



November 3, 2011

Colleagues,

As many of you are aware, there is now a commercially available maternal blood test to screen for Trisomy 21.

The four identified high risk populations for which this test is currently indicated are:

1. Positive prenatal screening
2. AMA
3. Family history of Down syndrome
4. Fetal US abnormality

In these high risk patients, the test has been shown to have a very high sensitivity (99.1%), and a very high specificity (99.8%). This means that the test picks up 99% of Down syndrome with only 2/1000 false positives. Therefore, this test is an outstanding screening test but it is **not a diagnostic test**. Positives must be confirmed with amnio/CVS and negatives are not a guarantee that the fetus does not have aneuploidy.

The test is not suitable for universal screening of low-risk women, because the test has not been validated in that population and the performance of the test cannot be assumed to be identical to that in high risk populations.

This new advanced screening test complicates our current screening process and makes informed consent quite complex. The International Society for Prenatal Diagnosis recommends **formal genetic counseling prior to ordering this test** and their statement (attached) outlines salient points in the counseling. To give you an idea of the complexity of the counseling, some of the key points are summarized on the attached page ("The Fine Print").

We agree with the ISPD recommendation that patients with any of the four risk factors should have genetic counseling because of the complexity of the screening and testing options.

We will be happy to accept referrals for genetic counseling for AMA patients and will of course recommend genetic counseling for US anomalies, abnormal first trimester results, and family history of T21.

We will be planning an educational evening meeting in the next few weeks for all of our interested referring providers. More information to come...

Please call me with any questions about this important new information.

Alan Fishman, MD



MaterniT21: “The Fine Print”

The MaterniT21 test from Sequenom must be drawn at the Hunter lab draw station in Campbell, and can be drawn any time after 10 wks. The turn-around time for test results is 8-10 business days (typically 12-14 calendar days, longer if intervening holidays).

A negative MaterniT21 decreases the risk for Down syndrome by 72-fold. This fact needs to be carefully explained to patients who are considering prenatal diagnosis. If the first trimester risk is 1:10 the risk with a negative MaterniT21 would be 1:720. Some patients may desire amnio despite a negative MaterniT21 test, in which case waiting 2 weeks and spending the money for the test may not be the best choice.

Women who are not interested in invasive testing at all may wish to have MaterniT21 testing as an alternative to California Prenatal Screening program 1st trimester screening and/or 2nd trimester screening, and others may want to have it in addition to California Prenatal Screening. Patients who decline invasive diagnostic testing (amnio or CVS) need to understand that MaterniT21 is not diagnostic.

Sequenom does not currently have any contracts with payors and their charges are as follows:

Retail price	\$2700
Cash pay price	\$1900

For patients with PPO insurance the charge is \$240 at time of draw and Sequenom will accept assignment of benefits and they will work with insurances to get paid/contracted.

For HMO or Medicaid patients unless there was authorization then the cost would be \$1900 to your patients.

Of course we will counsel all patients as to the availability of testing but it seems unlikely that many HMO or Medi-Cal patients will opt to pay \$1900. If they are unwilling to pay for the test themselves, as a practical matter, they will not realistically have this testing available until their respective payors decide about whether or not this will be a covered benefit.

The test is validated only for detection of Trisomy21. In theory, the technology should also detect Trisomy 18 and Trisomy 13. Sequenom’s CMO and lab director has told us, unofficially, that they will report a positive screen for these other trisomies, but we do not yet have validation of sensitivity, specificity or predictive values for this.

The test is validated only for singleton pregnancies at this time.