the presence of subtle clinical clues^[1].

Case Report

In our outpatient department came a 4 month old infant who was having simple partial seizures since last one month. As told by parents, child was second born full term male of a non consanguineous marriage with uneventful antenatal and immediate neonatal period. According to parents this baby was not in accordance with other newborns as

baby was a bit lethargic and had flabby muscles. As time passed by this baby was not able to achieve the milestones as per age as he has not attained head holding as yet , never tries to approach a red ring and no social smile and was not able to recognize parents. At three months of age this baby started having seizures which were in the form of focal twitching of facial muscles many a times confused as hiccups. For these seizures this infant was shown to various private pediatricians and also to another teaching hospital but got no relief in his seizures. Then this child came to Rama Medical College OPD. On examination child was lethargic, without any interest in its surroundings. This infant has very fair complexion, chubby cheeks, doughy skin, high arch palate and has sparse silvery

convoluted scalp hairs (figure no1). Head circumference was 40 cms and weight of the child was 6.0 kgs and length of the baby was. Developmentally child was grossly delayed. Fundus examination was normal and BERA for hearing could not be performed. This child showed no eye to eye contact and no visual fixation was observed. Systemic examination of respiratory, cardiovascular and abdomen was normal.



Fig 1: Showing sparse silvery convoluted scalp hairs and chubby cheeks with fair complexion.



Fig 2: Showing metaphyseal widening of long bones

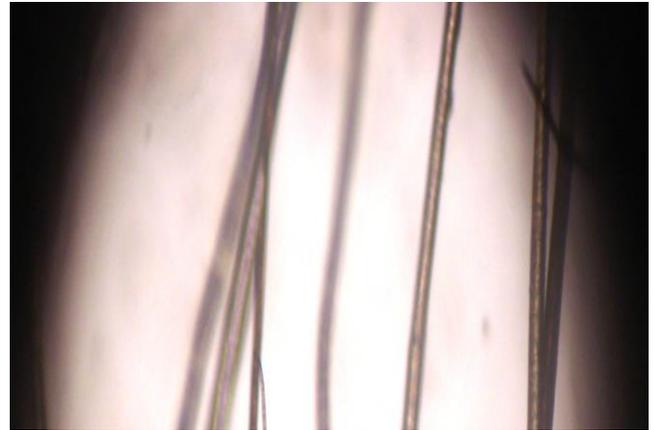


Figure 3(a): Showing pili torti in hairs on microscopic appearance



Fig 3(b): Showing pili torti on microscopic examination

We started treatment of infant with phenobarbitone and levetiracetam and started investigations for the cause of seizures. Complete blood counts, C-reactive protein, electrolytes, blood sugar, liver function test, and kidney function tests were performed, but all these investigations were normal. Keeping in mind the typical silvery sparse convoluted hair colour serum copper as well as serum ceruloplasmin was also done both of which were found very low. Serum copper levels were 19.7 µg/dl

(normal for this age being 20-70 µg/dl) and serum ceruloplasmin was 8.45mg/dl (normal range being 20.00-60.00mg/dl). Ultrasound abdomen and MRI brain with cerebral angiography was also performed. MRI and cerebral angiography was normal. Long end of bones were also seen using digital X rays which showed metaphysial widening (figure no). Therefore with typical appearance and low serum ceruloplasmin, low serum copper and metaphysial widening this child was diagnosed as having Menke Kinky Hair Disease.

Discussion

Menke Kinky hair disease is a disorder of copper metabolism which is inherited as X linked recessive trait with locus on Xq13.3, and presents at 2-3 months of age^[2]. This syndrome is rare, lethal and involvement is multisystemic. This syndrome was first described by Menkes and its association with copper was described by Drank et al later on ^[3,4,5]. In this disease although the intestinal copper intake is normal but its transport to other organs is affected. This defective copper transport is linked to the defect in the copper binding ATPase, ATP7A, which helps in distribution and metabolism of copper in tissues. Because of defective intestinal copper transport there occurs decreased levels of copper and ceruloplasmin in serum` which results in defective functioning of copper dependant mitochondrial enzymes. The enzymes which are affected include lysyl oxydase, cytochrome c oxidase, dopamine β-hydroxylase, tyrosinase, and superoxide dismutase. Tyrosinase deficiency causes depigmentation of hair and skin pallor, cytochrome c oxidase deficiency causes hypothermia , and lysyl oxidase deficiency causes tortuous arteries in brain, and progressive vascular changes predispose to

thrombosis and deficient blood supply to the developing brain^[4,6,7]. As per the literature available it is seen that there is considerable variability in the clinical presentation^[8]. Clinical presentation of Menky Kinky hair disease is with hypotonia and seizures in infancy associated with marked developmental delay. Along with this there is significant hair abnormality in this disease. Hair in this disease are sparse, depigmented, wiry and brittle. Skin is also depigmented, doughy, pale and mottled. Light microscopic examination of scalp hair shows pili torti (twisted hair), monilethrix (different diameter of hair shaft), and fracture of hair shaft at regular interval known as trichorrhexis nodosa^[9]. Baerlocher and Nadal published a paper in 1988 with the presenting signs and symptoms of 127 patients with Menkes kinky hair disease who had been reported in the medical literature before 1985^[10]. Radiologically long bones usually show osteoporosis, methaphyseal spurring, periosteal reaction and scalloping of the posterior aspects of the vertebral bodies^[11,12].

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