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CASE STUDY

A rare case report on Hurler syndrome with umbilical hernia

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Hurler syndrome is also known as MPH I. A hereditary sickness of the lysosome is a result of a deficiency in the enzyme alpha-L-iduronidase, which breaks down glycosaminoglycans (GAG or mucopolysaccharides). As a result, tissue harm from dermatan sulfate and heparin sulfate worsens over time until death. Umbilical hernias are ventral hernias that arise at or across the umbilicus. This situation of a case report describes the surgical treatment of an umbilical hernia in a 4-year-old pediatric patient, a known case of Hurler syndrome. The patient received treatment for an umbilical hernia, a confirmatory test was performed for Hurler syndrome, and the only patient complaint related to it—corneal clouding—was treated.

Keywords: Hurler syndrome, alpha-L-iduronidase, mucopolysaccharidosis, umbilical hernia

1. Introduction

Hurler also known as mucopolysaccharidosis type I, is one of the 11 mucopolysaccharidoses (MPS) diseases (MPH I). Hurler syndrome was formerly known as gargoylism. It is a lysosomal disorder that is inherited and is caused by a deficiency in the enzyme alpha-L-iduronidase, which breaks down glycosaminoglycans (GAGs or mucopolysaccharides). Heparin sulfate and dermatan build up as a result, causing numerous tissues to eventually degenerate and perish. The purpose of the multidisciplinary team in treating patients with Hurler syndrome is explained in this exercise, along with the pathogenesis, assessment, and therapy of the disorder (1). Hurler's disease is characterized by nearly normal-appearing newborns, and generic rhinitis and inguinal hernia are the most typical early signs (2).

An umbilical hernia is a tear in your abdominal muscles that occurs just below your belly button and allows a part of your intestine to protrude through the navel. Umbilical hernias are often benign and frequent. Umbilical hernias most usually occur in infants; however, they can also occur in adults (3). Children who have an inadequate closure of the fascia of the umbilical ring may develop an umbilical hernia, allowing intra-abdominal contents to protrude (4).

1.1. Co-relation between Hurler syndrome and umbilical hernia

Congenital umbilical hernias in Hurler syndrome (mucopolysaccharidosis I) are normally treated conservatively because the risks of surgical repair outweigh the rarity of complications like incarceration. Primary fascial closure was used to urgently treat the hernia; elective surgical hernia repair may be recommended in some Hurler syndrome instances (warning signals of rupture) (5). A major anesthetic danger exists due to the rising frequency of airway problems (6). Cardiopulmonary issues manifest later in life, and as a result, the majority of children pass away before turning 10 years old. Patients with MPSIH who received HSPC gene therapy experienced a considerable metabolic improvement in both their peripheral organs and central nervous system (7). Complications of umbilical hernia

Abbreviations: HSPC, Hematopoietic stem and progenitor cell; MPS I, mucopolysaccharidosis type I; USG, Ultrasonography.

surgery include issues with anesthesia and localized wound infections (8).

2. Case presentation

A 4-year-old male patient was admitted with complaints of swelling in the umbilical (since 2 years), approximately 10 × 10 cm. corneal clouding, broad saddle nose, thick lips, and large tongue. After 1 month, the patient was admitted for wound dehiscence in an o/c/o umbilical hernia.

2.1. Physical examination

Upon physical examination, the patient had a mild pallor. On general examination, the patient was fair, cooperative, and coherent (CNS: conscious and oriented; CVS: S1, S2 (positive), NO murmurs noted; RS: CLEAR (-); GIT: soft, non-distended).

Patient's body weight: 10 kg

Height: 80 cm.

2.2. Vitals

Respiratory rate (RR): 16 breaths/minute; pulse rate (PR): 59 beats/minute; temperature: 98°F; oxygen saturation (SpO_2): 98%.

2.3. Patient's past medical history

The patient has been diagnosed with Hurler syndrome.

2.4. Laboratory investigations

See **Table 1**.

2.5. Diagnostic tests

The patient has inflammation of the organ; swelling started appearing after 1–2 years of age, which is relatively small in size and has approximately increased by 7 × 7 cm until now (4 years) without a case of pain, fever, or vomiting.

- MPS screen: mucopolysaccharidosis – positive
- USG abdomen: Approximately 5.3 cm defect is noted in the anterior abdominal wall, in the umbilical region omentum, and bowel loops are seen to herniate.
- X-ray of the chest: The anterior ribs and clavicles have widened. Humeral diaphyseal widening with osteopenia is noticed. Osteopenia and diaphyseal expansion of the distal radius and ulna are observed. Hand metacarpals' proximal tapering is seen.
- X-ray of the cervical spine-lateral view: Reduced vertebral body height and intervertebral disc space noted involving the cervical spine, resulting in a short neck. A kyphotic deformity involving the thoracolumbar spine was noted.
- X-ray of the skull: Frontal bossing of the cranium was noted.

