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Case Report

# 4-Month-Old Girl with Unilateral Edema of the Back of the Foot

**Marta Folch-Benito*, Isabel M Salguero-Pérez, M Belén Caurín-Saboya and Marga Català-Puigbo**

*Paediatric Emergency Unit, Paediatrics Department, General Hospital of Granollers, Granollers, Barcelona, Spain*

***Corresponding Author:** Marta Folch-Benito, Paediatric Emergency Unit, Paediatrics Department, General Hospital of Granollers, Granollers, Barcelona, Spain.

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## Abstract

A 4-month-old female infant who consulted the emergency room for fever of a few hours and edema of the dorsum of the right foot (Figure 1), observed by the parents two days earlier, without other accompanying symptoms.

She has no pathological history of interest, she is a healthy girl, correctly vaccinated and with good weight gain. She exclusively breastfeeds. As a family history, it is noteworthy that the father has unaffiliated renal failure and HLA-B27-positive ankylosing spondylitis.

***Keywords:*** *Unilateral Edema; Back of the Foot; HLA-B27-Positive Ankylosing Spondylitis*

## Introduction and Case Report

On physical examination, only unilateral edema was observed in the right foot, without fovea, not painful and limited below the ankle. Good mobility of the joint without inflammatory signs and absence of skin lesions. No more edema is observed in other locations.

In the emergency room, due to the febrile condition, a urine test strip is carried out showing leukocyturia, without proteinuria. Few gram-negative bacilli are observed in the urine sediment and the blood analysis does not show any alteration in the infection parameters. It is oriented as a possible urinary tract infection and treatment with oral antibiotic therapy is initiated. On the other hand, given the recent appearance of edema, a follow-up appointment is made once the intercurrent condition has resolved.

After five days, the girl was afebrile and the urine culture was positive for *Escherichia coli*.

The patient is subsequently summoned and the persistence of the oedema is observed, so it is decided to complete the study to confirm the diagnostic suspicion.

### What is the diagnosis?

***Figure 1:*** *Unilateral edema of the dorsum of the right foot.*

## Discussion

It is a healthy infant, who presented an edema of the dorsum of the foot unilaterally, not painful, without fovea and without affecting joint mobility. This oedema was not observed at birth and had been discovered at the time of the intercurrent febrile condition.

The guiding symptom is unilateral edema, which once the usual causes such as soft tissue infections and trauma have been ruled out, initially directs us towards primary lymphoedema.

On the other hand, we cannot forget hereditary lymphoedema, which can be associated with genetic syndromes with alterations in the number of chromosomes. For this reason, it would be advisable to carry out a cytogenetic study in peripheral blood, as it is an innocuous and easy to carry out complementary examination. Another explanation that helps us diagnose alterations produced in primary lymphoedema is lymphatic scintigraphy.

The cytogenetics study showed a monosomy of the X chromosome, with karyotype 45, X (Figure 2). Faced with this diagnosis, the patient was evaluated by Cardiology, which ruled out cardiac involvement. The Endocrinology team also monitors thyroid growth and function, which is within normal range.

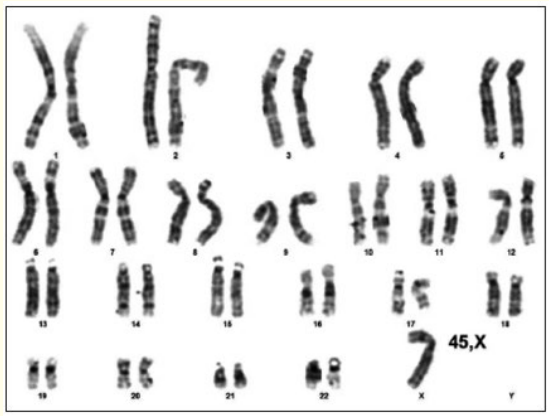


***Figure 2:*** *Image of the cytogenetics study showing the monosomy of the X chromosome.*

Currently, the patient is 3 years old, the edema has resolved and does not present the characteristic phenotype of monosomy 45, X.

Final diagnosis: Turner syndrome.

### Comment

Primary lymphoedema can be isolated or syndromic, and depending on the time of presentation, congenital (< 1 year) or late (> 1 year) [1,2].

Turner syndrome affects only women, due to the total or partial absence of the X chromosome and gives very variable clinical manifestations and possible involvement of different organs. Some patients do not present clinical symptoms or do so in a very mild way, as in the case presented. Prenatally, there may be intrauterine growth retardation and cardiac, renal or lymphatic vessel malformations (such as enlargement of the nuchal fold, hydrops fetalis and nuchal cystic hygroma, more characteristic) may be observed [3-5].

At birth, they may have low length and/or weight and a characteristic phenotype with low-set ears and scalp, winged and short neck, small jaw, wide chest and separated nipples, ogival palate and distal lymphoedema, among others [3].

Neonatal distal lymphoedema usually resolves during the first 18 months, may not be present at birth and may be established later [5].

Sometimes, the diagnosis is late due to short stature and late puberty or amenorrhea, due to gonadal dysgenesis.

Other malformations, especially cardiac and renal malformations, present in between 55% and 45% of patients, must be ruled out at diagnosis [4]. Auditory, ocular, autoimmune and endocrine abnormalities can also be found. Neurodevelopment is usually adequate, but with a risk of learning and emotional problems [3,4]. These patients need adequate follow-up at different stages of life.

## Conclusion

In this case, the distal lymphoedema of the lower limb was not detected at birth, and re-examining the parents in a directed way, they reported having observed it in photographs taken at home at approximately 2 months of age, but that it had gone unnoticed until now. The cytogenetics study allowed early diagnosis, despite the fact that the phenotype was normal.

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