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Nursing Care Plan for a Child Diagnosed with Lathosterolosis

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Abstract

Lathosterolosis is a very rare autosomal recessive cholesterol metabolism disorder. The disease manifests itself with developmental and mental delays, learning disabilities, microcephaly, facial dysmorphism, bilateral cataracts, and skeletal defects. It is caused by the deficiency of the enzyme sterol-C5-desaturase, which is involved in cholesterol biosynthesis. This deficiency prevents the synthesis of cholesterol, which acts as a precursor for lipid, bile acids, and steroid hormones. Until 2020, only 7 cases had been reported. In this case report, it was aimed to plan and implement the nursing care of a 2-year-old boy diagnosed with lathosterolosis with the diagnoses of the North American Nursing Diagnostic Association (NANDA).

Keywords

Lathosterolosis, Cholesterol Metabolism, Nursing Care Plan

1. Introduction

Lathosterolosis is an extremely rare disorder of cholesterol metabolism. It is an autosomal recessive disease. It is caused by the deficiency of the enzyme sterol-C5-desaturase, which is encoded by the SC5D gene, which catalyzes the conversion of latosterol to 7-dehydrocholesterol, the second final step of cholesterol biosynthesis. This deficiency prevents the synthesis of cholesterol, which acts as a precursor for lipid, bile acids, and steroid hormones. The disease manifests itself with developmental and mental delays, learning disabilities, microcephaly, facial dysmorphism, bilateral cataracts, and skeletal defects. Lathosterolosiswas first reported by Brunetti-Pierri in 2002. Only 7 cases have been reported by 2020 [1] [2] [3] [4].

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The limited information about the disease, the challenge for distinction of symptoms with other diseases, and the physician's lack of sufficient knowledge about rare diseases make the diagnosis process difficult. In the diagnosis process, detailed physical examination and anamnesis, genetic tests, laboratory tests are used. The aim of treatment is to relieve symptoms and increase the patient's quality of life. Lack of information about the disease, economic difficulties experienced in the treatment process, mood changes experienced during the adaptation process to the disease, social isolation and increased care responsibilities are the difficulties brought by the disease for the parent who is in the caregiver role [5].

In this case report, a boy with a diagnosed of lathosterolosis accompanied by immunodeficiency, adrenal insufficiency and cholestasis, who was treated at Balcalı Training and Research Hospital, was examined. While the case was being handled, it was aimed to plan and implement care with the diagnoses of the North American Nursing Diagnostic Association (NANDA).

Date of The Data Was Received: 02.01.2022

Name: D.B

Age: 2 Years 1 Month

Gender: Boy

Diagnosis: Lathosterolosis, Immunodeficiency, Adrenal Insufficiency, Cholestasis, Hepatosplenomegaly

Gestational age: 36 weeks

Date of Birth: 29.11.2019

Birth type: Cesarean Section

Birth Weight: 2400 gr

Birth Height: 45 cm

Allergy: Milk, Egg, Spices, Vinegar, Beef

Family History

Father: 38 years old, self-employed.

Mother: 38 years old, housewife, 4th pregnancy, parity 3, 3 living children. She was monitored during her pregnancy due to reason she was a carrier of hepatitis B. She also used insulin injection during her pregnancy. There is a cross-cousin marriage between parents.

Child's Vital Signs and Anthropometric Measurements: It is shown in Table 1.

Table 1. Child's vital signs and anthropometric measurements.

Body Weight	8 kg (under 3% percentile)
Height	74 cm (under 3% percentile)
Head Circumference	43 cm (under 3% percentile)
Body Temperature	36˚C
Pulse	131/min
Blood Pressure	100/60mmHg
Respiratory Rate	24/min

2. Medical History

The patient with immunodeficiency is receiving intravenous immunoglobulin (IVIG) treatment at a dose of 1 g/kg every 20 days. The patient, who was discharged 1 month ago, was hospitalized in the pediatric gastroenterology clinic for further examination and treatment after the polyclinic control due to the complaints of jaundice, fever, vomiting, resistant diarrhea, malnutrition, appetite and weight loss.

Genkort was added in treatment plan according to the Synacten test (ACTH) to the patient who was treated with teicoplanin, tazobactam and ganciclovir on the day of hospitalization. On the 9th day of hospitalization, the patient underwent endoscopy, colonoscopy and simultaneous phimosis operation. Biopsy was taken during the procedure and sent to pathology. After the phimosis operation, Thiocilline cream applied on the wound area.

