

# Menkes Syndrome (MNK): Know Everything About The Disorder That Affects Copper Levels In Body

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What are the symptoms of menkes disease? What are the alternative medicines and what it leads to when untreated? Here are all FAQs answered....

**OTHER DISEASES**  
Written by: Tavishi Dogra  
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**What is Menkes Syndrome?** Menkes disease is also called copper transport disease, kinky hair, trichothiodystrophy or x-linked copper deficiency. It is caused by a variation in the ATP7A gene, responsible for copper transport throughout the body. Many enzymes require copper for their normal functioning. Due to copper deficiency, these enzymes do not work naturally in Menkes disease, which ultimately affects hair, brain, bones, liver and arteries. Menkes syndrome is a rare inherited disorder affecting the male child below one year of age. Abnormal copper handling in the body results in low copper levels. **What are the causes behind Menkes Syndrome?** Copper is necessary for various cellular functions, but it is toxic when present in excessive amounts. Mutations in the ATP7A gene result in a poor distribution of copper to the body's cells.

**What are the symptoms and types of Menkes disease?**  
*Dr Advait Prakash Kulkarni, Consultant – Neurologist, Columbia Asia Hospital –Sajapur Road shares the following symptoms that indicate Menkes disease.*

- Low body temperature
- Developmental delay
- Intellectual disability
- Sagging facial skin
- Feeding difficulties
- Skeletal changes
- Failure to thrive
- Rosy cheeks
- Osteoporosis
- Tangled hair
- Hypotonia
- Seizures



**There are two types of Menkes disease-**

**Classical type** (As described above)  
**Occipital horn syndrome:** It is a milder form of Menkes syndrome with less severe neurological involvement. It is usually diagnosed often at five years of age. It is a lethal disease, leading to death by the time male child is 3-4 years of age. Death usually happens due to pneumonia of infection. There is no cure for Menkes disease.

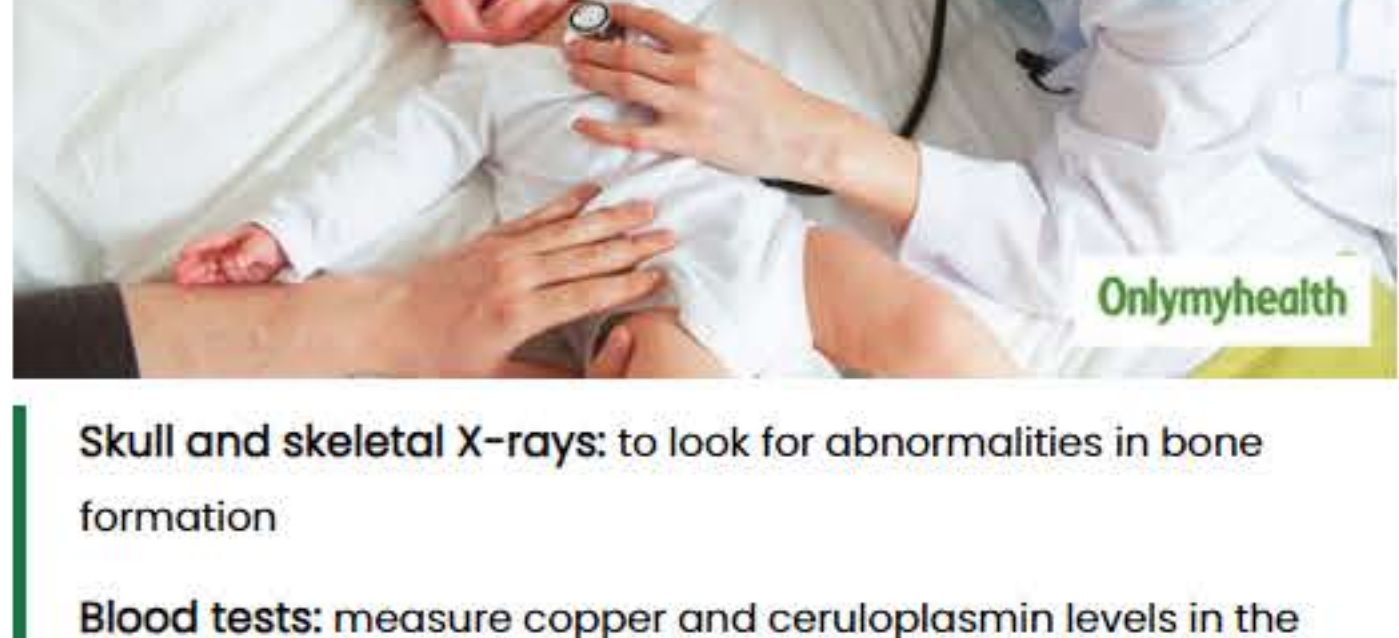
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**What is the diagnosis for Menkes Syndrome?**  
The diagnosis is usually suspected when infants exhibit typical neurologic changes and concomitant characteristic changes of the hair (short, sparse, coarse, twisted, and often lightly pigmented). **Diagnosis is confirmed by genetic testing.** Other types of tests that may help include analysis of catecholamines (chemicals sensitive to copper) and copper levels in the blood. Lack of metabolic use of congenital copper can be used for prenatal diagnosis of disease, determination of copper concentration in skin fibroblasts and amniotic fluid cell culture. **Differential diagnosis:**

