

# Milroy Disease: A Case Report

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## Abstract

**Hereditary lymphoedema type I is a congenital onset primary lymphoedema with autosomal dominant inheritance, which is characterized by the swelling of the lower body. In this article, the authors report a case of a 32-year-old woman with repeating episodes of swollen limbs. Imaging studies and genetic study were carried out and a Milroy's syndrome diagnosis was established. The patient started sintomatic treatment of the edema, with fitting stockings and descompressive massage, with symptomatic benefit.**

## Keywords

**Milroy Disease, Lower Limbs Edema, FLT4 Gene, VEGFR-3**

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## 1. Introduction

Milroy disease (hereditary lymphoedema type I) is a congenital onset primary lymphoedema with autosomal dominant inheritance [1] [2], but, occasionally, it can develop later in life [1]. It is clinically and genetically heterogeneous [3] [4], showing both inter- and intrafamily variability in the severity of the edema [1] [4]. FLT4 gene mutation in the q35.3 locus is the most common mutation associated in Milroy's disease [5] [6]. The FLT4 gene provides instructions for producing a protein called vascular endothelial growth factor receptor 3 (VEGFR-3), which regulates the development and maintenance of the lymphatic system. Mutations in the FLT4 gene interfere with the growth, movement, and survival of cells that line the lymphatic vessels (lymphatic endothelial cells). These mutations lead to the development of small or absent lymphatic vessels [2] [6] [7]. If lymph fluid is not properly transported, it builds up in the body's tissues and causes lymphedema.

## 2. Case Report

A 32-year-old woman with repeating episodes of swollen limbs (**Figure 1**) and no other associated complaints presents herself in a consultation. The patient was questioned about her past medical history, family medical

history, occurrence of cellulitis, venous problems as well as , age at onset of swelling, progression of the oedema over time and treatments of previous episodes. The patient reported a maternal aunt with lower limbs' edema as well as a personal history of varicose veins as well as three miscarriages. On examination, bilateral edema in the legs was present, with no associated inflammatory signs and no pain to palpation, the remaining examination was normal. Lower limbs venous ultrasonography was done, which proved normal. The patient was referred to a Vascular Surgery consultation. After lymphangiography showed lymphatic hypoplasia further study was carried out with magnetic resonance imaging (MRI)-revealing edema and a lymphocele-as well as molecular genetic study which identified a mutation in the gene FTL4 chromosome 5q35-responsible for vascular endothelial growth factor receptor (VEGFR-3) expression establishing a Milroy's syndrome diagnosis. The patient then began sintomatic treatment of the edema, with fitting stockings and descompressive massage. After one month of treatment, the sweeling was reduced (**Figure 2**). Two years after the diagnosis, with correct management of the disease and hipocoagulation, the patient got pregnant and had a full-term pregnancy.



**Figure 1.** Patient with lower limbs edema due Milroy's disease.



**Figure 2.** Patiente after treatment, with fitting stockings.

### 3. Conclusion

Clinical onset at birth usually is a requirement for the clinical diagnosis of Milroy disease [6]. Nevertheless, there are exceptions with some mutations leading to swelling later in life, like in our patient, although, in some cases, early changes may be missed or as a result of variable expression or reduced penetrance [1] [8]. Treatment is aimed at reducing swelling and preventing infection. Complete decongestive therapy (CDT) is a form of treatment in which specialized manual techniques (manual lymph drainage) is combined with multilayered compression bandaging, meticulous skin care, exercise, and the use of well-fitted compression garments [5] [7] [8]. Decongestive and conditioning exercises are important components of CDT. Prevention of secondary complications is also important [5] [8]. Frequency of cellulitis can be reduced through good skin hygiene, prompt treatment of infections with antibiotics, and prophylactic antibiotics for recurrent episodes [1]. Genetic counseling will benefit people with hereditary lymphedema and their families. It is very important adhere to lymphedema management recommendations to prevent progression the lymphedema and rehabilitation therapy may be even necessary in cases where extreme lymphedema impairs daily activities.

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