# WILSON DISEASE – FREQUENTLY ASKED QUESTIONS

## What is Wilson disease?

Wilson disease is a genetic disorder in which copper does not get excreted from the body. This excess copper builds up in the liver, brain, eyes, and other organs. If left untreated, Wilson disease can cause severe brain damage and neurological symptoms and liver failure.

## What are the symptoms of Wilson disease?

The presentation may occur from 3 to 55 years of age. It is suspected when a young patient presents with acute liver failure or cirrhosis.

Symptoms of Wilson disease depend on the predominantly affected organ and include:

- Recurrent jaundice,
- Abdominal distension (fluid in the abdomen)
- Swelling of feet
- Vomiting of blood or passing black stools
- Nose bleeds
- Easy bruisaibility
- Fatigue
- Anemia
- Bone or joint pains
- Deteriorating school or college performance
- Change in handwriting
- Speech and language problem
- Tremors and muscle weakness
- Psychiatric problems

## What causes Wilson disease?

Wilson disease is a genetic disorder due to mutation of the ATP7B gene. More than 300 mutations have been identified. Wilson disease is an autosomal recessive trait that means the affected individual must receive two copies of an abnormal gene one from each parent, both of whom are carriers of the disease.

## How is Wilson disease diagnosed?

- Eye examination showing corneal Kayser-Fleischer rings, golden brown rings seen at the edge of the cornea with help of a slit lamp
- Patients may have cataracts also
- Blood test showing decreased level of serum ceruloplasmin (a copper binding protein)
- Increase of copper content in the urine
- Liver biopsy for copper content in the liver
- Genetic testing is helpful for screening patients' family members if there is a known patient of Wilson disease in the family
- MRI of the brain may show copper deposits

Not all tests may be positive in all patients, but a family history of a close relative having Wilson disease or symptoms suggestive of Wilson disease is extremely helpful to make a diagnosis.

## How is Wilson disease treated?

- The treatment includes removal of excess copper, reduction of copper intake and to treat any liver or central nervous system damage.
- D-penicillamine or trientine hydrochloride help remove copper from tissue.
- In patients with neurological disease the dose should be gradually increased as these drugs can cause temporary worsening of the disease
- Zinc acetate interferes with copper absorption but is slower in action. It is inexpensive and is used as maintenance treatment or in treating patients who have been diagnosed with the disease but have no symptoms (pre-symptomatic)
- Patients will also need to follow a low-copper diet, which means avoiding nuts, chocolates, dried fruit, shellfish, organ meats and mushrooms
- Liver transplantation is needed in patients with acute liver failure due to Wilson disease.
- Patients with primarily neurological disease need aggressive physiotherapy and speech therapy.
- Early detection and lifelong treatment can ensure patient maintains a completely normal health.
- Psychological counselling may be needed
- Patients need to avoid alcohol and get vaccinated for Hepatitis A and B.

## How are patients monitored on treatment?

Those of d-penicillamine or trientene are monitored by 24 hour urine copper and protein, complete blood count, free copper (derived from serum copper and serum ceruloplasmin)

## Who is at risk for Wilson Disease?

All siblings and children and close family members of Wilson disease patients should be tested for the condition.

## Wilson disease in India

- Though the exact frequency is not known, the disease is not uncommon. Marriage within families (consanguinity) still takes place in certain communities and should be strongly discouraged.
- Some common mutations have been described in the Indian population but these cannot be used to conclusively to diagnose Wilson disease in the general population
- All tests for diagnosis are available including estimation of liver copper
- Free copper estimation is not very reliable given the methods available for ceruloplasmin estimation
- Trientene is not freely available and is almost 10 times more expensive than d-penicillamine

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