

CASE STUDY ON “WILSON’S DISEASE”

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Case Scenario

Mast. Deepak, 11 year old boy, got admitted to a private hospital with H/o yellowish discolouration of sclera, high coloured urine, generalised itching of the body, mild abdominal distension with pain, loss of appetite and loss of weight and fatigue

- Family History: Death of sibling 3 years back at the age of 6 years due to jaundice
- Nutritional history: Had Pica for mud since 7-8 months

Investigations

Table 1: Investigations.

Hb	7.3gm/dl	low
S. Iron	11 mcg/dl	low
Total Iron binding capacity	443 mcg/dl	low
SGOT	65/IU	Elevated
SGPT	72/IU	Elevated
Bilirubin	12/mg/dl	Elevated

- Treatment:** was given for Icterus but no improvement was seen in the patient. After a long phase of poor response to the treatment, Mast Deepak was referred to Tertiary care hospital.

In tertiary care Hospital, On examination

- K-Y Ring: (Kayser-Fleischer rings) was identified in the eyes.

Golden-brown eye discoloration



Investigations

USG: showed Altered echo texture of the Liver and cholelithiasis  
24 hours urine copper- 322 mcg/dl (Normal - 20-50mcg/dl).

Diagnosis: Based on the findings of classic sign of K-Y ring and other investigations, the final diagnosis of Mast Deepak was done as “Wilson’s Disease”

Table 2: Following treatment was given.

Drug	Action
Tb. Penicillamine 250 mg OD	Chelating agent
Tb. Zinconia 50mg	Zn supplement
Inj. Ferric carboxy maltose Inj Encicarb	Iron therapy

**Disease condition:** Wilson's disease (WD) is a rare Genetic disorder with impaired copper excretion leading to Excessive accumulation of copper in the liver, brain, eyes, kidneys, RBCs, heart and bones.

Pathogenesis

- The protein *ATP7B* is important in the pathway of hepatic copper transport into bile. In Wilson's disease, gene mutation occurs on chromosome 13, leading to the absence or diminished function of the hepatic protein *ATP7B*. Decrease in biliary copper excretion and ultimately leads to the hepatic accumulation of copper, leading to Hepatic failure.
- Decreased Ceruloplasmin (the primary copper-carrying protein in the blood)

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Leads to Increased Urine copper excretion (Hypercupricuria)

- Deficiency of ceruloplasmin, the copper transport protein which results in excessive inorganic copper in the blood circulation, much of it accumulates in red blood cells,

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Leads to Hemolysis

**Signs and symptoms**

- Hepatic disease-chronic active hepatitis, cirrhosis, fulminant hepatitis
- Neurologic disease-dystonia, dysarthria, tremors, involuntary choreiform movements (e.g., features of cerebellar or extrapyramidal disease)
- Psychiatric disease, particularly with associated organic neurologic disease
- Renal disease- Nephro calcinosis
- Arthropathy

- Ophthalmologic manifestations-Kayser-Fleischer rings, sunflower cataracts
- Hematologic manifestations-Hemolysis

**Priority nursing diagnosis:** following are the priority nursing diagnosis and its class is done as evidenced by frequent questioning and verbalisation of doubts and fear about the outcome

Domain	Class	Nursing Diagnosis
Domain 9- Coping stresstolerance	2- Coping Responses	Death Anxiety
11- Safety/ Protection	2- Physical Injury	Risk for Impaired skin integrity
2- Nutrition	1- Ingestion	Imbalanced Nutrition less thanbody requirement
2- Nutrition	5- Hydration	Excess fluid volume
12- Comfort	1- Physical comfort	Impaired comfort
4- Activity/Rest	4- Cardiovascular PulmonaryResponses	Activity Intolerance
4- Activity/Rest	4- Cardiovascular PulmonaryResponses	Risk for Ineffective tissue perfusion
2- Nutrition	4- Metabolism	Risk for Metabolic Imbalancesyndrome
6- Self perception	2- Self esteem	Risk for situational low self esteem
1- Health promotion	2- Health Management	Ineffective Health Maintenance
1- Health promotion	1- Health Awareness	Readiness for enhanced HealthLiteracy

**Nursing care plan based on the priority Nursing diagnosis was designed for Mast Deepak**

1. Death anxiety related to outcome of the condition/ impending death/death of sibling/uncertainty of prognosis as evidenced by frequent questioning and verbalisation of doubts and fear about the outcome

**Nursing outcome**

Identify, disclose and demonstrate techniques for psychological comfort.

