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Original Research Article

Natural history and outcome of patients with Wilson's disease from a tertiary care hospital in Odisha

Brijeshraj Swain, Naluri Mahesh*, Samir Ranjan Jena

Department of Medicine, MS and SUM Hospital, Bhubhaneswar, Odisha, India

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***Correspondence:**
Dr. Naluri Mahesh,
E-mail: maheshn.nmahesh@gmail.com

ABSTRACT

Background: Wilson's disease (WD) is a rare autosomal recessive disorder of defected copper metabolism in the liver. Excessive copper deposition causes oxidative stress, impaired mitochondrial function, and eventually leads to hepatic, neurologic, renal, and other organ dysfunction. Diagnosis of WD can be challenging due to its rarity, highly variable clinical manifestations, and non-specific diagnostic tests that are currently available.
Methods: It was a prospective observational study carried out in tertiary care hospital in Odisha during the period of 18 months. All the patients were subjected to a detail general and systemic examination as per the standard protocol. The past history of hepatic or psychiatric illness and family history of similar illness were taken from all patients. An ophthalmological examination for the presence of Kayser-Fleischer ring was carried out by examination with naked eye and by slit lamp evaluation.
Results: Of the 28 patients with Wilson's disease, 18% patients had history of consanguineous parentage and they had significantly younger age at onset than patients without history of consanguineous parentage. Out of 11 patients who had family history of Wilson's disease, 2 patients had death of sibling due to the disease.
Conclusions: It has been concluded from the study that features of chronic liver disease were the commonest manifestation of Wilson's disease. Screening of all asymptomatic siblings for Wilson's disease is a very important issue and must be carried out in all. Early and correct diagnosis and institution of proper treatment and lifelong continuation can prevent devastating consequences as the disease is treatable.

Keywords: Chronic liver disease, CTP score, Kayser-Fleischer ring, MELD score, Wilson's disease

INTRODUCTION

Wilson's disease (WD) is an uncommon autosomal recessive condition characterised by abnormal liver copper metabolism. Hepatic, neurological, renal, and other organ dysfunction are ultimately brought on by excessive copper deposition, which also results in oxidative stress and compromised mitochondrial function.[1] Due to its rarity, very diverse clinical presentations, and the current lack of definitive diagnostic tools, WD can be difficult to diagnose.[2,3] Treatments for WD have advanced significantly since the condition was originally described in 1912. The fight against this deadly illness began to change course in the 1950s with the introduction of copper chelating agents.[4] Many individuals can currently get appropriate medical therapy with copper antagonists and chelating drugs with satisfactory clinical response.[5] Patients with fulminant hepatic failure and end-stage liver disease can also benefit greatly from liver transplantation. As the healthcare system has improved during the past ten years, the long-term results following transplantation have also significantly improved.[6,7] Paediatric patients undergoing liver transplants for WD had an 81% 10-year survival rate, according to a recent European cohort research.[7] Whether exhibiting hepatic or neurologic symptoms, the classic Wilson's disease patient may be

evaluated between the ages of 6 and 40 if their serum ceruloplasmin level is less than 5 mg/dl (<50 mg/l) and they have distinct Kayser-Fleischer rings. Otherwise, Wilson's disease is strongly suggested by a low ceruloplasmin level (< 40 mg/dl) and an elevated basal 24-hour urine copper excretion (>40 ug/dl) when there is chronic liver disease or characteristic neurologic symptoms. By evaluating the amount of copper excreted in the urine 24 hours after D-penicillamine is administered, the D-penicillamine challenge test may be very conclusive. A percutaneous liver biopsy can be used to measure the concentration of parenchymal copper, which is considered by some to be the gold standard for diagnosing Wilson's disease, and to gauge the extent of liver damage. In patients without traditional symptoms, a thorough investigation is necessary; in the end, a gene mutation study can be the only valid and trustworthy diagnosis method. Most of the ATP7B mutations that have been found so far are missence mutations.[8] Wilson's illness is notorious for its high phenotypic and genotypic heterogeneity, which makes it difficult for pathologists and clinicians to diagnose, despite the fact that diagnostic criteria are updated on a regular basis.[9] Furthermore, whereas Wilson's disease pathology was thoroughly detailed in early treatises, more contemporary discourses have not been as thorough or shown a strong association between pathologic and clinical aspects.[10] Wilson's illness is always lethal if left untreated. Nonetheless, decoppering therapy has been around for almost 30 years. If a diagnosis is made promptly, this therapy has been shown to be effective. Thus, the clinician's primary responsibility is to promptly identify patients in order to prevent organ damage. These days, this isn't always done correctly, primarily because early signs aren't identified, but current diagnostic techniques can also fall short.[11] Wilson's disease patients visiting an Odisha tertiary care hospital were the focus of the current study, which sought to assess their clinical, biochemical, imaging, endoscopic, and treatment characteristics.