- Skeletal copper deficiency:** Skeletal changes are related to collagen synthesis defects, similar to scurvy, as both cause abnormal collagen and bone lesions.
- Phenylketonuria:** This disease should be distinguished from phenylketonuria.

**What are the procedures and laboratory tests for Menkes disease?**

*Dr Advait Prakash Kulkarni shares the following procedures and laboratory tests are used to detect Menkes disease:*



- Skull and skeletal X-rays:** to look for abnormalities in bone formation
- Blood tests:** measure copper and ceruloplasmin levels in the blood
- Skin biopsy:** examining the symptoms of Menkes disease

**Symptoms that indicate a complication of MENKES SYNDROME**

*Dr Dhanashree Peddawad, Consultant Neurologist, Jupiter Hospital, Pune, shares signs of this disease.*

The patient also shows progressive neurological worsening, which begins in the first year of life. Infants present with seizures, decreased muscle tone and developmental delay.  
The affected infant may also have jaundice, lower body temperature, osteoporosis (reduced bone density), bone fracture.  
The affected child will be brittle and silvery hair. Their skin appearance is doughy due to connective disturbance.  
Brain imaging shows cerebral and cerebellar atrophy with white matter changes.  
Cerebral blood vessels are usually tortuous and elongated.

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**What is an alternative medicine for the treatment of Menkes disease?**

*The following alternative therapies and treatments are known to help treat or manage Menkes disease:*

- Physical therapy reduces signs and symptoms of Menkes disease.
- Occupational Therapy: Improving Functional Efficiency



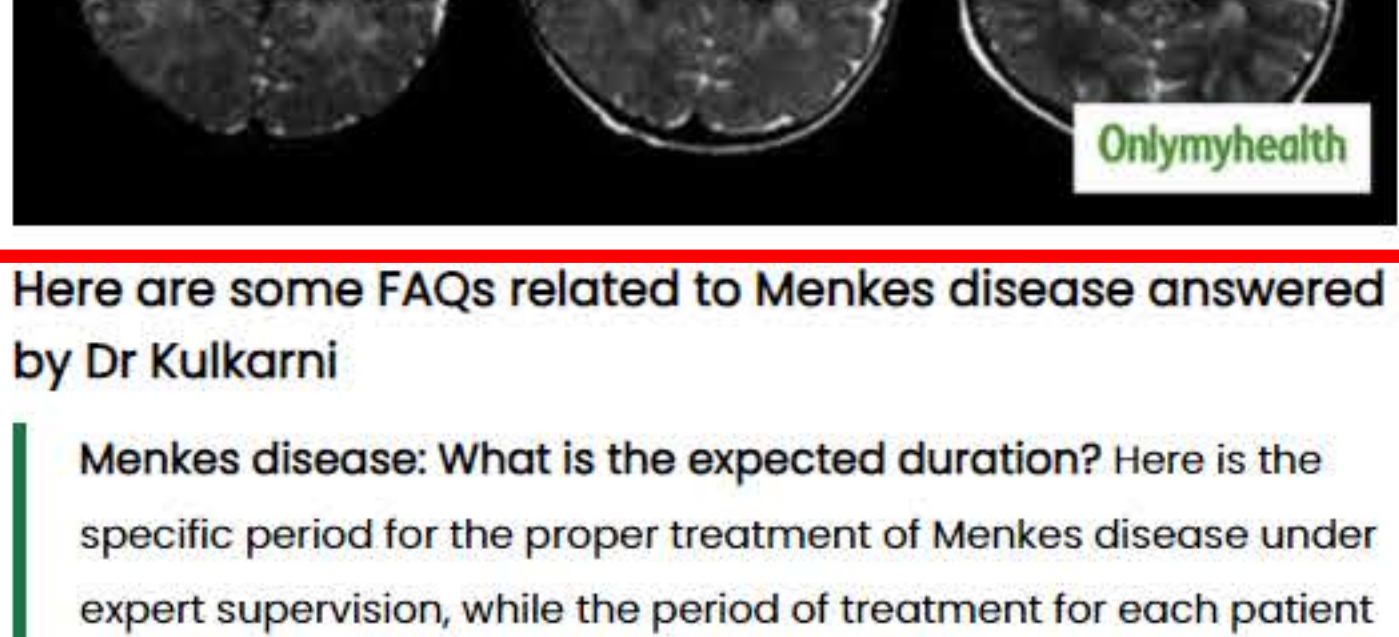
**What problems can Menkes disease cause if untreated?**  
Yes, Menkes disease causes complications if it is not treated. *The list below is of difficulties and problems that can arise from leaving Menkes disease untreated:*