**Nursing Interventions**

- Family counselling: to correct misinformation
  - Environmental safety: to discuss feelings
  - Reflection: on real and imagined threats to well-being.
  - Psychological support: diagnostic procedures.
  - Reference: the family to a spiritual counsellor/ social worker
  - Comfort and Relaxation techniques
2. Risk for Impaired skin integrity related to increased concentration of bilirubin in blood, excretion of bile through skin and Generalised itching

**Nursing outcome:** Patient maintains intact skin with relief from itching and skinbreakdown.

**Nursing Interventions**

- Skin assessment
- Skin care management: nails short, use of emollients, Avoid alkaline based soap
- Comfort measures : use of cotton balls or knuckles

for itching, cool showers, cotton clothes with loose fittings.

Diversional therapy: Play therapy

3. Excess fluid volume related to compromised regulatory mechanisms for sodium and water secondary to hepatic failure as evidenced by Abdominal distension, shiny skin.

**Nursing outcome**

Patient verbalizes awareness of causative factors and behaviours essential to correctfluid excess.

Patient maintains Normovolemic state with normal I/O, Normal abdominal girth, absence ofedema.

**Nursing interventions**

- Assessment: Monitor weight & I/O chart, abdominal girth
- Review serum electrolyte levels
- Fluid management
- Comfort measures /position
- Education on supportive therapy
- Pharmacotherapy and Management

4. Imbalanced nutrition less than body requirement related to Insufficient intake to meet metabolic demands: loss of appetite/Altered absorption and metabolism of ingested foods and nutrients /Eating of non nutritive substance as evidenced by loss of weight.

**Nursing outcome:** Balanced nutritional state is

maintained in accordance with activity level and metabolic needs.

#### Nursing interventions

- Nutritional assessment
- Serum value Monitoring
- Supportive therapy to Enhance appetite
- Fluid management
- Dietary consultation :for dietary modifications
- Behavioural therapy: Positive reinforcement for Pica
- Comfort Measures
- Environment modification and socialization
- Supplement therapy

5. Risk for Ineffective tissue perfusion related to Decreased haemoglobin concentration in the blood

Outcome criteria: The patient maintains adequate tissue perfusion, as evidenced by strong peripheral pulses, warm skin temperature with adequate capillary refill, normal I/O and stable Vital signs.

#### Nursing Interventions

- System wise Assessment: Integumentary, CNS, CVS, respiratory, Renal system
- Monitor O<sub>2</sub> saturation levels
- Monitor lab values of BUN, creatinine, Hb, serum lactate
- Respiratory support
- Warmth and comfort measures
- Fluid therapy
- Iron therapy
- Blood transfusion as per the order
- Pharmacotherapy: Tb. D Penicillamine- chelating agent (removes excessive copper) as prescribed and dose monitoring

Tb. Zinconia as prescribed (prevents absorption of copper from food)

6. Risk for Metabolic imbalance syndrome related to Multi organ accumulation of copper as evidenced by K-Y Ring, Hypercupricuria, Lowered ceruloplasmin levels and Elevated 24 hrs urine copper levels.

**Nursing outcome:** Maintains the copper levels in the body organs (serum and urine) within normal limits  
Demonstrates effective health maintenance

#### Nursing Interventions

- Assessment- system wise, serum and urine values
- Diet therapy: copper intake less than 1mg/day
- Avoid: dry fruits, liver, mushrooms, nuts, shellfish, chocolates
- Water demineralisation if copper levels are more than 0.1mg/dl
- Copper chelation therapy: D Pencillamine
- Adjuvant therapy: Zn Therapy: (impairs absorption

of copper from GIT)

**Take home message:** Self-management for health promotion is the key for treatment success. Nurses play a major role in providing awareness on copper rich food which have to be avoided, prevention from infections and regular follow up.

Dietary advice and compliance to chelating agents, Zn and iron supplements is important.

#### CONCLUSION

For poet "Eyes are the window to the soul" but for doctors and nurses "Eyes are the window to the diagnosis of various diseases like Wilson's disease". Mast Deepak's life was saved only by the intelligent observation of the health care team.

#### REFERENCES

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