

## Family-provider interactions surrounding the diagnosis of Down syndrome

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During the past 40 years, much has been written about parental satisfaction with family-provider interactions surrounding the diagnosis of Down syndrome (DS).<sup>1–14</sup> In addition, there have been numerous articles about how to break bad or difficult news to patients and families, with the unexpected diagnosis of DS frequently being used as an exemplar.<sup>15–25</sup> Also, a number of educational programs have been developed to help healthcare providers feel better prepared to deliver the diagnosis of DS.<sup>26–28</sup> Additionally, parents of children with DS throughout the world have devoted a great deal of time, effort, and money to the development of educational resources (e.g., books, pamphlets, and videos) that can be used by healthcare providers to inform expectant or new parents about life with DS in the 21st century ([http://www.ds-health.com/ds\\_sites.htm](http://www.ds-health.com/ds_sites.htm) for links to resources). Furthermore, the Prenatally and Postnatally Diagnosed Conditions Awareness Act (S 1810) was signed into law on October 8, 2008, and became Public Law No: 110-374.<sup>29</sup> One of the main goals of this legislation was to improve the quality of information and support services that pregnant women and new parents receive about DS and other prenatally and postnatally diagnosed conditions.

Unfortunately, despite all of this attention being paid to family-provider interactions surrounding the diagnosis of DS, reports of parental dissatisfaction with the informing process continue to appear in the popular literature, as do reports of healthcare providers giving parents inaccurate, out-dated information about life with DS. Moreover, anecdotal reports of parents feeling pushed or coerced to make unwanted choices, such as undergoing invasive testing or terminating a pregnancy after the diagnosis of DS, are becoming more common.<sup>30–33</sup> During a session on prenatal screening at the 10th World Down syndrome Congress in Ireland in 2009, many parents expressed concern that although advances in genomics may contribute to improved health and increased life span for individuals with DS, advances in genomics may also result in decreased support for individuals with DS and their families. A number of parents noted that once prenatal testing became a routine part of prenatal care in their country, there seemed to be a growing sentiment among healthcare providers that families who chose to continue a pregnancy after a prenatal diagnosis of DS are making the wrong choice. More importantly, because it is “their choice” to have a child with DS, questions are being raised about their government’s obligation to provide resources and support.

At the heart of most arguments concerning termination as the “right choice” after a prenatal diagnosis of DS is the underlying assumption that individuals with DS have a negative impact on their family, their community, and society as a whole. Unfortunately, this is a widely held assumption, despite growing evidence that many families adapt successfully to the challenges associated with raising a child with DS and some even thrive.<sup>34–44</sup> The continued existence of this assumption became very apparent during recent media coverage of a large-scale study by Chiu et al.<sup>45</sup> demonstrating the clinical efficacy and practical feasibility of using multiplexed maternal plasma DNA sequencing analysis to screen for DS among high-risk pregnancies clinically indicated for amniocentesis or chorionic villus sampling. In most of the media coverage, the new test was presented as a way to prevent the unwanted birth of a child with DS without endangering the life of a “normal child.”<sup>46</sup> Very little, if any, attention was paid to educating people about what life is like for individuals with DS and their families.

The main purpose of this commentary is to advocate for the inclusion of accurate, up-to-date information about the family experience of living with DS in early discussions with expectant or new families after the diagnosis of DS. Preliminary findings from an ongoing study concerning adaptation and resilience in families of children with DS being conducted by the authors of this commentary suggest that although parental satisfaction with the informing process continues to be less than ideal, many parents of children with DS are satisfied with how they were informed, especially those who were given information about what life is like for children with DS and their families.<sup>47</sup>

Of the 224 mothers who completed the online survey, 52% were satisfied with how they were informed of their child’s diagnosis and 48% were dissatisfied. Twenty-three percent of the mothers who were satisfied were very satisfied, 22% were moderately satisfied, and 7% were slightly satisfied. Of those who were dissatisfied with how they were informed, 25% were very dissatisfied, 12% were moderately dissatisfied, and 11% were slightly dissatisfied. The 60 fathers who completed the survey responded in a similar manner, 50% were satisfied with how they were informed (25% very satisfied, 18% moderately satisfied, and 7% slightly satisfied) and 50% were dissatisfied (23 very dissatisfied, 17% moderately dissatisfied, and 10% slightly dissatisfied).

