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Original Research Paper

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CASE REPORT ON WILSON DISEASE

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ABSTRACT

Wilson Disease (WD) is a rare autosomal recessive disease resulting in a systemic overload of copper. Wilson's disease is a rare inherited disorder that causes copper to accumulate in your liver, brain and other vital organs. Most people with Wilson's disease are diagnosed between the ages of 5 and 35, but it can affect younger and older people, as well. A 12 years old boy residing in community, her mother reported with the chief complaints of his son with poor academic performance, decreased concentration and lack of interest in outdoor activities, loss of appetite and later they have consulted the doctor and after the investigation he was diagnosed as an Wilson`s disease.

KEYWORDS : Wilson disease

INTRODUCTION

Copper plays a key role in the development of healthy nerves, bones, collagen and the skin pigment melanin. Normally, copper is absorbed from your food, and excess is excreted through a substance produced in your liver (bile).But in people with Wilson's disease, copper isn't eliminated properly and instead accumulates, possibly to a life-threatening level. When diagnosed early, Wilson's disease is treatable, and many people with the disorder live normal lives.

Wilson disease is a rare disorder that affects males and females in equal numbers. The disease is found in all races and ethnic groups. Although estimates vary, it is believed that Wilson's disease occurs in approximately one in 30,000 to 40,000 people worldwide. Approximately one in 90 people may be carriers of the disease gene. Although only about 2,000-3,000 cases have been diagnosed in the United States, other affected individuals may be misdiagnosed with other neurological, liver or psychiatric disorders. According to one estimate, there may actually be 9,000 people affected by Wilson's disease in the United States.

Liver disease is typically the initial feature of Wilson disease in affected children and young adults; individuals diagnosed at an older age usually do not have symptoms of liver problems, although they may have very mild liver disease. The signs and symptoms of liver disease include yellowing of the skin or whites of the eyes (jaundice), fatigue, loss of appetite, and abdominal swelling.

Nervous system or psychiatric problems are often the initial features in individuals diagnosed in adulthood and commonly occur in young adults with Wilson disease. Signs and symptoms of these problems can include clumsiness, tremors, difficulty walking, speech problems, impaired thinking ability, depression, anxiety, and mood swings.

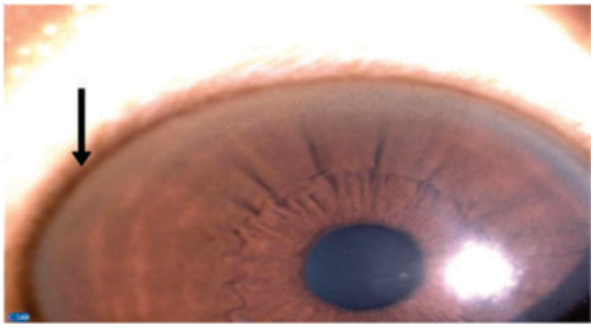
Wilson's disease is inherited as an autosomal recessive trait, which means that to develop the disease you must inherit one copy of the defective gene from each parent. If you receive only one abnormal gene, you won't become ill yourself, but you're a carrier and can pass the gene to your children.

Case Presentation

A 12 years old boy residing in community, her mother reported with the chief complaints of his son with poor academic performance, decreased concentration and lack of interest in outdoor activities, loss of appetite and so they consulted the doctor mainly for the complaints of poor academic performance, during the investigation of eye examination as well as MRI report he was diagnosed as an Wilson`s disease.

There is a history of third degree of consanguineous marriage. He was delivered by normal vaginal delivery and there was no postnatal complication and he has normal milestone development and his academic performance was normal during this early childhood period, but later his academic performance became very poor and lack of interest towards studies.

The boy was well oriented and on examination he looks dull, fatigue, inactive and thin. During the eye examination the copper rings are visible in both the eyes (figure 1) it looks like golden- brown eye discoloration.



MRI report shows the findings of bilateral lentiform nucleus and midbrain peri aqueduated region is present. Other blood investigation are done his hemoglobin level is 11.8 g/dl, TC level is 7000 cells and DC level is 54%. He is under the medication of tab. Penicitin 250 mg regularly.

He is adviced to take diet low in copper content foods in fruits and vegetables like pineapple, papaya, apple, melon, grapes. Adviced the mother to use the copper vessels for cooking and storage of water for drinking. Avoiding foods that are high in copper, such as chocolate, liver, mushrooms, nuts, and shellfish.

