ORIGINAL RESEARCH PAPER

General Medicine

A RARE CASE OF WILSON'S DISEASE

KEY WORDS:

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INTRODUCTION-:

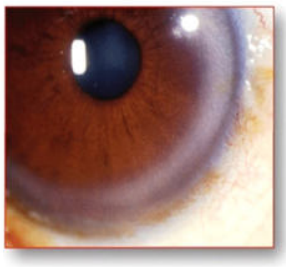
Wilson's disease is a rare inherited disorder of copper metabolism with deposition of copper in the liver, brain, and other tissues with an incidence of one in 30,000.

Initially there is deposition of the metal in the liver followed by its release into the circulation and thereafter chronic accumulation in the brain and other extra hepatic tissues. Liver cirrhosis occurs early in the disease progression In the nervous system basal ganglia and midbrain are affected most frequently. According to a German study the patients who presented in first decade show predominantly hepatic manifestations while the patients with neurological symptoms presented during the third decade.

Untreated Wilson's disease has a progressive course and may be fatal. The disease tends to be under diagnosed and timely diagnosis remains a challenge. Wilson's disease (WD), which results from the defective ATP7B protein product, is characterized by impaired copper metabolism and its clinical consequences vary from an asymptomatic state to fulminant hepatic failure, chronic liver disease with or without cirrhosis, neurological, and psychiatric manifestations.

A high grade of suspicion is warranted to not miss cases of WD, especially less florid cases with only mild elevation of transaminases, or isolated neuropsychiatric involvement.

Screening in first and second relatives of index cases is mandatory, and treatment must commence upon establishment of diagnosis



GENERAL DETAILS

CASE STUDY :- A 19 year old female, presented in medicine OPD, NCH, Surat, with the chief complaints of pain in abdomen on and off, mainly in right hypochondriac region since 1-2 months. There was no history of alcoholism, melena,, loose stools, bleeding from any site, abnormal body movements, breathlessness, vomiting. Patients vitals were with in normal limits. Respiratory examination was s/o right side lower zone air entry decreased and per abdomen was soft and non tender.

The patient was admitted for further evaluation.

Investigation

Usg (abdo.+Pelvis):
-Shrunken liver with irregular margin and altered echotexture, s/o
chronic liver parenchymal disease
-Severe ascites
Chest x-ray pa view:
-B/l cp angle blunted
Hb: 10.9 alt: 25 sgpt: 25
Pcv: 33.2 t.B.0.4 sgot: 63.45
Mcv: 67 d.B.0.2 alp: 219
Plt:96000 albumin: 2.3
Wbc: 4600 sodium: 140
M.P: no parasites potassium:3.71
Esr: 4 creatinine:0.7
Retic count: 0.8% ldh: 403
Ana screening was normal
Hiv/ hep a, e, d, b, c: non reactive
Lipid profile: normal
Stool and urine routine, micro: nad
Serum ceruloplasmin was low.
References: 1. Kaur, Harharpreet & Kaur, Kiranj

Management

Though patient didn't have any neuropsychiatric signs and symptoms, but it can progress to severe disease associated with irritability, tremors (specifically knowns as Wing beating tremors). So its very importance of early diagnosis and treatment. So Patient was immediately started on zinc and d-penicillamine therapy and has been followed back after one study.

CONCLUSIONS

As Wilson's disease is a rare disease the diagnosis is likely to be missed. There should be a high index of suspicion in all cases of liver cirrhosis with no clear cut etiology or an isolated neurological symptom such as tremor. It is also important to warn patients not to stop therapy. The patient had many unusual features which are being reported and shared for future reference.

REFERENCES:

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2. Rodriguez-Castro KI, Hevia-Urrutia FJ, Sturniolo GC. Wilson's disease: A review of what we have learned. World J Hepatol. 2015;7(29):2859-2870. doi:10.4254/wjh.v7.i29.2859