

# Having a Perfect Child

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As a perinatologist, I have spent a considerable amount of my time counseling patients on the many issues and risks of delivering a child with Down syndrome. I have tested thousands of women utilizing screening blood tests, ultrasound examinations, chorionic villi sampling and amniocenteses to achieve or rule out a diagnosis of Down syndrome to provide accurate information needed when making a critical decision concerning continuation of pregnancy. In August of 2003, my newly born grandson, Seth, was to help me understand the issues I have been discussing with patients during the past 35 years from a personal perspective. Seth was born with Down syndrome.

Down syndrome was first described by Dr. John Langdon Down in 1866 as a condition in which a child is short in stature with mild-to-moderate mental retardation, as well as often being associated with other physical problems such as heart defects. The risk of having a child with Down syndrome increases as women age. At 20 years of age, the risk is ap-

proximately one in 2,000 births and at 35 it is one in 365. At age 40, the risk of delivering a child with Down syndrome is one in 100. Seth's mother was 26.

Over the past several decades, our specialty has increasingly focused on antenatal detection of structural, metabolic, and chromosomal anomalies, the most frequent and common of which is Down syndrome. We have steadily improved the sensitivity and specificity of screening all women to establish those who are at increased risk

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of having a child affected with Down syndrome. Using certain blood markers and ultrasound testing in the first and second trimesters, we are now more able to detect women who are at a significantly higher risk of delivering a child with Down syndrome than ever before. Because of this, obstetricians and maternal-fetal medicine specialists are spending a relatively large amount of time counseling patients on the results of these tests in an attempt to obtain appropriate informed consent for further testing and patient care.

During the past 3 years of being involved in my grandson's growth

and development, along with learning more about the lives of children with Down syndrome, I wonder if I, as well as many of my colleagues, have been giving patients appropriate counseling. I now worry that we may be perceived as being on a search and destroy mission, not giving patients adequate information on all the possibilities that exist for families and children with Down syndrome.

My son, Tommy, and his wife, Lisa, were expecting twins, a girl and a boy. I vividly remember the excitement as my wife and I waited during the delivery and cried with joy as we each held a child in our arms in the recovery room. Marly and Seth were perfect. What dreams and expectations we held for these two precious bundles of joy.

Several hours later, however, we were confronted with the fact that Seth carried a diagnosis of Down syndrome while his sister Marly did not. Our family was filled with emotions, from the high that came with the birth of twins to a low at learning of Seth's disability and knowing that he would be unique and have certain anticipated learning and physical challenges.

Later that evening, holding Seth in my arms and gazing into his angelic face, I was overcome with unconditional love for my grandson. As the tears rolled down my cheek, I understood that, despite his diagnosis, Seth would be loved and nurtured, the same as his sister Marly. Our dreams and expecta-

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tions for him may now be different from those for his sister, yet they are dreams and expectations nonetheless.

To me, Seth is perfect. His smile lights up a room and his laughter brings warmth to all who are near. He loves to cuddle and gaze into the eyes of those who hold him and he embraces his sister with what can only be described as pure affection and love. Seth often walks around the house singing songs he has recently learned and smiling at those who enter the room. He is clearly behind in development when compared with his sister, but he is learning the alphabet and how to speak and his desire to learn is intense.

I have attended a number of meetings which help parents understand what to expect when raising a child such as Seth. I have met many young adults who have learned to cope with their disability and have been overwhelmed with how well some do in a world that generally looks negatively at those in society who develop atypically. I have read about many children and adults with Down syndrome who have excelled in the real world of sports, music, and the arts. I have seen heartache but I have also seen joy and love. It has been an eye-opening experience for me, and it has made me a more sensitive and knowledgeable person and physician.

Over the many years of practicing obstetrics, I have attempted to answer the many questions that each patient invariably wants answered. While these questions are often specific to each patient's pregnancy, one common question usually surfaces at some point in our discussion: "Is my baby OK?"

Because it is so difficult for me to define "OK," this question has always been a tough one for me to answer. At the heart of this question lies a pregnant patient's desire to know if her unborn child will be perfect (normal and healthy), a very reasonable concern. Once again, however, defining perfect is not an easy task.

Most pregnant women do not realize that approximately 2% of the 4 million births each year in this country involve the delivery of a child with a significant congenital anomaly. These birth defects include hundreds of different types of anomalies such as neural tube defects, cardiac and other organ abnormalities, as well as Down syndrome. When one considers the fact that each of us begins with the union of sperm and egg to create one cell and this one cell continuously divides to create the billions of cells that result in a living child, it is truly remarkable that any of us are born, much less that we are born without congenital anomalies.

It is with this in mind that I explain to patients that each birth is a miracle and that the uniqueness of each of us is what makes us perfect. I also attempt to explain that normal is in the eye of the beholder. The gift of life can be "perfect" even in the presence of serious problems.

Since Seth's birth, I am even more aware of the need to properly and thoroughly counsel patients on the full extent of what it means to begin the process of screening and testing for Down syndrome, as well as what to expect if the screening process is begun and a diagnosis of fetal Down syndrome is made.

In Nashville, when told of available tests to assess risk for Down

syndrome, many of our patients tell us that they would not undergo abortion if a diagnosis of Down syndrome was confirmed based on their moral and religious convictions and therefore decline testing. Others are not sure how they would respond and seek from us proper counseling on the risk/benefit ratio of screening tests and invasive confirmatory tests such as chorionic villi sampling or amniocentesis. It is not for the physician to tell a patient how to proceed; however, I now understand even more clearly that it is imperative that the counseling of those who wish to be screened and tested include all aspects of raising a child with Down syndrome, not just the negative ones.

When counseling a patient who is making a decision of whether to be tested or to continue a pregnancy, it is important to mention that many children with Down syndrome, especially those without other significant anomalies, can be a loving addition to a family and live a productive life that can be long as well as rewarding.

Seth is one of many children who are born with congenital anomalies and complications, yet like so many others, Seth has embarked on a journey of life. That life will be filled with challenges for him and his family, yet that is also true of each of us as we embrace life with its ups and downs.

As we enter a world in which more genetic information will be available for us to consider in selecting a "perfect" child, I hope we have room in our world and hearts for those like Seth who are challenged and different, because challenged and different can still be perfect.