The Journal of Nursing Trendz
Vol. 10, Issue 1, Jan - April 2019, pp-32-35

IndianJournals.com
DOI: 10.5958/2249-3190.2019.00008.7

MENKE'S KINKY HAIR DISEASE.

Tibin J., Lecturer, Dept. of Pediatric & Neonatal Nursing,
Dilla University Health & Science College, Dilla University, Ethiopia, East Africa.

Definition

Menke's disease is an X-linked lethal multi system disorder caused by alteration of copper distribution in different tissues due to genetic mutation of p ATPase7 gene.

Other names

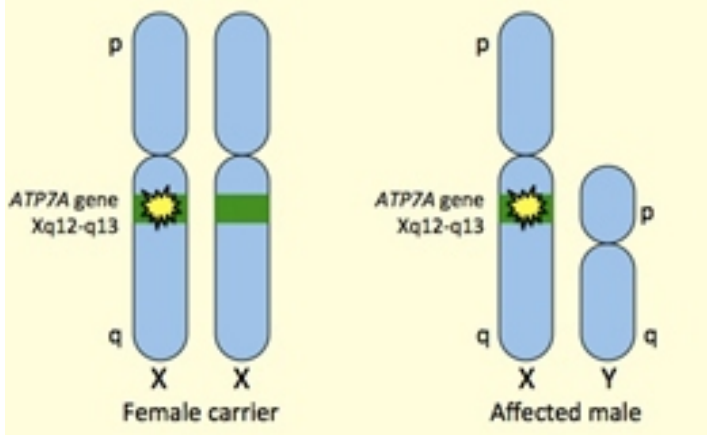
- Copper transport disease.
- Hypocupremia, Congenital.
- Kinky Hair Syndrome.
- Menkea syndrome.
- Menkes Disease.
- MK.
- MNK.
- Steely Hair Syndrome.
- X-linked copper deficiency.

Incidence

- Male infants are primarily affected, in rare cases female infants can also be affected.
- Females are carriers of the mutated gene.
- 30% of cases are spontaneous genetic mutations; 70% genetically inherited.
- Disease occurs in 1 in 100,000 to 250,000 live births.

Causes

This is caused by a genetic defect in the ATP7A gene. The ATP7A gene provides instructions for making a protein that is important for regulating copper levels in the body.

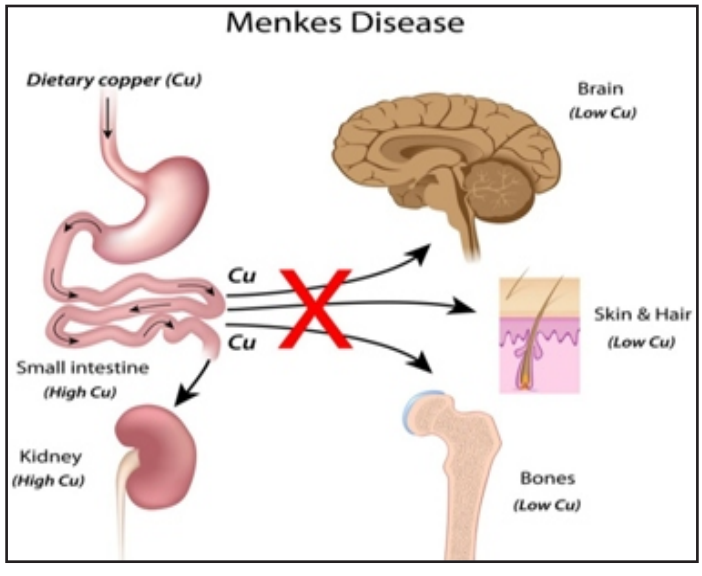


Pathophysiology

- Mutations in the ATP7A gene result in poor distribution of copper to the body's cells.
- Copper accumulates in some tissues, such as the small intestine and kidneys, while the brain and other tissues have unusually low levels of copper.
- The decreased supply of copper can reduce the activity of numerous copper-containing enzymes that are necessary for the structure and function of bone, skin, hair, blood vessels, and the nervous system.
- The defective protein is a copper-binding ATPase, ATP7A, responsible for

distribution and metabolism of copper in tissues. A defect in intestinal copper transport with associated low serum copper and ceruloplasmin levels results in defective functioning of copper-dependent enzymes like lysyloxydase, cytochrome c oxidase, dopamine β-hydroxylase, tyrosinase, and superoxide dismutase with subsequent clinical manifestations. Depigmentation of hair and skin pallor are due to tyrosinase deficiency, hypothermia is due to cytochrome c oxidase deficiency, and lysyl oxidase deficiency causes tortuous arteries in brain, and progressive vascular changes predispose to thrombosis and deficient blood supply to the developing brain.

Fig 1. Abnormal distribution of copper.



Clinical manifestation

- Developmental delay.
- Seizure.
- Weak and "floppy" muscle.
- Failure to thrive.
- Kinky hair (short, sparse, coarse and twisted hair, usually white or gray in color.)
- Pudgy, rosy cheeks.
- Irritability.
- Feeding problems.
- Hypothermia.
- Pale, pigmented skin color.
- Sagging facial muscles
- Frequent urinary tract infections (UTIs).
- Weak and/or brittle bones.
- Respiratory problems.

