

CASE REPORT

Menkes disease - A rare case report

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ABSTRACT

Menkes disease (MD) is a rare, lethal multisystemic disorder of copper metabolism characterized by cerebral and cerebellar neurodegeneration, fair skin with peculiar “kinky hair” and connective tissue abnormalities. It is an x-linked recessive disorder leading to copper deficiency which usually occurs in newborn babies which can even lead to death. The main characteristic features include low serum copper, developmental delay, osteopenia, tortuous vessels on imaging, hair changes, axial hypotonia, ceruloplasmin, elevated lactate levels, and recurrent respiratory tract infection. Here, we report the case of a 10-month-old male child presented with soft skin with wrinkling of skin, difficulty in holding neck, graying of hair, seizure, and respiratory tract infection. Thereby, he was diagnosed to have MD and was treated with copper supplements. However, the patient was expired at an age of 2 years. There are reports suggest that a critical period of 2 months of neonatal life within which the treatment showed a beneficial outcome with up to 50% successful cure rates. In India, very few cases are reported about this disease, which increases the relevance of this report. Further studies regarding this disease are necessary for establishing better treatment modalities. Early diagnosis and medical management of MD is crucial to improve the survival rate of patients.

KEY WORDS: Ceruloplasmin; Hypotonia; Kinky Hair; Menkes Disease; Osteopenia

INTRODUCTION

Menkes disease (MD) is a rare, lethal multisystemic disorder of copper metabolism characterized by cerebral and cerebellar neurodegeneration, fair skin with peculiar “kinky hair” and connective tissue abnormalities. This disease is usually varies in its severity and often associated with death in early childhood.^[1] MD is most frequently seen in males.^[2] At early stage of disease, low serum copper may not be reliable indicator of the disease, but the diagnosis can be made on the basis of plasma neurochemical measurements

of deficient dopamine β -hydroxylase. Treatment with copper and gene therapy along with supportive management are the treatment options for this disease.^[3] In India, very few cases are reported about this disease, which increases the relevance of this report. Further studies regarding this disease are necessary for establishing better treatment modalities.

CASE REPORT

A 10-month-old male child with a normal antenatal history, at 1st day of life, parents noticed child had soft skin with wrinkling of skin and at 3 months of age child had difficulty in holding neck and there was graying of hair. However, he developed eye contact and social smile by 3rd month. He was presented with developmental delay with milestones of growth not achieved. Thereafter, he developed three episodes seizure. Magnetic resonance imaging brain showed ectatic and prominent right basilar artery, also multiple flow voids

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in the spinal canal anterior to the spinal cord and diagnosed to have MD. He was managed with copper supplements. He developed high-grade fever with chills and rigor also breathing difficulty for 22 days. Later, he developed moist cough associated with fast breathing and chest retractions and was started on antibiotics. However, his condition worsened with increasing requirement of oxygen. X-ray report showed bronchopneumonia and antibiotics were given but repeat X-ray showed worsening of infection. In view of that higher antibiotics along with antiviral agents such as oseltamivir were used. However, the child was tachypneic with chest signs. Routine blood investigation showed leukocytosis, raised C-reactive protein and erythrocyte sedimentation rate. Chest X-ray showed left lower zone consolidation, collapsed right lower lobe. Bronchoalveolar lavage culture was sent and was sterile. His blood culture has grown coagulase negative staphylococci and antibiotics were given according to culture. He had recurrent desaturations and maintained on nasal oxygen. He was continued on copper injections and trihexyphenidyl was added for his dystonic posturing. Dermatology opinion was taken for his skin lesions and opined as intertrigo over neck and hands, with candidiasis over groin and managed with antifungals along with supportive care. On discharge, he is afebrile on nasogastric feeds and oxygen saturations maintained on 1 L oxygen by nasal prongs. He was discharged as per request of the parents with Injection copper histidinate and was followed up three times. He died at the age of 2 years.

DISCUSSION

Copper is a trace element which is essential in human body, which exists in two different oxidation states, Cu⁺ and Cu⁺⁺. Switching of these oxidation states is crucial in metabolic processes but can adversely affect the cellular events due to the release of free radicals. Therefore, it is important to maintain the copper levels balanced inside the body.^[4] MD is an X-linked recessive disorder leading to copper deficiency which usually occurs in newborn babies which can even lead to death.^[1,5] The main characteristic features include low serum copper, developmental delay, osteopenia, tortuous vessels on imaging, hair changes, axial hypotonia, low ceruloplasmin, elevated lactate levels, and recurrent respiratory tract infection. In MD, lack of functioning dopamine β-hydroxylase and peptidylglycine α-amidating monooxygenase results in extrapyramidal symptoms and seizures. Dysfunctioning of superoxide dismutase makes the patient vulnerable to infections.^[6] In previous studies, it is reported respiratory infections as the major cause of severe morbidity or mortality in MD.^[7,8] In our patient, greying of hair, seizures, vessel changes, respiratory tract infections, and extrapyramidal symptoms like dystonia were present.

Standard treatment guidelines for MD include parenteral copper L-histidine because there is no clinical betterment

seen with oral treatment with copper salts.^[9] There are reports suggest that a critical time period of 2 months of neonatal life within which the treatment showed a beneficial outcome with up to 50% successful cure rates.^[10] For this patient, the diagnosis was made only after the 3rd month and treatment provided with intravenous copper L-histidine but the patient was expired at an age of 2 years. Early diagnosis and medical management of MD is crucial to improve the survival rate of patients.

CONCLUSION

In India, very few cases are reported about this disease, which increases the relevance of this report. Further studies regarding this disease are necessary for establishing better treatment modalities. Early diagnosis and medical management of MD is crucial to improve the survival rate of patients.

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