METHODS

It was a prospective observational study carried out in IMS and SUM Hospital, Department of Medicine, Bhubhaneswar during the period of 1st January 2024 to 30th June 2025. Total 28 patients included into the study.

Inclusion criteria

Inclusion criteria were the patient with age group of 5-40 years, patients diagnosed as a case of Wilson’s disease, and patients willing to participate in the study.

Exclusion criteria

Exclusion criteria were patients who does not give consent for participation, patients aged <5 years or >40 years, and patients who have HIV or PTB or malignancy or an alcoholic.

Data collection

The study was conducted in the hospital with consent from the Institute's Ethical Committee. Every patient and parent gave their proper informed permission. In accordance with customary procedure, each patient underwent a thorough general and systemic examination. All patients were asked about any past medical or mental health conditions, as well as any family history of such conditions. The presence of the Kayser-Fleischer ring was investigated ophthalmologically using both slit lamp evaluation and examination with the unaided eye. The patients underwent routine blood counts, liver function tests, and measures of total serum copper, serum ceruloplasmin, and 24-hour urinary copper excretion. There was neuroimaging (brain MRI/CT scan). EEG was used in cases where seizure disorder symptoms were present. Upper G.I. endoscopy and abdominal ultrasound were performed. The normal value of serum ceruloplasmin, as determined by nephelometry, is between 20 and 60 mg/dl. Urine copper levels are measured by atomic absorption spectrometry with Zeeman correction; the typical range is 2-30 ug/L. Using spectrophotometry, the normal range for serum copper is 70-150 ug/dl for men and 80-155 ug/dl for women.

Neuroimaging: A light speed volume CT scan of the brain was performed using a 64-slice MSCT (multi-slice computer tomography) equipment. A 1.5 tesla-strong MRI scanner was used to perform the brain scan.

Electroencephalography (EEG): In accordance with international criteria, a sixteen-channel EEG recording was conducted using a grass-telefactor twin recording and analysis software system.

UGI endoscopy: An endoscope was used to perform the procedure.

USG abdomen: A convex-type ultrasonic transducer with a central frequency of 2.5 MHz to 7.5 MHz was used to perform the USG Abdomen.

Statistical analysis

Microsoft Excel was used to enter the data, while SPSS version 28 was used for analysis. Numbers and percentages were used to display the data. Where necessary, the mean and standard deviation were computed.

RESULTS

The majority of patients in this study (43%), followed by those over 15 (36%), were in the 11-15 age range. The oldest patient was twenty-five years old, while the youngest was six. With a range of 6 to 25 years, the study's mean age was 14.28±4.79 years. Nine (32%), and 19 (68%), of the 28 Wilson's disease patients were female. The ratio of males to females was found to be 2:1, with

