



Leiomyomatosis and Alport syndrome

“Leiomyomas” are benign (not cancerous) smooth muscle tumors that can be found in the esophagus, lungs, uterus, and other female reproductive organs. Some patients with X-linked Alport syndrome may be at risk to develop leiomyomas although this is rare. Symptoms may include difficulty swallowing, vomiting after eating, heartburn-type pain, recurrent bronchitis, shortness of breath, cough, and stridor (noisy breathing) that often appears in late childhood. Almost all patients with Alport syndrome and leiomyomatosis have a very specific mutation that is a deletion of the beginning part of the *COL4A5* gene that extends into the beginning part of the *COL4A6* gene that is next door. Patients with other types of *COL4A5* mutations such as missense mutations would not be expected to develop leiomyomas. Knowing your Alport mutation is important to be able to understand your risk of developing leiomyomas and to plan for whether you should have any screening for these benign tumors. Treatment depends on where the tumors are found and what kind of problems they are causing but may include surgical removal.

This information was graciously provided by Dr. Michelle Rheault of Alport Syndrome Foundation's [Medical Advisory Committee](#).