Case Report:

Wilson's disease: Case report from Maharashtra

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ABSTRACT

Wilson's disease is an autosomal recessive disorder of copper metabolism resulting from the absence or dysfunction of a copper transporting P-type ATPase encoded on chromosome 13. Presentation in childhood may include chronic hepatitis, asymptomatic cirrhosis, or acute liver failure. In young adults, neuropsychiatric symptoms predominate and include dystonia, tremor, personality changes, and cognitive impairments secondary to copper accumulation in the central nervous system. The laboratory diagnosis of Wilson's disease is confirmed by decreased serum ceruloplasmin, increased urinary copper content, and elevated hepatic copper concentration. Molecular genetic analysis is complex as more than 100 unique mutations have been identified and most individuals are compound heterozygotes.

Keywords: Wilson's disease, copper – liver

Background:

Wilson's disease is an autosomal recessive disorder of copper metabolism resulting from the absence or dysfunction of a copper transporting P-type ATPase encoded on chromosome 13.[1] Under physiologic circumstances, biliary excretion represents the sole mechanism for copper excretion, and thus affected individuals have progressive copper accumulation in the liver. When the capacity for hepatic storage is exceeded, cell death ensues with copper release into the plasma, hemolysis, and tissue deposition. Presentation in childhood may include chronic hepatitis, asymptomatic cirrhosis, or acute liver failure. In young adults, neuropsychiatric symptoms predominate and include dystonia, tremor, personality changes, and cognitive impairments secondary to copper accumulation in the central nervous system. [2]The laboratory diagnosis of Wilson's disease is confirmed by decreased serum ceruloplasmin, increased urinary copper content, and elevated hepatic copper concentration.

Case Report:

Herewith we reported a case of 15 yr old male came with chief complaints of: Tremors in right hand, associated with slurring of speech which was insidious in onset and gradually progressed over one year.

Initially patient could perform his routine daily activity without hesitation, but in the next 6 months subsequently patient found difficulty in bringing the food to mouth with the right hand. There was also h/o spilling of water from glass while bringing to mouth with right hand. Patient could not button and unbutton his shirt himself. Patient also complained of difficulty in speaking, non- fluently with breaking of sentence into 2-3 words.

No history of limb weakness, headache, seizures, vomiting.

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No history of difficulty in swallowing, chewing, drinking water.

No history of diplopia, blurring of vision .

No history of chest pain, dyspnea, palpitations.

Patient also had history of road traffic accident in past but no history of head injury, unconsciousness or bleed from ear or nose.

No past history of any major illness or any major surgery.

Birth history and vaccination history is normal.

On general examination: Pulse – 80/min, BP - 110/70 mm Hg. No evidence of pallor, icterus, clubbing, cyanosis, edema or lymphadenopathy.

Per abdomen soft and showed no organomegaly.

Cardio vascular examination showed normal heart sounds with no murmur. Respiratory examination was normal.

Central nervous examination revealed that patient was conscious, oriented to time, place and person, Speech – scanning in nature, Cranial nerve examination – normal, Motor system examination revealed normal nutrition, tone, power and deep tendon reflexes and bilateral flexor planter response.

Cerebellar signs:	Right	Left
Finger nose test	+++	+++
Dysdiadochokinesia	+++	+++
Heel to knee test	impaired	impaired
Romberg's test	positive.	

On Investigation Haemoglobin was 11.7 gm%, TLC – 4900/cmm, DLC – P60 L35 E02 M03, ESR 24 mm/hr, Platelet count 1.6 lakhs/cmm. PBS – normocytic, normochromic. LFT – S.bil – (T) – 0.7mg% (D) - 0.3 mg%, ALT – 24 IU /L, AST – 28IU/L, ALP- 79 IU/L, RFT-BUN – 28 mg%, S.creatinine- 1.2mg%, S.Na – 143mmol/l, S. K - 4.6 mmol/l. USG(abdomen & pelvis) – shows diffuse hepatyomegaly. Chest x-ray – PA view – normal. Following this, an MRI BRAIN was done which showed the following results: Abnormal signal intensity in thalami and gangliocapsular region bilaterally and also in the midbrain, suggestive of metabolic involvement. Following this a detailed ophhalmoscopic examination was undertaken which showed, Presence of KF (Kayser- Fleischer) ring. Fundus was within normal limits. EEG: normal.

To confirm the diagnosis of Wilson's disease, the following tests were done: 24 hr urinary copper -- 375.30 (ref range 7-12), Serum copper – 95.67 (ref range 30-150), Serum ceruloplasmin – 6.04 (ref range 20-60)

Treatment following these reports, the patient was put on T. Propranolol 20 mg BD, Cap zinc acetate (220) 2 tablets TDS and T. Pacitane 1mg TDS.

The patient was asked to follow up regularly to look for worsening or improvement of symptoms. The patient responded to treatment with regression of symptoms after 6 months of therapy.

Discussion

Wilson's disease or hepato-lenticular degeneration is an autosomal recessive genetic disorder in which copper accumulates in tissues; this manifests as neurological or psychiatric symptoms and liver disease.[1] Symptoms usually appear between the ages of 6 and 20 years, but cases in much older people have been described. The main sites of copper accumulation are the liver and the brain, and consequently liver disease and neuropsychiatric symptoms are the main features that lead to diagnosis.[3]

Some are identified only because relatives have been diagnosed with Wilson's disease; many of these, when tested, turn out to have been experiencing

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symptoms of the condition but haven't received a diagnosis. [4]

On examination, signs of chronic liver disease such as spider naevi (small distended blood vessels, usually on the chest) may be observed. Chronic active hepatitis has caused cirrhosis of the liver in most by the time they develop symptoms. While most people with cirrhosis have an increased risk of hepatocellular carcinoma (liver cancer), this risk is relatively very low in Wilson's disease.

About half the people with Wilson's disease have neurological or psychiatric symptoms. Most initially have mild cognitive deterioration and clumsiness, as well as changes in behavior. Specific neurological symptoms usually then follow, often in the form of parkinsonism (cogwheel rigidity, bradykinesia or slowed movements and a lack of balance are the most common parkinsonian features) with or without a typical hand tremor, masked facial expressions, slurred speech, ataxia (lack of coordination) or dystonia (twisting and repetitive movements of part of the body).Psychiatric problems due to Wilson's disease may include behavioral changes, depression, anxiety and psychosis.[Psychiatric symptoms are commonly seen in conjunction with neurological symptoms and are rarely manifested on their own. These symptoms are often poorly defined and can sometimes be attributed to other causes. Because of this, diagnosis of Wilson's disease is rarely made when only psychiatric symptoms are present .[5]

Eyes: Kayser–Fleischer rings (KF rings), a pathognomonic sign, may be visible in the cornea of the eyes, either directly or on slit lamp examination as deposits of copper in a ring around the cornea. [3,4]

Kidneys: renal tubular acidosis, a disorder of bicarbonate handling by the proximal tubules leads to nephrocalcinosis (calcium accumulation in the kidneys), a weakening of bones (due to calcium and phosphate loss), and occasionally aminoaciduria (loss of essential amino acids needed for protein synthesis).[5]

Heart: cardiomyopathy (weakness of the heart muscle) is a rare but recognized problem in Wilson's disease; it may lead to heart failure (fluid accumulation due to decreased pump function) and cardiac arrhythmias (episodes of irregular and/or abnormally fast or slow heart beat).[1,5]

Hormones: hypoparathyroidism (failure of the parathyroid glands leading to low calcium levels), infertility, and habitual abortion.[3]

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