article

Wilson`s Disease : Experience at Gastrointestial Disease & Hepatology Center

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***Abstract*: This is a retrospective study included 17 patients with Wilson's disease (WD) admitted to GIT Centre over 3 Years (1994-1997). The diagnosis of WD was suspected in any young patient with chronic liver disease (CLD). The age ranged from (6-30 years) with a mean of (15.8) years, there were 10 males and 7 females, 13 patients had positive family history (FH) for WD and 13 patients had positive Kayser-Fleischer (KF) ring; one third (6 patients) had osophageal varices indicating portal hypertension (PH), jaundice was seen in eight patients (47%), over the period of the study (3/17) patients died, 2 from hepatic encephalopathy and one from uncontrollable GI bleeding. The study concludes that WD is not uncommon in IRAQ, and often presents late, family screening of index patient is extremely useful.**

ÇáÎáÇÕÉ

åÐå ÏÑÇÓÉ ÇÓÊÑÌÇÚíÉ ÔãáÊ 17 ãÑíÖ ãÕÇÈíä ÈãÑÖ æáÓä Êã ÇÏÎÇåã Çái ãÑßÒ ¡ ÇáÌåÇÒ ÇáåÖãí ÎáÇá ÇáÓäæÇÊ 1994 - 1997 Êã ÇáÉÍÑí Úä ãÑÖ æáÓä Ýí Çí ãÑíÖ íÇÝÚ ãÕÇÈ ÈãÑÖ ÇáßÈÏ ÇáãÒãä ¡ ÚãÑ ÇáãÌãæÚÉ (ÊÑÇæÍ ãÇÈíä) 6 - 30 (ÓäÉ æÈãÚÏá) 8Ñ15 ÓäÉ . ßÇä ÇáãÑÖì ÇáÐßæÑ ÚÔÑÉ æÇáÇäÇË ÓÈÚÉ .13 ãÑíÖÇ)76% (ãä ÇáãÑÖì ßÇä áåã ÊÇÑíÎ ÚÇÆáí ãæÌÈ ááãÑÖ ¡ ßÇäÊ ÍáÞÉ ßÇíÓÑ ÝáíÔÑ ãæÌÈÉ Ýí ËáÇËÉ ÇÑÈÇÚ ÇáãÑÖì) 13 ãÑíÖÇ (¡ ÊÈíä Çä 3/1 ÇáãÑÖì) 6 ãÑÖì (áÏíåã ÏæÇái ÇáãÑÆ ÇáÏÇáÉ Úái ÇÑÊÝÇÚ ÖÛØ ÇáÏã ÇáÈæÇÈí . ßÇä) (ÇáíÑÞÇä ÇßËÑ ÇáÚáÇãÇÊ ÔíæÚÇ) 8 ãÑÖì æÎáÇá ÝÊÑÉ ÇáÎæá ÊæÝí ËáÇËÉ ãÑÖì (% 47 ÇËäÇä ãä ÚÌÒ ÇáßÈÏ æÇáËÇáË ãä äÒÝ ÇÚáì ¡ . ÇáÌåÇÒ ÇáåÖãí ÊÓÊäÊÌ ÇáÏÑÇÓÉ Çä ãÑÖ æáÓä áíÓ äÇÏÑÇ

Introduction:

Wilson's disease (WD); "Hepatolenticular degeneration" is a hereditary disorder with a recessive mode of inheritance [1,2,3] ; predominantly affecting young people, is due to toxic effect of accumulated copper in the body (especially in the liver, brain, kidneys and cornea) resulting from low biliary excretion of copper [1,2,3,4] .

We aimed in this study to identify the general features of WD diagnosed in GIT Center.

Patients and Methods:

This is a retrospective study carried in GIT Center; 17 patients with WD were admitted to GIT Center over 3 years (1994-1997). The diagnosis of WD was considered in any patients who fulfilled two of the following criteria: [1,2,3]

1. Serum Caeruloplasmin < 20 mg/dl.
2. 24-hour urine copper excretion > 100 mg / 24 hour.

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3. Presence of Kayser-Fleischer ring.
4. Reduced serum copper < 70 mg/dl.
5. Liver biopsy.

However, no single test can be used to diagnose WD in all circumstances. It's the constellation of clinical history, family history, physical exami-nation, and key laboratory test that establish the diagnosis.

