SKIN MANIFESTATIONS OF INTERNAL DISEASE

# MULTIPLE LEIOMYOMAS? THINK OF HEREDITARY LEIOMYOMATOSIS AND RENAL CELL CANCER!

*Angeliki Liakopoulou [(1)] - Philip Pavlou [(1)] - Emanuele Verga [(1)] - Roberto Verdolini [(1)]*

*Princess Alexandra Hospital, Dermatology, Harlow, United Kingdom [(1)]*

Background: Hereditary leiomyomatosis and renal cell carcinoma (HLRCC) is a rare autosomal-dominant hereditary syndrome, caused by germline mutations in the fumarate hydratase (FH) gene. HLRCC patients are predisposed to develop cutaneous leiomyomas and early onset aggressive renal tumors. Young female patients also develop multiple, symptomatic uterine fibroids. Recognition of the syndrome in patients with multiple cutaneous leiomyomas is therefore extremely significant since it will guide early interventions leading to decreased mortality.

Observation: A 52-year old woman presented to the dermatology department with multiple painful, flesh-coloured papules and nodules, effecting her chest, back and arms. Dermoscopy revealed a delicate pigment network without any vascular structures. The clinical and dermoscopic findings were suggestive of leiomyomas. The patient underwent two punch biopsies from the lesions that confirmed the diagnosis. Histology revealed dermal proliferation of irregular smooth muscle cell bundles. The lesional cells showed strong expression of SMA, desmin and vimentin. Upon further questioning, the patient revealed that she had her uterus removed at a young age for multiple fibromas. Family history was unknown, as she and her brother were adopted but she did mention that her brother also had similar lesions. Her brother was consequently examined and found to have multiple leiomyomas which were also histologically confirmed. HLRCC syndrome was highly suspected and genetic sequencing was performed that revealed FH gene mutations, confirming the diagnosis. Routine blood tests and urinalysis were normal and MRI scan of the kidneys did not reveal any abnormalities. Periodic surveillance for kidney tumors was started with annual MRI scans.

Key message: The presence of multiple leiomyomas should raise clinical suspicion for HLRCC syndrome. Genetic testing of the FH gene should be offered to any individual who presents with clinical manifestations of HLRCC or has a family history of HLRCC.