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Case Report

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Milroy syndrome: apropos of a clinical case

Abstract

Because Milroy syndrome is a rare disease, it aroused interest among pediatricians at the David Bernardino Pediatric Hospital in Luanda and for this reason we present this clinical case.

Keywords: milroy syndrome, David Bernardino Pediatric Hospital

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Introduction

It is a rare genetic disorder, autosomal dominant, with variable penetrance and expression, characterized by anaplasia/hypoplasia of the lymphatic vessels, due to mutation in the VEGFR 3 gene (endothelial growth factor receptor 3, culminating in congenital lymphedema.[1]

Case description

9-day-old newborn, daughter of a 24-year-old mother, G2 P2 and 1 stillbirth, with no prenatal complications, having performed only one ultrasound in the first trimester of pregnancy.[2] Born at term, dystocia by caesarean section, weighing 4,900 grams, late cry at birth without reference to the APGAR score. He presented edema of the lower limbs without other morphological alterations. She went to the David Bernardino Pediatric Hospital, in Luanda, Angola, on the ninth day of life, having stayed in the Neonatology service for 7 days. On physical examination, she showed hard edema of the lower limbs, symmetrical, up to the root of the thigh and without signs of inflammation. Report of maternal family history of possible congenital lymphedema (maternal grandfather). Complementary exams: X-ray of the chest and lower limbs, echocardiography and Doppler ultrasound of the lower limbs without alterations. Lymphatic drainage was indicated. As she did not present other clinical alterations, she was advised to be discharged and followed up at the outpatient clinic. She returned after seven (7) days for reassessment, showing a decrease in lymphedema and a decrease in consistency (Figure 1 & 2).

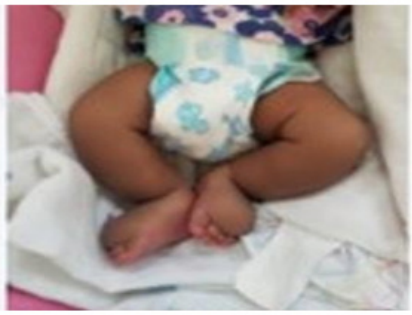

Figure 1 Congenital lymphedema (Diameter of right and left lower limbs: 10 and 9.7 cm respectively), before lymphatic drainage.

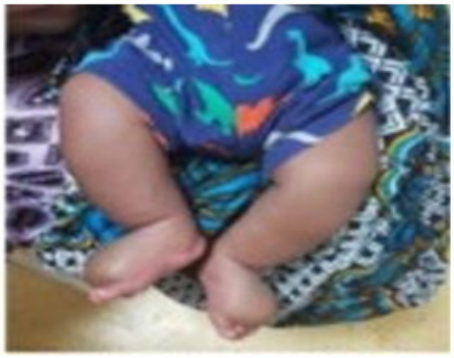

Figure 2 Congenital lymphedema (Diameter of right and left lower limbs: 8,2 and 8 cm respectively), 10 days after starting lymphatic drainage.

Discussion

Milroy syndrome is a rare genetic disorder that results from a mutation in the VEGFR 3 gene, causing congenital lymphedema. It was first described by Max Noone in 1891. A history of 250 generations revealed 22 people with the syndrome. It is more frequent in females (3:1) with 95% of cases related to family history. The diagnosis is essentially clinical, supported by imaging and genetic studies. The importance of prenatal care is highlighted, as the morphological ultrasound shows edema as early as the 20th week of gestation. The treatment is only supportive (lymphatic drainage and use of elastic stockings), with the aim of improving aesthetics and social reintegration. The present case corroborated the data described in the literature.

Conclusion

Management is supportive/ symptomatic only with the aim of avoiding complications (cellulitis, lymphangiosarcoma, and lymphangitis). Follow-up is multidisciplinary (pediatrician, dermatologist, physiotherapist and clinical geneticist) and aims at social reintegration.

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Conflicts of interest

The author declares no conflicts of interest.

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