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# A Study on Assessment of Wilson Disease Therapeutic Strategies and Clinical Implications for Prevention of Wilsons Disease

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**ABSTRACT**

**Introduction:** Wilson disease is an inherited disorder of copper metabolism caused by pathological copper accumulation in many organs, particularly the liver and brain, leading to a wide range of symptoms. Aim: The study aimed to assess wilson disease therapeutic strategies and clinical implications for prevention of wilsons disease. **Methods:** It was Prospective observational study. Present study was conducted for a period of six months. The Present study was conducted in General medicine department in a tertiary care hospital. The sample was 200 Patients. **Results and Discussion:** In our study 36-45 years age patients were more 60(30%) compared to other ages. Phenotype at Wilson disease diagnosis includes Acute hepatic presentation were more 85 (42.5 %) compared to other diagnostic categories. Clinical symptoms of Wilson disease includes neurologic symptom patients were more 151 (75.5) compared to other symptoms. Complications includes Kidney problems patients were more 85 (42.5%) compared to other complications. Drug prescribing pattern for wilson disease management includes Penicillamine prescribed patients were more 85 (42.5%) as compared to other drugs. **Conclusion:** Development of new therapeutic trends and clinical research studies and promotion of clinical guidelines to practicing physicians can shows better outcomes for Wilson disease patients. Advances in genetic testing and a better understanding of Wilson disease can prevent future complications of the disease.

**Keywords:** Wilson disease, neurologic symptom, diagnostic categories, therapeutic trends, clinical guidelines.

## INTRODUCTION

Wilson disease (WD) is an inherited disorder of copper metabolism caused by pathological copper accumulation in many organs, particularly the liver and brain, leading to a wide range of symptoms. The disease is caused by homozygous or compound heterozygous mutations (the presence of two different mutant alleles) in the ATP7B gene that encodes a transmembrane copper-transporting ATPase that mediates the excretion of copper into bile and delivers copper for the functional synthesis of ceruloplasmin (the major copper-containing protein in the blood). In WD, defective ATP7B function leads to copper overload in hepatocytes, with associated liver pathology. Excess copper is also released into the circulation with secondary pathological accumulation in other tissues, particularly the brain, which can lead to neurological symptoms and psychiatric disturbances[1-2]. Symptoms vary widely and present most commonly between ages 5 and 35 years. WD is rare, with the prevalence of symptomatic disease estimated to be 1 in ~30,000; however, a greater prevalence of genetic WD (based on two alleles with pathogenic mutations) has been observed according to recent molecular studies[3-6]. WD belongs to just a few genetic disorders which can be successfully managed if diagnosed early and correctly treated; however, if left untreated, WD is universally fatal.

### Epidemiology

Epidemiological studies from Germany and Japan in the 1970s indicated a WD prevalence of 29 per 1,000,000 and 33 per 1,000,000, respectively. In 1984, the general prevalence of WD was estimated as 1 in 30,000 in non-isolated populations with a mutation carrier frequency of one allele of 1 in 90 (almost 1% of the population) and these epidemiological data are still widely cited today. The prevalence of WD is higher in China (58.7 per 1,000,000) and Asian countries than in western countries. Epidemiological studies from isolated communities reported higher frequency due to consanguinity[7].

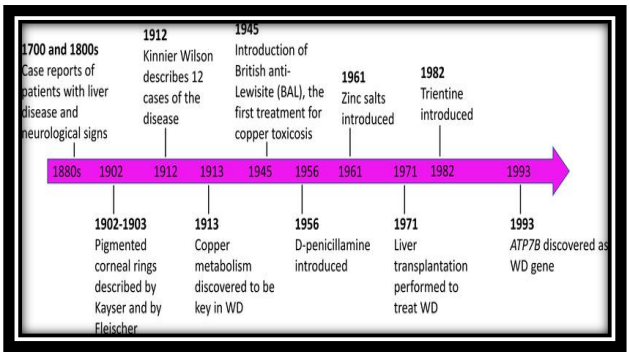


**Figure 1:** Discoveries in Wilson disease

### Etiology

Wilson disease is an autosomal recessive condition caused by 1 of several mutations in ATP7B gene which encodes the ATP7B protein transporter on the long arm of chromosome 13 (13q) responsible for excreting excess copper into bile and out

