

SPEAKER: The initial presentation can be any time-- for both pain and GI symptoms anytime from age 2 up into adulthood. Some of that variability is associated with the kind of mutation that the patient has. The lower the level of the enzyme that's present, the more likely they are to have earlier onset or more severe symptoms. But there's not a direct correlation with that.

So we will have people in the same family who have very different presentations as far as severity of pain, severity of GI symptoms, or even the other symptoms. So as the patients get older and they start to develop potential things like kidney disease or heart disease, which one will be more prominent will be influenced by the severity of the mutation, by the family history, by the sex of the patient, but none of those things are adequate by themselves to predict which symptoms will be taking the lead in any given person's life.