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Prenatal Test Puts Down Syndrome in Hard Focus

By AMY HARMON

DETROIT — Sarah Itoh, a self-described “almost-eleven-and-a-half,” betrayed no trace of nervousness as she told a roomful of genetic counselors and obstetricians about herself one recent afternoon.

She likes to read, she said. Math used to be hard, but it is getting easier. She plays clarinet in her school band. She is a junior girl scout and an aunt, and she likes to organize, so her room is very clean. Last year, she won three medals in the Special Olympics.

“I am so lucky I get to do so many things,” she concluded. “I just want you to know, even though I have Down syndrome, it is O.K.”

Sarah’s appearance at Henry Ford Hospital here is part of an unusual campaign being undertaken by parents of children with Down syndrome who worry about their future in the face of broader prenatal testing that could sharply reduce the number of those born with the genetic condition.

Until this year, only pregnant women 35 and older were routinely tested to see if their fetuses had the extra chromosome that causes Down syndrome. As a result many couples were given the diagnosis only at birth. But under a new recommendation from the American College of Obstetricians and Gynecologists, doctors have begun to offer a new, safer screening procedure to all pregnant women, regardless of age.

About 90 percent of pregnant women who are given a Down syndrome diagnosis have chosen to have an abortion.

Convinced that more couples would choose to continue their pregnancies if they better appreciated what it meant to raise a child with Down syndrome, a growing group of parents are seeking to insert their own positive perspectives into a decision often dominated by daunting medical statistics and doctors who feel obligated to describe the difficulties of life with a disabled child.

They are pressing obstetricians to send them couples who have been given a prenatal diagnosis and inviting prospective parents into their homes to meet their children. In Massachusetts, for example, volunteers in a “first call” network linking veteran parents to new ones are now offering support to couples deciding whether to continue a pregnancy.

The parent evangelists are driven by a deep-seated fear for their children’s well-being in a world where there are fewer people like them. But as prenatal tests become available for a range of other perceived genetic imperfections, they may also be heralding a broader cultural skirmish over where to draw the line between preventing disability and accepting human diversity.

"We want people who make this decision to know our kids," said Lucy Talbot, the president of a support group here who prevailed on the hospital to give Sarah and two teenage friends an audience. "We want them to talk to us."

The focus on the unborn is new for most parent advocates, who have traditionally directed their energy toward support for the born. But after broader testing was recommended in January, the subject began to hijack agendas at local support group meetings.

A dwindling Down syndrome population, which now stands at about 350,000, could mean less institutional support and reduced funds for medical research. It could also mean a lonelier world for those who remain.

"The impact of these changes on the Down syndrome community is going to be huge," said Dani Archer, a mother in Omaha who has set aside other Down syndrome volunteer work to strategize about how to reach prospective parents.

The 5,500 children born with Down syndrome each year in the United States suffer from mild to moderate mental retardation, are at high risk for congenital heart defects and a variety of other medical problems, and have an average life expectancy of 49. As adults, some hold jobs, but many have difficulty living independently.

"There are many couples who do not want to have a baby with Down syndrome," said Deborah A. Driscoll, chief of the obstetrics department at the University of Pennsylvania and a lead author of the new recommendation from the obstetricians' group. "They don't have the resources, don't have the emotional stamina, don't have the family support. We are recommending this testing be offered so that parents have a choice."

But the richness of their children's lives, parent advocates say, is poorly understood. Early medical intervention and new expertise in infant heart surgery stave off many health problems; legally mandated inclusion in public schools has created opportunities for friendship and fostered broader social awareness of the condition.

With no formal financing or organization, parents are arranging to meet with local obstetricians, rewriting dated literature and pleading with health care workers to give out their phone numbers along with test results. Medical professionals have for the most part responded with caution. Genetic counselors, who often give test results to prospective parents, say they need to respect patients who may have already made up their minds to terminate their pregnancy. Suggesting that they read a flyer or spend a day with a family, they say, can unnecessarily complicate what is for many a painful and time-pressured decision.

Their goal, parents say, is not to force anyone to take on the task of parenting a child with disabilities. Many participants in the ad-hoc movement describe themselves as pro-choice. Yet some see themselves as society's first line of defense against a use of genetic technology that can border on eugenics.

