CASE REPORT

A case study on menke's disease, does it really exist

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ABSTRACT

Copper is the trace element which cannot be synthesized by the human body. So it is absorbed from the ingested food. The normal value of copper is 32g±21mcg/dl, while ceruloplasmin (serum ferroxidase, contains 95% of plasma copper) is 6 to 12 mg/dl. Copper helps in the formation of RBCs, keeps the blood cells, nerves, bones and immune system healthy. Copper helps in absorption of iron. This absorption is enhanced by the gene called as ATP7A. A defective gene ATP7A, impairs the transport and absorption of copper. Menke's disease is a rare X- linked recessive disorder which causes deficiency of copper levels in the body. This case study elaborates on a child with Menke's disease and its management.

Key Words: *Ceruloplasmin; Copper; ATP7A gene; Ceruloplasmin*

INTRODUCTION

A 8 year old boy K/C/O Menke's Disease with developmental delay, was now admitted with complaints of difficulty in micturition, dribbling of urine, abdominal pain [1]. He was premature baby born by 7 months LSCS, and was normal, but with milestone delay up to 9 months of age, then after he developed limb dystrophy, hypotonia, dysphasia, failure to thrive, sagging facial features, kinky and brittle hair. Blood, urine test were conducted to rule out the cause. X ray of the skull and skeleton were conducted to look for bone abnormalities [2]. A Genetic test was performed and was confirmed to have ATP7A gene defect, and was diagnosed to have MENKE'S DISEASE. The child was treated conservatively with symptomatic management and very frequently develops the Urinary Tract infection, Blood pressure elevation, and poor oral intake. Now he was treated for UTI, catheterized, on IV fluids and with antibiotics [3].

CASE STUDY

About the disease condition

Menke's disease also known as Menkes syndrome is a X – Linked recessive disorder caused due to mutation of the genes coding for copper transporting protein ATP7A leading to copper deficiency [4], characterized by sparse kinky hair, failure to gain weight, and deterioration of nervous system [5].

Epidemiology

1 in 35,000 male livebirths worldwide. Females are usually carriers.

Etiology

- Mutation of ATP7A gene.
- Point mutation and skewed inactivation of X chromosome.

Pathophysiology

ATP 7A Gene is an active Copper transporter, transmembrane protein present in the Golgi bodies at enterocytes, placenta, central nervous system except liver

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In small intestine, controls the absorption of ingested copper from blood

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in other cells, protein travels between the Golgi apparatus and the cell membranes to maintains copper concentration

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ATP7A protein helps in modifying the other proteins, including the enzymes These enzymes helps in the formation of bone, skin,, hair, nervous system, blood vessels

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Neuro denegeneration in the grey matter of the brain

- Arteries in the brain may be twisted with frayed and split inner wall
- Blockage of the arteries of the brain
- Osteoporosis (weakened bones) bone spurs
- Feeding difficulties
- Pudgy, rosy cheeks
- Irritability

Diagnostic evaluation

- Blood test- copper and ceruloplasmin levels in the blood
- Skin biopsy
- Optic microscopic examination of the hair
- Xrays of the skull and the bones of the limbs
- Urine Homovanillic acid and vanillylmandelic acid ratio
- Genetic testing

TABLE 1
Difference between Menkes and Wilson disease

Parameter's	Menkes disease	Wilson disease
Cause	Copper deficiency	Copper overload
Inheritance	X linked recessive	Autosomal recessive
	ATP7A loss of function in the enterocytes, BBB	ATP7B loss of function in the hepatocytes
Age of onset	1-5 months	20 to 30 years
Presentation	Neurodegeneration(seizures)	Neurodegeneration (ataxia, dystonia)\
	Connective tissue disorder such as kinky, sparse hair	Hepatitis, liver failure
	Lack of pigment	Psychiatric symptoms such as cognitive disorders and Psychosis
	Hypothermia	

Management

Copper supplements with acetate or glycinate at earlier periods Symptomatic treatments (Table 1):

- Pain medications
- Anti- seizure drugs
- Feeding tube
- Physical and occupational therapy.

DISCUSSION

Copper helps in the formation of RBCs, keeps the blood cells, nerves, bones and immune system healthy. Copper helps in absorption of iron. This absorption is enhanced by the gene called as ATP7A. A defective gene ATP7A, impairs the transport and absorption of copper. Menke's disease is a rare X- linked recessive disorder which causes deficiency of copper levels in the body.

CONCLUSION

Menke's disease is an under diagnosed entity, being familiar with its manifestations are essential for its earlier detection and prompt treatment.

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