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A RARE CASE OF CIRRHOSIS – WILSONS DISEASE : A CASE REPORT

KEYWORDS

Wilsons Disease , Hepatolenticular Degeneration , Nazar Prognostic Index(NPI), Ceruloplasmin, Kayser Fleischer Ring(KF)

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ABSTRACT A 22 Year Old Male patient, presented with pedal edema, facial swelling, jaundice and neuropsychiatric symptoms. On subsequent investigations a rare inherited disorder - WILSONS DISEASE or HEPATOLENTICULAR DEGENERATION was diagnosed. On medical management with Anti-copper therapy and supportive measures condition of the patient improved. Early diagnosis and early institution of Anti-copper therapy in this disorder improves the outcome with recovery of liver function, although the residual liver damage is usually present.

INTRODUCTION

Wilsons disease or hepatolenticular degeneration is an autosomal recessive metabolic disorder caused by mutations in the ATP7B GENE, a membrane Bound , copper-transporting ATPase. clinical manifestations are caused by copper toxicity due to impaired clearance from organs and primarily involve liver and the brain.Because effective treatment is available,it is important to make this diagnosis early.The frequency of wilsons disease in most population is about 1 in 30,000-40,000,and the frequency of carrriers of ATP7B mutation is 1%

CASE STUDY

A 22yr old unmarried male Non-alcoholic,
Non-smoker born to Non-consanguineous parents ,
Presented with the Swelling of legs since 2 weeks,
Facial puffiness since 1 week ,Yellowish
discoloration of eyes, palms, and urine since 1week.
No h/o dyspnoea, PND , Orthopnea, No h/o Oliguria , malena, hematemesis

In the PAST HISTORY He Had Jaundice at The Age of 10years for a Period of 1-2months(approx)which Resolved Spontaneously Without Treatment, at The Age Of 16years he Had Jaundice For The Second Time Which Resolved In 1month. He also Had h/O- Learning Difficulties, so he Discontinued Education From 8th Standard & he also had Loss of Emotional Control. He had no h/o fever , blood transfusions and drug intake . He had No siblings and no similar complaints in his family members. PHYSICAL EXAMINATION at the time of the admission revealed Icterus, B/L Pedal Edema and Puffiness of Face, no skin pigmentation with normal Vital parameters .He had no Scratch Marks & Except Jaundice other Signs of Liver Cell Failure were Absent. On Slit lamp examination of eye – Kayser Fleischer (KF) ring was seen. On abdominal examination- Spleen was palpable with no distension & tenderness. Respiratory, cardiovascular & central nervous system were normal on examination with No focal motor / sensory deficits & No involuntary movements with B/l plantars flexors.

A Provisional diagnosis of CHRONIC LIVER DISEASE with PORTAL HYPERTENSION was made with Wilson's disease or Chronic Viral hepatitis, or Autoimmune hepatitis as the probable cause .

His routine lab investigations revealed CBP : Hb -11 g/dl, WBC-4400/cu.mm, Platelet – 1.7 lac/cu.mm, CUE: colour - yellowish, no proteins, no bile salts/pigments & others normal , RBS: 130mg/dl, Blood group – A positive, LIVER FUNCTION TEST-Tot bilirubin-10mg/dl, Direct – 8 mg/dl, SGOT- 54 IU/L, SGPT – 68IU/L, ALKphosphatase–140 IU/L,Total proteins–6.3gm/dl, Albumin- 2.8 gm/dl , Globulin – 3.5 gm/dl,
PROTHROMBIN TIME –22 Seconds& INR – 2.0,
Blood urea - 18mg/dl, S.creatinine - 0.6mg/dl ,
HBsAg –NEGATIVE , ANTI-HCV –NEGATIVE,
SERUM CERULOPLASMIN–12 mg/dl↓ (18-35 mg/dl) and SERUM COPPER (TOTAL)- 40 mg/dl ↓ (80-140) with an ELEVATED 24 HOUR
URINE COPPER- 116 ug/24hrs ↑(20-50 ug) ,
serum iron, % saturation of transferrin & serum ferritin were normal, ANA & ANTI-LKM1- NEGATIVE .
CXR–was normal,
USG ABDOMEN & PELVIS - s/o CHRONIC PARENCHYMAL LIVER DISEASE with mild splenomegaly & prominent portal vein. MRI BRAIN was NORMAL.
UPPER GI ENDOSCOPY - s/o PORTAL HYPERTENSIVE GASTROPATHY with no varices.

