# HUGE UMBILICAL HERNIA WITH SKIN ULCERATION AND BOWEL PROTRUSION IN A PATIENT WITH MUCOPOLYSACCHARIDOSIS: A CASE REPORT

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## ABSTRACT

Mucopolysaccharidosis (MPS) is a rare inherited storage disease caused by defect or absence of a lysosomal enzymes that result in systemic deposition of MPS recently called glycosaminoglycans (GAG) with development of a spectrum of diseases of different presentations and severity. Hurler disease is a subtype of (MPS I) caused by α L-iduronidase enzyme deficiency in the lysosomes.

A thirteen-year-old female mentally retarded patient, short stature with multiple congenital abnormalities admitted to Sulaimaniyah emergency hospital in the Kurdistan of Iraq. She had a huge irreducible umbilical hernia with ulceration and protrusion of the omentum and small bowel from the hernia sac diagnosed after admission as Hurler syndrome. She was treated successfully as an emergency case and the patient discharged well following a challenging operation.

**Keywords:** *Hernia, Hurler disease, Mucopolysaccharidosis, Ulcerated umbilical hernia, Anesthetic risks in mucopolysaccharidosis.*

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## INTRODUCTION

Mucopolysaccharidosis (MPS) is a rare type of storage disease with incidence equals or less than 1:100,000 births with no ethnical difference.The age of onset and the progression is variable among the involved patients[1].

It results form defect or deficiency in one of the lysosomal enzymes causing a spectrum of progressive disorders with different tissues and organ involvement [2] Most of the patient's appearance is normal early in life then gradually worsen with advancement of the age, phenotype of those patients shows short stature, dysostosis multiplex (skeletal deformities), coarse facial features, hepatosplenomegaly, airway narrowing, cardiovascular abnormalities and mental retardation[1] .

MPS results from accumulation of Glucosamine glycan a macromolecule (GAG) metabolite's, in the lysosome with gradual and progressive impairment of cell physiology, (GAG) is shown to have a major role in the elastic and micofibril structure of the extracellular matrix and it is part of cell wall material [3] .

MPS has many types, Hurler disease is one of MPS I group inherited as an autosomal dominant caused by deficiency of α-L-iduronidase enzyme [1] .

Around 75% of patients with MPS I undergo at least one surgical intervention the median of 3-4 operations per patient across all MPS types. Among the common surgeries are repair of different types of hernias, tonsillectomy, carpal syndrome myringotomy,

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orthopedics procedures cardiac valve surgeries and tracheostomies [2].

## CASE PRESENTATION

A thirteen-year-old girl was admitted to Sulaimani teaching hospital on the 7th of October 2013 her stature was short, mentally retarded with coarse facial features, prominent forehead, and an enlarged protruded tongue.

She presented with a large irreducible multiloculated umbilical hernia protruding from a 4 cm defect in the abdominal wall with distortion of the umbilicus, the overlying skin was atrophied with visible dilated veins and two by three cm ulcer, a four cm long segment of the omentum was protruding from the ulceration. She was complaining from attacks of abdominal pain but no vomiting nor constipation.

She also had a 2 years older sister with the same features underwent elective surgery for umbilical hernia in another center but she died during the anesthesia, the cause of death was not clear.

She has been admitted to the Emergency Room kept nil-by mouth iv fluid and broad-spectrum antibiotics was given.

Her condition was discussed with pediatrician, anesthetist, plastic surgeon and otolaryngologist. Diagnosis of Hurler syndrome was made and precaution for possible complication was taken.

While preparing the patient for surgery a 15 centimeter segment of small bowel protruded outside the skin defect with the omentum after the patient had a bout of cough.

The patient was taken to the operation room with otolaryngologist team stand by for possible tracheostomy in case of failure of establishing airway. Fortunately endotracheal intubation was successful which performed with great precaution and minimal hyperextension of the neck.

An elliptical incision was made around the hernia the bowel dissected and the protruded omentum excised, the bowel washed with warm saline, then reduced and returned back to the peritoneal cavity the defect closed with unexpectedly no tension as the tissues was quite lax then the wound close.

The patient discharged after 48 hours with smooth post operative period apart from mild atelectasis which was treated successfully with physiotherapy, expectorant, mucolytic, After 10 days the stiches removed sent to pediatrics hospitalfor enzyme replacement therapy and follow up, she is also been followed up by our surgical team for possible development of incisional hernia.

## DISCUSION

Patients with mucopolysaccharidosis have high incidence of different types of hernias among those are inguinal hernia which should be treated surgically with high possibility of recurrence, and umbilical hernia which should be left untreated unless it's too large or complicated [4].

