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Study Shows: Physicians Must Improve How They Deliver a Prenatal Diagnosis of Down Syndrome

7 Specific Recommendations Offered

BOSTON, MA—A survey of mothers in the March issue of the *American Journal of Obstetrics and Gynecology* found that obstetricians and genetic counselors are falling short when it comes to delivering a prenatal diagnosis of Down syndrome to pregnant women. Mothers who have children with Down syndrome, diagnosed prenatally, reported that 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. Further yet, the mothers report that all of these shortcomings are happening at an emotional time when women have to decide whether or not to continue their pregnancies. This study remains the largest and most comprehensive study on prenatally diagnosed Down syndrome, to date.

One mother in the study reported 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.”

The study was done by Brian Skotko, a student at Harvard Medical School (HMS) and Harvard's John F. Kennedy School of Government, and supported by the Tim White Fund from Children's Hospital Boston and a part-time research grant from HMS.

Skotko mailed an 11-page survey to nearly 3,000 members of five Down syndrome parent organizations in California, Colorado, Massachusetts, North Carolina, and Rhode Island. Of the 1,250 responses, approximately 140 were from mothers who had received a definitive prenatal diagnosis through amniocentesis or chorionic villus sampling.

“Doctors continue to find it very challenging to deliver a diagnosis like Down syndrome to an otherwise happy expectant mom,” says Skotko, who has a 24-year-old sister with Down

syndrome and co-authored the award-winning book, *Common Threads: Celebrating Life with Down Syndrome* (Band of Angels Press). “But the results of this study are conclusive: Delivering a prenatal diagnosis of Down syndrome does not have to be a gloomy affair. In fact, mothers in this study have now written the prescription on how best to explain the diagnosis in a loving manner.”

Based on the mothers’ comments, Skotko offers a 7-point “prescription” for communicating a diagnosis of Down syndrome:

- Results from the prenatal screening should be clearly explained as a risk assessment, not as a “positive” or “negative” result.
- Results from the amniocentesis or CVS should, whenever possible, be delivered in person, with both parents present.
- Sensitive language should be used when delivering a diagnosis of Down syndrome.
- If obstetricians rely on genetic counselors or other specialists to explain Down syndrome, sensitive, accurate, and consistent messages must be conveyed.
- 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.
- Up-to-date information on Down syndrome should be available.
- Contact with local Down syndrome support groups should be offered, if desired.

Skotko published a companion paper in the January issue of *Pediatrics* summarizing responses from women who received the Down syndrome diagnosis postnatally. He also has conducted the same pair of studies in Spain to get a cross-cultural perspective.

Dr. Allen Crocker, Skotko's faculty advisor on the project and director of the Down Syndrome Program at Children's Hospital Boston, says that the survey findings echo his nearly 40 years of experience working with families. "Physicians have consistently been inadequate and incomplete, and, on occasion, offensive," he says. "These two studies offer the most searching review of parents' experiences of postnatal and prenatal presentation of a diagnosis of Down syndrome ever published, and they have been done with considerable statistical care. This is clearly a case of families teaching physicians."

Approximately one of every 1,000 children in the U.S. is born with Down syndrome, meaning that approximately 5,000 parents receive the diagnosis for their child each year. Of the mothers who receive the diagnosis, about 12.5 percent find out before birth, suggesting that 625 newborn infants with Down syndrome will be diagnosed prenatally each year.

All pregnant women over the age of 35 are now 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 two maternal serum markers and ultrasonographic findings. With a 5 percent false-positive rate, 69 percent of fetuses with Down syndrome are correctly detected with triple screening, 75 percent with quadruple screening, and 79 percent with the recent first-trimester screening involving 2 maternal serum protein markers and ultrasonographic findings. For a definitive prenatal diagnosis, mothers have one of two options: chorionic villus sampling (CVS), typically between the 10th – 12th weeks of pregnancy or amniocentesis, typically after the 15th week of pregnancy. Neither procedure, however, is without risk; both carry an approximately 0.35 percent to 0.30 percent additional chance of causing a spontaneous miscarriage.

To identify parents of children with Down syndrome who are willing to be interviewed about their experiences, contact the Massachusetts Down Syndrome Congress (617-308-8067; Suzanne Shea, President), National Down Syndrome Congress (1-800-232-NDSC; David Tolleson, Executive Director), or the National Down Syndrome Society (1-800-221-4602; Suzanne Elliott Armstrong, Director of Communications).

CHILDREN'S HOSPITAL BOSTON

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