LETTER TO EDITOR (VIEWERS CHOICE)

MILROY DISEASE MANIFESTING IN ONE OF THE TWINS: UNIQUE PRESENTATION

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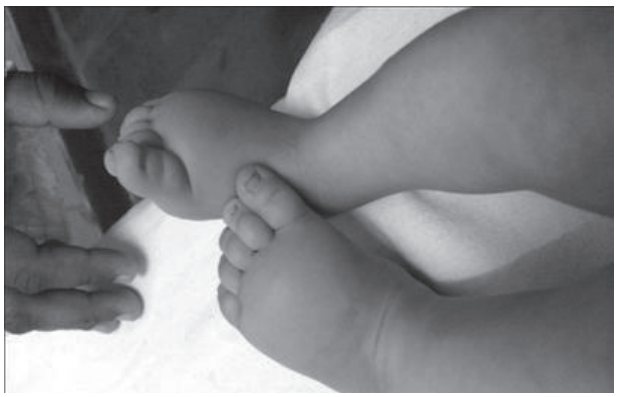
Keywords: Milroy disease, congenital lymphedema, rare autosomal dominant disorder.

A 4 months old female infant presented with swelling of both feet and legs since birth. She was born as one of twins (the other twin was a male child) by lower segment caesarean section (LSCS) at full term to the mother who was 2nd gravida. Although immediate perinatal period was uneventful for both neonates, this child was noticed to have swelling of both feet during first few days of life which had gradually increased (Figure 1). There was no maternal illness during pregnancy. On examination, the child had asymmetrical edema of lower limbs upto knees and feet. There was upslanting toenail (ski jump toenail). Other examination findings were normal. Hemogram, renal and liver function tests were normal. Urine examination was also normal. Later lymphoscintigraphy done showed absent uptake of radionuclide substance in draining lymph nodes in lower limbs (Figure 2) indicating absent or dysfunctional lymphatics confirming the diagnosis of Milroy disease.

Figure 1: Bilateral primary congenital lymphedema in lower limbs.



Lymphedema is abnormal build-up of fluid in extremities due to malfunction or malformation of lymphatic system. Lymphedema is classified as primary or secondary. Hereditary lymphedema is also known as primary lymphedema. Primary lymphedema is classified according to time of onset. Congenital lymphedema which is present at birth or soon thereafter is known as Milroy disease and primary lymphedema presenting between 1-35yrs of age is called lymphedema praecox or Meige disease which is mostly sporadic. (1) Actual incidence of Milroy disease is not exactly known. Milroy disease shows autosomal dominant pattern of inheritance with reduced penetrance 80-90%. (2) 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. If lymph fluid is not properly transported, it builds up in the body's tissues and causes lymphedema. (3) Clinically affected children were having edema mostly asymmetrical and more commonly in lower limbs. Other conditions associated with Milroy disease include cellulitis (23%), large calibre leg veins (23%), papillomatosis (10%), up slanting toenail (10%). (2) Milroy disease is rarely associated with complications like cellulitis, lymphangitis, septic arthritis, chylous ascites, and chylothorax. (1) Differential diagnosis include Turner syndrome, Noonan syndrome, lymphedema distichiasis syndrome, lymphedema & ptosis syndrome and yellow nail syndrome. (4,5) Management is mainly supportive including elevation of affected limb, skin care to reduce risk of cellulitis & lymphangitis. Manoeuvres to enhance lymph drainage such as compression bandages are recommended. (5) Our case is a rare and unique case of Milroy disease manifesting in one of twins with no family history.

Figure 2: Lymphoscintigraphy showing lack of tracer migration in lower limbs

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