CASE REPORT

Musculoskeletal manifestations in syndrome Klippel-Trenaunay

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ABSTRACT

Klippel-Trenaunay syndrome is a rare congenital disease with undefined etiology characterized by the presence of the triad: port wine stains, venous malformations or varicose veins, and bone and/or tissue hypertrophy. It affects the lower limbs more commonly. Treatment is generally conservative, with interventions limited to the treatment of complications. **Objective:** To present a case report of a child with musculoskeletal manifestations of the syndrome evaluated by a multiprofessional team, composed of social work, psychology, physiotherapy, occupational therapy, phonoaudiology, nursing and a physiatrist. **Methods:** After evaluation, it was decided that awareness and correction of posture as well as body perception, performance of orthostatic activity, balance training, dissociation of waists, and postural changes would be approached by the multiprofessional team. **Results:** The patient received multiprofessional care for two months, obtained better gait stability, and had independent gait in the community, with higher speed and lower number of falls. **Conclusion:** She was discharged after achieving the goals and after her parents were sensitized regarding the importance of maintaining the multiprofessional follow up and follow the goals set for homecare.

Keywords: Klippel-Trenaunay-Weber Syndrome, Hypertrophy, Hemangioma, Syndactyly

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INTRODUCTION

The Klippel-Trenaunay syndrome is characterized by the occurrence of capillary malformations associated with venous malformations or varicose veins and bone or tissue hypertrophy. It occurs sporadically, although some family cases have been reported, and it is commonly observed in the lower extremity of the body, but the trunk or face may also be affected.[1]

In most cases both haemangiomas and varicose veins may be present at birth, and generally they become more prominent until adolescence.[2] Lymphatic changes are seen in 70% of patients, and venous anomalies are characterized by: agenesis, hypoplasia, atresia, valvular incompetence, and occlusion of the deep venous system secondary to fibrosis.[3] Among the manifestations of the musculoskeletal system described in the orthopedic literature, there are hip dysplasia, syndactyly, macrodactyly,[3] polydactyly,[4] metatarsal varus foot, congenital ankle-cavus varus foot, anisomyelia, and scoliosis and angular deformities of the lower limb. Hypertrophy may be secondary to bone involvement such as discrepancy or increase of soft tissues, what compromises limb circummetry. The diagnosis is essentially clinical,[5] and there is no curative treatment, and the therapeutic goals are intended to diminish the patient's symptoms

CASE REPORT

MRF is a white female patient of one and a half years of age that was referred to the Institute of Physical Medicine and Rehabilitation of the University of Sao Paulo (IMREA-HCFMUSP) and admitted at the outpatient facility of Pain and Palliative Care of the *Itcaci Hospital.* She had Klippel-Trenaunay syndrome diagnosis and was unable to hold orthostatic position and to walk without assistance.

She was born after 34 weeks of pregnancy, weighing 1,720g, with 40cm in height and 26.5cm of head circumference, APGAR 7 and 8 in the first and eighth minute respectively. Since her birth port-wine stain, venous ectasia and hypertrophy of lower limbs soft tissues were observed. She acquired neuropsychomotor development by acquiring cervical control at four months, capacity to sit without support at six months, crawl at seven months, stand with support at eight months, and finally she was able walk with assistance at twelve months. She underwent recurrent urinary tract infection, with more than four episodes since birth. At physical examination, the patient wore socks with non-slip soles and lower limbs bandaged with cutaneous venous fragility and risk of bleeding. The presence of a port-wine stain extending in both lower limbs, associated to a significant increase in limb volume, in addition to the presence of venous ectasia (Figure 1) and lower limb discrepancy associated with syndactyly of the 2nd and 3rd fingers the left and 2nd, 3rd, 4th, 5th fingers to the right (Figures 2 and 3). No change in range of motion in the lower limbs was observed and the strength was preserved. In the dynamic evaluation of gait, external rotation of the left lower limb, with broad base, valgus of bilateral ankles, tendency to use manual support in objects and people around was present. Imbalance and propensity to fall with increasing speed was also observed.

The patient was evaluated by a multiprofessional team, composed by Social Worker, Psychologist, Physiotherapist, Occupational Therapist, Speech Therapist, Nursing and Physiatrist. After evaluations, it was decided to engage awareness and correction of posture as well as body perception, performance of orthostatic activity, balance training, and dissociation of waists and postural changes. Other approaches included stimulating task sharing, joint protection and energy conservation, training of activities of daily living for independence gains, ergonomic sitting posture, use of children table, chair and footrest, encouragement for graphics such as pinch, fine and overall motor coordination in addition to cognitive activities involving attention, reasoning and planning. The patient parents received guidelines on skin care and skin lesion prevention, and learned about the

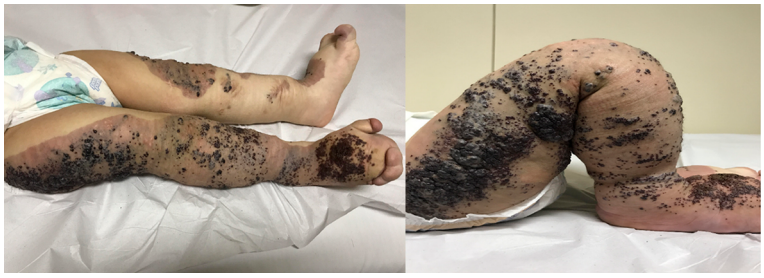

Figure 1. Port wine stain, venous ectasia and hypertrophy of the lower limbs.

