

A Case Report

Management of Wilson's disease in a 5 year old patient

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Abstract

A rare inherited disorder known as wilson's disease is characterized by excess copper accumulation in liver, brain and other vital organs in the body. Present case was a 5 year old child showing symptoms of jaundice, shortness of breath, fatigue, anorexia, ascitis, pain abdomen, Kayser Fleischer's ring etc. after which he was diagnosed to have wilson's disease. On assessment, the patient was found to have anemia, low ceruloplasmin, hypoalbuminemia underweight and low calorie intake. As per protocol, the patient was recommended a low copper diet in which beef liver, cashews, black eyed peas, vegetable juice, shellfish, mushroom and cocoa were excluded. At the time of discharge, the patient was found to be improved in terms of ascitis, shortness of breath, jaundice and food intake.

Keywords: Copper, ceruloplasmin, Wilson, ascitis, CLD, jaundice, keyser Fleischer's ring

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Introduction

A 5 years old boy belonged to Sangaria town in Hanumangarh, Rajasthan was admitted to the hospital. The patient belonged to middle income group joint family including sixteen members. By occupation, his father was a farmer and he just started to going school.

Case history

At the time of admission, the patient had symptoms of jaundice i.e. yellow discolouration of eyes and nails. He was admitted in a hospital at Sriganganagar for 3 days where he was advised liver transplant. Then he was admitted to a government children's hospital at Jaipur for 11 days and then was referred to Mahatma Gandhi College and Hospital under Department of Hepatology.

Family history

Grandmother was found to be hypertensive.

Investigations

Parameters	Result (date of admission)	Reference value
Complete blood count (CBC)		
White blood cells	4.9 10^3 /ul	5.0 – 15.0 10^3 /ul
Red blood cells	3.18 10^3 /ul	4.0 – 5.2 10^3 /ul
Platelet count	104 10^3 /ul	200 – 490 10^3 /ul
Hemoglobin	8.6g/dL	11.0 – 14.0 g/dL
Electrolytes		
Serum sodium	138 mmol/L	137.0 – 145.0 mmol/L
Serum potassium	3.2 mmol/L	3.5 – 5.1 mmol/L
Serum chloride	107 mmol/L	98.0 – 107 mmol/L
Renal function test		
Blood urea nitrogen	23.4 mg/dL	Child 15.0 – 45.0 mg/dL
Serum creatinine	0.4 mg/dL	Child 0.5 – 1.25 mg/dL
Serum uric acid	2.7 mg/dL	3.5 – 8.5 mg/dL
Liver function test and protein		

Serum Bilirubin total	0.7 mg/dL	0.2 – 1.3 mg/dL
Serum Bilirubin Direct	0.5 mg/dL	<0.3 mg/dL
Serum Bilirubin Indirect	0.2 mg/dL	0.1 – 0.9 mg/dL
SGOT / AST	69.9 U/L	15.0 – 46.0 U/L
SGPT / ALT	56.5 U/L	13.0 – 69.0 U/L
Serum Total proteins	6.5 g/dL	6.3 – 8.2 g/dL
Serum albumin	2.3 g/dL	3.5 – 5.0 g/dL
Ceruloplasmin	10 mg/dL	18 – 46 mg/dL
Thyroid function tests		
Triiodothyronine (T3)	0.6 ng/ml	0.97 – 1.69 ng/mL
Thyroxine (T4)	3.4 ug/dL	5.53 – 11.0 ug/dL
Thyroid stimulating hormone (TSH)	2.66 uIU/mL	0.46 – 4.68 uIU/mL

Table 1: Investigations of the patient

Eye Test: Provider uses a special lamp (a slit lamp) to check for brown Kayser-Fleischer rings in eyes.

CT Scan: CT Scan was done to check the copper accumulation in liver and ascitic fluid level.

Final Diagnosis: CLD, Ascites and Wilson's disease

- Eye test showed positive Kayser - Fleischer rings in his eyes.
- CT Scan - Fluid in peritoneal cavity was 150 ml., an indicator of mild ascites
- Copper accumulation was found in the liver
- Ceruloplasmin was below normal levels

Medical management

Medicines	Dosage	Action
Cap Ciamine 250 mg	1 in afternoon	To treat wilson's disease
Tab Usodic 300 mg	½ - ½ tablet in morning and night	Antioxidant/ anti-inflammatory
Tab Rifadin 400 mg	½ - ½ tablet in morning and night	Antibacterial

Syp looz 15 mg	1 in night	Laxative
Tab larizole junior 30 mg	1 in morning	Antacid
Lamino GI Plus	2 scoop/day	Nutraceutical

Table 2: Medical treatment

Nutritional management

1. Assessment of nutritional status

1.1 Anthropometric measurements at the time of admission

Parameters	Current parameters	Reference value
Weight	16 kg	18.3 kg (RDA 2024)
Height	116 cm	100.3 – 117.8 cm
Mid upper arm circumference	18.4 cm	>13.5cm

Table 3: Anthropometric measurements

1.2 Biochemical reports: These have been previously mentioned as a part of medical assessment.

1.3 Clinical Signs & symptoms: As stated earlier, the patient had yellow discoloration of eyes and nails, ascites, weakness, fatigue, anorexia, shortness of breath, nausea etc.