of the body. The primary route of copper excretion is through the liver. The excess copper first accumulates in the liver and then accumulates in the blood, where it is distributed to other organ systems. Furthermore, the excess copper leads to the generation of free radicals, which in turn cause the oxidation of vital proteins and lipids. Early cellular changes typically occur in the mitochondria, nuclei, and peroxisomes. Approximately 30% to 50% of patients will have neuropsychiatric symptoms, including an asymmetric tremor. Other symptoms may include ataxia, hypophonia, dysarthria, and clumsiness. Renal symptoms are due to varying degrees of tubular dysfunction with a spectrum including hydroelectric acid-base disorders, nephrolithiasis, urolithiasis, bile cast nephropathy, and Fanconi syndrome. Physical examination may demonstrate hepatosplenomegaly[8].

**Treatment Monitoring**

The main goal of treatment monitoring is to stabilize clinical progression and reversal of biochemical abnormalities with adequate pharmacotherapy and to look for drug-induced adverse effects. Regular tracking of copper levels in blood and urine, along with monitoring of liver functions, is essential to assess the effectiveness of treatment and educate the family regarding treatment adherence. It is also more critical to monitor patients who are on D penicillamine and trientine, which can cause paradoxical neurological worsening[9].

## MATERIALS AND METHODS

**Study Design**: It was Prospective observational study.

**Study Period**: The Present study was conducted for a period of six months.

**Study site**: The Present study was conducted in General medicine department in a tertiary care hospital.

**Sample size:** It was 200 Patients.

**Inclusion criteria**

- Patients with age of more than 18 years.
- Recently diagnosed with Wilson disease.
- Patients who are willing to give consent.
- Patients receiving treatment for Wilson disease.

**Exclusion criteria**

- Patients below 18 years.
- Patients who were not willing to join in the study.
- Psychiatric abnormalities.

**Institutional ethics committee (IEC) consideration:**

The research protocol was submitted to ethical committee and ethical Committee was permitted to perform the research work in the selected department of a tertiary care hospital.

**Patient data collection and management:**

The data collection form contains information regarding age, sex, diagnosis, past medical history, medication history, laboratory data, and diagnosis, dose and frequency of administration and duration of therapy was collected from the patients treatment chart.

**Statistical analysis:** The collected data was represented as percentages.

## RESULTS AND DISCUSSION

**Table 1: Age wise distribution**

In our study 20-25 years age patients were 55(27.5%), 26-35 years age patients were 41(20.5%), 36-45 years age patients were 60(30%), 46-55 years age patients were 44(22%).

| S.No | Age | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | 20-25 | 55 | 27.5 |
| 2. | 26-35 | 41 | 20.5 |
| 3. | 36-45 | 60 | 30 |
| 4. | 46-55 | 44 | 22 |
| | **Total** | **200** | |

**Table 2: Gender:** In our study male patients were 117 (58.5 %), female patients were 83 (41.5 %).

| S.No | Gender | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Male | 117 | 58.5 |
| 2. | Female | 83 | 41.5 |
| | **Total** | **200** | |

**Table 3: BMI:** In our study BMI category includes Underweight patients were 39(19.5%),Normal patients were 44(22%),Overweight patients were 62(31%), Obese patients were 55(27.5%).

| S.No | BMI | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Underweight | 39 | 19.5 |
| 2. | Normal | 44 | 22 |
| 3. | Overweight | 62 | 31 |
| 4. | Obese | 55 | 27.5 |
| | **Total** | **200** | |

**Table 4: Locality status:** In our study rural area patients were 115 (57.5 %), urban area patients were 85 (42.5 %).

| S.No | Locality status | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Rural | 115 | 57.5 |
| 2. | Urban | 85 | 42.5 |
| | **Total** | **200** | |

**Table 5: Education:** In our study Education wise patients includes SSC were 95(47.5%), Degree were 70(35%), Post-graduation were 35(17.5%).

| S.No | Education | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 2 | SSC | 95 | 47.5 |
| 3 | Degree | 70 | 35 |
| 4 | Post-graduation | 35 | 17.5 |
| | **Total** | **200** | |

**Table 6: Professional status:** Professional status includes Employee were 115(57.5%), Not working were 55(27.5%), Retired were 30(15%).

| S.No | Professional status | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Employee | 115 | 57.5 |
| 2. | Not working | 55 | 27.5 |
| 3. | Retired | 30 | 15 |
| | **Total** | **200** | |

