Introduction

5

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Lymphedema is a progressive and highly disabling disease that affects an estimated 200–300 million people worldwide [1]. Lymphedema can be either primary (hereditary) or secondary (acquired) and develops as tissue fluid retention and swelling. Primary lymphedema is caused by lymphatic system dysplasia or developmental dysfunction of unknown cause. The leading cause of secondary lymphedema worldwide is filariasis [2]. Tumor treatment is the main cause of secondary lymphedema in other regions. Peripheral lymphedema is the most common type and is caused by superficial lymphatic system disorders. Lymphedema may have serious consequences if it is neglected or not treated in time. Once lymphedema occurs, the edema fluid, which is rich in macromolecules, stays in the tissue, the fibrous tissue and fat continue to deposit, and the affected limb becomes deformity. Lymphedema is often accompanied by frequent attacks of erysipelas and cellulitis. Each infection exacerbates edema, creating a vicious circle. Currently, peripheral lymphedema is an incurable disease because the pathological changes in the affected tissue and the lymphatic system itself induced by lymphedema are generally irreversible. Secondly, the mechanisms underlying abnormal lymphatic system development and defective postnatal lymphatic regeneration remain largely unclear. This has led to a lack of effective and targeted treatments. To date, more than 20 underlying genes have been identified in lymphedema, and these are linked to different phenotypes of primary lymphedema [3], providing

valuable insight into the molecular mechanisms that regulate the development and function of the lymphatic vasculature. Additionally, a new multi-modal imaging examination for the lymphatic system that was developed over the last decade is now available in many centers [4, 5]. Therefore, future studies into correlations between phenotype and genotype will help obtain a better understanding of this disease and explore new treatments.

References

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