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Clinical Insights into Milroy Syndrome: A Case Study Perspective

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Abstract

Given the rarity of Milroy syndrome, it has become a focal point of interest for pediatricians at the David Bernardino Pediatric Hospital in Luanda. This clinical case serves as a valuable contribution to understanding the manifestations of Milroy syndrome in a pediatric context.

1. Introduction

Milroy syndrome, an autosomal dominant genetic disorder, exhibits variable penetrance and expression characterized by anaplasia/hypoplasia of lymphatic vessels. This condition results from mutations in the VEGFR 3 gene, leading to congenital lymphedema [1,2].

2. Case Description

A 9-day-old newborn, born to a 24-year-old mother with a history of 1 stillbirth, presented with lower limb edema. The pregnancy had no significant complications, with limited prenatal evaluations. Delivery occurred at term via cesarean section, and the newborn, weighing 4,900 grams, experienced dystocia with a delayed cry at birth.

Receiving care at the David Bernardino Pediatric Hospital in Luanda, Angola, the infant exhibited symmetrical, non-inflammatory lower limb edema reaching the thigh roots. Maternal family history included a possible congenital lymphedema case in the maternal grandfather. Comprehensive exams, including chest and lower limb X-rays, echocardiography, and Doppler ultrasound, revealed no abnormalities. Lymphatic drainage was recommended.

Upon reassessment after seven days, a noticeable reduction in lymphedema and consistency was observed, prompting outpatient clinic follow-up (Figures 1 and 2).

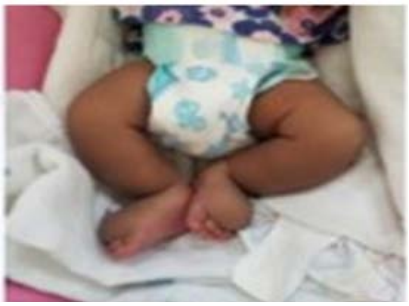

Figure 1: Congenital lymphedema (Diameter of right and left lower limbs: 10 and 9.7 cm respectively), before lymphatic drainage.

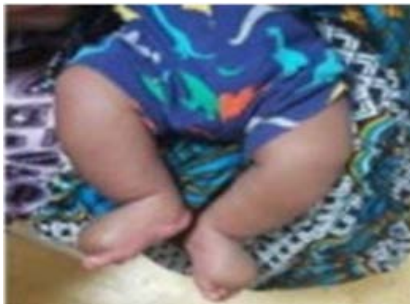

Figure 2: Congenital lymphedema (Diameter of right and left lower limbs: 8,2 and 8 cm respectively), 10 days after starting lymphatic drainage.

3. Discussion

Milroy syndrome, stemming from a VEGFR 3 gene mutation, presents as congenital lymphedema and was initially described by Max Noone in 1891. With a predominance in females (3:1) and a significant association with family history, prenatal care gains importance due to early ultrasound detection of edema around the 20th week of gestation. Diagnosis relies on clinical assessment, complemented by imaging and genetic studies. The rarity of Milroy syndrome accentuates the significance of reported cases in enhancing our understanding.

4. Conclusion

Management of Milroy syndrome focuses on supportive and symptomatic measures, aiming to prevent complications such as cellulitis, lymphangiosarcoma, and lymphangitis. Treatment involves a multidisciplinary approach, including pediatricians, dermatologists, physiotherapists, and clinical geneticists, emphasizing social reintegration. This case aligns with existing literature, underscoring the importance of supportive care in addressing the cosmetic and social aspects of this rare genetic disorder.

3. References

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