

## **Mothers Speak Again: Physicians Must Improve How They Deliver a Prenatal Diagnosis of Down Syndrome**

by Brian Skotko

I have recently reported to you the results from my study on how physicians could better convey a diagnosis of Down syndrome after a child is born. As many of you know, however, Down syndrome can now be diagnosed prenatally, as early as the first trimester of pregnancy. The process of conveying a prenatal diagnosis is decidedly different: obstetricians or genetic counselors are typically the professionals delivering the prenatal news whereas neonatologists or pediatricians customarily convey postnatal diagnoses. Further, mothers who receive a prenatal diagnosis have the option of deciding whether or not to continue their pregnancies.

Two years ago, I asked mothers who have children with Down syndrome to respond to a survey, examining how they had received the diagnosis for their child. Of the 1,250 responses that poured in, 141 were from mothers who had received definitive diagnoses of Down syndrome before their child was born. I am delighted to inform you that the reflections and recommendations of these mothers will now have a national voice in the leading medical journal, *American Journal of Obstetrics and Gynecology*, where my study has been published in the March issue.

This purpose of my study was to ask mothers who had children with prenatally detected Down syndrome: How did your physician convey the information and what, if anything, could have been better? Five parent support groups—the Massachusetts Down

Syndrome Congress (Massachusetts), the Mile High Down Syndrome Association (Colorado), the Triangle Down Syndrome Network (North Carolina), the Down Syndrome Association of Los Angeles (California), and the Down Syndrome Society of Rhode Island (Rhode Island)—distributed copies of my research survey to all of the mothers on their mailing lists. Thanks to generous responses, mothers have made this the most comprehensive and robust study on the topic of prenatally diagnosed Down syndrome, to date.

As you might know, all pregnant women over the age of 35 are offered prenatal testing for Down syndrome, and younger women are increasingly requesting such tests on their own. Typically, although not necessarily, mothers will begin with a prenatal screening test like the triple screen, quadruple screen, or the newest combination of maternal serum markers and ultrasound findings. The results from these tests simply give a pregnant woman an odds-ratio as in, “You have a 1 in 847 chance of having a child with Down syndrome.” Based on these numbers, many mothers will go on to have one of the two definitive prenatal tests: chorionic villus sampling (CVS) or amniocentesis. The results from these tests are conclusive.

The majority of mothers sampled in my study felt anxious and scared after receiving the results from the amniocentesis. While mothers who received prenatal care within the past five years seemed especially satisfied with the care that they received, they reported that the doctors did not tell them about the positive potential of people with Down syndrome nor did they feel like they received enough up-to-date information or contact information for parent support groups.

Through their collective responses, mothers have now asked obstetricians and genetic counselors to adopt 7 recommendations when delivering a prenatal diagnosis of Down syndrome:

- (1) **Results of the prenatal screening should be clearly explained as a risk assessment, not a “positive” or “negative” result.** Many mothers understood the triple or quadruple screen to be an all-or-nothing statistic. Physicians need to explain, in plain English, what the odds ratios mean so that mothers can make an informed decision on if and how to proceed with more definitive testing.
- (2) **Results from the amniocentesis of CVS should, whenever possible, be delivered in person, with both parents present.** Mothers who had to learn of the diagnosis by telephone reported intense resentment for their obstetricians and/or genetic counselors. Ideally, physicians should ask that all persons receiving prenatal testing to return, in person, to hear the results.
- (3) **Sensitive language should be used when delivering a diagnosis of Down syndrome.** Mothers requested that physicians not begin by saying, “I’m sorry,” or “Unfortunately, I have some bad news to share.”
- (4) **If obstetricians rely on genetic counselors or other specialists to explain Down syndrome, sensitive, accurate, and consistent messages must be conveyed.** One mother wrote that her genetic counselor “showed a really pitiful video first of people with Down syndrome who were very low tone and lethargic-looking and then proceeded to tell us (in 1999) that our child would never be able to read, write, or count change.”

- (5) **Physicians should discuss all reasons for prenatal diagnosis including reassurance, advance awareness before delivery of the diagnosis of Down syndrome, adoption, as well as pregnancy termination.** Physicians must now realize that many mothers who opt for prenatal testing have no intentions of terminating their pregnancies and are offended by their physicians' assumptions that they would do so.
- (6) **Up-to-date information on Down syndrome should be available.** Mothers have requested two kinds of information: accurate clinical knowledge and "success stories" that demonstrate the potential and possibilities for children with Down syndrome. The Healthcare Guidelines for infants and toddlers with Down syndrome was cited for the clinical information, and books such as *Common Threads: Celebrating Life with Down Syndrome*, *Babies with Down Syndrome: A New Parent's Guide*, and *Choosing Naia: A Family's Journey* were all found to be helpful as success stories.
- (7) **Contact with local Down syndrome support groups should be offered, if desired.** Mothers consistently and overwhelmingly appreciated when their physicians provided contact information for local support groups. One mother reported that after talking to other parents, "I felt 100% better and positive about having my daughter."

Interestingly, mothers who received prenatal diagnoses were generally happier over the birth of their infants with Down syndrome than mothers who had received the news postnatally (as I had reported to you previously). This difference might stem from the fact that mothers who had prenatal testing tended to resolve any grief before the child

was born. Nonetheless, 141 mothers who have had prenatal testing have now spoken, and their recommendations are constructive ones. Delivering a prenatal diagnosis of Down syndrome should no longer be viewed as a gloomy affair. In fact, with the appropriate sensitivity and explanation, obstetricians can make the births of children with Down syndrome celebratory experiences for those mothers who choose to continue their pregnancies after receiving prenatal diagnoses.

You can ensure that this is so by sharing the complete study with the obstetricians and genetic counselors in your surrounding area. (The complete report is available online at <<list parent support group web page>>.)

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*Brian Skotko is a joint-degree student at Harvard Medical School and Harvard's John F. Kennedy School of Government. He has a 24-year-old sister with Down syndrome and has co-authored the book, Common Threads: Celebrating Life with Down Syndrome. He can be contacted at [Brian\\_Skotko@student.hms.harvard.edu](mailto:Brian_Skotko@student.hms.harvard.edu).*

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