PULSE PARA VOLVER AL ÍNDICE

Síndrome de Klippel - Trenaunay – a propósito de un caso

Klippel - Trenaunay Syndrome – case report

INTRODUCTION

Klippel - Trenaunay Syndrome (KTS) is a rare condition that can be classified as a combination of vascular malformations affecting the arterial, venous and lymphatic systems. It is characterized by a clinical triad that includes cutaneous capillary malformations, venous malformations and bone/soft tissue extremity hypertrophy[1]. The etiology in unclear but it is presumed a disruption into mesoderm that compromises angiogenesis[2]. Some investigators suggest that deep venous obstruction/atresia causes chronic venous hypertension, leading to the onset of hemangiomas, varicose veins and limb hypertrophy. Although KTS is a sporadic condition, studies report familial cases that were not inherited from Mendelian pattern, suggesting a multifactorial inheritance[3]. Studies conducted later by Happle suggested that inheritance of a single defective gene could explain the development of KTS as well as the occurrence of sporadic and familiar cases. KTS has an equitable geographical, racial and gender distribution[4]. Clinically there may be changes in the upper or lower limbs, rarely the trunk. Patients may have symptoms ranging from moderate bone hypertrophy, hemangiomas and varicose veins. Occasionally they may have hematuria and hematochezia[5]. Treatment should be adjusted individually for each patient as well as clinical course and prognosis.

CASE REPORT

A 31 year-old male diagnosed with KTS appealed to the emergency department with asthenia and hematochezia (with dejections) with 2 months of evolution. He denied nausea, vomiting or weight loss. At the examination he presented cutaneous hemangioma in the left shoulder, left flank and left thigh (Images 1 and 2). He also had exuberant varicose veins in the left lower limb. The rectal touch was performed without evidence of blood. The study performed showed hemoglobin of 6.7 g/dL, VCM 81 fL, iron 28 g/dL, total iron fixing capacity 287 g/dL, transferrin 220 mg/dL, ferritin 51.4 ng/mL. He was hospitalized due to the needed of transfusion support and to clarify the clinical situation. The study carried out at the hospital revealed an anuscopy with large, congestive and friable internal hemorrhoids that needed an elastic ligation. At the colonoscopy there were no significant endoscopic changes. Upper digestive endoscopy and capsule enteroscopy revealed erythema of the duodenal bulb mucosa and a small ulcer/erosion of congestive edges in the terminal ileon (histologic study revealed no malignancy). The patient underwent thoracic, abdominal and pelvic computed tomography without significant changes (without visceral involvement of KTS). For iron deficiency anemia began oral iron supplementation with progressive improvement.

DISCUSSION

Klippel - Trenaunay Syndrome usually affects a body segment and has a wide range of clinical manifestations[6]. The presence of two of the abnormalities initially described are sufficient to the diagnostic; however, all changes are commonly present, in most patients, at birth or during childhood. The lower limb is the most common site of presentation but there are cases of involvement of more than one body segment, like our patient. Visceral involvement also occurs in about 20% of cases[7]. The involvement of gastrointestinal system by

Figure 1

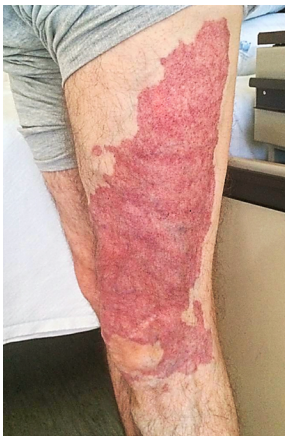
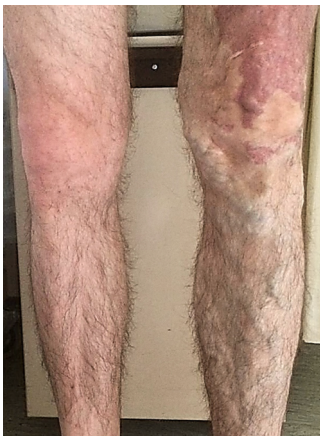


Figure 2



KTS is characterized by the presence of varicose veins (especially in the rectum and sigmoid) that may bleed, as described for our patient[9]. The diagnosis of this pathology is clinical; however some complementary diagnostic tools help to investigate the involvement of various systems and organs by KTS.

REFERENCES

1. Sung HM, Chung HY, Lee SJ, et al. Clinical Experience of the Klippel-Trenaunay Syndrome. Arch Plast Surg. 2015;42(5):552-558.
2. Jacob AG, Driscoll DJ, Shaughnessy WJ, et al. Klippel-Trenaunay syndrome: spectrum and management. Mayo Clin Proc. 1998;73:28-36.
3. Aelvoet GE, Jorens PG, Roelen LM. Genetic aspects of the Klippel-Trenaunay syndrome. Br J Dermatol. 1992;126(6):603-607.
4. Jacob AG, Driscoll DJ, Shaughnessy WJ, Stanson AW, Clay RP, Gloviczki P. Klippel-Trénaunay syndrome: spectrum and management. Mayo Clin Proc. 1998;73(1):28-36.
5. Husmann DA, Rathburn SR, Driscoll DJ. Klippel-Trenaunay syndrome: incidence and treatment of genitourinary sequelae. J Urol. 2007;177(4):1244-1249.
6. Kotze PG, Soares AV, Lima MC, Balidn-Junior A, Sartor MA, Bonardi RA. Síndrome de Klippel-Trenaunay: Uma causa rara de hemorragia digestiva baixa. Rev Bras Coloproct. 2002;22:109-112.
7. Cha SH, Romeo MA, Neutze JA. Visceral manifestations of Klippel-Trénaunay syndrome. Radiographics. 2005;25(6):1694-1697.

Palabras clave: síndrome de Klippel-Trenaunay, malformaciones capilares cutáneas, malformaciones venosas, anemia, hematoquecia.

Keywords: Klippel-Trenaunay syndrome, cutaneous capillary malformations, cutaneous capillary malformations, anemia, hematochezia.

Maria J. Marelo Tavares[1], Gonçalo C. Branco[1], Magda Fernandes[1], Jorge Cotter[1]

[1]*Serviço de Medicina Interna, Hospital Senhora da Oliveira, Guimarães, Portugal*

Correspondencia: mjmt_b@hotmail.com

Cómo citar este artículo: Marelo Tavares MJ, Branco GC, Fernandes M, Cotter J Síndrome de Klippel - Trenaunay – a propósito de un caso. Galicia Clin 2021; 82-1: 59

Recibido: 20/9/2019; Aceptado: 15/10/2019 // https://doi.org/10.22546/60/2093