

SPEAKER: Fabry disease is a heterogeneous disease. On the one hand, we have the gender effect. Males will develop fully the disease while in females, it may not be so penitent. Females who are very unlucky, they will inactivate mostly their healthy X chromosome, and they will have this disease as severe as in males. This will be at around 10% of females.

But some females, they are very lucky, and they inactivate the pathological X chromosome so all their cells will be healthy. And they will be the true carriers. Absolutely no evidence of Fabry disease. And in between, you have females with more severely affected or less severely affected. So we will discuss Fabry kidney disease in males. But we know that some females will have the full-blown disease, some females will have not noticed this whatsoever, and some of them may have a milder form of the disease.

The other source of heterogeneity is the severity of the mutation. Severe mutations will give rise to classic Fabry disease, and this is the one that we will be discussing. Less obvious mutations give rise to later onset, non-classic Fabry disease, and these patients may-- even if they are males, they may or may not have kidney disease. OK. So what we are going to explain now is the male with classic Fabry disease. And we know that females or later onset patients may have or not kidney disease, and if they do, it may be more or less severe.