**Table 7: Duration of Hospital stay:** Duration of Hospital stay includes 1-7 days patients were 115 (57.5%), 8-12 days patients were 45 (22.5%), 13-15 days patients were 40(20%).

| S.No | Duration of hospital stay | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | 1-7 days | 115 | 57.5 |
| 2. | 8-12 days | 45 | 22.5 |
| 3. | 13-15 days | 40 | 20 |
| | **Total** | **200** | |

**Table 8: Phenotype at Wilson disease diagnosis**: Phenotype at Wilson disease diagnosis includes Acute hepatic presentation 85 (42.5 %), Chronic hepatic presentation 35 (17.5%), Neurological presentation associated with symptomatic liver disease 42 (21%), Neurological presentation associated with asymptomatic liver disease 38 (19%).

| S.No | Phenotype at WD diagnosis | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Acute hepatic presentation | 85 | 42.5 |
| 2. | Chronic hepatic presentation | 35 | 17.5 |
| 3. | Neurological presentation associated with symptomatic liver disease | 42 | 21 |
| 4. | Neurological presentation associated with asymptomatic liver disease | 38 | 19 |
| | **Total** | **200** | |

**Table 9: Diagnosis duration (years):** Diagnosis duration of Wilson disease includes 1-4 years patients were 100 (50%), 5-7 years patients were 40 (20%), 8-9 years patients were 60 (30%).

| S.No | Diagnosis duration (years) | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | 1-4 | 100 | 50 |
| 2. | 5-7 | 40 | 20 |
| 3. | 8-9 | 60 | 30 |
| | **Total** | **200** | |

**Table 10: Clinical symptoms:** Clinical symptoms of Wilson disease includes neurologic symptom patients were 151 (75.5), Psychiatric symptom patients were 28(14), Hepatic symptom patients were 21(10.5).

| S.No | Clinical symptoms | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Neurologic | 151 | 75.5 |
| 2. | Psychiatric | 28 | 14 |
| 3. | Hepatic | 21 | 10.5 |
| | **Total** | **200** | |

**Table 11:** Risk factors

| S.No | Risk factors | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Family history of the disease | 25 | 12.5 |
| 2. | Consanguineous Marriages | 41 | 20.5 |
| 3. | Genes | 28 | 14 |
| 4. | High Dietary Copper Intake | 45 | 22.5 |
| 5. | Alcohol | 61 | 30.5 |
| | **Total** | **200** | |

**Table 12: Comorbidities:** Comorbidities includes Osteoporosis patients were 63(31.5%), UTI patients were 38 (19%), Migraine patients were 47 (23.5%), Hernia patients were 52 (26%).

| S.No | Comorbidities | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Osteoporosis | 63 | 31.5 |
| 2. | UTI | 38 | 19 |
| 3. | Migraine | 47 | 23.5 |
| 4. | Hernia | 52 | 26 |
| | **Total** | **200** | |

**Table 13:** Lab test

| S.No | Lab test | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | **Eye exam** | 95 | 47.5 |
| 2. | **Liver biopsy** | 65 | 32.5 |
| 3. | **CT/MRI scan** | 25 | 12.5 |
| 4. | **Blood test** | 15 | 7.5 |
| | **Total** | **200** | |

**Table 14:** Complications: Complications: **Complications** includes Liver failure patients were 25 (12.5%), Kidney problems patients were 85 (42.5%), Brain problem patients were 35(17.5%), Hemolysis patients were 55 (27.5%).

| S.No | Complications | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Liver failure | 25 | 12.5 |
| 2. | Kidney problems | 85 | 42.5 |
| 3. | Brain problem | 35 | 17.5 |
| 4. | Hemolysis | 55 | 27.5 |
| | **Total** | **200** | |

**Table 15:** Drug prescribing pattern for Wilson disease: Drug prescribing pattern for wilson disease management includes Penicillamine prescribed patients were 85 (42.5%), Trientine prescribed patients were 24 (12%), Zinc Acetate prescribed patients were 57 (28.5%), Vitamin B6 prescribed patients were 34 (17%).

| S.No | Prescribed drugs | Total (N=200) | Percentage (%) |
|---|---|---|---|
| 1. | Penicillamine | 85 | 42.5 |
| 2. | Trientine | 24 | 12 |
| 3. | Zinc Acetate | 57 | 28.5 |
| 4. | Vitamin B6 | 34 | 17 |
| | **Total** | **200** | |

