

Dear Wisconsin Upside Down Members,

As you may have heard, there has been a major development for the Down syndrome community in the last 48 hours. On Monday, October 17th a San Diego-based biotechnology company called [Sequenom released a non-invasive pre-natal test for Trisomy 21](#). It is now available in 20 cities. At least two other companies are preparing to roll out similar tests in the next two years.

The significance of this news cannot be overstated for the Down syndrome community. Amniocentesis and chorionic villus sampling, the current standards for detecting chromosomal conditions during pregnancy, involve risks to the fetus, which many expectant parents choose not to take. The result is that the vast majority of Down syndrome diagnoses happen after the child is born.

However, the emergence of this new simple blood test, which can be administered in the first trimester, means that a prenatal diagnosis will almost certainly become the norm in the years ahead. The promise of earlier, largely accurate detection at no risk to the pregnancy will no doubt be welcomed by expectant parents as positive advancements.

The Dilemma

As a [New York Times article](#) on the subject notes, the new tests raise a number of troubling ethical issues. Dr. Brian G. Skotko points out in the story that these tests could encourage more people to end their pregnancies, causing a decline in the numbers of people with Down syndrome and leading to diminished support for all people with Down syndrome.

Behind this concern is the issue of whether expectant parents, told that their child is likely to have Down syndrome, and facing a daunting decision, have all the necessary information to make the best choice for their family.

In the case of the Sequenom test, dubbed MaterniT21, it is essential that parents are clear on the limitations. First, the test only detects trisomy 21, not other forms of Down syndrome, such as translocation or mosaic Down syndrome. A "negative" result could still mean that the fetus has trisomy 18, trisomy 13, or other genetic conditions, which would have been picked up by an amniocentesis or CVS. Second, the Sequenom test for trisomy 21, while extremely accurate, is not foolproof. According to a clinical study, a "positive" result will be wrong 1.8 percent of the time, a "negative" result .2 percent.

Even more importantly is the question of whether expectant parents truly understand what it means to have a child with Down syndrome. With early, accurate detection becoming commonplace, will expectant parents reflexively choose not to have a baby with Down syndrome without fully understanding the joy and wonder that people with Down syndrome offer?

The Truth

If the answer is yes to the question above, it misses something central about the Down syndrome experience that we in the Down syndrome community know, but that the general public does not yet grasp. Namely, that for all the challenges of living with Down syndrome, parents, siblings, even people with Down syndrome overwhelmingly report that they are enriched by the experience.

Until a few weeks ago, this was an unproven truth. However, earlier this month, Dr. Skotko, along with Susan Levine of Family Resource Associates, published a study in the [October issue of the American Journal of Medical Genetics](#) that backed it up with hard numbers. According to the study:

- 79 percent of parents or guardians felt their outlook on life was more positive because of their child. Only 5 percent felt embarrassed by their child.
- 94 percent of siblings age 9 and older expressed feelings of pride about their sibling. 88 percent said they felt they were better people because of their sibling with Down syndrome. Only 4 percent said they would "trade their sibling in" for another.
- 99 percent of people with Down syndrome said they were happy with their lives. 97 percent liked who they are and 96 percent liked how they look. Only 4 percent expressed sadness about their life.

When talking about life with Down syndrome, it is also critical to note the medical advances in recent decades that have led to a dramatic increase in life expectancy. Today, people with Down syndrome live, on average, to 60 years old, with many living into their seventies.

The Response

Fortunately, the Wisconsin Upside Down is perfectly positioned to advocate on behalf of all people with Down syndrome, their families and loved ones in these complex times. With years of experience behind us - and a wealth of connections and expertise that we have built over this time - we are uniquely equipped to serve current and future families as these and other technological developments unfold.

Given the changes the new tests will engender, more expectant parents than ever will need support and guidance to make informed decisions. Wisconsin Upside Down is prepared to meet these families' needs on a number of levels through both longstanding, proven programs and bold new initiatives.

On a federal level, Wisconsin Upside Down has joined forces with Down syndrome leaders across the country in anticipation of these new noninvasive tests hitting the market. We have sought to reach consensus on the difficult issues the tests gives rise to, and we have collectively lobbied our leaders on Capitol Hill.

Just last week, our national affiliate, the National Down Syndrome Society, released [a position statement on prenatal tests](#), which Wisconsin Upside Down board of directors has endorsed. Among other things, the statement declares that for a test to be called a "Down syndrome test," all forms of Down syndrome must be evaluated. Also, the decision whether to have a test performed must be that of the pregnant woman and her significant other, and the decision should remain strictly confidential.

In closing, we recognize the emotional resonance this issue holds for our friends and families in the Down syndrome community. We fully understand the mix of emotions this news may give rise to, even as the full implications for our members remain, to some degree, unknown. That said, we hope you do know that Wisconsin Upside Down will continue to be here to help in whatever way possible. We are more committed than ever to ensuring that all individuals in Wisconsin with Down syndrome are valued, included, and given every opportunity to pursue fulfilling lives.

Sincerely,
Robbin Thomas Lyons
President **Wisconsin Upside Down** www.wiusd.org

Down syndrome is down right beautiful
[262-443-8690](tel:262-443-8690)