On the 12th day of hospitalization stools was green and diarrhea continued. In the stool examination Giardia and Vancomycin-Resistant Enterococci were grown therefore Metronidazole was added to the treatment.

In order to start total parenteral nutrition (TPN) for the child whose diarrhea continued, central venous catheterization was performed on the 15th day of hospitalization and TPN was started as 12 × 5 cc per day. In the following days, the dose of TPN was gradually increased and Peptamer was added to the diet enterally.

According to colonoscopy results grade 2 esophageal varices were detected on the 16th day of hospitalization. Hepatosplenomegaly was diagnosed according to portal doppler.

Minimal pericardial effusion was detected in the echocardiography taken on the 17th day of hospitalization.

Since the INR was 1.33 in the blood tests performed on the 21st day of hospitalization, a single dose of vitamin K was administered. Since his diarrhea continued, probiotics were added to the patient's treatment. Gentian violet (triphenylmethane compound) was started on the patient with gland dermatitis findings.

Periorbital edema was observed on the 26th day of hospitalization. Orbital MRI was planned after the ophthalmology consultation.

Erythrocyte Suspension was given on the 28th day of hospitalization due to low HTC after the examinations made because the patient's body temperature increased.

On the 31st day of his hospitalization, tachypnea developed and he was aspirated. A chest x-ray was taken and blood gas analysis was performed. Oral intake was stopped and O_2 support was dissolved with a high-flow nasal cannula. When the respiratory rate reached the normal limit, O_2 support was stopped with a high-flow nasal cannula and O_2 support was continued with a reservoir mask.

Central venous catheter was removed on the 35th day of hospitalization. Diaper dermatitis continues with bleeding. A loading dose of Keppra was administered due to leg tremors that continued for three days. Trombocyte suspension

loading was performed due to decreased platelet level.

On the 45th day of hospitalization, the patient continued to be fed with formula, received IVIG treatment and was discharged after service consultation.

Physical Examination Finding and System Examination

Skin

The patient has jaundice and hyperpigmentation that has been going on for 2.5 months, and there is dryness in the scalp and hair. The mother of the patient said that she is using olive oil and vaseline for dryness and itching in his bod. The patient has progressive diaper dermatitis.

Head-Neck

The patient has red eye and orbital edema. Common candida albicans (mikostatin is applied) and 4 - 5 tooth decay were seen in the mouth. Her mother stated that the patient had gingival bleeding and epistaxis 3 - 4 times a month.

The respiratory system

The child has intermittent cough with wheezing, vomiting, hemoptysis, and tachypnea.

Genitourinary System

Color of the urine is yellow, pH: 7

Gastrointestinal System

The child has diarrhea 5 - 7 times a day and vomit which including mucus once a day. The child has loss of appetite, weight loss, abdominal pain, and abdominal edema. His stool is yellow and watery.

Neuromuscular System

The child has a joint pain, weakness, fatigue, hypotonia and inability to walk.

Extremities

Pain and edema in the joint: The child does not allow to touch and is agitated.

Laboratory Results

Hemoglobin 7.8 g/dl (12 - 16)

Hematocrit 22.6% (35 - 49)

Leukocyte $3.2 \times 10^3/mm^3$ (4 - 12)

PLT 22 $10^3/\mu l$ (176 - 373)

INR 1.33 (0.85 - 1.2)

Uric acid 3.9 mg/dl (3.03 - 5.8)

Bilirubin (total) 3.33 mg/dl (0.2 - 1.2)

Creatinine 0.17 mg/dl (0.3 - 1)

AST 106 U/L (0 - 40)

ALT 87 U/L (8 - 39)

BUN 29 mg/dl (5 - 18)

CRP 23 mg/dl (0 - 5)

Potassium 4.5 mEq/L (3.7 - 5.5)

Calcium 9.15 mEq/L (7.0 - 12.0)

Sodium 136 mEq/L (135 - 145)

Nursing Diagnoses

It is at the end as a table (Table 2).

Table 2. Nursing diagnoses and care plan according to the patient's data (arranged according to NANDA Diagnostic Taxonomy) [7].

