Case Report

Management of Klippel-Trenauny syndrome with multiple organ involvement

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ABSTRACT

Klippel-Trenauny syndrome is a disturbance in the development of the mesodermal and ectodermal tissues occurring in utero, which is characterized by vascular nevi, varicose veins, soft tissue, and occasionally, bone hyperplasia. Our patient is a 6-year-old boy with presentation of left lower extremity overgrowth, abdominal mass, abdominal pain, bilateral buttock mass, rectal bleeding, and skin hemangiomatosis. The major problems of this case were involvement of the levator ani, external anal sphincter, and encasement of the sciatic nerves within the buttock mass. We concluded that the use of muscle and nerve stimulator for detection and saving sphincters and the nerves in these cases could improve the results of surgical resection.

KEY WORDS: Hemangioma, Klippel-Trenaunay syndrome, port-wine stain, soft-tissue hypertrophy, venous system abnormalities

INTRODUCTION

Klippel-Trenauny syndrome (KTS) was first reported in 1900 by Klippel and Trenauny. The KTS is a disturbance in the development of the mesodermal and ectodermal tissues occurring in utero that is characterized by vascular nevi, varicose vein, soft tissue, and occasionally bone hyperplasia, which usually affects one extremity.[1] The rectum and urinary bladder are rarely involved in KTS, but may occasionally be the source of severe blood loss.[2] Here, we report this case of KTS because of 1- multiple organ involvement, 2- benefit of staged operation, and 3- using of muscle and nerve stimulator could facilitate saving the sphincters and nerve branches.

CASE REPORT

A 6-year-old boy, hospitalized on May 12, 2001 with abdominal enlargement, severe hypertrophy of the left lower limb [Figure 1], multiple skin hemangioma, mass in both buttocks and intermittent rectal bleeding. The main problems of the patient were inability to walk and sit normally, intermittent rectorrhagia, and abdominal pain. Rectosigmoidoscopy revealed mucosal hypervascularity and vein tortoises from the dented line up to 20 cm proximally. In laboratory tests, the only positive finding was low Hb (Hb = 8.5 gm/dL).

Abdominal sonography and CT-scan both revealed a huge multiloculated cystic and solid mass in retroperitoneal area prominently in the left side. Staged operation was planned. Indications of surgical treatment were as follow:
1. Abdominal mass and abdominal pain
2. Intermittent rectal bleeding
3. Inability to sit due to the mass of the buttocks.

In the first operation, the retroperitoneal mass that had encased the whole left renal pelvis and left ureter associated with vesical wall invasion, was removed, the ureter and pelvis remained intact. In the second operation, the huge mass of the left and right buttocks were resected. These mass had involved the levator ani muscles and external sphincter of the rectum. The siatic nerves were also encased within the mass in both sides. The muscles and the siatic nerves were dissected safely by using of muscle and nerve stimulator. In the third operation the rectosigmoid colon was removed by the Sweson's procedure. The patient recovered well after all operations. The only complication was collection of serum in the left buttock after 3 weeks of operation,
which was managed by drainage and pressure dressing. For the left lower limb, the patient referred to an orthopedic center. They planned below knee amputation, and using prosthesis for him. In the last visit two years after the first operation, the patient had no complain of abdominal pain, he had no rectal bleeding any more, and he was able to sit normally and was able to walk with his artificial leg.

Histopathology examination showed varicose veins within the bladder muscle coat, and in the intestinal submucosa, respectively [Figure 2]. Histopathologic examination of resected mass of buttocks and retroperitoneal showed cystic lymphatic malformation [Figure 3]. The skin sample had changes like lymphangioma circumscriptum [Figure 4].

**DISCUSSION**

The reported cases had all three features of the syndrome; in this case, there were multiple involvements of different organs, such as bladder, rectosigmoid colon, and retroperitoneal lymphatic tissue and involvement of left lower extremity. KTS has a spectrum of symptoms and signs. The etiology of KTS is not clearly known but few hypothesis on its hereditary source has been suggested.[3] Non-invasive imaging strategy, including color duplex ultrasonography, MRI, lymphoscintigraphy, and plain radiographs usually are used for evaluation of KTS.[4] Most patients with KTS may be managed conservatively. Sclerotherapy is an alternative method of treatment for venous malformations. In one report, Color duplex-guided sclerotherapy was effective in 72% of the patients with involvement of head and neck by venous malformation.[5] The definite indication for operation is a leg length discrepancy projected to exceed 2.0 cm at skeletal maturity, which can be treated with epiphysiodesis in the growing child. Surgery was undertaken in selected cases for complications of the hemangioma, or for cosmetic reasons.[6] In patients with limb overgrowth and the diagnosis of Klippel-Trenaunay-
Weber or Proteus syndromes, surgical treatment consisted of epiphysiodesis, osteotomies, debulking procedures, and amputation can be selected.[7] Here in this case the abdominal mass was composed of pure lymphatic malformation. Rectorrhagia during defecations had been made the patient anemic. Bladder had also vascular malformation but without history of hematuria. Management of the disease is usually conservative, but sometimes surgery is inevitable. In this case because of some functional problems, we had to plan surgical management as mentioned above. Because of a long segment involvement of the rectosigmoid, endoscopic sclerosis did not apply for him. The patient had a good outcome after multiple operations. Conclusion, when there is multiple involvements associate with functional problems, the patient will have benefit of staged operation. Use of muscle and nerve stimulator helps the surgeon to save the involved sphincters and nerves (i.e., sciatic nerve, facial nerve). Swenson's procedure may be used safely if there is diffuse involvement of mid and lower rectum.

REFERENCES


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