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Ehler Danlos Syndrome, Surgical Challenges - A Case Report

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I. INTRODUCTION

The first detailed description of Ehlers-Danlos syndrome was provided by A.N. Tschernogubow in 1891. Thirty years later, Edward Ehlers and Henri Alexander Danlos named the condition. In the 1960s, the Berlin nosology identified 11 subtypes of EDS. In 1998, the Villefranche nosology revised this classification to six types with descriptive names. More recently, the EDS International Consortium identified 13 subtypes. The prevalence of EDS is about 1 in 5000 people. Among the subtypes, the hypermobile type affects 1 in 5000 to 20000 people, and the classical type occurs in 1 in 20000 to 40000 people[1].

Ehlers-Danlos syndrome encompasses a group of inherited disorders affecting connective tissue, classified into 13 subtypes. The hypermobility subtype is the most prevalent, while the vascular subtype poses the greatest risk. This syndrome impacts genes that code for connective tissues, such as collagen, fibrillin and matrix proteins resulting in loose ligaments, tissue fragility and poor healing. It primarily manifests through joint hypermobility and its complications, abnormal skin texture and repair and various hollow organ and vascular dysfunctions[2]. Although it may be of minor concern to physicians, surgeons find operating on these patients challenging due to issues like unexpected bleeding and handling of delicate tissues.

Surgical complications frequently observed in patients with Ehlers-Danlos syndrome include poor healing, postoperative bleeding, and hematoma formation. Patients who undergo surgery for inguinal hernias often experience recurrences, requiring multiple operations[3]. Inguinal hernias in these patients resemble those seen in elderly individuals, characterized by a thin transversalis fascia and a significantly dilated internal ring. Mesh repair has shown a low chance of recurrence in these cases[3]. Due to prior surgical complications, surgeons often anticipate unsatisfactory outcome in future surgeries, leading to many procedures being deferred. Sometimes, the diagnosis of the syndrome is made after the patient has already undergone surgery and complications arise. Early diagnosis can help surgeons and anesthetists take necessary precautions and follow specific protocols when managing such cases.

Even vein cannulation is difficult in such patients as there observed tearing of vessel wall and extravasation of blood and fluid[4]. Relatively minor trauma may result in rupture of a major blood vessel with subsequent death due to uncontrollable hemorrhage[5].

II. MATERIAL AND METHODS – CASE REPORT

A 10-year-old male child with Ehlers-Danlos syndrome and CTEV presented with bilateral non palpable undescended testes and right inguinal hernia. He underwent a right herniotomy 3 years ago, which was uneventful with no intraoperative or postoperative complications, and there was no recurrence. 1year ago, he had a laparoscopy for bilateral intrabdominal testis and bilateral Fowler-Stephens stage 1 surgery was done. Post-surgery, he developed cellulitis at the umbilical port site, healing was delayed and had to take dermatology opinion finally it healed after taking 4 months.

After a year , the patient was taken up for laparoscopic bilateral Fowler-Stephens stage 2 surgery. During the incision for port introduction tissues were found to be easily torned. Fortunately, there was no significant bleeding, and extra care was taken during the surgery. A week later, the patient was reviewed, the wound site appeared healthy, and the child was doing well.

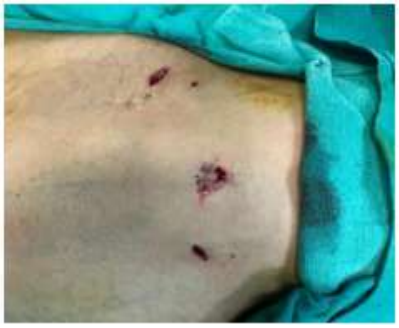

Figure 1 Skin Tear

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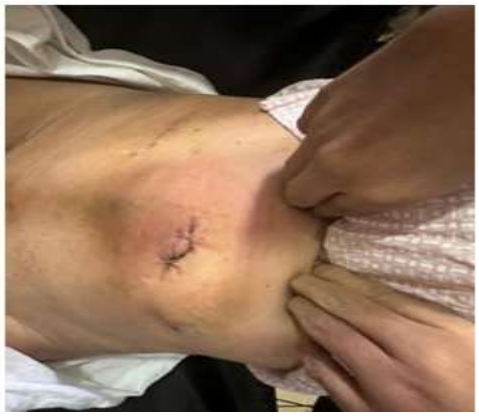
Figure 2 Healed Umbilical port (After 1 week)

III. DISCUSSION

Ehlers-Danlos syndrome (EDS) is an inherited autosomal dominant connective tissue disorder characterized by a wide spectrum of manifestations, ranging from mild symptoms to severe, life-threatening complications. Clinical features include skin hyperelasticity, joint hypermobility, atrophic scarring, and fragility of blood vessels[6]. Approximately 50% of individuals diagnosed with the classical type of EDS have mutations in the COL5A1 and COL5A2 genes, which encode the alpha 1 and alpha 2 chains of type 5 collagen, respectively[7]. The vascular type is primarily caused by pathogenic variants in the COL3A1 gene [8].

Diagnosis of EDS relies on comprehensive history-taking, including family history, physical examination, and additional investigations such as biochemical tests. Advanced DNA analysis techniques, including targeted DNA analysis and next-generation sequencing, are also employed [9].

In addition to surgical complications associated with the disease, EDS itself presents numerous complications, with vascular and organ ruptures being the most severe and potentially fatal [6,10]. Patients with connective tissue disorders planning for any surgical procedure should first be evaluated for anaesthesia risks, the expected outcome of the surgical procedure and any complications that could arise [4].

In our study, a 10-year-old child, a known case of EDS underwent multiple surgeries. The child experienced poor postoperative wound healing of the umbilical port site which took about 4 months for complete healing. During a later surgery, a skin tear was observed during port introduction, but no other complications were noted. The child was doing well, and the wounds of laparoscopy healed well. Major complications often reported in EDS patients, such as haemorrhage, wound dehiscence, unsuccessful surgery and recurrence of hernia, were not found in this case. Although poor wound healing and a skin tear occurred, they were managed with precise care. Similar outcomes, with no major surgical complications, have been reported in other literatures also [10, 11].

IV. CONCLUSION

Ehlers-Danlos Syndrome (EDS) is often diagnosed when complications arise during surgical procedures. Surgical management of these patients can be challenging and surgeons often defer surgical interventions. In the case of this patient, who had undergone multiple surgeries, initial wound healing was delayed after the first surgery, and a skin tear was observed intraoperatively. However, systematic management yielded good results, and no major complications were encountered in subsequent surgeries. Therefore, a diagnosis of EDS should not be considered a contraindication for surgery. If the benefits outweigh the risks, surgeries can be performed meticulously with proper precautions in patients with EDS.

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