Nursing Diagnoses	Specifications	Aim	Nursing Interventions	Evaluation
Deficient Fluid Volume: Fluid loss due to loss of appetite, malnutrition and diarrhea	Poor oral intake, dryness of skin and mucous membranes, loss of body weight	Regulation of fluid intake in accordance with age and metabolic requirements. Regulation of signs and symptoms of dehydration.	- Intake and output was monitored. Noted incerased and decreased urinary output. - Infusion rate of parenteral fluids was monitored closely. - The patient was weighed daily. - Vital signs was monitored regularly. - Hydration and dehydration status were evaluated (fontanelle, mucous membrane, skin turgor, tears). - Laboratory findings and blood gas tests were checked.	The signs of dehydration tend to decrease, the oral mucous membrane is slightly moist.
Impaired Oral Mucous Membrane: Due to lack of fluid volume, common oral candida albicans, decayed teeth and O_2 support with reservoir mask.	Dryness and redness of the oral mucosa, common oral candida albicans	Ensuring the integrity and moistness of the oral mucosa.	- The infant's oral mucous membrane was evaluated daily. - Redness in the mouth and sores on the tongue, tissue integrity, moistness were observed. - Moisturizers were applied to the rim and lips to prevent chapping and drying. - Oral care was applied with sodium bicarbonate diluted with physiological saline solution every six hours. - Mikostatin was administered as ordered by the doctor. - The mother was informed about oral care and its importance. - The child was hydrated.	There is still oral candida albicans and mild redness. The situation continues.
Imbalanced Nutrition: Less Than Body Requirements: Associated with anorexia and oral candida albicans	Losing weight, eating less than the recommended daily amount and switching to parenteral nutrition	Reaching the ideal nutritional level.	- The child was weighed every morning at the same time and in the same clothes. - Intake and output was monitored. Noted incerased and decreased urinary output. - Head circumference, height and weight percentile values were followed and recorded. - The child underwent central venous catheterization and TPN was started. - In addition, Peptamer food was added to the diet. - The amount of food given was recorded regularly. - Laboratory results were followed.	TPN and parenteral nutrition were discontinued on the 35th day of hospitalization. She continues to be fed with Peptamer formula.
Delayed Growth And Development: Immunodeficiency associated with adrenal insufficiency	Body weight and height below the expected percentile (height, body weight and head circumference below the 3% percentile)	Ensuring the child's age-appropriate growth and development.	- A nutrition plan was created according to the needs of the child. - Height, body weight and percentile values were checked at regularly. - Its care is planned in the same time period. Frequent rest periods were planned.	Height, body weight and head circumference are below the 3% percentile. There is developmental delay. The child does not grow and develop in accordance with his age.

Continued

İmpaired Tissue (Skin) İntegrity: Because of edema, phimosis operation and invasive procedures	Damaged skin surface	Protection of skin integrity.	- Central venous catheter site and operation area were checked daily and wound care was performed. - The probe was changed every 3 - 6 hours when the patient had a vital monitor probe. - Unnecessary invasive intervention is avoided. - The position of the child was changed every 6 hours.	The patient does not have erythema or redness that disrupts skin integrity.
Fear: Due to the effort to cope with the rare and incurable disease, lack of knowledge, the abundance of painful interventions and prolonged hospitalization.	Child cries during treatments or sees healthcare worker and parent seems nervous	Ensuring the child's adaptation to the healthcare worker and the hospital environment. Informing parents and supporting their participation in their child's care.	- A calm and safe environment was provided for the child and family. - Parent was informed before all procedures. - It was ensured that the parent was with the child during the procedure. - It was tried to ensure that the child was taken care of by the same nurse as much as possible. - It is ensured that the child's favorite toys are with him. - Parents were encouraged to voice their fears and concerns.	The child's crying time during interventions decreased. It has been observed that the family trusts the health workers and exhibits more adaptive behaviors.
Diarrhea	Frequent (at least 3 times a day), watery and misshapen stools, increased intestinal sounds, continued antibiotic therapy	Normal consistency and number of stools.	- The child underwent central venous catheterization and TPN was started. In addition, Peptamer was added to the diet. - The amount of food given was recorded regularly. - Probiotic and Zinco syrup added to the treatment. - The child was hydrated. - The child was weighed every morning at the same time and in the same clothes. - Hydration and dehydration status were evaluated (fontanelle, mucous membrane, skin turgor, teardrop). - Laboratory findings and blood gas tests were checked regularly.	Diarrhea continues.
Acute Pain: Because of the painful procedures such as CVP and IV catheter insertion, endoscopy, colonoscopy, and diarrhea	Score of visual pain scale is 3, anxious look and groaning	To reduce or eliminate existing pain.	- Vital signs were checked regularly - Pain was evaluated with visual analog scale (VAS) and recorded. - Nonverbal pain behaviors were observed in the child. - Pain was tried to be reduced by using therapeutic techniques such as distracting the child's attention and keeping favorite toys with him. - When necessary, analgesics were administered.	The child's vital signs are stable. VAS pain score was evaluated as 2. The child continues to look anxious but there is no groaning.
Fatigue: Because of the immunodeficiency, adrenal insufficiency and hospitalization and persistent diarrhea	Weakness, constant fatigue, hypotonia, inability to walk	Relieving fatigue.	- The amount of light, heat and sound of the environment has been adjusted. - Unnecessary invasive procedures were avoided. - Interventions were planned to provide resting periods for the child.	Weakness, fatigue, hypotonia continues in the child.

