Genetic counselling, an example of a patient meeting:

Genetic counsellor = GC

Patient = P

GC: Hi, welcome P. How are you?

P: Hi. I´m ok. Since my mother passed away in breast cancer I haven't been able to think about much else.

GC: I understand, I´m sorry for your lost. When did she pass away?

P: 2 months ago.

GC: I understand you mother tested positive for a gene variant in a gene called BRCA1. Is that correct?

P: Yes. This keeps me up at night. I´m very worried that I inherited the same mutation. I'm very anxious.

GC: I understand your worry, it is very normal for people in the same situation as you to experience anxiety. This gene variant is inherited in an autosomal dominant way. Are you familiar with this concept?

P: No, I'm not.

GC: Humans have 23 pairs of chromosomes in our cells. These chromosomes contain our DNA which are our genes. One chromosome is inherited from our mother and one chromosome is inherited from our father. Meaning we have double set of gene copies. Autosomal dominant inheritance means that the gene in question is located on one chromosome. "Dominant" means that a single copy of the mutated gene (from one parent) is enough to cause the disorder. A child of a person affected by an autosomal dominant condition has a 50% chance of being affected by that condition via inheritance of a dominant allele.

P: So, this means that I have a 50% risk of getting breast cancer?

GC: Good question. Not exactly, though, this means that you have a 50% probability of having inherited the same gene variant as your mother had. Note, having the same gene variant doesn't mean you will develop cancer. A gene mutation in BRCA1 means that you have an increased risk of developing breast cancer and ovarian cancer.

P: I understand… How do I live with this risk?

GC: If you have inherited the same gene variant as you mother had, you will be included in control programs and have the option to undergo risk reducing surgery to minimize the risk.

P: That is good. How does this work?

GC: You will be offered to take a blood sample that we analyse for this specific gene variant. If you want to?

P: Yes, I want to get tested.

GC: Do you have any further questions?

P: No.

The patient takes a blood sample and wait for the test result.