- Cerebral artery thrombosis
- Cerebral artery thrombosis
- Osteoporosis
- Can be fatal

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## Menkes Syndrome: Know How It Affects Your Mental Health?

**Dr Malini Saba, a Psychologist,** says that "Menkes disease or the defect in the ATP7A gene affects mental, physical and chemical wellbeing of the whole body; it is more common in male infants and those with a family history. Menkes syndrome creates learning, mood, anxiety and emotional problems, ADHD, memory loss, schizophrenia and others. There is no recognised method to stop Menkes syndrome, but the condition could be managed by regular counselling and therapies. If there is a family history of the disorder, one needs to get proper counselling when deciding whether to have children."



**Here are some FAQs related to Menkes disease answered by Dr Kulkarni**

**Menkes disease: What is the expected duration?** Here is the specific period for the proper treatment of Menkes disease under expert supervision, while the period of treatment for each patient may vary: The condition cannot be treated but is only maintained, or the effect is minimal.

**Menkes Syndrome: When should one seek medical advice?** Seek medical advice in case of dry skin in newborn and abnormal hair that is often brittle, tangled, sparse, or kinky and is usually white ivory or grey

**Menkes disease: What is the recovery rate?** The following actions may help Menkes disease: Genetic counselling: beneficial for affected individuals and their families.

**How to prevent Menkes Syndrome?** Menkes syndrome may be controlled by genetic counselling and avoiding marriage with close relatives (consanguineous marriage).

**Doctor for diagnosis of Menkes disease:** Patients should visit the following specialists if they have symptoms of Menkes disease: Endocrinologist and Neurologist

**The total number of cases:** The following are the number of cases of Menkes disease seen worldwide every year: Rare between 10 - 50 patients.

