NIPT Accuracy

~ Trisomy 18 and Trisomy 13 Screening ~

Non Invasive Prenatal Testing (NIPT) is not as accurate as advertised

by Dr Marty McCaffrey, neonatologist
Taken from a SOFT Facebook Group post 10/29/2015
https://www.facebook.com/groups/TrisomySOFT/

You may have heard about new testing that can be done on maternal blood to see whether a baby has Trisomy 21 (Down Syndrome), Trisomy 18 (Edwards Syndrome) or Trisomy 13 (Patau Syndrome). These tests are marketed as having specificity and sensitivities that are 98% or so. Sounds great right? These numbers lead some to believe that whether the test is positive or negative, it is right 98% of the time. That would be…WRONG!
I won’t belabor the statistics here but what you care about with a screening test like the new testing of maternal blood for infant DNA is positive (PPV) and negative (NPV) predictive value. If the test is positive, what % of those positives are true positives.? If the test is negative, what percent of those negatives are true negatives? 

So, look at these slides from a recent New England Journal publication on the accuracy of the new “cell free DNA” tests for screening unborn babies for trisomies. If you are negative on one of these tests the chances are virtually 100% (almost; see last parent comment below – editor) that you do not have one of the trisomy disorders.

Not the same story for positives. The reason is that the less likely a screened population is to have a baby with trisomy, the higher the chance for a false positive.

How high is the chance for a false positive? VERY HIGH. For Trisomy 21, when high risk patients (>35 years) are included, the PPV is only 80% for
Down Syndrome, 90% for Edwards Syndrome and 50% for Patau Syndrome. This is the reason that these tests are NOT to be used as diagnostic tests by maternal providers. These tests are SCREENING tests to be used in high risk populations, not low risk women. After such testing, for those with a positive desiring a more definitive diagnosis, amniocentesis or chorionic villous sampling should be undertaken.

Sadly what will likely happen is many obstetricians will offer these tests, without proper genetic counseling and some mothers will make a decision to abort a pregnancy based on an inaccurate screening diagnosis.

Comments from parents

I’m lucky that my doctor was very honest about these tests. She told me it was just an indication that further tests and ultrasounds were needed. I can’t imagine how many pregnancies have been ended with false positives on these blood tests.
Also, think about what this does to skew the statistics on live births of these children.

So, I’m in Vermont. The docs here do not offer termination based on any screening test (quad screen or others) or markers on an ultrasound. An amnio is required for a definitive diagnosis. And mom is sent to our largest state hospital for genetic couns...

And honestly, in our current medical model, genetic counseling and additional testing equals huge profits and less lawsuits.

I (a parent who is a Nurse Practioner) just did a literature review on cell free DNA and aneuploidy. The false positive rates are higher than the false negative rates, which, when being used to counsel couples on termination, is quite scary!

I took the Harmony test and it came back negative! However my son has FT18. These tests are definitely not 100% or even close