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Clinical Spectrum and Outcomes in 21 Patients with Wilson's Disease: A Single Center Experience

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KEYWORDS

Wilson's disease, Case series, Kayser–Fleischer ring, autosomal recessive disorder

ABSTRACT:

The case-series aimed to understand the Wilson's disease diagnosed in Madurai Medical College and Government Rajaji Hospital, Madurai, Tamil Nadu. A total of 21 individuals who were diagnosed with Wilson's disease were included in the study. The mean age of the study participants was 24.8 ± 11.3 years. The minimum and maximum age of the study participants were 12 years and 57 years respectively. Majority of the study participants were females (n=11, 52.4%). All the study participants had Kayser–Fleischer (KF) ring (n=21, 100%). All the study participants had reduced serum ceruloplasmin (n=21, 100%). In all the study participants, WD diagnostic score was >4 (n=21, 100%). And all the study participants were treated with Chelating agents and zinc salts (n=21, 100%). Wilson's disease is a rare, but potentially treatable inherited disorder of copper metabolism which can affect primarily the central nervous system and liver. Early diagnosis is crucial and helps in better prognosis of the disease. Characteristic findings such as Kayser-Fleischer ring and reduced ceruloplasmin levels plays vital role in diagnosis of the disease. Neurological manifestations like tremors and ataxia were commonly presented symptoms.

INTRODUCTION

Wilson's disease is a rare autosomal recessive disorder occurring due to gene mutations, which causes an abnormal accumulation of copper in organs like liver, brain and cornea [1]. Neurological or hepatic symptoms are the primary presentation symptoms. Children and young adults are the high-risk groups [2]. However, it can affect or present at any age [3,4]. In India, ATP7B mutation in p.C271X in India. Wilson's disease (WD) was more prevalent among males and osseomuscular symptoms predominated neurological and hepatic presentations [5].

The present study aimed to understand the Wilson's disease diagnosed in a tertiary hospital in Tamil Nadu.

CASE SERIES

The present study was designed as a case-series of 21 patients visited the OPD of a tertiary hospital in Tamil Nadu and was diagnosed with WD.

A total of 21 individuals who were diagnosed with Wilson's disease were included in the study. The mean age of the study participants was 24.8 ± 11.3 years. The minimum and maximum age of the study participants were 12 years and 57 years respectively. Majority of the study participants were females (n=11, 52.4%) (Table 1).

All the study participants had Kayser–Fleischer (KF) ring (n=21, 100%). All the study participants had reduced serum ceruloplasmin (n=21, 100%). In all the study participants, WD diagnostic score was >4 (n=21, 100%). And all the study participants were treated with Chelating agents and zinc salts (n=21, 100%).

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Prevalence of neurological symptoms were akinetic rigidity in 28.6% (n=6), seizure in 9.5% (n=2), tremor in 52.4% (n=11), ataxia in 42.9% (n=9), dystonia in 28.6% (n=6), cognitive impairment in 19% (n=4), and changes in MRI in all patients (n=21, 100%).

Table 1: Sociodemographic details and symptoms of Wilson's disease among study participants

PARAMETER	VALUE
Mean age	24.8 ± 11.3 years
Gender	
Male	10 (47.6%)
Female	11 (52.4%)
WD diagnostic score >4	21 (100%)
Symptoms	
Akinetic rigidity	6 (28.6%)
Seizure	2 (9.5%)
Tremor	11 (52.4%)
Ataxia	9 (28.6%)
Dystonia	6 (28.6%)
Cognitive impairment	4 (19%)
MRI changes	21 (100%)

To explain in detail about individual cases, the patient number1 was 26-year-old male with insidious onset slowly progressive tremulousness with unsteadiness of gait for 3 years, on clinical examination patient had KF ring, postural tremor, cerebellar signs like ataxic speech, limb incoordination, wide based gait. Patient had normal LFT and USG. Serum ceruloplasmin levels was 3mg/dl. On CT brain hypodensity in basal ganglia was seen. In MRI brain t2w hyperintensity in caudate, putamen, globus pallidus and midbrain was seen (Figure 1).