**Who is at risk of developing MENKES Syndrome?** Males are more frequently affected and are at higher risk than women.

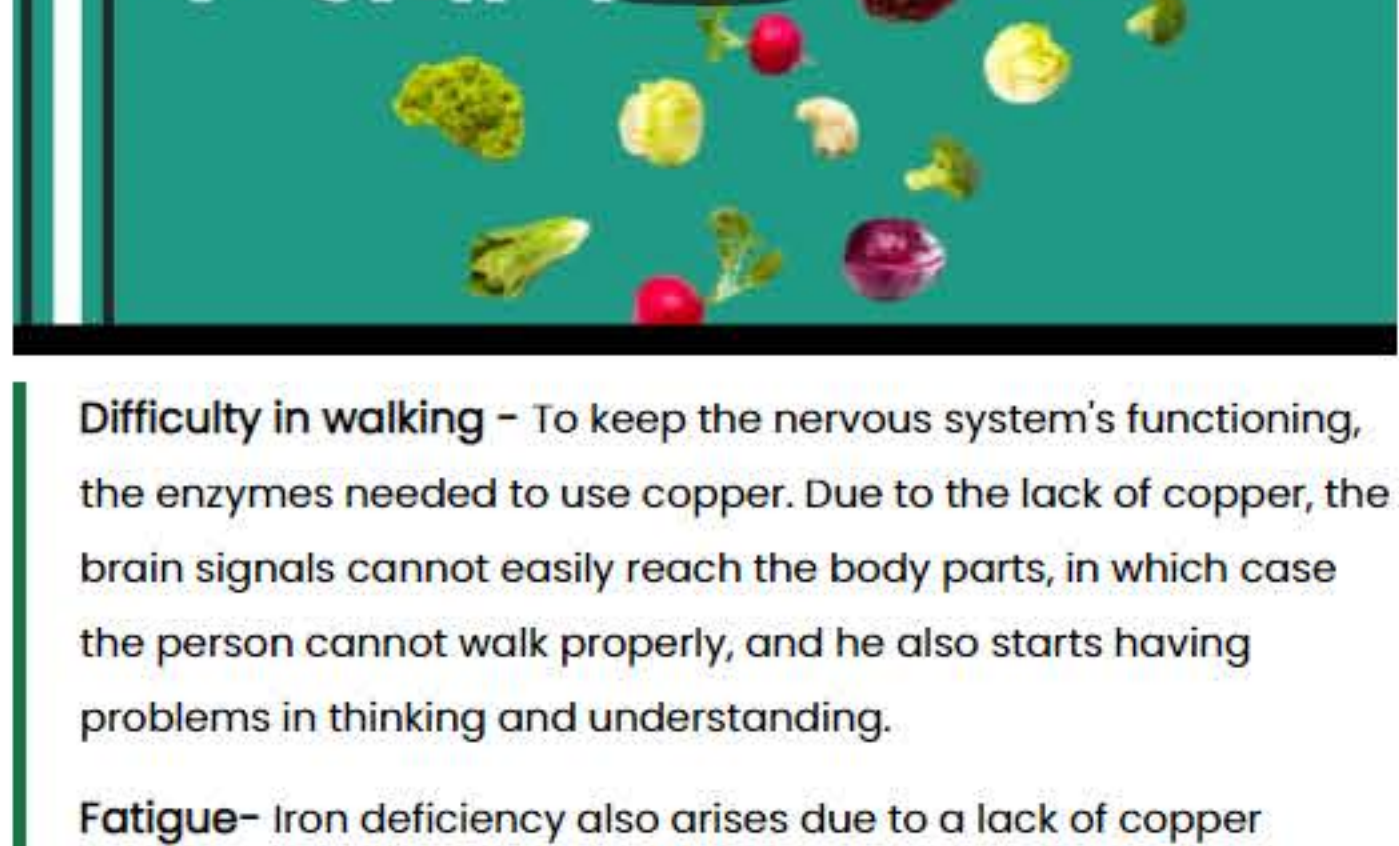
**What is the general age group?** Most Menkes disease occurs in the following age groups: At birth.

**What is th typical gender?** Menkes disease can occur in any gender.

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### Conclusion

Since it is complicated to determine whether your body is deficient in copper or not, people suffering from Menkes disease and are undergoing treatment have a regular checkup of their urine. So that it can be accurately ascertained how much copper is coming out of the urine. This is the easiest and most important test to check the amount of copper in the body. Due to the medicines being taken to treat this disease, copper and protein in the form of urine are released out of the body. Based on the urine copper and urine protein test, the doctor determines the amount of medicine to be given to the patient so that the amount of copper in the body is controlled. **Signs of copper deficiency:**



**Difficulty in walking** – To keep the nervous system's functioning, the enzymes needed to use copper. Due to the lack of copper, the brain signals cannot easily reach the body parts, in which case the person cannot walk properly, and he also starts having problems in thinking and understanding.

**Fatigue**- Iron deficiency also arises due to a lack of copper because copper is also necessary to absorb iron. Therefore, copper deficiency can cause anaemia, which makes you feel tired all the time.

**Hair whitening**- Melanin affects not only the skin but also the hair colour. Copper is also an essential element for secretion from melanin, so hair starts to turn white due to a lack of copper.

**The weakness of bones**- Not only calcium but lack of copper can be also harmful to bones. Due to the lack of copper, bone tissue is weakened, which can lead to osteoporosis problem.

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