2.6. Final diagnosis

Correlating with the obtained subjective and objective data along with the history, the final diagnosis was confirmed to be an umbilical hernia in a known case of Hurler syndrome.

2.7. Plan of analysis

See **Table 2**.

3. Discussion

A 4-year-old patient presented to the hospital with the symptoms of swelling in the umbilical, corneal clouding, a broad saddle nose, thick lips, and a large tongue. The patient was previously diagnosed with Hurler syndrome based on the diagnostic test (**Table 1**). The patient was successfully operated on for an umbilical hernia, and after a follow-up of 1 month, the patient was again admitted for wound dehiscence. The patient was admitted again, and wound dressing was done with the ointment sofradex (Framycetin and Dexamethasone) and Betnovate-N (Betamethasone valerate and Neomycin).

By aiding early identification and treatment, newborn screening for mucopolysaccharidosis type I (MPS I) has

TABLE 1 | Laboratory findings.

Parameters	Obtained value	Normal range	Interference
Hemoglobin	9.0 g/dl	11.5–14.5 g/dl	Anemia
WBCs	5400 cells/cmm	5.0–14.5 cells/cmm	Normal
Neutrophils	56%	40–80%	Normal
Lymphocytes	42%	20–40%	Normal
Total rotein	5.60 g/dl	6.0–8.0 g/dl	Malnutrition
Serum Globulin	2.10 g/dl	2.3–3.6 g/dl	Malnutrition
Potassium	5.40 mEq/L	3.5–5.5 mEq/L	Hyperkalemia
Urea	11.00 mg/dL	5–25 mg/dL	Malnutrition
Creatinine	0.54 mg/dL	0.12–1.06 mg/dL	Malnutrition

TABLE 2 | Drug management.

Sr. No	Formulation	Generic name	Brand name	Dose	Route	Frequency
1.	Syrup	Ibuprofen	Ibugesic	5 ml	Oral	TDS
2.	Syrup	Amoxicillin + clavulanic acid	Augmentin	5 ml	Oral	TDS
3.	Suspension	Timolol eye drop	Timol		Topical	BD
4.	Inj.	Ceftriaxone	Rocephin	600 mg	IV	12-hourly
5.	Inj.	metronidazole	FLAGYL	200 mg	IV	8-hourly
6.	Tab.	pantoprazole	Pantop	20 mg	Oral	1 BD
7.	Syrup	Multivitamin	Becosules	5 ml	Oral	BD

the potential to enhance outcomes. However, the ability of MPS I diagnostic tests to predict whether a child would have the severe central nervous system disease linked to Hurler syndrome or the minimal or absent central nervous system involvement linked to the attenuated phenotypes is limited (Hurler-Scheie and Scheie syndromes). The lack of a trustworthy predictive biomarker makes therapeutic decision-making for infants diagnosed through newborn screening more difficult because the best course of treatment varies between Hurler syndrome and the attenuated MPS I phenotypes. The management of newborns with Hurler syndrome (**Table 2**), input into treatment choices, and evaluation of treatment efficacy and prognosis may all benefit from the knowledge of the condition's natural history. The typical tiny neck of umbilical hernias will increase the hazard of confinement and strangulation. The herniated sac can cause spinal problems if it bulges or irritates the spine. A sciatic hernia is a condition where the herniated sac compresses the sciatic nerve. Sciatica is the term used in an article that describes the pain brought on by sciatic nerve inflammation (9). Hurler's syndrome-related umbilical hernias have a tendency to grow significantly, which makes surgical correction extremely challenging and increases the risk of recurrence. The accompanying spinal abnormalities make spinal anesthesia contraindicated.

4. Conclusion

Hurler syndrome is a rare diagnosis in children; however, allogeneic bone marrow transplantation is successful in treating Hurler syndrome. Early bone marrow transplantation in children is likely to be very beneficial. But because there are no matching donors, patients/medical professionals do not consider treatment plans. Therefore, in this case, only umbilical hernias were treated using the open hernia repair technique and were monitored until the condition returned to normal. Hurler syndrome was not adequately treated. The complaint of corneal clouding received treatment.

Author contributions

All authors contributed equally to the study. AP has collected the data from the patient and drafted it. GC assisted with the development of the manuscript. RS paraphrased the document. SN edited and drafted the final manuscript.

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