DISCUSSION

Wilson disease is a genetic disorder that prevents the body from removing extra copper, causing copper to build up in the liver, brain, eyes, and other organs. Without treatment, high copper levels can cause life-threatening organ damage. Wilson disease is a rare disorder that affects approximately 1 in 30,000 individuals.

Wilson Disease occurs with equal frequency in men and women. For a child to inherit it, both parents must carry and pass on a specific gene. Two abnormal genes are required for the child to have the disease. All siblings and children of Wilson disease patients should be tested for the condition. Other relatives who have had symptoms or laboratory tests that indicate liver or neurological disease should also be tested.

Wilson disease is caused by mutations in the ATP7B gene. This gene provides instructions for making a protein called copper-transporting ATPase 2, which plays a role in the transport of copper from the liver to other parts of the body. Copper is necessary for many cellular functions, but it is toxic when present in excessive amounts. The copper-transporting ATPase 2 protein is particularly important for the elimination of excess copper from the body. Mutations in the ATP7B gene prevent the transport protein from functioning properly. With a shortage of functional protein, excess copper is not removed from the body. As a result, copper accumulates to toxic levels that can damage tissues and organs, particularly the liver and brain.

Research indicates that a normal variation in the PRNP gene may modify the course of Wilson disease. The PRNP gene provides instructions for making prion protein, which is active in the brain and other tissues and appears to be involved in transporting copper. Studies have focused on the effects of a PRNP gene variation that affects position 129 of the prion protein. At this position, people can have either the protein building block (amino acid) methionine or the amino acid valine. Among people who have mutations in the ATP7B gene, it appears that having methionine instead of valine at position 129 of the prion protein is associated with delayed onset of symptoms and an increased occurrence of neurological symptoms, particularly tremors. Larger studies are needed, however, before the effects of this PRNP gene variation on Wilson disease can be established.

Children with Wilson disease are usually normal at birth and may remain healthy for a variable period of time; most cases present in the second and third decade of life. Our patient had conjugated hyperbilirubinaemia, ascites and severe dysfunction of the synthetic activity of his liver. Although he had proteinuria and generalized edema, which informed the initial diagnosis of nephrotic syndrome, the proteinuria never reached nephrotic range and the progression of the edema (ascites preceding pedal edema) did not support a renal aetiopathogenesis for the generalized edema. Indeed, it is now known that persons with Wilson disease could have some degree of proximal tubulopathy, which may be partial (as may be the case in our patient) or generalized (Fanconi syndrome).

The long-term treatment of symptomatic cases of Wilson disease entails the chronic use of copper chelators and zinc, while liver transplantation provides a cure. The copper chelators commonly used for Wilson disease are penicillamine and trientine hydrochloride.

Treatment for Wilson disease includes three types of medications. First those that remove (chelate) copper from the body by urinary excretion such as penicillamine (Cuprimine) and trientine dihydrochloride (Syprine), second, zinc salts to prevent the gut from absorbing copper from the diet, and third, tetrathiomolybdate which both prevents absorbing copper and binds up toxic copper in the blood making it nontoxic. Patients who present symptomatically with mild to moderate liver failure can be effectively treated with a combination of trientine and zinc for 4-6 months, and then go on maintenance therapy with zinc or trientine alone.

A second choice would be penicillamine and zinc, but penicillamine has more side effects than zinc. Patients with severe liver failure may require liver transplantation. Patients who present neurologically can best be treated with tetrathiomolybdate, but it is not commercially available as yet. The second choice is zinc alone. Zinc is rather slow acting but doesn't cause the drug catalyzed worsening so common with trientine and penicillamine. Trientine and penicillamine are poor choices to treat neurologically presenting patients because of the high frequency of neurological worsening, from which many patients never recover.

If the Wilson disease is untreated, it can be fatal. Serious complications include: Scarring of the liver (cirrhosis), Liver failure. Persistent neurological problems, kidney problems, psychological problems and blood problems.

Wilson's disease is an inherited gene that's passed down from parents to their children. If parents have a child with Wilson's disease, they could potentially have other children with the condition as well.
Although we can't prevent Wilson's disease, when it is on early onset of the condition. If we find out anyone having Wilson's disease early on, we may be able to prevent the symptoms by taking medications like zinc. A genetic specialist can help parents determine their potential risk for passing Wilson's disease to their children.

CONCLUSION

Wilson's disease tends to become progressively worse and is eventually fatal. With early detection and treatment, most of those affected can live relatively normal lives. Liver and neurologic damage that occurs prior to treatment may improve, but it is often permanent. Patients with acute liver failure due to Wilson disease should be considered for liver transplantation. Liver transplantation effectively cures this disease, with a long-term survival rate of about 80%.

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