

## Renal disease (or renal abnormalities) in primary lymphoedema

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**Aims:** In the follow-up of a cohort of patients with primary lymphoedema (PLE), a recurrence of renal abnormality/pathology was noted in patients with the CELSR1 stop codon mutation. We will clinically characterise this group to identify the signs of call and set up the necessary follow-up.

**Method:** This is a descriptive single-centre cohort study approved by the local ethics committee.

**Results:** We studied exomes of our cohort (n=445) and identified 5 patients with kidney anomaly/disease. From this group it was identified, 14 carriers (9 women and 5 men) out of 5 families carrying the CELSR1 mutation. Average age of onset is 15 years (standard deviation=15). Twelve subjects were explored with clinical examination, lower limbs lymphoscintigraphy, MRI, Doppler ultrasound of abdomen. Nine carriers presented with a lymphoedema, three carriers did not have a lymphoedema (CNL) and two members of the same family, carriers of the mutation, did not want to be examined. For the carriers with lymphoedema, abdominal ultrasound was either normal (n=4), pathological (n=4; atrophy/hypotonia), and there was one enuresia in childhood. Lymphoscintigraphy showed functional deficiencies (delayed drainage, deep node, insufficient number of nodes). In unilateral case, the contralateral leg was normal (n=1) and pathological (n=3). By MRI, 80% (n=4/5) had rich lymphatic tissue in the pelvis, 25% had an atretic thoracic duct (1 man). In mutation carriers without PLE, abdominal ultrasound was normal in two, and one patient was operated for mega ureter. Lymphoscintigraphy showed functional deficiencies or anatomical variants for one or both lower limbs. By MRI, 75% (n=3/4) had rich lymphatic tissue in the pelvis and 50% had a dilatation of thoracic duct (men).

**Conclusions:** General characteristics of our cohort with stop codons in CELSR1 is in accordance with the literature. We found that rich pelvic tissue appears recurrently on MRI. Lymphoscintigraphy showed that the lymphatic involvement is systematic and that men are less symptomatic, probably due to compensation through the deep pathway but the first main message, is the discovery of associated renal abnormalities made it possible to set up targeted screening.