

Diagnostic Tests in Medicine

Keywords: combinatorics, probability and statistics, conditional probability, frequency of occurrence

Every person undergoes many diagnostic tests during their lifetime, either at the doctor's office or at home. It doesn't matter whether it is a test for COVID-19, celiac disease, general mammogram screening, or a home pregnancy test. In any case, we want to know whether the test result is reliable. For each such test, two basic pieces of information are provided:

- *Test Sensitivity*: the probability that the test will detect the disease when I have it.
- *Test Specificity*: the probability that the test will be negative when I do not have the disease.

The so-called *prevalence of the disease*, i.e., the ratio of the number of individuals who have the disease to the number of all individuals in the population, is also important for evaluation.

Test Result Interpretation

A number of home tests for allergies and intolerances are sold in pharmacies and drugstores, including COVID-19 or pregnancy tests. For example, the leaflet of a test for celiac disease (gluten intolerance) states that the sensitivity of the test is 96,3 % and the specificity of the test is 89,7 %. We know that the number of celiacs in the population is roughly 1 %, so the prevalence of the disease is 1/100.

We will be primarily interested in the question: If I test positive, what is the probability that I actually have celiac disease? Try to guess the answer first. In the following exercise, we will perform the exact calculation.

Exercise 1. The test sensitivity is 96,3 %, the test specificity is 89,7 % and the prevalence of the disease is 1/100. What is the probability that a patient who tests positive for celiac disease actually has the disease?

Exercise 2. What is the probability that a patient who tests negative is really negative?

Interpretation Without Using Probability

Let's try to solve the first exercise again, but we will reframe the assignment to avoid the concept of conditional probability.

Exercise 3. Out of 100 000 people, about 1 000 have celiac disease. Of these 1 000 people, the test detects celiac disease in 960 cases. Of the remaining 99 000 people who don't have celiac disease, 10 200 people will also test positive. Let's imagine the group of those people who test positive. What proportion of them actually have celiac disease?

Literature

1. Singh P., Arora A., Strand T.A., Leffler D.A., Catassi C., Green P.H., Kelly C.P., Ahuja V., Makharia G.K., *Global Prevalence of Celiac Disease: Systematic Review and Meta-analysis*. Clin Gastroenterol Hepatol. 2018 Jun; 16(6):823-836. doi: 10.1016/j.cgh.2017.06.037.
2. Gigerenzer, G., *Calculated risks*, Simon and Schuster, 2003.