males outnumbering females. Six (21%), out of the 28 individuals with Wilson's disease, had siblings who had a comparable ailment in the family. Five (18%) of the 28 patients with Wilson's illness had a history of consanguineous parenthood, and their onset age was noticeably younger than that of patients without such a history. Two of the eleven individuals with a family history of Wilson's disease lost a sibling to the illness. The most prevalent presentation in our study was jaundice, which occurred in 22 (79%), followed by abdominal distension, pedal oedema, anorexia, and weakness, which occurred in 20 (71%), of the patients. Fifteen patients (54%) reported vomiting, ten patients (36%) reported abdominal pain, and eight patients (29%) reported UGI haemorrhage. Jaundice was the most prevalent symptom in our study, occurring in 22 (79%), followed by ascites and pedal oedema in 20 (71%), respectively. Nineteen (68%) of the patients had a KF ring. Twelve patients (43%) had hepatomegaly, three patients (11%) had encephalopathy, and thirteen patients (46%) had splenomegaly. The most prevalent hepatic presentation in our sample was jaundice, which occurred in 22 (79%), followed by ascites and pedal oedema in 20 (71%), respectively. Twelve patients (43%), and thirteen patients (46%), exhibited hepatomegaly and splenomegaly, respectively. UGI haemorrhage was observed in 8 (29%) of the individuals. Five patients (18%) had dystonia and poor academic performance, which was the most common neuropsychiatric presentation. Two patients (7%) each had tremor, chorea, seizure condition, and psychiatric manifestation.

Table 1: Type of presentation in different age group.

Age (years)	Hepatic only	Neurological only	Both hepatic and neurological
6-10	6	0	0
11-15	8	1	3
>15	4	2	4
Total, N (%)	18 (64)	3 (11)	7 (25)

Table 2: KF RING.

KF Ring	No. of patients	Percentage
Present	19	68
Absent	9	32
Total	28	100

At least one of the following criteria was used to identify hepatic dysfunction: total protein <5.5 g/dl, albumin <3.5 g/dl, serum glutamic pyruvic transaminase (SGPT) >100 IU/l, serum glutamic oxaloacetic transaminase (SGOT) >100 IU/l, or bilirubin >2.0 mg/dl. The mean in our sample was 5.78±6.28 mg/dl for total bilirubin, 181.42±154.41 IU/l for SGOT, 152.85±125.51 IU/l for SGPT, 6.37±0.98 gm/dl for serum protein, 2.9±0.69 gm/dl for serum albumin, and 13.18±3.51 for PT.

Serum copper levels in our study ranged from 32.14 to 102.42 ug/dl, with 21 (75%) patients having levels below 75 ug/dl and 7 (25%) having levels above 75 ug/dl.

Table 3: Serum copper and serum ceruloplasmin.

	No. of patients	Percentage
Serum copper		
<75 ug/dl	21	75
>75 ug/dl	7	25
Total	28	100
Serum ceruloplasmin		
<20mg/dl	22	79
>20 mg/dl	6	21
Total	28	100

Table 4: Child Turcotte Pugh score.

CTP score	No. of patient	Percent
A	4	14
B	7	25
C	14	50

Table 5: Treatment.

Treatment	No. of patients	Percentage
D-penicillamine+zinc	24	86
Zinc	4	14

24 hours urinary copper

Three patients (11%) had 24-hour urine copper levels between 40 and 100 ug/day, while 25 patients (89%) had levels greater than 100 ug/day, with a range of 62.34 to 1742.56 ug/day. Three (11%), whose levels were between 40 and 100 ug/day, underwent the panicillamine challenge test, which involves giving 0.5 g D-penicillamine at the start of the 24-hour urine collection and 12 hours later. The results showed that the 24-hour urinary copper was greater than 1600 ug/day. Thirteen (46%), twenty (71%), twelve (43%), and twenty-five (89%), patients in our study had splenomegaly, ascites, hepatomegaly, altered echotexture, dilated portal veins, and three (11%), normal abdominal ultrasonography studies. Three patients (11% each) had Gr-I and Gr-IV esophageal varices, four patients (14% each) had Gr-II and Gr-III esophageal varices, five patients (18%) had portal hypertesive gastropathy (PHG), and fourteen patients (50%) had no varix/PHG on UGI endoscopy. In our investigation, seven individuals with Wilson's illness had MRI results of elevated T2 intensities

in the thalamus, cerebellum, and basal ganglia (globus pallidus, putamen, and caudate nucleus). One of our instances displayed the classic "giant panda face." In two cases where there were signs of seizures, an EEG was conducted. Two patients had theta backgrounds, localised and generalised discharges, and aberrant EEG. Four (14%), seven (25%), and fourteen (50%) of the patients in our study had Class-A, Class-B, and Class-C CTP scores, respectively. Three individuals (11%) with neurological Wilson's disease had a questionable CTP score.