The patient number 2 was 20-year-old female with insidious, progressive difficulty in swallowing with drooling of saliva and aggressive behavior for past 2 years, on clinical examination patient had KF ring, Pseudobulbar features, drooling of saliva, open jaw, oropharyngeal dystonia and extrapyramidal rigidity was seen. Patient had normal LFT and USG. Serum ceruloplasmin levels was 2mg/dl. In MRI brain t2w hyperintensity in both basal ganglia, thalamus and midbrain was seen.

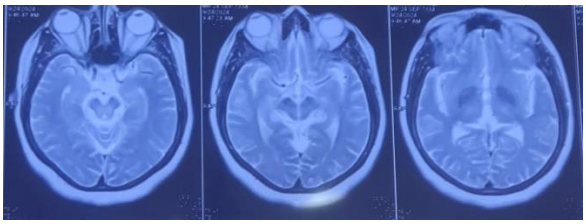

Figure 1: T2 Hyperintensity noted in midbrain (sparing bilateral red nucleus) and pons with hypointense central tegmental tract- giving double panda sign

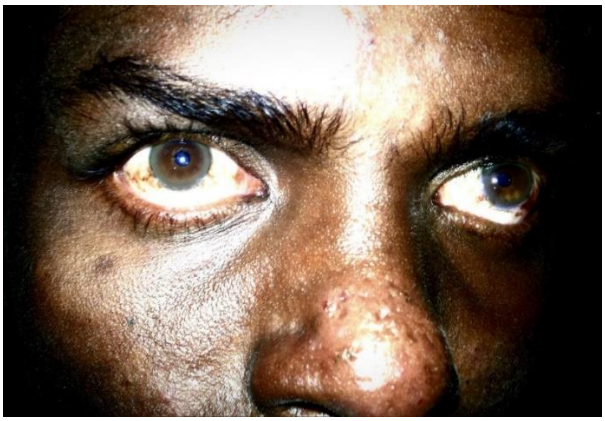

Figure 2: Keyser Fleisher Ring seen in one patient

The patient number 3 was 20-year-old male with difficulty in writing for 1 year, on clinical examination patient had KF ring, writing dystonia. Patient had normal LFT and USG. Serum ceruloplasmin levels was 7mg/dl. In MRI brain t2w hyperintensity in caudate, putamen and globus pallidus was seen.

The patient number 4 was 23-year-old male with insidious onset progressive involuntary jerky movements involving entire body and limbs for 4 years, on clinical examination patient had KF ring, mild MR and choreoathetosis. Patient had normal LFT and on USG hepatomegaly was seen. Serum ceruloplasmin levels was 6 mg/dl. On CT brain hypodensity in both basal ganglia was seen. In MRI brain t2w hyperintensity in both caudate nucleus and putamen was seen.

The patient number 5 was 57-year-old male with insidious onset slowly progressive slowness in all activities for 2 years 6 months, on clinical examination

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patient had KF ring, cognitive impairment, akinetic rigid syndrome. Patient had normal LFT and USG. Serum ceruloplasmin levels was 5mg/dl. On CT brain hypodensity in basal ganglia was seen. In MRI brain- t2w hyperintensity in caudate, putamen, globus pallidus and thalamus was seen.

The patient number 6 was 18-year-old female with progressive difficulty in swallowing, slurring of speech for 4 years, on clinical examination patient had KF ring, oropharyngeal dystonia, dysarthria, vacuous smile, open mouth, drooling of saliva. Patient had normal LFT and USG. Serum ceruloplasmin levels was 5mg/dl.In MRI brain t2w hyperintensity in basal ganglia, thalamus and midbrain was seen.

The patient number 7 was 40-year-old female with insidious onset, progressive ataxia for 4 years, on clinical examination patient had KF ring, postural tremor, wing beating tremor and gait ataxia. Patient had normal LFT and USG. Serum ceruloplasmin levels was 6mg/dl. In MRI brain t2w hyperintensity in caudate, putamen, globus pallidus and cerebellum was seen.

