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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 12mg/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.

KEYWORDS

Ceruloplasmin, Copper, ATP7A gene and X linked recessive disorder.

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INTRODUCTION

Case history

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. 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. 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.

About the disease condition Definition

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, characterized by sparse kinky hair, failure to gain weight, and deterioration of nervous system.

Epidemiology

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

Etiology

Mutation of ATP7Agene.

Point mutation and skewed in activation of Xchromosome.

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.

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.

Management

Copper supplements with acetate or glycinate at earlier periods Symptomatic treatments:

Pain medications

Anti-seizure drugs

Naso gastric tube feeding to meet the nutritional needs and to prevent aspiration.

Physical and occupational therapy.

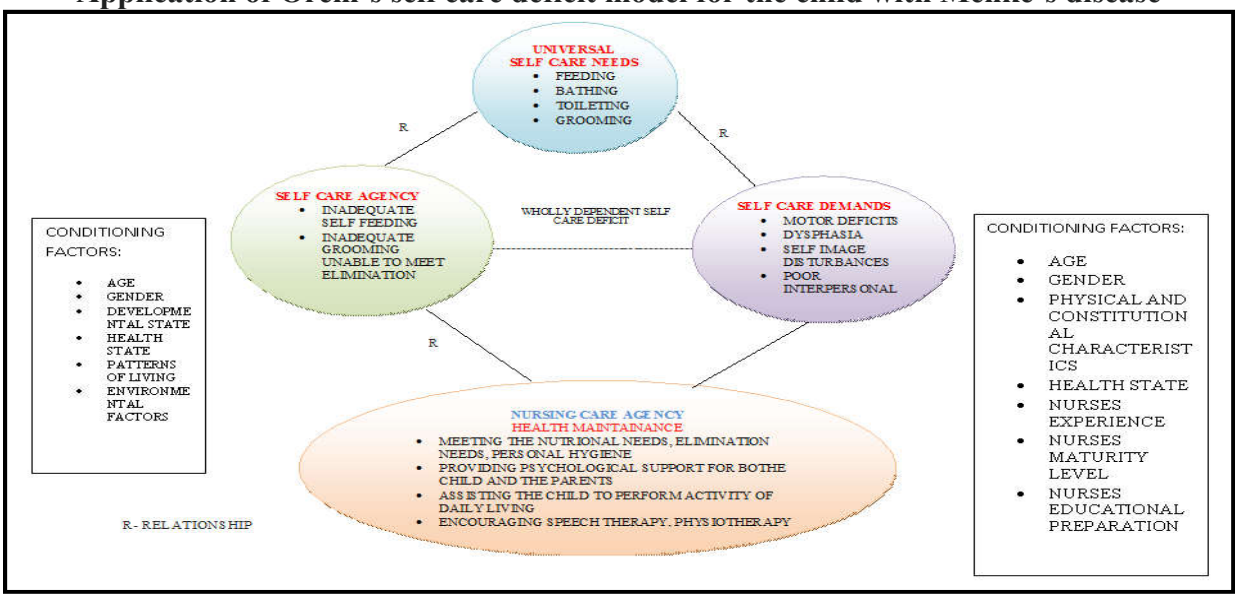
Nursing management

Nursing care is the utmost entity for the child with Menkes Disease. As the child has self care deficit. The nursing management can be incorporated by applying Orem's Self Care Deficit Model.

Difference between Menkes and wilson disease

S.No	Symptoms	Menkes disease	Wilson disease
1	Cause	Copper deficiency	Copper overload
2	Inheritance	X linked recessive	Autosomal recessive
3	-	ATP7A loss of function in the enterocytes, BBB	ATP7B loss of function in the hepatocytes
4	Age of onset	1-5 months	20 to 30 years
5	Presentation	Neurodegeneration (seizures) Connective tissue disorder such as kinky, sparse hair Lack of pigment hypothermia	Neurodegeneration (ataxia, dystonia)\ Hepatitis, liver failure Psychiatric symptoms such as cognitive disorders and Psychosis

Application of Orem's self care deficit model for the child with Menke's disease



CONCLUSION

Menke's disease is an under diagnosed entity, being familiar with its manifestations are essential for its earlier detection and prompt treatment. As the child with Menkes disease is a special child with wholly dependent self care deficit, the nurses has to be well equipped in handing the special child and help the child to lead the normal life.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

BIBLIOGRAPHY

1. Barnes N, Tsivkovskii R, Tsivkovskaia N, Lutsenko S. The copper – transporting ATPases, Menkes and Wilson disease proteins, have distinct roles in adults and developing cerebellum, *J Biol Chem,* 280(10), 2005, 9640-9645.
2. Bertini I, Rosato A. Menkes disease, *Cell Mol Life Sci,* 65(1), 2008, 89-91.
3. Ojha R, Prasad A N. Menkes disease: What a multidisciplinary approach can do, *J Multidiscip Healthc,* 9, 2016, 371-385.
4. Kaler S G, Ferreira C R, Yam L S. Estimated birth prevalence of Menkes disease and ATP7A-related disorders based on the Genome Aggregation Database (gnomAD), *Mol Genet Metab Rep,* 24, 2020, 100602.
5. Kaler S G, Holmes C S, Goldstein D S, Tang J, Godwin S C, Donsante A, Liew C J, Sato S, Patronas N. Neonatal diagnosis and treatment of Menkes disease, *N Engl J Med,* 358(6), 2008, 605-614.

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