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Klippel Trenaunay Syndrome with Angiokeratoma Circumscriptum Naeviforme and Bilateral Congenital Anorchia: A Rare Association

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Abstract

Klippel-Trenaunay syndrome (KTS) is not a common congenital vascular abnormality. A trio of capillary malformation, venous varicosities, and bony or soft-tissue hypertrophy define this syndrome. Significant morbidities associated with this illness include bleeding, deep vein thrombosis, and embolic consequences. Angiokeratoma circumscriptum naeviforme (ACN) is indeed a congenital variant of angiokeratoma that appears as a hyperkeratotic plaque on the lower extremity. Bilateral congenital anorchia (BCA) is the total lack of testicular tissue in a male with a normal phenotype and karyotype. KTS has been linked to ACN. Here we presented an 8-year-old male child who came with a swollen left thigh and the right side of his face with overlying blackish nodules on his left thigh and scrotum. The patient was diagnosed as KTS with angiokeratoma circumscriptum naeviforme and bilateral congenital anorchia based on his history, imaging studies and the typical clinical features of the disease.

Keywords

Klippel-Trenaunay Syndrome, Angiokeratoma Circumscriptum Neviforme, Bilateral Congenital Anorchia

1. Introduction

Klippel-Trenaunay syndrome is an uncommon condition characterized by soft tissue overgrowth, varicose veins and port-wine stains. It is induced by a congenital defect of the veins, capillaries, and/or lymphatics. These three descriptions

highlight the key characteristics of the condition, with two of the three confirming the diagnosis. The etiology of KTS is unclear, but it is thought to be caused by an intrauterine insult during early pregnancy [1]. It is an uncommon multisystem illness with a frequency of around 1:100,000 with zero preference for gender, race, or geographical location, and the majority of cases are sporadic [2]. The severity of this condition manifests itself in a wide range of symptoms, from asymptomatic cosmetic abnormalities to severe limb hypertrophy [3]. Angiokeratomas are uncommon vascular lesions characterized by dilated papillary dermal blood vessels and subsequent epidermal alterations such as acanthosis and/or hyperkeratosis. Angiokeratoma circumscriptum naeviforme (ACN), is the rarest kind of angiokeratoma. Clinically, it is characterized by unilateral dark red to blue-black nodules or plaques on the lower legs, thighs, or buttocks [4]. ACN has been observed infrequently in conjunction with KTS [5]. Bilateral congenital anorchia (BCA) is a very uncommon disorder. This condition is also known as testicular regression syndrome or vanishing testis syndrome. The lack of testis in a 46XY person with a male phenotype characterizes the condition [6]. As far as we know, no literature has yet revealed a link between BCA and KTS or ACN. Here, we report a case of KTS with ACN and BCA in an 8-year-old male child.

2. Case Report

An 8-year-old male child of a non-consanguineous marriage presented with a large swollen left thigh and right side of the face with overlying blackish discoloration from birth, slowly progressing to the present size. He also complained of the appearance of multiple small nodules over his left thigh and scrotum. There was also occasional bleeding from the lesions after modest trauma. There was no family history of comparable lesions or a personal history suggestive of a bleeding problem. His developmental milestones were all within the usual range. There was no substantial prenatal and neonatal morbidity or evidence of trauma prior to the development of the lesion.

On examination, the circumference of the left thigh (34 cm) was greater than that of the right thigh (23 cm). There were dark blanchable patches suggestive of port wine stain present over the left thigh, scrotum and right side of the face (**Figure 1**). Examination revealed multiple, small, dark red to black, hyperkeratotic, nodular lesions on the left thigh extending from the left side of the waist up to the lower part of the left thigh and scrotum (**Figure 1**). These were non-pulsatile, tender, and non-compressible when palpated. Auscultation revealed no bruit. The supra-pubic region has a few dilated and tortuous veins. There was also hypertrophy of the left thigh and right side of the face (**Figure 1**). The systemic examination was unremarkable.

Blood investigations including complete hemogram and serum biochemistry profiles were within normal limits except for the hemoglobin level, which was 8.1 g/dl. Radiological examination of the pelvis, including both thighs revealed

soft tissue swelling in the left thigh and inguino-scrotal region with multiple small nodular opaque shadows suggestive of arterio-venous or only venous malformation (Figure 2). The Color Doppler study of the affected limb showed visible multiple tiny swellings in the anterior aspect of left thigh which sonographically is anechoic diffuse tubular structures. There was no evidence of arteriovenous fistula or deep venous thrombosis (Figure 3). Ultrasonography of both testes revealed a markedly thickened and edematous scrotal wall having anechogenecity. There were no testes either in the scrotal sac or abdomen (Figure 4).

Based on these features and consultation with a dermatologist, a diagnosis of KTS with ACN and BCA was made. Parents were counseled regarding the disease, supportive management was given. Also advised biopsy of a skin lesion from the left thigh for histological confirmation of Angiokeratoma circumscriptum naeviforme, but was not possible due to economic constraints.

