

# MetDESK News

MetLife's Division of Estate Planning for Special Kids

*Information and Support for the Special Needs Community*



## MetLife®

### In the Public Eye –Prenatal Testing for Down Syndrome



2008 was a fast-paced year for the Down syndrome community as ABC News and Time Magazine featured a genetic test for Down syndrome as one of the top ten medical stories of the year. While genetic testing companies worked on developing safer, earlier and more accurate prenatal blood tests for the genetic condition, many local Down syndrome organizations

were increasing their efforts to offer doctors and patients better support and more comprehensive information for women seeking prenatal diagnosis services.

As commercial laboratories continued science's march of progress in delivering a diagnosis ever earlier in a pregnancy, volunteer organizations were attempting to bridge the gap between the knowledge provided by science and the information needed by doctors and families.

In September 2008, Congress recognized the need for families to receive more support and information when it passed the Prenatally and Postnatally Diagnosed Conditions Awareness Act. The measure, which has yet to be implemented, has set the stage for more meaningful conversations about the humanity of people who have a genetic diagnosis. A diagnosis can provide a scientific result and a general description of a condition but often leaves many questions unanswered.

Here are just a few:

-- "What is life like for people with Down syndrome and their families?"

-- "Will my child ever be able to live on their own, get married, have a job?"

-- "Can I afford to take care of a child with a disability, and where can I find help?"

Local Down syndrome organizations can help answer these important questions by offering meaningful resources to doctors and genetic counselors as well as parent-to-parent support for those couples who may wish to talk with someone who has life experience.

As genetic testing companies announce the development of tests for even more genetic conditions, the Down syndrome community is also providing leadership for other disability groups on how to give support to families and physicians responding to a growing number of clinical tests.

In a recent report by CNN Money, Sequenom, Inc. Chief Executive Harry Stylli was quoted as saying that "the market for testing for prenatal chromosomal disorders like Down syndrome could be \$3 billion to \$5 billion worldwide. But a larger opportunity may exist in testing for tiny variations in the genetic code that can cause inherited diseases like autism, heart ailments, cystic fibrosis and birth defects. Charles Cantor, Sequenom's Chief Scientific Officer, said in a telephone interview, "Down's is the single most common fetal defect, but it's the tip of the iceberg."

With the expected arrival of a huge international market for prenatal genetic testing also comes a growing need for education about the conditions being tested. Down syndrome organizations stand in the forefront of the disability community as they learn to provide support and education for the delivery of a prenatal diagnosis.

Although prenatal testing for Down syndrome has been available for over 30 years, the need for collaborative dialogs with medical professionals became more apparent in 2007. At that time, the American College of Obstetricians and Gynecologists (ACOG) issued recommendations that prenatal screening and diagnostic tests for Down syndrome should be offered to all pregnant women regardless of age.

Down syndrome organizations anticipated these recommendations would cause more expectant mothers to opt for testing, which in turn would increase the need for accurate information and support both before the testing takes place and after a test produces an unexpected result. While ACOG itself did not provide information resources to doctors, it acknowledged the need and recommended that physicians refer patients to Down syndrome organizations where they can learn the “natural history” of Down syndrome.

Women currently receive information about a possible prenatal diagnosis of Down syndrome in the following ways:

1. Prenatal screening blood tests provide a numerical chance that a fetus has Down syndrome. For example, “You have a 1/350 chance of having a child with Down syndrome.” The results cannot predict with certainty whether or not Down syndrome is present, but rather provide only a probability score.
2. Invasive diagnostic tests, including an amniocentesis or a chorionic villus sampling (CVS), can tell a woman for certain whether her fetus has Down syndrome. These tests carry a small but significant chance of causing the woman to miscarry her fetus..

Some laboratories, including those at Stanford University and for-profit Sequenom, Inc., say they are now developing tests that could deliver definitive results with just a simple blood draw in the first trimester of pregnancy, thereby avoiding the risk of a testing-related miscarriage. If these safer and more accurate tests come to market, combined with ACOG’s recommendations, it seems likely that more women will be tested than ever before, and that the need for support and reliable information will increase dramatically.

Since studies indicate that 90% of American women who receive a confirmed diagnosis of Down syndrome choose to terminate a pregnancy (Mansfield, et al., 1999), Down syndrome advocates have wondered whether prospective parents were being given accurate and up-to-date information about Down syndrome when they received a diagnosis.



A 2005 study of women who received a prenatal diagnosis of Down syndrome found that “about half [of surveyed mothers] felt pressured into making a decision.” The women also reported that some doctors “emphasized only the negative aspects of Down syndrome” and tried to change their

patient’s decision to continue the pregnancy. The women said they needed relevant, up-to-date information and referrals for support, because “nearly all of [the respondents thought their obstetricians had failed to provide enough up-to-date printed material on Down syndrome” and “obstetricians did not appear to explain Down syndrome adequately.” (Skotko 2005)

The research suggested that accurate and up-to-date materials were largely unavailable, and that negative stereotypes about Down syndrome still lingered in the medical community.

This is perhaps not surprising, given that the life circumstances of people with Down syndrome have been changing dramatically in recent years. Thanks to advances in medical care, education and social services, people with Down syndrome are now living longer, healthier lives and achieving more than ever before.

See our next issue for Part 2.

MetDESK®, MetLife’s Division of Estate Planning for Special Kids, works with national non profits to help support the mutual goal of helping individuals with special needs. MetDESK® is dedicated to helping families plan for the future of children and/or dependents with special needs. Helping you answer those questions, directing you to the right resources, and just being there as a resource and advocate for you, is an important part of what we do. If you’d like to find out more, or you’d like to be referred to a local MetDESK® Specialist, please call 1-877-MetDESK, or 1-877-638-3375, or visit our website at [www.metlife.com/DESK](http://www.metlife.com/DESK).

This article was written as a collaborative effort by the Down Syndrome Affiliates in Action ([dsaia.org](http://dsaia.org)) which provides resources and a conference specifically for leaders of Down Syndrome organizations across the country. For more information about your local affiliate, please refer to [dsaia.org/affiliatelist](http://dsaia.org/affiliatelist) which provides the name and website addresses of organizations providing direct parent support.



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**Produced by MetLife**  
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