A Diagnosis of WILSONS DISEASE was made with the clinical features s/o CHRONIC PARENCHYMAL LIVER DISEASE, NEUROPSYCHIATRIC symptoms, presence of KAYSER FLEISCHER RING with DECREASED SERUM CERULOPLASMIN, SERUM COPPER & an ELEVATED 24 HOUR URINARY COPPER LEVELS.

Nazar prognostic index score for this patient was 3 hence he was initiated on Medical Management With Anti-copper therapy - TRIENTINE & ZINC, along with supportive treatment the condition of the Patient Improved subsequently .

DISCUSSION

Wilsons disease is an autosomal recessive disorder caused by mutations in ATP7B Gene (WD Gene), located on the long arm of chromosome 13q14.1. WD gene encodes a copper-transporting P-Type ATPase which is expressed predominantly in the liver with minor expression in brain. ATP7B protein deficiency Impairs Biliary Copper Excretion, resulting in positive copper balance with hepatic accumulation & oxidant stress due to copper toxicity , leading to liver damage. This is followed by Accumulation of Free Copper in other parts of the body (e.g., in the brain, with consequent neurologic and psychiatric disease).

The clinical presentation of Wilson disease is extremely variable. The age at onset of symptoms generally ranges from 6 to about 40 years. Patients may present with chronic or fulminant liver disease, a progressive neurologic disorder without clinically prominent hepatic dysfunction, isolated acute hemolysis, or psychiatric illness. The clinical variability often makes confirmation of the diagnosis difficult.

HEPATIC PRESENTATION -

A hepatic presentation of Wilson disease is more common in children than in adults. Pts may present with a self-limited clinical

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illness - acute hepatitis or with severe, established chronic liver disease or with cirrhosis & portal hypertension.

NEUROLOGIC PRESENTATION - Patients may present with movement disorder or with rigid dystonia. About 20% of patients may present with purely psychiatric symptoms like depression, phobias, compulsive behaviour, aggressive & antisocial behaviour.

OCULAR SIGNS -
The Classic Kayser-fleischer Ring Is Caused By Copper Deposition In Descemet's Membrane Of The Cornea. KF ring is present in 30-50% patients with hepatic presentation & in 99% patients presenting with a neurologic or psychiatric presentation of WD. Some Extrahepatic disorders like Hemolytic anemia, Arthritis, Rhabdomyolysis, Pancreatitis, Nephrolithiasis, Cardiomyopathy, Hypoparathyroidism, Amenorrhea and Testicular problems & Infertility can be present.

Diagnosis of WILSONS DISEASE is done by estimating Serum ceruloplasmin and Serum copper (Levels decrease), With an elevated 24 hour urine copper. The gold standard for diagnosis remains liver biopsy with quantitative copper assays. If the condition is Untreated, progresses to cirrhosis hence Early diagnosis and early institution of Anti-copper therapy is very important. Treatment is life long with Anti-copper therapy like TRIENTINE, ZINC, TETRATHIOMOLYBDATE & PENICILLAMINE. Nazer prognostic index is used to establish disease severity in patients presenting with hepatic decompensation

- It is calculated by taking serum bilirubin, aspartate aminotransferase and prothrombin time into consideration
- Pts with scores < 7 needs medical therapy.
- Pts with scores >9 needs Liver Transplantation , & Pts with score between 7 - 9 require clinical judgment between medical therapy & liver transplantation

Conclusions
-Any person with recurrent hepatic disease and unexplained neurologic symptoms should be investigated for Wilson's disease As the Early diagnosis and early institution of anti copper therapy improves the outcome with recovery of liver function although the residual liver damage is usually unchanged.

- The most important investigation is liver biopsy with the assessment of hepatic copper.

- Genetic analysis may help in doubtful cases.

- The faster the Diagnosis & introduction of the anticopper therapy is, the better the results are.

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