The patient number 8 was 29-year-old male with insidious onset slowly progressive tremulousness with slowness of all activities for 3 years, on clinical examination patient had KF ring, postural tremor, wing beating tremor, cog wheel rigidity and Parkinson gait. Patient had normal LFT and USG. Serum ceruloplasmin levels was 4mg/dl. In MRI brain t2w hyperintensity in both basal ganglia was seen.

The patient number 9 was 20-year-old female with insidious onset slowly progressive difficulty in writing, swallowing difficulty and drooling of saliva for 3 years, on clinical examination patient had KF ring, pseudobulbar features, vacuous smile, drooling of saliva and writing dystonia. Patient had normal LFT and USG. Serum ceruloplasmin levels was 8mg/dl. In MRI brain t2w hyperintensity in caudate, putamen, globus pallidus and thalamus was seen.

The patient number 10 was 26-year-old male with insidious onset slowly progressive tremulousness for 2 years, on clinical examination patient had KF ring, postural tremor, wing beating tremor, cogwheel rigidity and reduced arm swing. Patient had normal LFT and USG. Serum ceruloplasmin levels was 4mg /dl. In MRI brain t2w hyperintensity in both basal ganglia was seen.

The patient number 11 was 27-year-old male with insidious onset slowly progressive tremulousness with ataxia for 3 years, on clinical examination patient had KF ring, postural tremor was seen. Patient had abnormal LFT and normal USG. Serum ceruloplasmin levels was 5mg/dl. In MRI brain t2w hyperintensity in caudate, putamen and midbrain were seen.

The patient number 12 was 28-year-old female with tremor and ataxia for 2 years, on clinical examination patient had KF ring, postural tremor, cerebellar signs like ataxic speech was seen. Patient had abnormal LFT and normal USG. Serum ceruloplasmin levels was 4mg/dl. In MRI brain- t2w hyperintensity in caudate, putamen was seen.

The patient number 13 was 21-year-old male with signs of ataxia for 4 years, on clinical examination patient had KF ring and ataxic gait were present. Hepatic involvement was seen. Serum ceruloplasmin levels was 5mg/dl. In MRI brain t2w hyperintensity in thalamus was seen.

Patient number 14 was 13-year-old female with signs of dystonia, ataxia, tremors, cognitive impairment, drooling of saliva and seizure for 3 years. On Clinical examination, she had KF ring and dystonia, ataxia, dysarthria, tremors, vacuous smile, drooling of saliva. Hepatic involvement in form of Chronic liver disease. Serum Ceruloplasmin was low (0.3mg/dl). In MRI brain- t2w hyperintensity in caudate, putamen, thalamus was seen.

Patient number 15 was 48-year-old female with signs of ataxia, tremors for 3 months. On Clinical examination, she had KF ring and ataxia, dysarthria, voice and hand tremors. Hepatic involvement in form of Fatty liver disease. Serum Ceruloplasmin was low (0.6mg/dl). In MRI brain- t2w hyperintensity in bilateral Thalami and hyperintensities in bilateral globus pallidus with T2 hyperintensity in midbrain (sparing bilateral red nucleus) and pons giving giant panda sign.

Patient number 16 was 15-year-old female with signs of ataxia, tremors, cognitive impairment and seizure for 3 years. On Clinical examination, she had KF ring and ataxia, dysarthria, tremors, cognitive impairment. Hepatic involvement in form of parenchymal liver disease with splenomegaly. Serum Ceruloplasmin was low (9.75mg/dl). In MRI brain- t2w Flair hyperintensity

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in bilateral basal Ganglia, thalamus and mild signal abnormality in brainstem.

Patient number 17 was 15-year-old male with abdominal distension, and yellowish discoloration of skin, slowness of movements, on clinical examination patient had KF ring, bilateral cog wheel rigidity, jaw tremors were seen. Patient had abnormal LFT and USG parenchymal liver disease and gross splenomegaly. Serum ceruloplasmin levels was 5 mg/dl. In MRI brain t2w hyperintensity in caudate, putamen was seen.