1.4 Nutritional History: Patient's 24 hour diet recall ^[1] was converted to nutrient intake using Indian food composition tables (2017) ^[2] that has been showed as below -

24 hours dietary recall

Time	Menu	House hold measurement
Food intake		
9.00 am	Milk	½ glass
	Biscuits	4-5
12.00 pm	Chapati	4
	Dal	1 bowl
3.00 pm	Chapati	3
	Dal	1 bowl
5.00 pm	Fruits (apple)	1
Nutrient intake		
Energy	1099 kcal	

Protein	43.08 gm
Fat	29.2 gm
Carbohydrate	203 gm
Sodium	439.3 mg
Copper	5.9 mg
Calcium	2527 mg

Table 4: Home diet recall and nutrient intake

2. **Nutritional diagnosis:** Patient was found at risk of malnutrition due to poor diet intake as evidenced by SGA rating B.
3. **Nutritional Intervention:** High protein low salt low copper low fluid diet was recommended ^[1].

Goals of Medical Nutrition Therapy (MNT)

Short term goals:

- To improve food intake
- To relieve ascites, anorexia, fatigue, gastro esophageal reflux

Long term goals:

- To educate patient and family for low copper diet.
- Maintain the overall nutritional quality in diet.
- To impart education about hygiene and hygienic foods after discharge

Nutritional requirements (according to the RDA Table, 2024)

Energy: 1632 Kcal (EAR x stress factor of 1.2)

Protein: 24 g/day (RDAs x stress factor 1.5)

Fat: 36 g (20% total calories)

Carbohydrate: 300 gm (74% of total calories)

Sodium: 1300 mg

Copper: 1.7 mg (For adults as per RDA, 2024, No recommendation has been given for pediatric population). So it has been taken as reference maximum limit.

Liquids: 1-1.5 L

Prescribed diet in hospital

Time	Meal	Menu - Day 1	Menu - Day 2	Menu - Day 3
6:00 am	Early morning	Milk (100ml) Biscuits- 2	Milk Biscuits	Milk (100ml)
9.00 am	Breakfast	Milk (100ml) Breakfast	Milk Breakfast	Milk (100ml) Breakfast
1.00 am	Lunch	Salad Chapati – 1 Khichdi – 1 bowl Dal – ½ bowl Vegetables – 1 bowl Curd – 1 bowl	Salad Chapati Khichdi Dal Vegetables – 1 bowl Curd – 1 bowl	Salad Chapati – 1 Khichdi – 1 bowl Dal – ½ bowl Vegetables – 1 bowl Curd – 1 bowl
4.00 pm	Evening	Egg white – 1	Egg white – 1	Egg white -1
7.00 pm	Dinner	Salad Chapati – 1 Dal – ½ bowl Vegetables – 1bowl Dalia – 1 bowl	Salad Chapati – 1 Dal – ½ bowl Vegetables – 1bowl Dalia – 1 bowl	Salad Chapati – 1 Dal – ½ bowl Vegetables – 1 bowl Dalia – 1 bowl
9.00 pm	After dinner	Milk – 100ml LGI plus 1scp/day	Milk- 100ml LGI plus 1sc/day	Milk (100ml) LGI plus 1sc/day
Nutrient intake				
Energy (Kcal)		1058	1058	1008
Protein (g)		47	47	46
Carbohydrates (g)		186	186	167
Fat (g)		40.5	40.5	40
Sodium (mg)		1684	1684	1680
Copper (mg)		7.24	7.24	6.75

Table 5: Hospital diet for 3 days

4. Monitoring & Evaluation

4.1 Recall of the prescribed 3 day's hospital diet:

	Energy (kcal)	Protein (gm)	CHO (gm)	Fats (gm)	Sodium (mg)	Copper (mg)
Day 1	1058	47	186	40.5	1683.7	7.24
Day 2	1058	47	186	40.5	1683.7	7.24
Day 3	1008	46	166	40.1	1683.1	6.75
Total	3124	140	538	121.1	5050.1	21.23
Average	1041	46.6	179.3	40.3	1683.3	7.07
% intake of the requirements	64	194	60	112	129	415%

Table 6: Average nutrient intake in comparison to requirements during hospitalization

The monitoring of hospital diet intake showed quiet high protein and copper intake as the child was eating some nuts, roasted chana and chocolate also from outside which were not allowed. Therefore, the patient and the attendants were counseled again and discharge diet was given accordingly.