Continued

Risk for Secondary Infection: Associated with presence of central venous catheter, gland dermatitis, surgical operation and prolonged hospitalization	CRP: 23 mg/dl (<0.5%)	Preventing the child from having a secondary infection other than the current infection.	- He was checked in terms of signs and symptoms (fever, chills, local or general skin discoloration) that may develop due to infection. - Hands were washed before and after each contact with the patient. Aseptic technique rules were followed in all invasive procedures. - Visitor restriction applied. - The family was informed about daily hygiene and diaper dermatitis. - Unnecessary invasive procedures were avoided. - The patient was started on topical treatment due to diaper dermatitis. (gentian violet)	The child has an infection. Intravenous antibiotic therapy is administered. No secondary infection was observed in the patient.CRP: 8.88 mg/dl (<0.5%)
Risk For Bleeding: INR: 1.33 and decrease in platelet level	Risky value of laboratory tests showing signs of bleeding	Absence of signs and symptoms of bleeding.	- Bleeding signs and symptoms were checked. - Vital signs were measured at regular intervals. - Laboratory findings were checked for Pt, Aptt, Inr and platelet count. Platelet transfusion was performed according to the results. - Unnecessary invasive procedures were avoided and the interventions were done gently. - The patient was protected against bumps and falls. - The caregiver parents were informed about the symptoms of bleeding and ways to protect themselves. - Kvit was applied at the order of the physician.	No signs of bleeding were observed in the child.
Risk of electrolyte ımbalances: Associated with resistant diarrhea and malnutrition	Caused by resistant diarrhea and malnutrition	Fluid electrolyte values have to be within normal limits, fluid intake and output have to be in balance	- Vital signs were checked regularly. - The daily fluid balance was checked. - Serum electrolyte levels were checked regularly.	For now the fluid intake and output are in balance.

3. Discussion

Lathosterolosis is an inherited disorder in the cholesterol synthesis mechanism. Until 2020, only 7 patients have been reported. While the age at diagnosis varies from 22 months to 10 years, some cases were diagnosed after death.

In this case report, there is a nursing care plan for the solution of the current problems of a 2-year-old boy diagnosed with latosterolosis. Nursing diagnoses suitable for NANDA diagnostic taxonomy were determined in the study. These diagnoses are; delayed growth and development, imbalanced nutrition: less than body requirements, deficient fluid volume, impaired tissue (skin) integrity, impaired oral mucous membrane, fear, diarrhea, acute pain, fatigue, risk for secondary ınfection, risk for bleeding, fluid and electrolyte ımbalances. Interventions suitable for the diagnoses were planned and the results were evaluated (Table 2).

When our case was compared with a 7-year-old boy who was diagnosed with lathosterolosis for the first time in 2002; portal hypertension, cholestasis and neurological hypotonia are observed in both cases. Although bilateral cataract

was seen in the first reported case, in our case, eye redness and orbital edema were present [1].

Our case was diagnosed with hepatosplenomegaly according to portal dopplers taken on the 16th day of hospitalization. Abnormal liver tests and hepatosplenomegaly were also observed in the third and sixth reported cases [3] [6].

Since there are no case reports involving nursing care in the literature about the disease, it is anticipated that the case will contribute to the literature and nursing care plans.

4. Result

Evaluation of the cases that we do not see frequently in the clinics within the framework of the nursing care plan is important in terms of increasing the standard and quality of nursing care. Case reports guide the use of evidence-based nursing approach in clinics. For this reason, there is a need for nurses to carry out studies that include nursing care, which is adapted to cases in uncommon diseases.

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Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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