Patient number 18 was 26-year-old male with insidious onset progressive slurring of speech and slowness of activities, sustained contraction of upper limbs for 1 years, on clinical examination patient had KF ring, mild MR, dystonia, bradykinesia. Patient had normal LFT and on USG hepatomegaly seen. Serum ceruloplasmin levels was 7 mg/dl. On CT brain hypodensity in bilateral basal ganglia was seen. In MRI brain t2w hyperintensity in both caudate nucleus, putamen and subtle signal abnormality in brainstem were seen.

Patient number 19 was 17-year-old female with insidious onset slowly progressive difficulty in walking in form of swaying and dysarthria in the past 6 years, on clinical examination patient had KF ring, rest tremors, cerebellar involvement were seen. Patient had abnormal LFT and USG showed cirrhosis and portal hypertension. Serum ceruloplasmin levels was 10 mg/dl. In MRI brain t2w hyperintensity in bilateral caudate, putamen and midbrain were seen.

Patient number 20 was 12-year-old female with insidious onset slowly progressive difficulty in writing, slowness in walking, slurring of speech, drooling of saliva in the last 1 year, on clinical examination patient had KF ring, ataxia, hypokinesia, cog wheel rigidity in upper limbs. Patient had normal LFT and USG showed changes of chronic liver disease and portal hypertension. Serum ceruloplasmin levels was 8mg/dl. In MRI brain t2w and flair hyperintensity in bilateral globus pallidus and thalamus and posterior midbrain was seen.

The patient number 21 was 19-year-old female with signs of jaundice 6 years back, bilateral tremors, and cognitive impairment, on clinical examination patient had KF ring and vacuous smile, bilateral wing beating tremors, cerebellar signs were present. Hepatic involvement was seen in form of deranged LFT, and chronic liver disease on USG. Serum ceruloplasmin levels was 8 mg/dl. In MRI brain t2w hyperintensity in bilateral basal ganglia, thalamus and midbrain were seen.

Wilson's disease is an autosomal recessive disorder affecting copper metabolism, often occurring without any evident family history. Early symptoms are usually unclear and non-specific, which can lead to diagnostic delays and uncertainty in clinical assessment. In this study, 21 patients were evaluated ten males and eleven females, presenting with diverse clinical features. A slightly higher proportion of females were reported in the present study. However, other studies all presented with male predominancy [2, 6, 7]. The mean age at diagnosis was 21.9 ± 10.8 years (ranging from 12 to 52 years), with males comprising 61% of the cases. Similar mean age was reported in a study conducted by Ramu S, 2020(2). All patients showed reduced serum ceruloplasmin levels and the presence of Kayser–Fleischer rings. Additionally, every patient had a Wilson's disease diagnostic score exceeding four, confirming the diagnosis. All patients were treated with chelating agents and zinc salts.

DISCUSSION

Prevalence of neurological symptoms in the present study were akinetic rigidity in 28.6% (n=6), seizure in 9.5% (n=2), tremor in 52.4% (n=11), ataxia in 42.9% (n=9), dystonia in 28.6% (n=6), cognitive impairment in 19% (n=4), and changes in MRI in all patients (n=21, 100%). In Ramu S, 2020 study, predominant neurological symptoms were tremor and ataxia which is similar to the present study (2). The findings of our study were consistent with another study conducted by Czlonkowska A *et al*, 2018, which reported that the most frequent neurological manifestations included dysarthria (73.6%), postural tremor of the arms (approximately 70%), impaired finger tapping (66%), abnormal posture (66%), and reduced facial expression (66%). Among the neurological syndromes, ataxia/tremor was the most prevalent (62.3%), followed by dystonia (15.1%) and parkinsonism (11.3%). A minor subset of patients (11.3%) exhibited only mild or isolated neurological signs and were categorized as unclassified [8].

Follow-up of the study participants was not done, which is an important limitation of the case series.

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CONCLUSION

Wilson's disease is a rare, but potentially treatable inherited disorder of copper metabolism which can affect primarily the central nervous system and liver. Early diagnosis is crucial and helps in better prognosis of the disease. Characteristic findings such as Kayser-Fleischer ring and reduced ceruloplasmin levels plays vital role in diagnosis of the disease. Neurological manifestations like tremors and ataxia were commonly presented symptoms.

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