Table 6: Follow up study at 6 months.

Parameter	At presentation	At 6 months
Clinical		
Jaundice	22	15
Ascites	20	15
Pedal edema	20	14
KF Ring	19	18
Laboratory		
Total bilirubin	5.78±2.28	4.12±2.17
SGOT	181.42±154.41	126.57±94.44
SGPT	152.85±125.51	110.28±79.16
Sr. albumin	2.9±0.69	3.1±0.65
PT/INR	13.8±3.51	11.62±3.28
24 hours urinary copper	588.90±158.01	278.22±159.06

Table 7: Clinical parameters.

Parameter	At presentation	At 6 months
Imaging		
Ultrasound	Ascites-20	Ascites-15
Endoscopy		
Esophasial varices + PHG	14	13
CTP score		
A	4	4
B	7	7
C	14	13
MELD score		
<15	8	9
>15	20	19

DISCUSSION

With a gene frequency of 0.56% and an estimated disease incidence of 1 in 30,000, Wilson's illness is an uncommon autosomal recessive condition. The current study found that 18 patients (64%) had hepatic illness onset, 3 patients (11%), and 7 patients (25%), had mixed disease onset (both hepatic and neurological). In almost 60% of the patients in the Walshe et al series, the primary presenting symptom was hepatic, with 40% experiencing neurological symptoms.[12] According to research by Seyhan et al, 22 (59.6%) of the 37 patients with Wilson's disease presented hepatically.[13] In research by Thijeel et al, Of the 30 patients, 23 (76.6%) presented with a hepatic presentation, 4 (13.3%) with a neurological presentation, and 3 (10%) with a mixed appearance (hepatic+neurological).[14] Similar to our analysis, Stephania et al.'s study found that the hepatic type was the most common (65%).[15] In their investigation, Raiamani et al discovered that 11 individuals had hepatic form.[16] Yuce et al found that among 33 children with Wilson's illness, six of them had fulminant hepatitis.[17] Jaundice was the most prevalent hepatic presentation in our sample, occurring in 22 (79%), followed by ascites and pedal oedema in 20 (71%), respectively. Twelve patients (43%) had hepatomegaly, while thirteen patients (46%) had splenomegaly. UGI haemorrhage was observed in 8 patients (29%). Dystonia and poor academic performance were the most common neuropsychiatric presentations in our analysis, occurring in 5 patients (18%) each. Tremor, chorea, seizure disorder, and psychiatric manifestations were found in 2 patients (7% each). Tremors (31.6%), dysarthria (15.6%), jaundice (12.4%), abnormal gait (8.8%), abdominal distention (7.8%), musculoskeletal symptoms (5.2%), seizure (4.9%), behavioural issues (4.6%), dystonia (3.6%), clumsiness (2.6%), salivary drooling (2.6%), generalised weakness (2.3%), poor academic performance (1.9%), altered sensorium (1.3%), bleeding diathesis (1.3%), dysphagia (0.9%), chorea (0.3%), and poor vision (0.3%) were all noteworthy clinical features in a large series of 307 patients from NIMHANS.[20] Niraj Kumar et al.'s study a total of 31 patients with Wilson's disease were examined.[21] Of these, 20 (64.5%) had dystonia, the most common initial neurological feature. Other symptoms included dysarthria (41.9%), salivary drooling (38.7%), parkinsonian features (38.7%), abnormal gait (25.8%), abnormal behaviour (22.6%), tremors (16.1%), and declining academic performance (9.7%). The dystonic group accounted for 83.9 percent of the patients with the primary neurological traits, followed by the parkinsonian group (64.5%), cerebellar group (22.6%), and choreoathetoid group (9.7%). Aravind et al.'s study, Jaundice was the presenting style: 13. Abdominal pain: 2, UGI bleed: 2, ascites: 2. Cognitive: 6 K.F. ring: 18 (90%) Normal: Nine (47%) of the 28 patients had hepatic onset of disease, three (16%) had neurological onset of disease, and seven (37%) had mixed (hepatic + neurological) onset of disease.[8] Of these, 19 (68%) exhibited KF ring. KF rings were found in 74.7% of cases in research by Pooya et al, of which 73% had a hepatic onset of the disease and 96% had a neurological onset.[22] 76% of the patients in research by Rukunuzzaman et al had KF rings; of them, 84% had hepatic onset and 90% had neurological onset.[23] According to El Karaksy et al, 62.5% of children had a neurological manifestation and 33% had liver illness.[24] According to a research by Shakya et al, 17 (89.5%) of the 19 individuals with Wilson's disease exhibited a KF ring in their cornea.[25] All neurological Wilson's disease patients had KF rings, according to Kumar et al.[21] A research by Thijeel et al found that 16 (53.3%) of the 30 patients had a KF ring.[14] According to Stephania et al, KF rings were found in 7 cases (41%), all of which had severe or chronic liver