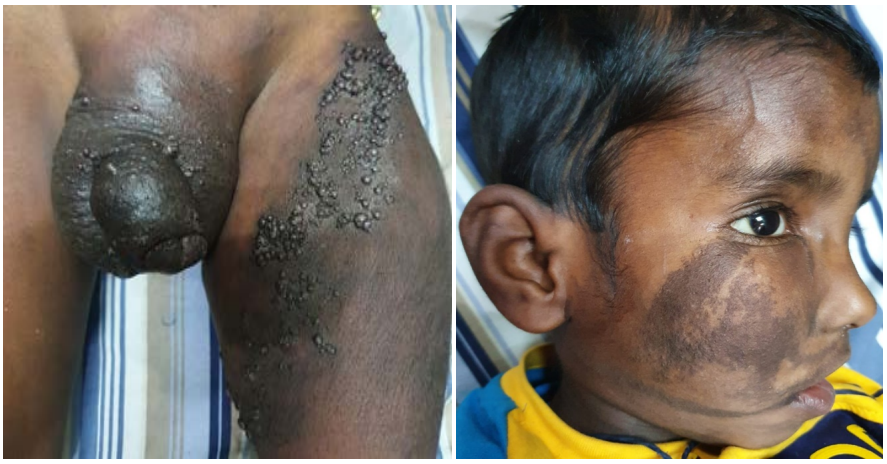

Figure 1. Port wine stains, soft-tissue hypertrophy of the left thigh, and right-sided face. Multiple small blackish nodules over the left thigh and scrotum, few dilated and tortuous veins over the supra-pubic area.

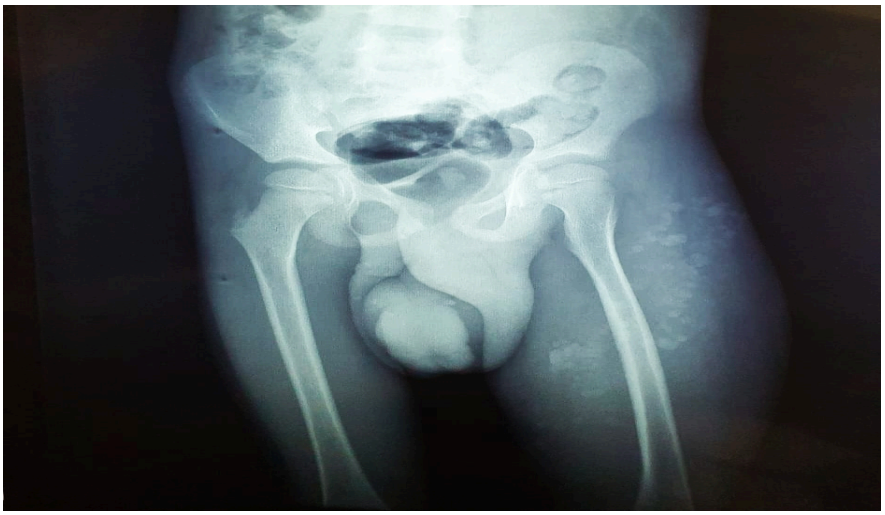

Figure 2. X-ray pelvis including both thighs suggestive of arterio-venous or only venous malformation.

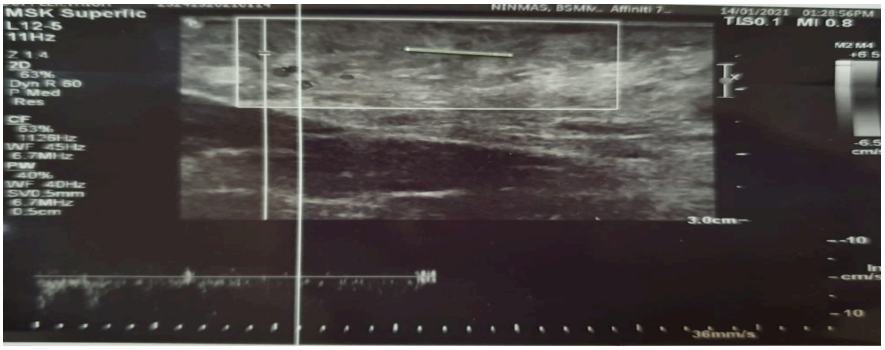

Figure 3. The Color Doppler study of the affected limb showed visible multiple tiny swellings in the anterior aspect of the left thigh which sonographically are anechoic diffuse tubular structures.

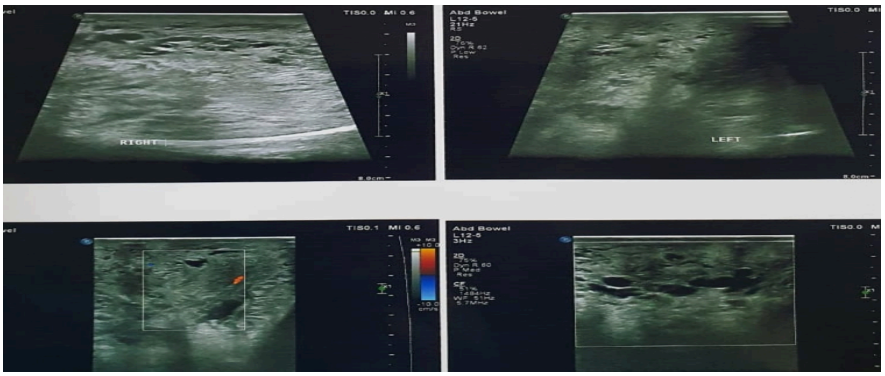

Figure 4. Ultrasound revealed no testes either in the scrotal sac or abdomen.