"For me, it's just faces disappearing," said Nancy Iannone, of Turnersville, N.J., mother to four daughters, including one with Down syndrome. "It isn't about abortion politics or religion, it's a pure ethical question."

Others admit freely to a selfish motive for their new activism. "If all these people terminate babies with Down syndrome, there won't be programs, there won't be acceptance or tolerance," said Tracy Brown, 37, of Seattle, whose 2-year-old son, Maxford, has the condition. "I want opportunities for my son. I don't know if that's right or wrong, but I do."

Ms. Brown has taken it upon herself to serve as a community resource on Down syndrome for prospective parents. She was encouraged when a counselor at the University of Washington Medical Center sent her an e-mail message recently with a question from a patient.

What developmental age equivalent, the patient wanted to know, do most people with Down syndrome reach?

For parents on an e-mail list where Ms. Brown solicited answers, the question underscored the difficulty in conveying the pleasure of parenting a child with Down syndrome to someone who has the option to reject it.

"Verbally," wrote one mother of her teenager, "she's at a 6-month level, but what 6-month-old do you know who can climb out a window and dance on a roof?!?!? We joke that she could climb Mt. Everest."

"If someone had told me Sam would still be in diapers at age 5 — ugh — I probably would have died," wrote another. "Living through it, not such a big deal. Because you don't give birth to a 5-year-old, you grow with and love this kid for five years."

Doctors have long recommended an amniocentesis test for pregnant women 35 and over, whose age puts them at greater risk for chromosomal defects. But because it carries a small risk of miscarriage, it has not been routinely offered to younger women, who give birth to the majority of children with Down syndrome.

Now, with a first-trimester sonogram and two blood tests, doctors can gauge whether a fetus has the extra 21st chromosome that causes Down syndrome with a high degree of accuracy and without endangering the pregnancy.

But many parents see expanded testing as a step toward a society where children like theirs would be unwelcome. The Newsweek columnist George F. Will labeled it a "search and destroy mission" for a category of citizens that includes his adult son, Jon Will.

Dr. Brian Skotko, a medical resident who has studied how mothers were told of prenatal diagnoses, found a high level of dissatisfaction. He said that most doctors have little or no training on how to relay a prenatal diagnosis of Down syndrome.

When he talked to obstetricians, geneticists and medical students at Massachusetts General Hospital in Boston about the subject last month, though, he was questioned sharply.

One doctor asked about studies suggesting there is a higher risk of early-onset Alzheimer's disease in people with Down syndrome, potentially saddling parents with another caretaking burden as they themselves age. Others take issue with the notion that they do not give parents a balanced portrayal of the condition.

"It's a mistake to say 'your baby is going to be mentally retarded, you should have a pregnancy termination,' "

said Dr. Allan Nadel, director of prenatal diagnosis at the hospital. "By the same token, I don't think it's quite fair to say 'these are wonderful lovely human beings, you can deal with all of their problems and it's not that big of a deal.' We strive to have the proper balance."

Parent advocates have some advice: don't begin with "I'm sorry," or "I have bad news," as many of their own doctors did.

Weeks after Patricia Lanter decided to continue her pregnancy, having learned that Down syndrome had been diagnosed in her fetus, her doctor reminded her that she could still get an abortion in Kansas if an ultrasound indicated the baby would need heart surgery. Ms. Lanter, an emergency physician from Norwich, Vt., has secured an invitation to lecture the obstetricians in her hospital this summer.

In Wilmington, Del., Kristin Pidgeon recalled her doctor's gloomy forecast for a local hospital audience: "She may be able to count change for the bus," he had said of her as-yet-unborn daughter. "But what's going to happen when the bus doesn't come?" (Her daughter Aliza, now 5, does not yet take the bus, Ms. Pidgeon said, but she does ride horses as part of her therapy.)

In the Detroit suburbs, Ms. Talbot is still working out the best strategy to drive her points home to medical professionals. When one doctor suggested she had chosen to show them only "high-functioning kids" like Sarah and her own daughter, Megan, she asked Trevor Taylor, who lacks the ability to communicate verbally, to join the lineup.

At the Henry Ford visit, Mr. Taylor, 19, a natural ham, acted out his speech as Megan, 18, read it, before hitting the music and signing along to "What a Wonderful World."

At the end, he blew a kiss to the audience. Then he hugged his mother.

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