

A Case Report

Nutritional management of a patient suffering with Gaucher's disease

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Abstract

Gaucher disease is a rare inherited lysosomal storage disorder with multisystem involvement, commonly affecting the haematological, hepatic, and metabolic systems. This case report describes a 28-year-old Hindu female admitted to Mahatma Gandhi Medical College and Hospital, Jaipur, presenting with oral ulcers, fever, lower respiratory tract infection, weight loss, joint pain, pancytopenia, and impaired liver function, along with a past history of tuberculosis. Biochemical investigations revealed leukopenia, thrombocytopenia, anaemia, elevated liver enzymes, hypoalbuminemia, and electrolyte imbalance. Based on these findings, a beta-glucosidase leukocyte test was performed, confirming the diagnosis of Gaucher disease. The patient received symptomatic medical management, including antimicrobials, hepatoprotective agents, antiemetics, and nutritional supplements. Nutritional assessment showed undernutrition with a body mass index of 17.32 kg/m², low protein status, and excessive intake of empty calories. The patient was classified as Subjective Global Assessment grade B, indicating moderate malnutrition. A personalized nutrition intervention comprising a high-calorie, high-protein, and low-fat diet with micronutrient supplementation was implemented to address hypermetabolism, thrombocytopenia, impaired liver function, and bone health concerns. Monitoring during hospitalization demonstrated gradual improvement in dietary intake. At discharge, the patient was advised to continue the prescribed diet, lifestyle modifications, and regular follow-up. This case emphasizes the importance of early diagnosis and individualized nutritional management in improving clinical outcomes and quality of life in Gaucher disease.

Keywords: Gaucher's disease, bone health, hyperlipidemia, anemia, thrombocytopenia, nutritional management

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Introduction

A 28 years old housewife Hindu female was admitted to Mahatma Gandhi Medical College and Hospital, Jaipur who belonged to middle income group and number of her family members was seven.

Present complaints: Her present complaints were oral ulcer, fever, lower respiratory tract infection, pancytopenia, history of tuberculosis, joint pain, weight loss, blood stained sputum, dry cough, hepatitis. Clinicians get her investigations done as mentioned below:

Parameters	Result values		References
	DOA- 06/12/21	DOD 13/12/21	
WBC	2.19 $10^3/\mu\text{L}$	3.47 $10^3/\mu\text{L}$	4.0-10.00 $10^3/\mu\text{L}$
RBC	4.41 $10^3/\mu\text{L}$	4.39 $10^3/\mu\text{L}$	4.50-5.50 $10^3/\mu\text{L}$
HB	11.7 g/dl	11.8 g/dl	13.0- 17.0 G/DL
HCT	37.5 %	35.3 %	40- 50 %
MCV	85.0 FL	80.4 FL	83.0-101 FL
MCH	26.5 PG	26.9 PG	27. 0- 32 .0 PG
MCHC	31.2 g/dl	33.4 g/dl	31.5- 34.5 G/DL
Platelet count	110 $10^3/\mu\text{L}$	70 $10^3/\mu\text{L}$	150- 410 $10^3/\mu\text{L}$
RDW- CV	16.2 %	18.0 %	11.6- 14.0 %
PDW	----- FL	---- FL	11- 16 FL
MPV	----- FL	---- FL	6- 11 FL
PCT	---- %	----- %	0.15- 0.50 %
Polymorphs	68 %	87 %	40-80%
Lymphocytes	18 %	05%	20.0-40%.0
Monocytes	08 %	08%	2.0-10.0%
Eosinophils	00 %	00%	1.0-6.0%
Basophils	00 %	00%	0.0-2.0%
ESR	38 %		-

Prothrombin time	17.9 second	-	CONTROL PT 12.0 SECONDS
Creatinine	0.6 mg/dL	-	0.52-1.25 mg/dL
LFT			
S. Bilirubin Total	1.6 mg/dl	1.2 mg/dl	0.2- 1.3 mg/dl
S. Bilirubin Direct	1.3 mg/dl	0.6 mg/dl	< 0.3 mg/dl
S. Bilirubin Indirect	0.3 mg/dl	0.8 mg/dl	0.1- 0.9 mg/dl
SGOT/ ALT	90.9 U/L	85 U/L	13.0- 69.0 U/L
SGPT/ AST	355.9 U/L	218 U/L	15.0- 46.0 U/L
Total protein	4.9 gm	6.6 gm	6.3 – 8.2 gm
Albumin	2.1 gm	3.2 gm	3.5 – 5.0 gm
Globulin	2.8 gm	3.1 gm	2.0 – 3.5 gm
A/G Ratio		0.8	1.2-2.5:1
Electrolytes			
Serum Sodium	127.4 mmol/L	139 mmol/L	137- 145 mmol/L
Serum Potassium	4.12 mmol/L	4.69 mmol/L	3.5-5.1 mmol/L
Serum Chloride	98.2 mmol/L	101 mmol/L	98.0- 107 mmol/L

