

Delivering a Prenatal Diagnosis of Down Syndrome

In the last *DSN* (Vol. 28, No. 1), the cover story reported results of a research survey by Brian Skotko, a joint-degree student at Harvard Medical School and the John F. Kennedy School of Government, that demonstrated mothers believe physicians need to do a better job in delivering a postnatal diagnosis of DS. More than 1,200 mothers responded to that survey, including 141 who received a prenatal diagnosis of DS. The responses of mothers who received a prenatal diagnosis were analyzed separately, are reported in the current issue of the *American Journal of Obstetrics and Gynecology* and summarized below.

It's probably not surprising that most mothers were scared and anxious when they learned of the diagnosis and didn't feel that their obstetricians had explained DS adequately. About a quarter learned of the results in person and nearly 75 percent didn't have their partner present. About half felt rushed or pressured to decide whether or not to continue the pregnancy. Most mothers liked the printed material that they received, but did not believe their obstetricians provided enough information or phone numbers of other parents who have children with DS.

Results suggest seven ways that physicians can improve how a prenatal diagnosis of DS is delivered to patients.

Results of the triple screen should be clearly explained as a risk assessment, not a "positive" or "negative" result. Many mothers understood the triple screen to be an all-or-nothing diagnostic test, even after their obstetrician had given them the results. The weak sensitivity and positive predictive value of the test should be explained in terms that each mother can understand. In addition, mothers requested that DS be first explained after the screening test rather than waiting for the results of an amniocentesis or chorionic villus sampling (CVS) to begin a discussion.

1. Results of the amniocentesis or CVS should, whenever possible, be delivered in person, with both parents present. Mothers who had learned of the diagnosis by telephone reported intense resentment for their obstetricians and/or genetic counselors. Ideally, physicians should ask that all persons receiving definitive prenatal testing return in person to hear the results. If a personal visit is not possible, physicians should offer each couple the option of returning or receiving the results over the telephone. If the latter, physicians should note that women who have children with DS wish they had learned the results in person, with their partner present. If the diagnosis is delivered on the telephone, the physician should arrange for a follow-up in person visit as soon as possible.
2. Sensitive language should be used when delivering a diagnosis of DS. Mothers requested that physicians not begin by saying, "I'm sorry," or "Unfortunately, I have some bad news to share." In addition, several mothers, including some who had

children as recent as 1997, reported their obstetricians had used the word “mongoloid” in describing DS, a term that is reprehensible in today's society and should not be used by today's physicians.

3. If obstetricians rely on genetic counselors or other specialists to explain DS, sensitive, accurate, and consistent messages must be conveyed. In 1999, one mother reported that her genetic counselors “told my husband and I that our child may not be able to complete school, will have limited cognitive abilities, and may remain a child, emotionally and mentally for life. Her information didn't include any possibilities of the lowest to highest range of functioning at all.” Another mother wrote, “[the genetic counselor] showed a really pitiful video first of people with DS 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.”
4. Discuss all reasons for prenatal diagnosis including reassurance, advance awareness before delivery of the diagnosis of DS, and adoption, as well as pregnancy termination. Many of the mothers who responded to this survey never planned to terminate the pregnancy and were upset when their physicians provided detailed descriptions of pregnancy terminations without knowing whether they would like those options discussed.
5. Up-to-date information on DS should be available. Respondents requested clinical information on the health concerns for infants with DS and “success stories” that demonstrated the potential and possibilities for children with DS. A list of current and relevant resources can be found on the NDSC's Web site, www.ndsccenter.org.
6. Contact with local DS support groups should be offered, if desired. Respondents appreciated providers who gave them contact information for local DS support groups. One mother reported that, after talking to other parents, “I felt 100 percent better and positive about having my daughter.” Another mentioned, “I regret that I didn't get involved with any support groups in the beginning. I thought everyone would sit around and cry on each other's shoulders, and I wasn't ready for a pity party. I only wish that physicians, nurses, and hospitals were better informed about the wonderful opportunities that are out there to help parents.”

Finally, most mothers reported that when they knew in advance that their baby had DS, the birth experience was a positive one. (JA)

Henry

Henry was born in August and had heart surgery to repair AV canal in December. Now he's moved on to other important things — like learning to sit up by himself. Henry's mother, Liz, had an amniocentesis because she and her husband, Tony, didn't want any unexpected surprises when their baby arrived. She got the results of her amnio over the phone while she was at work. Liz works in a school and was surrounded by people —

although her husband wasn't one of them — when the genetic counselor began, “We don't like to see this result.” When she complained to the clinic about how the diagnosis was delivered, her concerns were dismissed. The one thing the clinic did right, according to Skotko's research, was to give them an up-to-date packet of information from a DS parent group.

Liz switched clinics and was rewarded to find herself in a birth prep class with two other expectant mothers who also knew they were carrying babies with DS. She later met a third mom and they've all been invaluable support to one another.

In addition to the DS diagnosis, Liz and Tony learned that their baby boy had a heart defect. The pediatric cardiologist had been concerned that Henry had a serious heart defect that would not be possible to repair. However, they were relieved to learn that was not the case. When their little boy was born — “a joyous event” — his parents were delighted.

Happy parents must be a rarity in a special care nursery. Hospital staff kept telling Henry's parents that those feelings would pass. Not so far. Liz says, “My pregnancy was so filled with fear — we kept getting scary news. The doctors had us prepared to welcome a very ill baby into the world. From the moment he popped out, Henry has been a vibrant and strong boy. He reassures us daily. He's just a joy. The word I think best describes how Tony and I feel is lucky.”