disease and ranged in age from 7 to 12 years (mean: 10.6 years).[15] 55.6% of patients had Kayser-Fleischer rings, according to Bem et al.[26] In several Indian series, the reported frequency of KF rings has been about the same, ranging from 86.6 to 97.1%. In line with our findings, Gow et al.'s study revealed that KF rings are found in only 50% to 62% of individuals with mostly hepatic illness at the time of diagnosis.[27] Patients with neurological findings are more likely than those with hepatic findings to exhibit KF rings. The KF ring's development is mostly correlated with the amount of time that has elapsed since the tissues' copper accumulation began. The fact that neurological abnormalities appeared at later ages may be the reason why KF rings are observed more commonly in patients with neurological indications. Serum protein was 6.37±0.98 gm/dl, serum albumin was 2.9±0.69 gm/dl, PT was 13.18±3.51, SGOT was 181.42±154.41 IU/l, SGPT was 152.85±125.51 IU/l, and the mean total bilirubin was 5.78±6.28 mg/dl in the current study. The mean total bilirubin was 3.86±3.73 mg/dl, the SGOT was 124.60±72.84 IU/L, the SGPT was 108.05±102.74 IU/l, the serum albumin was 3.09±0.88 gm/dl, and the PT was 21.93±8.67 in research conducted by Rukunuzzaman et al in 100 patients with Wilson's illness.[23] In a research by Thijeel et al, 16 patients (69.5%) had extended PT, while 19 patients (82.6%) had increased liver enzymes.[14] The mean total bilirubin was 3.25±1.57 mg/dl, the SGPT was 101.52±65.61 IU/l, the serum albumin was 2.61±1.05 gm/dl, and the PT was 17.96±5.12 in a study conducted by Farhana Bayes et al in 31 patients with Wilson's illness.[28] Melanie Johncilla[29] found that 14 patients with Wilson's disease had elevated AST levels (mean = 97±67 IU/l), elevated ALT levels (mean = 65±67 IU/l), elevated ALP levels (mean = 118±80 IU/l), and elevated total bilirubin (mean = 3.65±0.95 mg/dl). Serum albumin was 3.1±1 gm/dl, PT was 27±13, SGOT was 333±60 IU/l, SGPT was 189±825 IU/l, and the mean total bilirubin was 11.8±14.8 mg/dl in 111 individuals with Wilson's illness, according to a study by Pooya et al.[22] Serum copper levels in our study ranged from 32.14 to 102.42 ug/dl, with 21 (75%) patients having levels below 75 ug/dl and 7 (25%) having levels above 75 ug/dl. According to research by Kumar et al, 64.5% of patients had low serum copper levels (75 ug/dl), with a range of 31.7 to 135.8 ug/dl, which is in line with our findings.[21] Twenty-two (79%), and six (21%), of the patients in our research had serum ceruloplasmin levels below 20 mg/dl and over 20 mg/dl, respectively, with a range of 3.25 to 38.12 mg/dl. Serum ceruloplasmin was low (20 mg/dl) in 93.54% of patients in research by Kumar et al, with a range of 4.1 to 26.4 mg/dl, which is in line with our findings.[21] According to Eve et al, 10-20% of heterozygotes may have decreased serum ceruloplasmin, while 5-15% of WD patients may have normal or perhaps slightly decreased ceruloplasmin.[30] Serum ceruloplasmin levels below 20 mg/dL have been found to be diagnostic for WD and to be consistent with KF rings, as seen in our patients. Three (11%), and twenty-five (89%), of the patients in our study had 24-hour urine copper levels between 40 and 100 ug/day, with a range of 62.34 to 1742.56 ug/day. A 24-hour urine copper test revealed that 3 (11%) of the individuals with values between 40 and 100 ug/day had levels greater than 1600 ug/day. In research by Rukunuzzaman et al, 17 patients had 24-hour urine copper levels between 40 and 80 ug/day, while 83 patients had levels greater than 80 ug/day.