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A CASE OF PRIMARY CONGENITAL LYMPHEDEMA

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ABSTRACT

Primary congenital lymphedema is a rare disorder of childhood which presents from birth and progresses into late childhood. It can be either unilateral or bilateral. Here we are presenting an eight year old boy with primary congenital lymphedema who was referred to us with progressive non-pitting edema of left lower limb since birth. The purpose of presenting this case is its rarity.

KEYWORDS: Lymphedema , Non-pitting , Childhood , Congenital.

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CASE HISTORY

An eight year old male child was brought to our centre with complains of swelling of entire left lower limb since birth. The swelling was progressive as the child grew and now has become static. There was no history of locomotory difficulty. No history of pain over the swelling. No history of fever with chills. The mother was very much concerned about the cosmetic disfigurement associated with the swollen left lower limb. There was no similar history in the family. On examination the child's left lower limb was uniformly swollen when compared to the right lower limb and swelling of the scrotum and penis was also noticed. Non pitting pedal edema of left lower limb was seen. There were no signs of inflammation. There was no neurovascular deficit. Right lower limb was normal. The child's anthropometry and other systems were normal. There were no external syndromic features. Thus a diagnosis of Primary Congenital lymphedema was made.

DISCUSSION

Primary congenital lymphedema is a rarest form of lymphedema [1]. Lymphedema is caused by an increase in the interstitial protein rich fluid, which results in insufficient lymphatic transport and drainage [2]. Primary congenital lymphedema can present at birth or within 2 years of age. It most commonly affects the feet and usually extends up to the knees but sometimes associated with cellulitis, papillomatosis, upturned toe nails and hydrocele [3]. Congenital lymphedema may have a familial distribution. This condition commonly presents as autosomal dominant inheritance but alternative patterns of inheritance have also been seen in few cases. Primary congenital lymphedema is also known as Milroy disease or type I lymphedema. This autosomal dominant disorder is linked to 5q35.3, caused by missense mutations in the tyrosine-kinase domain of the vascular endothelial growth factor receptor-3 (VEGFR3), also known as FLT-4 [4]. Congenital lymphedema may be associated with various syndromes like Turners syndrome and Nail patella syndrome [5]. The diagnosis is often made through history taking and a good physical examination. Lymphography is sometimes used to diagnose and prognosticate primary lymphedema. But the most reliable, objective and non invasive tool for the diagnosis of lymphedema is isotopic lymphoscintigraphy [6]. Various treatment modalities such as intermittent pneumatic compression which effectively removes excess fluid from the affected limb, compressive garments which prevents re-accumulation of fluid after decompression and application of heat to the lymphedematous limb are available [7]. Rarely surgical excision of the fibrotic tissue is done and this is indicated only in very advanced cases. Sometimes microsurgical techniques are also used to achieve lymphatic drainage in cases of lymphedema[8].

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