

Fabry disease is a lysosomal storage disease. It is one of two or three X-linked conditions. So there is some differences between the way it impacts men and women.

The classical form of Fabry disease presents in childhood. It is due to a deficiency of alpha-galactosidase A. And the initial presentation is usually with pain or gastrointestinal symptoms, which can include both GI pain as well as recurrent diarrhea. That progresses over time to involve other organ systems, eventually, resulting in significant risk for progressive kidney disease, heart disease, and stroke, and a number of other kinds of symptoms.

With the X-linked inheritance, it was initially assumed that men would be severely affected and women would be either not affected or mildly affected. That, however, has not turned out to be the case. So there are some differences in the way it impacts men and women.

Men, it tends to be primarily a kidney disease as far as the first life threatening complication, followed by heart disease, followed by risk for stroke. For women, it presents a little bit later and the life threatening components of it are heart disease, followed by stroke, followed by kidney disease. So it's a slightly different pattern and we can, potentially, go into that a little later.

The inheritance pattern is a little bit unique as well because a man with Fabry disease can only pass the disease onto his daughters, so all of an infected man's children-- all of the daughters will have Fabry disease, but none of the sons will. For a woman who passes it on, she has two X chromosomes. One would have the mutation, the other, presumably would have normal function. And the chance for a son or a daughter to inherit Fabry disease would be 50-50 and with equal chance for passing the disease on to both sons and daughters.