

Data Analysis

Data Set 1: General population

10,000 pregnant women were tested with the NIPT early in pregnancy. After birth, 10 of the newborns were found to have Trisomy 21; of these 10, all 10 had tested positive for Trisomy 21 using the NIPT test early in pregnancy and 0 tested negative. 9,990 were unaffected by Trisomy 21; of these, 20 had tested positive for Trisomy 21 using the NIPT test early in pregnancy and 9,970 tested negative.

	Trisomy 21 +	Trisomy 21 -	Total
NIPT +	10	20	30
NIPT -	0	9,970	9,970
Total	10	9,990	10,000

Prevalence = $10/10,000 = 0.1\%$

Sensitivity = $10/10 = 100\%$

Specificity = $9,970/9,990 = 99.7\%$

PPV = $10/30 = 33.3\%$

NPV = $9970/9970 = 100\%$

Data Set 2: Women 40 years and older

10,000 pregnant women age 40 and older were tested with the NIPT early in pregnancy. After birth, 100 of the newborns were found to have Trisomy 21; of these 100, 99 had tested positive for Trisomy 21 using the NIPT test early in pregnancy and 1 tested negative. 9,900 were unaffected by Trisomy 21; of these, 20 had tested positive for Trisomy 21 using the NIPT test early in pregnancy and 9,880 tested negative.

	Trisomy 21 +	Trisomy 21 -	Total
NIPT +	99	20	119
NIPT -	1	9880	9881
Total	100	9900	10,000

Prevalence = $100/10,000 = 1\%$

Sensitivity = $99/100 = 99\%$

Specificity = $9,880/9,900 = 99.7\%$

PPV = $99/119 = 83.2\%$

NPV = $9,880/9881 = 99.9\%$

The sensitivity and specificity of this test are both very high. There are extremely high rates of both true positives and true negatives which makes this an excellent screening test, especially because it is non-invasive. There are some false positives with this screening test which could bring unnecessary stress and emotional trauma to clients and the possible need for further diagnostic testing. I could not find any specific information for my state on testing recommendations, but I will highly recommend this test to my own clients if they want this type of testing done. I know midwives in my state do offer NIPT to their clients.