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Genetics Research at Lymphoedema Patients: Why is it Important?

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ABSTRACT

Lymphoedema is a congestion of a protein rich fluid in the tissue because of the impairment of the lymphatic drainage. Lymphatic drainage may be disrupted primarily or secondarily. Primary lymphoedema can be found in 10% of cases as hereditary genetic disorders and in 90% of cases after sporadic mutations. Identification of genetic mutations in patients with primary lymphoedema is particularly important because: prompt and patient-oriented therapy prevents progression of swelling and their complications. Together with advised lifestyle changes can improve the quality of life of patients and their families. When patients are planning the family, genetic counseling can also improve the quality of life of the whole family.

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