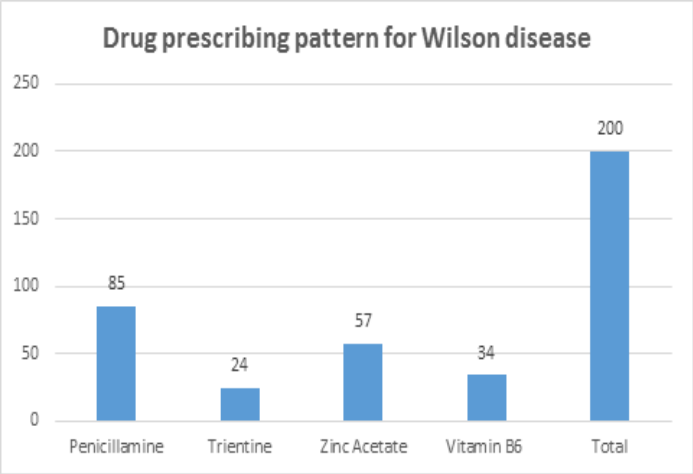


**Figure 1:** Drug prescribing pattern for Wilson disease

## DISCUSSION

- In our study 36-45 years age patients were more 60(30%) compared to other ages.
- In our study male patients were more 117 (58.5 %) compared to female patients were 83 ( 41.5 %).
- In our study BMI category includes Overweight patients were more 62(31%) compared to other BMI groups.
- In our study Rural area patients were more 115 (57.5 %), compared to urban area patients were 85 (42.5 %).
- In our study Education wise patients includes SSC were more 95(47.5%), compared to other education groups.
- Professional status includes Employee were more 115(57.5%) compared to other categories.
- Duration of Hospital stay includes 1-7 days patients were more115 (57.5%) compared to other categories.
- Phenotype at Wilson disease diagnosis includes Acute hepatic presentation were more 85 (42.5 %) compared to other diagnostic categories[10].
- Diagnosis duration of Wilson disease includes 1-4 years patients were more 100 (50%) compared to other diagnosis duration [11].
- Clinical symptoms of Wilson disease includes neurologic symptom patients were more 151 (75.5) compared to other symptoms.
- Risk factors of Wilson disease includes Alcohol patients were more 61(30.5) compared to other risk factors.
- Comorbidities includes Osteoporosis patients were more 63(31.5%) compared to other Comorbidities
- Lab test **includes Eye exam** patients were **more** 95(47.5%), compared to other symptoms.
- **Complications** includes Kidney problems patients were more 85 (42.5%) compared to other complications[12].
- Drug prescribing pattern for wilson disease management includes Penicillamine prescribed patients were more 85 (42.5%) as compared to other drugs13-14.

## CONCLUSION

Microspheres are mono- or multi-nuclear drug delivery systems in which the active pharmaceutical ingredient is embedded within a spherical polymeric matrix. These solid particles, generally ranging in size from 1 µm to 1000 µm, are prepared using biodegradable synthetic polymers or modified natural products such as starches, gums, proteins, fats, and waxes. Microsphere-based formulations offer the advantages of controlled release, improved bioavailability, and site-specific delivery. Cyclobenzaprine is a centrally acting skeletal muscle relaxant structurally related to tricyclic antidepressants. It is primarily prescribed for the management of acute musculoskeletal conditions associated with muscle spasms, pain, and stiffness. However, its short half-life and systemic side effects limit its therapeutic efficiency, creating the need for a sustained release formulation. The entrapment efficiency of the formulations was found to be F1: 34.75%, F2: 60.92%, and F3: 87.45%. The drug loading values were F1: 18.10%, F2: 20.25%, and F3: 22.65%. The percentage yield obtained was F1: 44.8%, F2: 55.7%, and F3: 63.2%. In vitro dissolution studies showed drug release of F1: 80.5%, F2: 77.8%, and F3: 72.9% at the end of 8 hours. Dissolution profile graphs (percentage drug release vs. time) were plotted for all formulations. Among them, F3 demonstrated the most consistent and reliable results. Hence, F3 formulation was considered promising and may be further subjected to stability studies, as well as pre-clinical and clinical investigations.

## CONFLICT OF INTERESTS

The authors declare no conflict of interest

## FUNDING

This study received no specific funding from public, commercial, or not for profit funding agencies.

## AI TOOL DECLARATION

The authors declare that no AI and related tools are used to write the scientific content of this manuscript.

## DATA AVAILABILITY

Data will be available on request

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