Results:
Seventeen patients were admitted over a period of 3 years (1994-1997), their age ranged from (6-30) years with a mean of (15.8) year; there were 10 males and 7 females. Thirteen patients (13/17) had positive family history of WD (76%), thirteen patients (13/17) had positive K.F. ring (76%), one third had oesophageal varices (6 patients), 3 patients died during follow up (18 %).

Table I : Demonstrates The Mode Of Presentation

No	Presentation	No	%
1	Jaundice	8	47
2	NeuroIogicI abnormaIity	4	23.5
3	EncepheIopathy	2	11.8
4	Asymptomatic (famiIy screen)	2	11.8
5	HemoIytic anemia	1	5.9

Table II : show the results of investigations

Lab. Test	Range	Mean
S. BiIirubin	3-25	10.8 (mg/dI
SGPT	6-420	86 (I.U./L.)
AIKaIine phosphtase	7-25	14 (K.A.U.)
Hb	7-13	10.I (mg/dI)

Table III : show the results of specific Lab. Test

Lab. Test	No of patients tested	Change in test
S. CaeruIopIasmin	10/16 (62.5%)	Decrease
24 h. Urine Cu	4/14 (28%)	Increase
S. Cu	3/3 (100%)	NormaI

***Discussion*:**
WD is not rare; it's rather a common cause of liver disease in a young age group [2,5,6] The age range in this study was 6-30 years, which emphasizes the importance of screening all patients below 40 years of age with liver disease for this disease[4].

WD is an autosomal recessive disorder that affects both sexes equally and increase with intermarriage [1] ; that's why WD is probably more frequent in community with high incidence of consanguinity like our community [1,3]. Thirteen patients (76%) had positive family history for WD; therefore

diagnosis of WD should benefit the patient's family by earlier screening of other members for WD [1,2]. Thirteen patients (76%) had positive Kayser Fleischer ring; overall the patients with negative K.F. ring was a younger age group (< 10 years) than other patients whom they mostly presented with hepatic manifestations of WD, this is compatible with pathophysiology of the disease in which Cu^{+} accumulation increases initially in the liver and later in life involves other organs like central nervous system and cornea[1,2]

***Table I*;** demonstrates the mode of presentation where at least more than half - two third (> 1/2 - 2/3) presented with hepatic manifestations, while less than a quarter (< ¼) presented with neurological manifestation; the neurological abnormalities includes both pyramidal and extra pyramidal features like chorea, dysarthria, dystonia and tremor and usually affects older age group than those presented with hepatic manifestations [1,2].

***Table II*;** In patients with hepatic involvement there are a discordance in the presence of high serum bilirubin level with a modest increase of serum transaminase, low serum alkaline phosphatase which may help in the diagnosis of Wilsonian fulminant hepatitis, although it's absence does not exclude the diagnosis[1,7].

***Table III*;** show the results of specific lab. test, the low sensitivity of these specific labs. test were due to variability of phenotypic expression of WD [1].

There is high proportion (1/3) of patients with S. Caeruloplasmin concentration > 20 mg/dl which is higher than what is encountered in western series (10%), possible explanations are that (especially in hepatic WD), S. Caeruloplasmin is acute phase protein and increase with hepatic inflammation or might due to type of lab. test used in measuring S. Caeruloplasmin[1,2,3].

Establishing the diagnosis of Wilson's disease may be problematic, but it is essential if copper chela-ting therapy is to be instituted as early as possible in the course of the disease ; for instance if the patient is in oliguric renal failure it is not possible to quan-tify urinary copper excretion, alternatively; plasma ceruloplasmin values in Wilson's disease may be elevated into the low normal range during acute hepatitis. Severe coagulopathy prevent us from doing percutaneous liver biopsy, therefore; the specific criteria used to establish the diagnosis of Wilson's disease must be tailored to the patient's clinical presentation. Liver biopsy (in our circum-stances) can in best situations suggest the diagnosis of Wilson's disease, but does not confirm it because the quantification of dry weight is unavailable, due to condition of the embargo. [1,2,3]

In any young patient with acute hepatitis and Coomb's negative hemolytic anemia; WD should be kept in mind [1].

Conclusion:

WD is not uncommon in IRAQ and often presents late. Family screening of index patient should be carried out for early diagnosis and treatment and better outcome.

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