Emergency operation on a mucopolysaccharidosis patient with complicated huge umbilical hernia is a challenge for both the surgical and the anesthetic team and needs multidisciplinary approach with involvement of pediatrician, otolaryngologist and possibly plastic surgeon [5] as MPS patients have more potential operative mortalities and anesthetic risk due to multiple associated inherited abnormalities including chronic obstructive airway diseases with pneumonia, respiratory failure, cardiac valve diseases mainly involving mitral and aortic valves, hypertension and neurological disorders like epilepsy [6].

Among those upper airway obstruction is well known serious risk as establishing and maintaining air way by mask and endotracheal intubation can be challenging due to craniofacial abnormalities, large tongue, narrow air upper airways, for these reasons laryngeal mask airway can be more appropriate than the formal ways, though airway can be established best with the aid of fiber-optic scope.

It's also advisable to time the elective operation properly, or avoid the operative interventions requiring general anesthesia if possible,as the operative mortality is high [2, 5-7]. Other studies showed that patients who have received L-iduronidase enzyme replacement or who underwent hematopoietic stem cell transplantation earlierwould have less perioperative complication rate, as it would prevent the progression of the disease [8, 9].

Long-term follow up is required for possible development of incisional or other types of hernias and to diagnose new arising conditions.

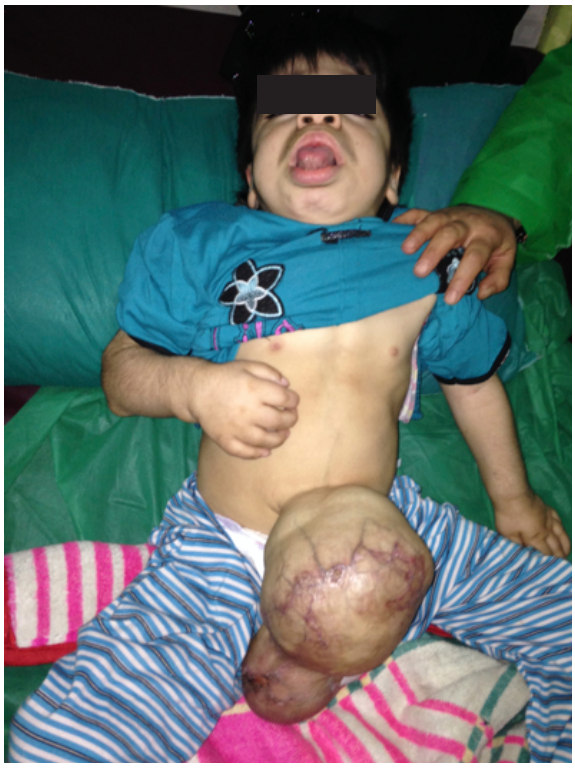

**Figure 1 . On admission.**

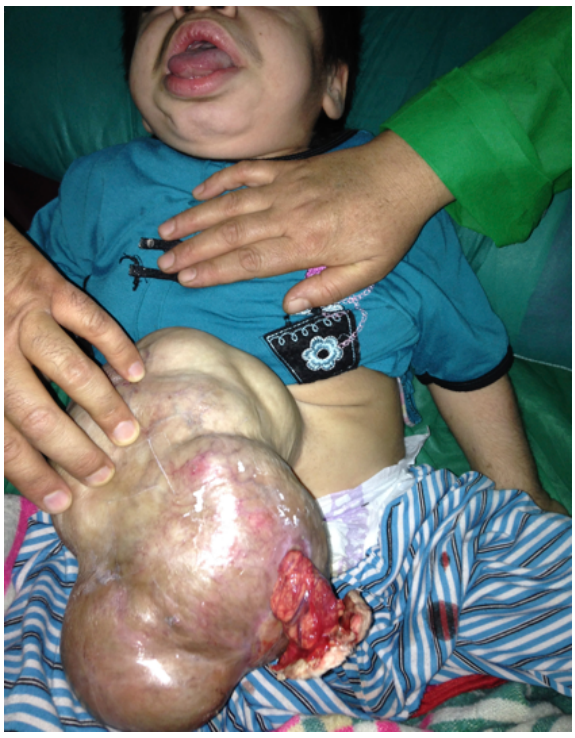

**Figure 2. On admission lower part of the hernia omentum out through ulcerated skin.**

**Figure 3. 24 hours after admission small bowel and omentum out after a bout of cough.**

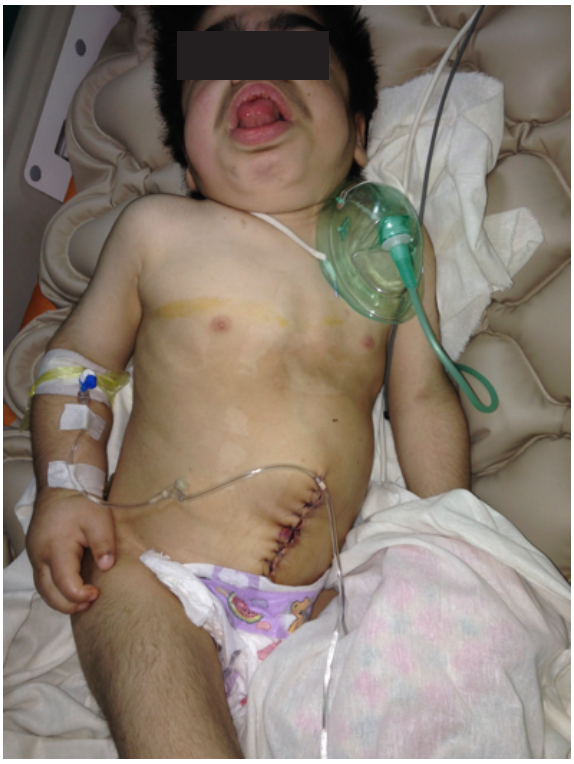

**Figure 4. 24 hours after operation.**

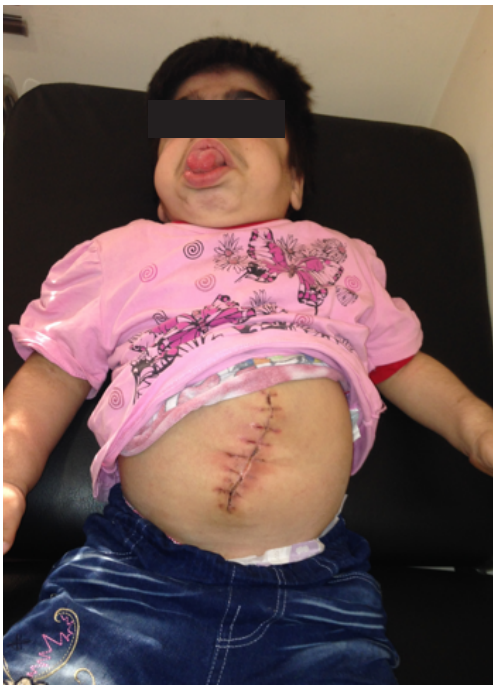

**Figure 5. 10 days after operation.**

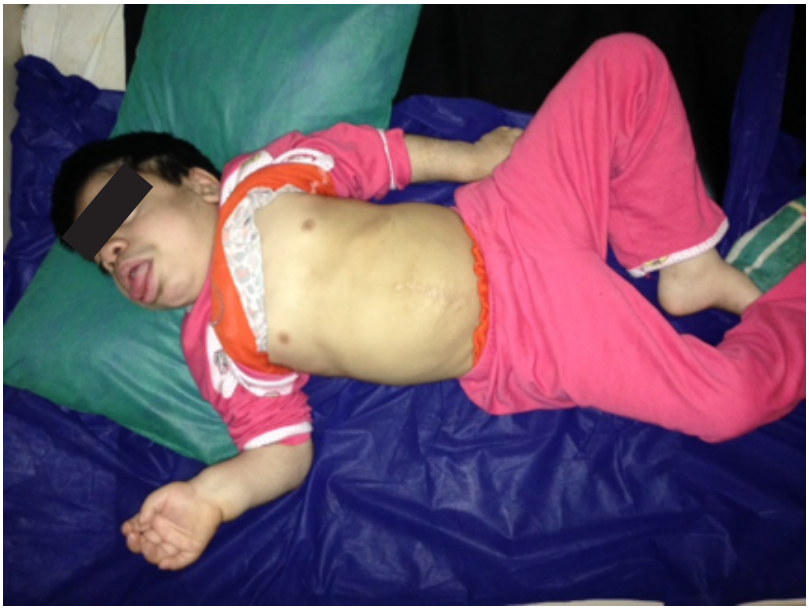

**Figure 6. One year after the operation.**

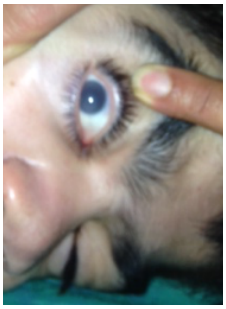

**Figure 7. Cloudy eye.**

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