

Images in paediatrics “swollen foetal feet-antenatal diagnosis”

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Citation

P Nikila, M Anne, C Lee. *Images in paediatrics “swollen foetal feet-antenatal diagnosis”*. The Internet Journal of Pediatrics and Neonatology. 2008 Volume 11 Number 1.

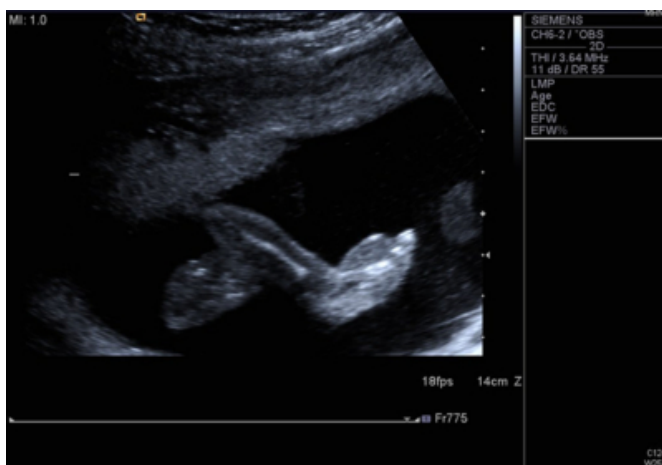
Abstract

Lymphedema of the lower extremities is a diagnostic challenge. Primary lymphedema is classified into idiopathic and familial subgroups. Hereditary lymphedema can be nonsyndromic. A comprehensive family history and clinical examination are the mainstays of medical assessment. Primary lymphedema can be classified depending hereditary or idioapathic. We present an antenatal diagnosis of a Milroy’s disease on antenatal Ultrasound scans.

Antenatal clinic liaison between the paediatrician/ultrasonography and obstetrician are vitally important for counselling of parents. This antenatal USG was brought to our notice for discussion. The mother was not surprised when advised about the scan but informed about a similar pathology with her and her family. Her father and brother were affected with Milroy’s disease.

The photograph (Figure 1) clearly reveals foetal pedal lymphedema.

Figure 1



The mother had similar features and edema of feet. (Figure 2)

Figure 2



The male infant was born by caesarean section had the presence of pedal edema as shown in the antenatal scans. No other abnormalities were noted. No features of Noonan’s syndrome were noted. The cord bloods have been sent for DNA for the VEGFR gene. The cord bloods have been sent for DNA for the VEGFR gene. VEGFR3/exons17-26 genetics report confirms the patient is heterozygous for the c.3122G>C(p.Arg1041Pro) mutation which is consistent for Milroy’s disease

DISCUSSION

Primary congenital lymphoedema (Milroy disease) is a rare autosomal dominant condition for which a major causative gene defect has recently been determined. Mutations in the vascular endothelial growth factor receptor 3 (VEGFR-3) gene have now been described in 13 families world-wide.

Other symptoms and signs included cellulitis (20%), large calibre leg veins (23%), papillomatosis (10%), and up slanting toenails (10%). In males, hydrocele was the next most common finding after oedema (37%). Thorough clinical examination of these patients indicates that there are few clinical signs in addition to lower limb oedema. Rigorous phenotyping of patients produces a high yield of VEGFR-3 mutations. ¹

Hereditary lymphedema type 1, Milroy’s disease is present at birth and mostly affects the dorsal aspects of feet. It is a lifelong condition but does not affect longevity. Complications are rare except for chronic discomfort and warmth of the affected areas. PCL is an autosomal dominant disease with incomplete penetrance due to mutation in the gene locus encoding for VEGFR3 with resultant dysgenesis of microlymphatic vessels. ²

Typical Milroy disease with family history (group I), typical Milroy disease with no family history (group II), atypical Milroy disease (group III), and complex primary lymphoedema (group IV). Results demonstrated that with rigorous phenotyping the likelihood of detecting VEGFR3 mutations is optimised. Mutation prevalence is 75% in typical Milroy patients with a family history (group I) and 68% if positive family

history is not a diagnostic criterion. A positive family history is not essential in Milroy disease. The likelihood of detecting VEGFR3 mutations in patients who have a phenotype which is not typical of Milroy disease is very small (<5%). ³

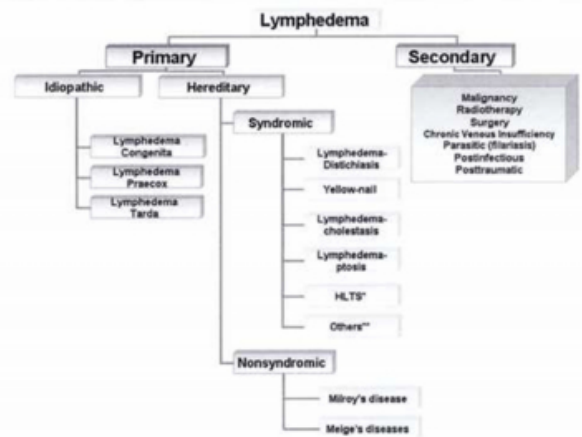
Management of primary lymphedema is conservative and usually successful. Skin care, manoeuvres that enhance

lymphatic drainage such as compression and directed exercises. In the neonatal period observation may be sufficient. The role of parents is crucial providing the necessary input. ⁴

A flow chart for diagnostic purposes is useful. ⁵

Figure 3

Flow chart



References

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