Discharge diet

Exchanges in discharge diet

Food Group	Exchange	Energy (kcal)	Protein (gm)	CHO (gm)	Fat(gm)	Sodium (mg)	Copper (mg)
Cereals	3 + 2 biscuits	444.2	9.48	76.8	1.6	2.44	0.96
Pulses	½	48.85	3.58	7.85	1.5	1.5	0.42
Milk	3	168	9.78	14.7	13.2	75.3	0.6
Egg	1	11.2	2.1	-	0.01	41.5	0.023
Veg	2	46.36	1.6	8	1.2	17.6	0
Fruits	1	62.3	0.2	13.1	0.6	1.4	0.1
Fats	4	180	-	-	20	-	-

Sugar	6	120	-	30	-	-	-
Sago	2	210	0.12	52.6	0.12	-	-
Total		1360	39	167	39	1341	2.90

Table 7: Food exchanges in discharge diet prescription

Detailed menu of discharge diet

Timings	Meal	Menu	Food item	Options	Amount	Household measures
7:00 am	Early Morning	Milk with 2 parle-G biscuits	Milk	-	50ml	1 cup
			biscuits	-	2	
8:00 am	Breakfast	Milk with poha + veg	Milk		100 ml	½ glass
			Poha	Upma/ khichdi/ rabadi/	200 gm	1 bowl
			Veg (cooked)	Lauki/ tinda/ kaddu	100 gm	½ bowl
11:00 am	Mid morning	Fruits + sago khichdi	Any fruit	Apple/pomegrante/ papaya	100 gm	1 bowl
			Sago(socked)	-	100 gm	1 bowl
1:00 pm	Lunch	Chapati 1+ veg+ rice+dal+curd	Chapati	-	30 gm	1
			Veg	Lauki/turai/ tinda/ gobhi	50 gm	½ bowl
			Dal	Moong dal/ chana dal/ arhar dal	25gm	¼ bowl
			Rice	-	30 gm	1 bowl
			Curd	-	100 gm	1 bowl
3:00 pm	After lunch	Sago khichdi	Sago (socked)	-	100 gm	1 bowl
5:00 pm	Evening Tea	Egg white	Egg	-		1
7:00 pm	Dinner	Chapati 1+ veg+dal+rice	Chapati	-	30 gm	1

			Veg	Lauki/turai/ tinda/ gobhi	50 gm	½ bowl
			Dal	Moong dal/ chana dal/ arhar dal	50 gm	¼ bowl
			Rice	-	30 gm	1 bowl
9:00 pm	Bedtime	Milk	Milk	-	50 ml	1 cup
		LGI Plus				2 scoop

Table 8: Discharge diet prescription

Counseling points:

Patient and his parents were counseled for the following dietary recommendations:

- Limit intake of wheat products in limited amount
- Take white bread, mango, papaya, pineapple, spinach, tomato in limited amount
- Eat foods that are rich in protein such as milk and milk products, egg white
- Drink limited amount of water in a day
- Eat Calcium rich foods such as milk and milk products, etc.
- Take 2-3 gm salt per day
- Take liquids only 1-1.5 L/day

Foods to be avoided:

- Soy and soy products, fish, organ meat, dry fruits (raisin, munakke)
- Chocolate, chocolate products, cocoa, chocolate cream
- Cold drinks, soup, juice
- Processed foods such as sauces, pasta, noodles, foods, pickles, etc.

Patient was asked to come for follow up after 1 month and re-evaluation at the time of discharge will be done at following criteria

- Relevant Anthropometry: changes in weight
- Biochemical Parameters: all the reports to check the current parameters
- Diet Compliance: Patient was on high protein low salt low fluid diet

Discussion

Liver functions are progressively deteriorated in chronic liver disease. It is an ongoing process of inflammation of liver parenchyma that leads to fibrosis and cirrhosis [5]. Many symptoms were apparent in the present case such as shortness of breath, fatigue, anorexia, that may be attributed to ascitic fluid accumulation. As he was diagnosed with Wilson's disease, which is a rare inherited disorder of accumulation of excess amount of copper in liver, brain and other organs. This disease is mostly seen in children and young adults between the age of 5 and 35 years [6]. Symptoms of jaundice and pain abdomen were due to Wilson's disease. Kayser Fleischer's ring was a typical symptom of Wilson's disease [7]. Wilson's disease is also associated to liver cirrhosis, liver failure, persistent neurological problems, kidney problems, psychological and blood problems. To relieve all these symptoms, a low copper diet was recommended in which beef liver, cashews, black eyed peas, vegetable juice, shellfish, mushroom and cocoa were excluded.

Conclusion

The study concluded that nutritional needs are drastically increased in Wilson's disease and CLD with ascites. It can be prevented or treated through early diagnosis of nutritional status, which allows timely decisions to be made regarding the right nutritional intervention of the patient. The patient's nutritional intake was improved with his physical health and diet compliance was seen as per dietitian's advice.

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