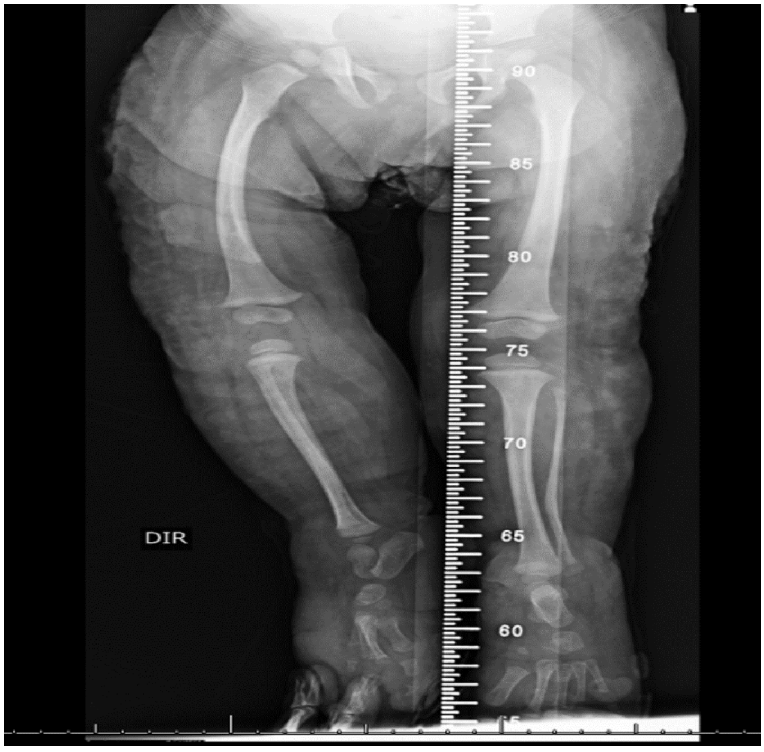

Figure 2. Right lower limb asymmetry in relation to left, with all right bones stretching compared to the right bones.

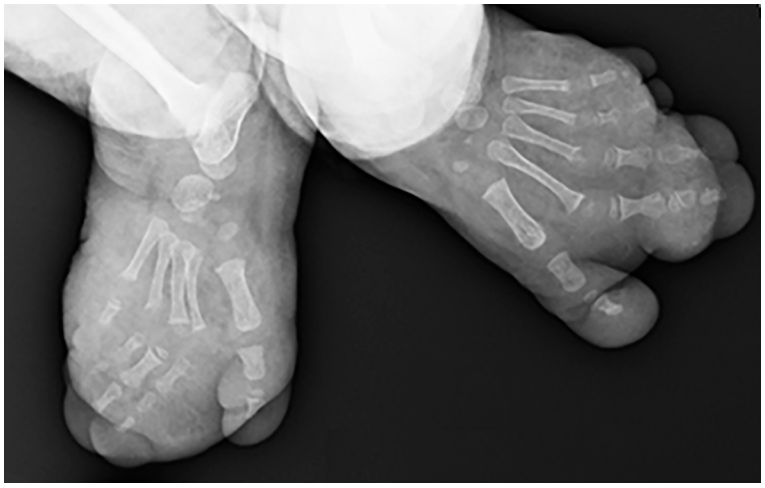

*Image collected with load (standing).

Figure 3. Syndactyly of the 2nd and 3rd fingers to the left, and 2nd, 3rd, 4th, 5th to the right.

importance of school inclusion. The patient participated in multiprofessional counseling for two months, achieved better gait stability and started to have independent gait in the community, with greater speed and lower number of falls. She was discharged after achieving these goals and after the parents were sensitized regarding the importance of maintaining the multiprofessional follow up and follow the goals set for homecare.

DISCUSSION

In 1900, French doctors Klippel and Trenaunay described this syndrome, in which the lower limbs are the most affected (90%) as well as the most frequent involvement is unilateral (85%). In this case report, the patient had bilateral lower limb involvement, and limb discrepancy, defined as abnormal asymmetry (5% difference), in length and in circumference, what may also be called hemi-hypertrophy or hemi-hypotrophy. At examination, one must look for vascular malformations associated with finger malformations or macrodactyly, which are usually associated with conditions of growth enhancement. Hypotrophic musculature, neurological changes, cognitive deficit or movement abnormalities may be associated with growth deficit. In this case report syndactyly was bilaterally present. Regarding the discrepancy of the lower limbs, epiphysiodes and tibia are only indicated when the discrepancy between the limbs is large enough not to allow the use of corrective insoles or adapted shoes to reduce the difference.[6] Some authors discuss the surgery to reduce the foot size to allow the use of footwear and amputations to promote functionality and/or to enhance appearance.[7]

Due to the peripheral vascular fragility, the use of orthosis to improve the stability of the ankles in gait was not indicated. As future goals for the development and growth of this child, computerized podobarometry examination and manufacture of appropriate shoes may be carried out. This syndrome is rare and has complex manifestations, patients need qualified multidisciplinary care. The therapeutic goals are intended to improve patient's symptoms, to prevent associated deformities, to stimulate independence for activities of daily living, and to promote quality of life.

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