

Phenotypes and Genotypes

Video description: Kevin (a male with short brown hair and dark grey shirt) sits in front of a dark blue background, looks into the camera and signs.

TRANSCRIPT: Kevin signing: Hi! Today I'm going to describe two important research terms: phenotypes and genotypes. What's the difference between the two? First, we'll look at diagnosis by "phenotype." Imagine a person who meets with a doctor, teacher, etc. Just by observing the person and learning a few details about them, the doctor determines that the person has Usher syndrome. He may further assume that since this person communicates in ASL, that they have Usher type 1. And when he learns that their family is from Louisiana, he decides that they probably have Usher type 1C. This is all assumption. It's not fact. Now let's look at an example of diagnosis by "genotype." Let's look at this same person and that person goes to a doctor who takes a blood sample to do genetic testing to scientifically determine what type of Usher syndrome the person has. Maybe the test results show that the person DOES have Usher type 1 BUT 1B not 1C! That's phenotype (observation) and genotype (genetic testing). I encourage you to get your bloodwork done for genetic testing if you want to know whether you have Usher type 1, 2 or 3, and the subtype - A, B, C, etc. It's so important. I also want to let you know that researchers who are testing treatments - and are looking for people to participate in clinical trials - require genetic confirmation that you have Usher syndrome. They need proof of the type of Usher syndrome you have in order for you to qualify to participate. Without that proof, you can't participate. So, genetic testing is something to think about if you want to find out your own identity. Plus, knowing your identity can be a very cool thing. Thanks!