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How to Deliver the News? New Advice for Doctors Diagnosing Prenatal Down Syndrome

BOSTON —September 28th, 2009 New prenatal tests for Down syndrome are soon to be offered to all pregnant women across the United States, yet telling an expectant couple that their child will be born with Down syndrome is a task very few physicians are trained for, claims research published in the *American Journal of Medical Genetics*. The study, which reviewed decades of surveys and interviews, offers several recommendations for how physicians can best deliver the news.

A 29-member research team, led by Dr. Brian Skotko from Children's Hospital Boston, supported by the National Down Syndrome Society and informed by experts from across the field reviewed surveys and research ranging from 1960 to present day to consider how prepared physicians felt they are to deliver a diagnosis. They also studied the opinions of couples who had received the diagnosis to determine the best way of delivering the news.

"Down syndrome (DS) remains the most common chromosomal condition. It occurs in one out of every 733 live births," said Skotko. "Nearly every obstetrician can expect to have a conversation with expectant parents about the realities of life with DS, but very little research has been dedicated to understanding how physicians should communicate the news."

The team found that in a 2004 survey approximately 45% of obstetric fellows rated their training as "barely adequate or nonexistent"; a similar survey four years later found little change as 40% thought their training was "less than adequate." In 2005 a survey of 2,500 medical students showed that 81% believed they were "not getting any clinical training regarding individuals with intellectual disabilities."

To improve this scenario the team set out to answer five critical questions which every physician should consider before delivering a diagnosis: Who is the best person to communicate the news? When is the best time to share the news? Where should the news be delivered? What information should be offered? How should the diagnosis be communicated?

The team found that while many sources are available, from trained counsellors to midwives, expectant couples prefer to receive the news from the health care professional with the most knowledge, the physician.

Also, women who decided to undergo definitive prenatal testing for DS prefer to receive the diagnosis as soon as possible in the company of their husbands or partner, while women who had arranged for the diagnosis to be delivered by a phone call were better prepared for the news than those who received the news from an unarranged call. Women who received the diagnosis through an unscheduled call expressed intense resentment towards their obstetricians and counsellors.

Regarding the amount of information a couple should be given, mothers emphasised that they should be provided with up-to-date information about DS, its causes and the expectations for a child living with DS today. This information should include descriptions of common or anticipated health conditions seen in infants and young children.

On top of this, parents found that they benefited from personal stories that demonstrate the potential and possibilities for children with DS and if possible if possible contact information for other parents who have children with DS should be made available.

Mothers emphasized that at the time of a diagnosis, physicians should discuss all options available to them, including continuing the pregnancy, offering the baby up for adoption after birth, or pursuing termination. In a survey of 71 women from the Netherlands who terminated their pregnancy after a diagnosis of DS, 34% indicated that the option of continuation was not raised.

Finally physicians should be mindful of how they communicate the news. In the largest study most mothers requested that physicians should not begin a conversation by saying "I'm sorry" or "unfortunately I have some bad news," instead they should use neutral and nondirective language.

Unsurprisingly the team also suggests that outdated and offensive language such as "mongolism" should be avoided and instead phrases such as "a fetus with Down syndrome" should be adopted.

Ultimately the research review found that mothers who received the diagnosis prenatally and continued their pregnancy were happier with the birth of their child than those who received the diagnosis after the baby had been born. Receiving the diagnosis in advance seems to allow parents the needed time to overcome the shock and initial grief of the diagnosis and begin preparing and celebrating the upcoming birth of a child.

"Of the studies reviewed nearly all mothers reported feelings of initial shock, anger and fear following the diagnosis," concluded Skotko. "Yet, these same mothers indicated that if physicians were to implement a few simple measures, as research suggests, the experience could be more sensitive to their emotions and needs."

This paper is published in the *American Journal of Medical Genetics: Part A*. To request a copy of this paper or for other media enquiries, please contact Ben Norman on Benorman@wiley.com +44(0) 1243 770 375

A companion paper by Dr Skotko has been published today in the Journal *Pediatrics*. For more information on this paper or to request a copy of this paper please contact Benorman@wiley.com

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9 New Steps Recommended in Delivering a Prenatal Diagnosis of Down syndrome

The 29-member Down Syndrome Diagnosis Study Group recommends the following steps for all physicians making a prenatal diagnosis of Down syndrome

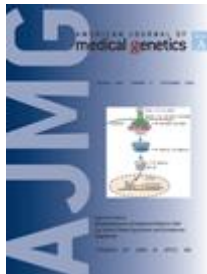
- Obstetricians should clearly outline the differences between prenatal *screening* tests and prenatal *diagnostic* test
- Prior to undergoing prenatal diagnostic tests, obstetricians should ask pregnant women if they have already formed a definitive personal decision on how they would proceed with the pregnancy. If they have not, they should mention all three options, depending on timing: continuing the pregnancy; placing the baby up for adoption after birth, terminating the pregnancy
- Obstetricians should work jointly with a health care professional knowledgeable about Down syndrome (e.g., geneticist) to share the diagnosis.
- This joint presentation should happen ideally in person or at a pre-scheduled time over the phone.
- During the conversation, the health care professionals should answer: What is Down syndrome? What causes the condition? What health care conditions go along with the condition?

- During the discussion, the health care professions should also answer: What are realistic expectations for a child with Down syndrome living today? Offering contact with other parents and support groups is of paramount importance.
- The health care professionals should use nondirective language.
- The health care professionals should offer an up-to-date bibliography of Down syndrome resources.
- The health care professionals should arrange for a follow-up appointment with the parents, including any desired meetings with subspecialists.

About the Author

The corresponding author for this paper is Brian Skotko, M.D., M.P.P. Dr. Skotko is a genetics fellow at Children's Hospital Boston, Massachusetts General Hospital, and Brigham & Women's Hospital. He has focussed his research on children with cognitive and development disabilities. Dr. Skotko is a prolific author and has co-authored the books, *Common Threads: Celebrating Life with Down Syndrome* and *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*. He currently serves on the Board of Directors for the Massachusetts Down Syndrome Congress, the National Down Syndrome Society, the Band of Angels Foundation and the Professional Advisory Council to the National Down Syndrome Congress. He has a 29-year-old sister, Kristin, who has Down syndrome. Dr. Skotko's web page is www.brianskotko.com.

To request an interview, or for questions related to this paper, contact Keri Stedman at Children's Hospital Boston: 617-919-3114 or keri.stedman@childrens.harvard.edu



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