Table 1: Biochemical investigations of the patient

On the basis of biochemical investigations, that showed pancytopenia, impaired LFT with lower total protein and albumen levels, the medical team get her beta glucosidase leukocyte test after which final medical diagnosis was Gaucher disease. Gaucher disease may also lead to cardiovascular problems, hypertension and syndrome X in later life. These factors should be taken care of at the time of management. Enzyme replacement therapy (ERT) or substrate reduction therapy (SRT) are another line of treatment besides diet and lifestyle related modifications ^[1].

Medical management: Medical team started following medications to treat and relieve symptoms:

MEDICATION	DOSAGE	MECHANISM OF ACTION
TAB RAGLAN	10 GM	Antiemetic
INJ PANTOP	40 MG	Antacid
TAB FOLVITE	5 MG	Nutritional supplement
INJ OPTINEURON	1 AMP	Nutritional supplement
TAB MONTAIR	1 TAB	Anti- allergic
TAB FABIFLU	400 MG	Anti - viral
TAB URSOCOL	300 MG	Hepatoprotective
INJ TRANEXAMIC	500 MG	Antifibrinolytic
CAP BECASUL	1 TAB	Nutritional supplement
INJ EMSET	4 MG	Antiemetic
TAB MOXIFLOXACIN	400MG	Antibiotic

Table 2: Prescribed medications for the patient

Nutrition care process

The nutrition care process includes four steps:

1. Nutritional assessment
2. Nutritional diagnosis
3. Nutrition intervention &
4. Monitoring and evaluation

1. Nutritional assessment: It is done on the basis of anthropometric measurements, biochemical reports, clinical signs and symptoms and dietary recall.

1.a Anthropometric measurements

Anthropometric Measurements for adults	
Weight	45.4 kg
Height	162 cm
BMI	17.32 kg/m ²
Target/Adjustable body weight	57cm
Triceps Skin fold thickness	22cm
Waist circumference	67cm
Hip circumference	105cm
W/H ratio	0.63cm
BMR/REE	1106.6 kcal
Physical activity (calculate as per NIN charts)	
	1.04

Table 3: Nutritional status assessment through anthropometry

1.b Patients biochemical reports at the time of admission and discharge have been mentioned in above table 1.

1.c Patients clinical signs and symptoms were oral ulcer, fever, lower respiratory tract infection, weight loss, anorexia that affected her food intake and were important from nutrition point of view.

1.d Patient's 24 hours home diet recall ^[4] showed her nutrient intake as below –

- Energy: 2345 kcal
- Proteins: 64 gm
- Carbohydrates: 351 (60%)
- Fat: 65 gm (25%)
- Empty calories: 585 kcal

2. Nutritional diagnosis: Patient was found to be at risk of malnutrition due to excess fat and empty calorie intake as evidenced by SGA grading B and BMI in the category of underweight grade I, pancytopenia, anemia, low protein levels, impaired liver function and impact of gaucher's disease.

3. Nutrition Intervention: As gaucher disease is characterized with marked hypermetabolism, insulin resistance and dislipidemia. Weight loss, celiac disease and cholelithiasis are other complications ^[2] that may occur in near future or in later life. Bone health is also affected, ^[3] therefore a low fat, high calorie and high protein diet was recommended with vitamin D supplementation as per following goals -

Short term goals

1. To control fluid & electrolytes losses from vomiting.
2. To overcome the abdominal pain.
3. To maintain low platelet count and stop thrombocytopenia.
4. To control elevated Liver Enzymes.
5. To maintain Hemoglobin as normal level.
6. To provide small frequent soft meals to reduce discomfort.

Long term goals

1. To prevent macronutrient & micronutrient deficiency.
2. To enhance overall quality of life.
3. To provide nutrition education to the patient and the care giver
4. To treat symptom & prevent complication.

