Math 107 Probability Project

In this project you will be given the opportunity to apply your knowledge of probability and contingency tables to two current public issues in medicine

Genetic testing and pregnancy

Due to advances in technology it is now possible to test at 16 weeks into a pregnancy for a number of possible abnormalities. The test is called a cell-free DNA test (cfDNA), a non-invasive-prenatal screen (also called NIPS, NIPD or NIPT). These tests take blood from the mother and can screen for

a) the sex of the fetus
b) the father of the child
c) the presence of a number of abnormalities.

In this project, we will be looking for the presence of Edward's syndrome, a serious (fatal) genetic defect.

cfDNA is a **screening** test. It can tell you if there is a higher or lower chance that a given chromosome condition is present in the baby. It does not give you yes or no answers because the PPV and NPV are too low.

Amniocentesis and CVS are considered **diagnostic** tests and can give you a yes or no answer with a very high degree of certainty because the PPV and NPV are extremely high. Amniocentesis and CVS are invasive tests and present some risk of miscarriage to the pregnancy (usually reported around 0.5%).

A cfDNA looks for an abnormal ratio of a specific chromosome. If an abnormal ratio occurs, the tests results are positive. In our case, an abnormal ratio of chromosome 18 might be caused by trisomy 18, a triplicate of the 18^{th} chromosome and the fetus would have Edwards's syndrome. However, the abnormal ratio might be caused by something else, creating a false positive.

Create three contingency tables, one for each age group. Use the following information.

- The test for an abnormal ratio of chromosome 18 has a specificity of 99.87% and sensitivity of 96.3%
- Your first table will be for pregnancy at age 21 where the prevalence of Edward's syndrome is 1 out of 4821. Assume the population has 250,000 pregnant 21 year olds
- Create another table for pregnancy at age 30 – sensitivity and specificity remain the same but prevalence is 1 out of 2727. Assume the population has 150,000 pregnant 30 year olds
- Finally make another table for pregnancy at age 39 – again sensitivity and specificity remain the same but the prevalence is 1 out of 438. Assume the population has 25,000 pregnant 39 year olds.

- For each age group, calculate the PPV, and NPV and determine the probability of a false positive given a positive test result and determine the probability of a false negative given a negative test result.

There are some additional facts about these tests you should know

- If an abnormal ratio is present, an amniocentesis is recommended. In this procedure the risk of fetal death is 1 in 200 (regardless of age of mother)
- The cfDNA costs about $1500 per test. It is usually not covered by public or private insurance. Amniocentesis costs around $4500 per test.
- These tests are being heavily marketed directly to public (you see the ads on TV – ask your doctor…). Now more pregnant women are asking for the test, especially to know the sex of their baby (so they can have a blue cupcake, pink cupcake party!)
- FYI: A third possibility of tests results is called a no-call. When a no-call is returned, a doctor usually recommends the mother has an amniocentesis.

Suppose you are working for a regional public health department for a large city. Your job is to write a report for your boss recommending the policy on when this test should be administered. Because you will have to defend your report you need to include all the data in the report you calculated. Make sure you take into consideration the additional facts above in your recommendation.