[23] Following a penicillamine challenge test, the 24-hour urine copper level was >200 ug/day in 99 individuals and >1600 ug/day in 58 patients. All patients (100%) had 24-hour urine copper levels greater than 100 ug/day in research by Shakya et al.[25] 91.4% of patients in a research by Ali Akbar Asadi Pooya et al.[22] had 24-hour urine copper levels greater than 100 ug/day. Basal 24-hour urine copper excretion may be less than 100 g at presentation in 16% to 23% of patients with WD, according to research by Sanchez-Albisua et al.[31] According to a research by Kumar et al, all 31 (100%) patients had elevated 24-hour urine copper levels (>100 ug/day), with a range of 110.10 to 971 ug/day, which is in line with our findings.[21] Thirteen patients (46%) had splenomegaly, twenty patients (71%) had ascites, twelve patients (43%) had hepatomegaly, twenty-five patients (89%), fourteen patients (50%) had dilated portal veins, and three patients (11%), had normal abdominal ultrasound studies. According to Stephania et al, 64% of the 14 patients who received abdominal ultrasonography experienced problems such cirrhosis and hepatosplenomegaly.[15] Six individuals exhibited CLD/splenomegaly, seven patients had coarse hepatic echotexture, one patient had a fatty liver, two patients had gallstones, and six patients had normal abdominal ultrasounds, according to a study by Aravind et al.[8] Three patients (11%) in our study had Gr-I and Gr-IV esophageal varices, four patients (14%) had Gr-II and Gr-III esophageal varices, five patients (18%) had portal hypertesive gastropathy (PHG), and fourteen patients (50%) had no varix/PHG on UGI endoscopy. According to Stephania et al.'s study, 7 individuals who had upper GI endoscopies reported portal hypertension symptoms, and 43% of them had esophageal varices.[15] Two patients had esophageal varices, five had portal hypertensive gastropathy, and thirteen had normal UGI endoscopy, according to a study by Aravind et al.[8] In our investigation, seven individuals with Wilson's illness had MRI results of elevated T2 intensities in the thalamus, cerebellum, and basal ganglia (globus pallidus, putamen, and caudate nucleus). In their investigation on neuro Wilson's disease, Panagariya et al found that 86% of patients had aberrant basal ganglia results.[19] In their investigation, Bem et al discovered that 72.2% of these individuals had abnormalities in their neuroradiological imaging.[26] Two patients in our investigation had theta backgrounds, localised and generalised discharges, and aberrant EEGs. Six participants in research by Kumar et al had aberrant EEG readings.[21] Four patients (Child A) received zinc as part of our trial, while the remaining twenty-four patients (Child B, Child C, and Neurological WD) received zinc with penicillamine. Zinc + penicillamine was used to treat every patient in a research by Bayram et al.[32] Zinc, penicillamine, and pyridoxine were used to begin treatment in a research by Nuzhat Noureen et al.[33] All of the patients in a study by Thijeel et al had penicillamine

