Understanding the implications of prenatal testing for Down syndrome

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With new prenatal tests for Down syndrome on the horizon promising to be safer, more accurate, and available to women earlier in pregnancy, the medical community must come together and engage in dialogue about the impact of existing and expected tests, argues a new leading article published Online First by *Archives of Disease in Childhood*.

Authored by Brian Skotko, MD, MPP, clinical genetics fellow at Children's Hospital Boston, the article shows a steady decrease in the number of babies being born with Down syndrome since the introduction of prenatal testing and poses the question: "As new tests become available, will babies with Down syndrome slowly disappear?"

Research reviewed by Skotko showed a 15% decrease in births of babies with Down syndrome between 1989 and 2005 in the United States. In the absence of prenatal testing, researchers would have anticipated the opposite - a 34% increase in births- due to the trend of women waiting longer to have children; known to increase the chances of having a baby with Down syndrome.

Currently, expectant women have two options if they would like to receive a definitive diagnosis of Down syndrome - chorionic villus sampling (CVS) and amniocentesis - both of which are invasive and carry a risk, however small, of causing a spontaneous miscarriage. New tests expected to be introduced next year will offer a simple blood test that poses no risk to the fetus and delivers a definitive diagnosis of one or more of the genetic variants of Down syndrome - trisomy 21,

translocation, or mosaicism.

Prior research conducted by Skotko found that expectant mothers who received a prenatal diagnosis felt their physicians provided them with incomplete, inaccurate, and oftentimes offensive information about the condition. Other studies have shown physicians themselves feeling unprepared and uninformed to deliver a diagnosis.

"Unless improvements are made prior to the arrival of new prenatal tests, a true collision is on its way," says Skotko. "More women will be going through the testing process, which could lead to a lot of difficult, uncomfortable conversations between physicians and expectant parents."

In anticipation of these tests, which could make Down syndrome the first genetic condition to be definitively diagnosed in the first trimester on a population basis, Skotko calls on the medical community to:

- Develop guidelines around how health professionals should deliver a diagnosis of Down syndrome.
- Assemble information packets that give accurate, current information on Down syndrome and give them to all expectant parents who receive a definitive diagnosis.
- Create a standardized training program for all healthcare professionals involved in prenatal care and for volunteer parents - such as those involved in First Call programs - to complete.
- Train not only the doctors of today but the doctors of tomorrow. Medical students, nurses and genetic counselors should be educated beyond the textbook and have interactions with people who have Down syndrome so that they may better understand the

realities of living with the disorder.

Recently, the National Down Syndrome Society and the National Down Syndrome Congress announced the selection of a booklet ("Understanding a Prenatal Down Syndrome Diagnosis") as a gold standard packet of information about Down syndrome, fulfilling Skotko's second recommendation.

"The ultimate goal is to ensure families receive accurate, up-to-date, information so they are well-informed and can make decisions that are right for them," says Skotko.

Source: Children's Hospital Boston (news : web)