SPOT DIAGNOSIS (IMAGE GALLERY)

Figure 1 : Swollen limbs in mother

Figure 2 : Lower limb edema in baby with single umbilical artery

LOWER LIMB EDEMA IN MOTHER AND CHILD

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A 28 year old primigravida with repeating episodes of swollen limbs (Figure 1) and no other associated complaints presented to obstetrics department for consultation. The patient reported her father also had similar lower limb edema. 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 doppler was normal. The patient was hospitalized and she delivered a female baby of 3.2 kg with similar lower limb edema which was non-pitting. On detailed examination, baby had single umbilical artery (Figure 2). Ultrasonography of abdomen and lower limbs was normal. Colour doppler of lower limbs of the newborn was normal.

What is the diagnosis?

Milroy disease is a hereditary lymphedema (type I) with autosomal dominant inheritance which presents at birth but occasionally can develop later in life. (1,2) It is clinically and genetically heterogeneous showing both inter and intra-family variability in the severity of the edema. Milroy disease should be suspected in individuals with lower limb swelling that is usually but not always bilateral and present at birth or develops soon after. In neonates the swelling predominantly affects the dorsum of the feet. With age, the swelling may improve or progress to affect the whole lower leg). (3) It may be associated with hydrocele (37 percent of males), prominent veins (23 percent), upslanting toe nails (14 percent), papillomatosis (10 percent), urethral abnormalities in males (4 percent) and in some cases cellulitis (20 percent). (4) Diagnosis is done by clinical findings and confirmed by molecular genetic testing. Mutations in the FLT4 gene lead to the development of small or absent lymphatic vessels. Lymphoscintigraphy reveals lack of uptake of radioactive colloid in the ilioinguinal lymph nodes caused by a paucity of lymphatic vessels in the lower limbs. (5) Treatment consists of stockings and massage to improve the cosmetic appearance or decrease the size of the limb and reduce the risk of complications. In our patient, lymphangiography and genetic analysis could not be done due to non-availability. The patient was advised fitting stockings and decompressive massage. After one month of treatment, the swelling markedly reduced.

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