treatment, either with or without zinc.[14] In their study of Wilson's illness, Seyhan et al discovered that 24 patients had been receiving zinc and d-penicillamine therapy for 3 months to 7 years (mean, 2.4±2.6 years), whereas 13 (35.1%) were newly diagnosed.[13] In our study, a follow-up investigation was conducted six months following the initial presentation. Jaundice, ascites, pedal oedema, total bilirubin, SGOT, SGPT, PT/INR, 24-hour urinary copper, and serum albumin levels all decreased, indicating improvement. After undergoing liver transplantation, one patient in our study demonstrated improvement in liver function tests, ascites, pedal oedema, KF ring, and jaundice. Follow-up studies by Socha et al, Eve et al, Roberts et al, and Schilsky et al measured 24-hour urine copper excretion during treatment to assess treatment adequacy.[34,30,9,35] Urinary copper excretion peaked right after therapy began, and during chronic (maintenance) treatment, it was between 200 and 500 ug per day. Research conducted by Beinhardt et al examined 175 patients with Wilson's illness.[36] Only 162 (71%) of the patients who received chelation therapy between January 2012 and April 2013 were able to have their long-term treatment outcomes examined because 12 patients required emergency liver transplants and one patient passed away shortly after diagnosis. Of those 162, 40 patients (25%) had stable disease at the time of examination, 42 patients (26%) had entirely healed, 39 patients (24%), and 24 patients (15%) had deteriorated despite therapy. In research conducted by Shakya et al on 19 cases of Wilson's disease, the KF ring vanished entirely or in part in 38.8% (7/18) of the patients at the 6-month follow-up, and in 14 cases within a year.[25] Five instances failed to show up for follow-up after a year. Patients who attended for follow-up showed good response to the D-pencillamine treatment. When symptoms went away or the biochemical markers improved, it was considered a favourable reaction. In research conducted by Bem et al, D-Pencillamine was administered to 94.2% of all patients for an average duration of 129.9±108.3 months.[26] The overall survival rate after starting treatment was 90.1%, and 78.8% of the patients experienced a stable or improved prognosis. In a study by Lowette et al, D-Pencillamine was used to treat 24 Wilson's disease patients between 1969 and 2009, and follow-up was conducted.[37] Overall, 91.6% of people survived. 22 out of 24 patients had liver disease when they were first seen, and 17 out of 24 patients (71%), 11 of them had cirrhosis-related problems. Three out of eleven of these patients received transplants, one of eleven passed away, and one of eleven stopped follow-up. Six out of eleven of these patients had hepatological improvement (five out of six) or stabilisation (one out of six). Improvement (four out of six) or stabilisation (two out of six) occurred in the six out of seventeen cirrhotic individuals who did not experience any problems. Only one patient experienced hepatological worsening as a result of poor therapy compliance, and one of seven patients stopped follow-up. Of the other seven patients, five of seven either demonstrated improvement (three of five) or stabilisation (two of five). 13 out of 24 patients had neuropsychiatric symptoms upon presentation; these symptoms were cured in 1 out of 13 patients, decreased in 7 out of 13, stabilised in 4 out of 13, and deteriorated in 1 out of 13 patients as a result of noncompliance.

CONCLUSION

The study found that the most prevalent sign of Wilson's disease was characteristics of chronic liver disease. It's crucial to screen all asymptomatic siblings for Wilson's illness; this needs to be done for everyone. Since the disease is curable, severe outcomes can be avoided with early and accurate diagnosis, appropriate therapy, and lifelong continuation. Wilson's illness is most likely more widespread than Indian reports indicate. Due to the high level of consanguinity in various regions of India, further epidemiological research as well as multicentric studies are required. The medical community's knowledge of the illness might also be raised by a national register.

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