3. Discussion

The classical Klippel Trenaunay syndrome was described first by Klippel & Trenaunay in 1900. The trio of port wine stain, venous and lymphatic malformation, and soft tissue hypertrophy of the affected extremity describe Klippel-Trenaunay syndrome (KTS) [7]. Men and women of all ethnic groups are equally affected by KTS. The cause of the condition is unknown and several ideas have been postulated. KTS is a congenital disease in which blood and lymph vessels do not form properly during intrauterine development, according to most experts. Patients with KTS present with a wide range of symptoms, ranging from minor port wine stains and a few varicose veins that cause merely aesthetic deformity to debilitating condition caused by enormous limb overgrowths, chronic pain syndrome, skin infections, and arthritis. A study by Jacob AG showed 246 patients (98%) had capillary malformations (port-wine stains), 182 (72%) had varicosities or venous malformations, and 170 had limb hypertrophy (67 percent). In 159 patients (63 percent), all three KTS characteristics were present, and 93 patients (37 percent) exhibited two of the three criteria [8]. Our patient had capillary abnormality-port wine stain, limb hypertrophy and dilated, tortuous vein over suprapubic area. Pain, edema, limb heaviness, and mobility problems are common complaints among KTS patients. Our case had limb heaviness with mobility problem and occasional bleeding after trauma from angi-

okeratoma circumscriptum naeviforme. Complications from vascular abnormalities include bleeding, thrombophlebitis, cellulitis, and ulcerations. Deep venous thrombosis (DVT), pulmonary embolism (PE), congestive heart failure, and recurrent internal bleeding from aberrant vessels in the gastrointestinal system, kidney, and genitalia are all linked to KTS [9]. Klippel-Trenaunay syndrome is diagnosed largely by a clinical interview, previous history, current patient symptoms, and physical examination. Duplex ultrasonography, magnetic resonance imaging (MRI), computed tomography (CT), lymphoscintigraphy, and conventional radiography are all helpful imaging procedures for confirming a diagnosis and assessing the severity of the disease and its co-morbidities [10].

Angiokeratomas are a group of diseases marked by benign vascular ectasias of the papillary dermis and epidermis hyperkeratosis, papillomatosis, and acanthosis. At birth, red-colored macules are the most common symptom. After a few years, the lesion changes into warty and/or hyperkeratotic violaceous nodules or plaques with dark red to blue violaceous characteristics [11]. They are most often found unilaterally on the lower extremities, although they have also been identified in other places, such as the trunk, neck, scrotum, and tongue. The affected regions are usually sensitive to palpation, well-circumscribed, firm, nonpulsatile, and non-compressible on physical examination. Angiokeratoma circumscriptum lesions have no tendency to heal on their own [12]. Our case had same features with bleeding tendency after trauma. Angiokeratoma circumscriptum lesions have dilated dermal papillary capillaries that are emptied by dilated venules on histological examination. The accompanying epidermis is acanthotic and hyperkeratotic, with elongated rete ridges, and may surround the dilated arteries. Clinically and histologically, verrucous hemangiomas and angiokeratoma circumscriptum are extremely similar. However, angiokeratoma circumscriptum is confined to the papillary dermis, whereas verrucous hemangiomas typically include the deep dermis and subcutaneous tissue. To get an accurate diagnosis, a thorough biopsy of the lesion is required [13]. In this case biopsy was not possible as parents denied for the procedure.

As previously stated, bilateral congenital anorchia is characterized by the total lack of testicular tissue. Because prospective investigations have not been conducted, the real prevalence of this disease is unclear. However, it is believed that there are about 0.5 - 1.0 cases per 20,000 males [14]. Although it is difficult to distinguish between an enlarged inguinal lymph node and the testes, ultrasonography is typically helpful in confirming the anorchia diagnosis. The sensitivity of the treatment is further reduced when the testes are located in the lumbar region. Magnetic resonance imaging (MRI) may be the most accurate way of identifying pelvic or abdominal testes. Anorchia can be diagnosed by undetectable plasma quantities of AMH and inhibin B, as well as an increased plasma FSH and a 46XY on karyotyping [15].

4. Conclusion

We presented an 8-year-old male child with hypertrophy of left thigh and the

right side of his face with overlying blackish nodules on his left thigh and scrotum. The patient was diagnosed as KTS with angiokeratoma circumscriptum naeviforme and bilateral congenital anorchia. There are just a few case reports of KTS and ACN in the worldwide literature. This case report highlights the clinical features and necessity of multidisciplinary approach for management and following up the case.

Ethical Issue

Parents provided informed consent for the publication of this case report.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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