Nutrient requirements

- Energy: 2340 kcal ($IBW \times 30 \times \text{Stress factor} = 65 \times 30 \times 1.2$)
- Proteins: 78gm ($IBW \times 1.2 = 65 \times 1.2$)
- Carbohydrates: 351 gm (60%)
- Fats: 52 (20%)

- Sodium: 2000mg
- Potassium 3500 mg
- Fluid requirements: 2.275 Lt (2275ml= IBW*35= 65*35) 35ml/ kg ABW/day
- NNC: 2028 kcal
- Cal: N² ratio: 2340: 12.48
- Consistency and frequency of meals: High Protein Normal diet (3 major & 3 minor meals)

Exchanges of the prescribed diet

Food group	Exchanges	Energy (Kcal)	Protein (g)	Carbohydrates (g)	Fat (g)	Sodium (mg)	Potassium (mg)
Cereals	12	1152.96	38.04	230.4	4.8	7.32	1119.6
Pulse	3.25	317.52	23.27	51.02	1.3	9.75	1236.3
Vegetables	4	92.72	3.2	16	0.4	35.2	1012
Milk	5.5	308	17.6	25.3	16.5	242	0
Egg	2	134	13.2	0	9	123	138
Fat	3	135	0	0	15	0	0
Salt	1 tsp	0	0	0		2000	0
Total		2140.20	95.31	323.72	47	2417.2	3505.9
Percentage		91.45	122.19	92.22	90.38	120.86	100.16

Sample diet included following meals

Time	Meal	Menu	Household measures
6 am	Early morning	Tea	1 cup
		Marie biscuits	2
8 am	Breakfast	Milk	1 glass

		Upma	200 g
11 am	Mid morning	Eggs	2
1 pm	Lunch	Salad (cucumber)	100 g
		Chapati	4
		Chana dal	1 bowl
		Tinda veg	100 g
		Khichdi	1 bowl
		Curd	1 bowl
3 pm	Evening	Milk	1 glass
		Marie biscuits	2
7 pm	Dinner	Salad (cucumber)	100 g
		Chapati	4
		Mix dal	200 g
		Capsicum1 veg	100 g
		Dalia	1 bowl

4. Monitoring and evaluation

Patient's 3 day's hospital diet intake¹ recall was as below-

	Energy (Kcal)	Protein (g)	Carbohydrate (g)	Fat (g)	Sodium (mg)	Potassium (mg)
Day 1	1500	62.7	203.6	49.1	1400	1650
Day 2	1217	52.93	181.56	34.63	1878	2079
Day 3	2251	96.93	335.55	52.01	2450	3717
Total	5022	212.56	720.71	135.74	5728	7446
Average	1674	70.85	240.23	45.24	1909.33	2482

Percentage	71.54	90.83	68.44	87.01	95.46	70.91
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Discharge diet: Patient was discharged at a high calorie, high protein, low fat normal diet with following dietary recommendations:

- Consume balanced diet that should be full of cereals, pulses, vegetable, fruits, milk & milk product.
- Increase intake of Iron rich foods such as- Vegetarian- Cereals, beans, green leafy vegetables, beetroot, tomato, dried apricots, pomegranate & dates etc. Non vegetarian- liver, kidney, brain, white egg, lean meat, fish
- Consume good fat omega 3 & omega 6 fatty acids such as- canola oil, walnuts, wheat germ oil, sunflower oil, soybean oil & coconut oil etc.
- Avoid intake of containing high levels of saturated and trans-fat such as- Fried foods, sauce, cookies, cakes, pastry, poori, bhature, paratha, chips samosa and kachori etc.
- Increase intake of liquid for preventing dehydration. Fruits juice, vegetable soup, buttermilk, coconut water & lemon water.
- Consume protein rich food such as- Milk, white egg, paneer, chhena, curd, sprouts, fish & lean meat
- Take 25ml of papaya leaf juice 1 weak for increase platelets count.
- Increase intake of kiwi fruits in the diet.

Patient was advised to come for follow-up after 1 month for evaluation of efficacy of the prescribed diet at the time of discharge and to monitor diet compliance.

Conclusion

The present study shows that adequate and timely nutritional assessment and care can aid in faster recovery of a patient even with multiple health ailments. As in the present case, who was suffering with Gaucher's disease, which was associated with poor bone health, anemia, pancytopenia and impaired liver function test, still a team of clinicians and

dietitians maintained patients nutrient intake close to her requirements, therefore patient revealed faster recovery and was discharged on a full diet.

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