Diagnosis

Early diagnostic measures:

- Genetic testing of mother will show a mutation in the ATP7A gene.
- Urine homovanillic acid/vanillylmandelic acid ratio has been proposed as a screening tool to support earlier detection.

Final diagnostic measures:

- **Blood tests** will reveal low serum copper (54 µg/dl)(ref. 70-155) and ceruoplasmin levels (8.3 mg/dl)(ref. 187-320 mg/l)
- **Copper level** in placenta will be high and can be used to diagnose newborns.
- Abnormal level of **catechol** in blood and cerebrospinal fluid, even in a newborn

- **Skin biopsy** will test copper metabolism.
- **Ultrasound** may show bladder diverticula or "out pouching" in most cases and thickening of aortic valve will occur in severe case.
- **X-ray** of skeleton/skull indicates wormian-twisted bones or metaphyseal widening, metaphyseal widening of the femur and ribs, tibial and femoral spurs, wormian bones
- **MRI scan** shows hyper intensity of the basal ganglia similar to that of chronic hepatic encephalopathy.
- **MR angiography** reveals tortuosity of the cerebral vessels with hairpin like bending.
- **Light microscopic examination** of the scalp hair showed pili torti (twisted hair shafts).

Treatment

Medical management

- **Copper supplements***:* Subcutaneous or intravenous injections of copper supplements (in the form of acetate salts) can be administered.
- **Antioxidant therapy:** Vitamin E supplements has also been suggested as therapy for individuals with Menkes kinky hair disease, presumably for its antioxidant property, which may reduce the effects of Cu/Zn2OD deficiency.
- **Diet:** Formula supplements (Polycose or MCT oil) or by emphasizing high-calorie foods, such as cheese and yogurt can be given to improve the nutritional status and weight of the child.

Surgical management

Certain surgical situations commonly arise in patients with Menkes kinky hair disease.

- ***Myringotomy*** tubes are needed for chronic otitis media.
- ***Gastrostomy*** tube placement is required for feeding problems.

Nursing Management

- Counsel the parents.
- Rewarm the child.
- Instruct the parents to give copper rich foods. The best dietary sources include seafood (especially shellfish), organ meats (e.g., liver), whole grains, legumes (e.g., beans and lentils) and chocolate. Nuts, including peanuts, pistachios and pecans, are especially rich in copper, as are grains such as wheat and rye, and several fruits including lemons and raisins. Other food sources that contain copper include cereals, potatoes, peas, red meat, mushrooms, some dark green leafy vegetables (such as kale), and fruits (coconuts, papaya and apples). Tea, rice and chicken are relatively low in copper, but can provide a reasonable amount of copper when they are consumed in significant amounts.
- In case of seizure
  a) Ensure safety and initiate seizure precautions for patients at-risk for seizures. This includes having suction set up and working, having an ambu-bag in the room, padding side rails, do not restrain them or putting anything in their mouth if a seizure occurs, and keeping all side rails up, and so forth.
  b) Maintain airway.
  c) Assess, monitor and document seizure activity.
  d) Administer antiepileptics (PRN and scheduled) medications per orders.
  e) Re-evaluate any medications that may lower the seizure threshold (some antibiotics, antidepressants, narcotics, and many more may do this).
  f) Educate patient and family on hospital procedures, and when to notify staff
  g) Provide emotional support.

Prognosis

Most of the children with this condition die within the first few years of life. However, symptomatic management will help the children to live up to 5 years.

Complications

- Seizures.
- Death.

References

- Donsante, A., Tang, J., Godwin, S. C., Holmes, C. S., Goldstein, D. S., Bassuk, A., Kaler, S. G. (2007) Differences in ATP7A gene expression underlie intrafamilial variability in Menkes disease/occipital horn syndrome. *J Med Genet.*;44(8):492-7.
- Bierings, M., Clayton, P., Houwen, R. (2012) Disorders in the Transport of Copper, Iron, Magnesium, Manganese, Selenium, and Zinc. In: *Saudubray J-M, van den Berghe G, Walter JH, eds. Inborn Metabolic Diseases: Diagnosis and Treatment* . 5th ed., New York, NY: Springer.
- Sanjiv, V. Choudhary, Rutuja, W. Gadegone, and Sankha, Koley (2012) Menkes Kinky Hair Disease. *Indian J Dermatol*. Sep-Oct; 57(5): 407–409.doi: 10.4103/0019-5154.100503**,** PMCID: PMC3482811.

STUDY IDENTIFIES HOSPITAL PLUMBING AS RESERVOIR FOR SPHINGOMONAS INFECTIONS.

A study published in Dec. 27, 2018 in the New England Journal of Medicine has identified Sphingomonas koreensis, a rare opportunistic waterborne pathogen in a group of healthcare-associated infections in a hospital. Two isolates of S. Koreensis obtained from the six patients identified in the 2016 cluster were unrelated, but four isolates shared more than 99.92% genetic similarity and were resistant to multiple antibiotic agents. The reservoir was found in the hospital plumbing.

(eMediNexus, 27 December 2018.)

"Success is not final, failure is not fatal:
it is the courage to continue that counts."

- Winston S. Churchill