Given the amount of attention that has been devoted to improving parental satisfaction with the informing process, one would hope to see improvement in parental satisfaction over time. However, for the current sample of parents, this was not the case. The relationship between age of child with DS and level of satisfaction was not significant. Parents of younger children with DS did not report higher levels of satisfaction than parents of older children with DS. Instead, levels of satisfaction remained fairly constant over the past 20 years, with approximately one half of the parents being satisfied and the other half being dissatisfied. Also, in terms of when parents first became aware of their child’s diagnosis (prenatally or postnatally), the relationship between timing of the diagnosis and satisfaction with how they were informed was not significant.

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A review of parental responses to two open-ended questions on the survey (i.e., please indicate at least one thing that healthcare providers did or said that you found helpful and please identify at least one thing that you would have liked healthcare providers to have done differently) revealed a fairly close match between parent preferences and recent guidelines concerning how best to inform parents of their child's diagnosis of DS.<sup>21,22</sup> Parents wanted to be told together in a joint meeting with a healthcare provider they knew and trusted. They wanted the conversation to take place in a private setting as soon as possible after the diagnosis of DS was suspected. They wanted providers to be upfront, honest, and forthcoming with accurate, up-to-date information. If the diagnosis of DS was made postnatally, parents wanted to have their child with them during the informing process.

Things parents found to be most helpful were (1) unbiased attitude of providers (e.g., presented balanced information and did not try to influence parental decisions), (2) positive comments about the child and what the child is likely to accomplish, (3) emotional support (e.g., caring, sensitivity, and hope), (4) material support (e.g., provision of up-to-date resources and information about local support groups), and (5) the provision of information in a timely manner (e.g., not too soon and but shortly after the diagnosis was suspected). Parents clearly appreciated being informed in a caring, supportive manner. According to one parent,

*"The afternoon he was born we spoke to a geneticist, who was the most warm and compassionate doctor I have ever met. He told us that we did all of the right things (it wasn't our fault) and that he would be more like other kids than not. I'm not sure exactly what he said, but after we talked with him, we knew our lives would be different but everything was going to be all right."*

Parental comments regarding what they would have liked healthcare providers to have done differently clearly reflect use of a less than desired approach by some healthcare providers. That is, the approach followed by some healthcare providers varied greatly from the approach recommended in recent guidelines concerning the best way to inform parents of a DS diagnosis. A mother who was felt pushed to make unwanted choices wrote,

*"When we were given our high-risk ratio of having a baby with DS based on the first trimester screening, the OB emphasized all the negatives of a child with DS and seemed to be encouraging a termination despite our clearly stated commitment to having the baby regardless of diagnostic outcome. Eventually after we persisted he began to acknowledge that 'some people choose to have babies like this in situations like these'."*

One parent said, "they need to present the information gently and personally—not a sterile statement void of any human emotion." Another indicated that she wished providers had not acted as if they were delivering "bad" news. A father wrote, "I wish we had known right from the start what a positive experience we could have with a child who has Down syndrome."

One possible explanation for why parents of children with DS may not receive accurate, up-to-date information about the family experience of living with DS at the time of diagnosis is that most of the existing guidelines for how to deliver the diagnosis of DS do not specifically address this issue. Rather, the focus is on giving parents "the facts" about DS and health-related conditions associated with DS. Little, if any, attention is paid to how individual family members or the family as a whole respond to the experience

of raising a child with DS. This is unfortunate, because as noted previously, there is growing evidence that negative consequences are not inevitable. In fact, many individuals and families not only adapt but also they thrive after the birth of a child with DS.<sup>34–42</sup> In the first author's ongoing program of research concerning adaptation and resilience in families of children with DS, many participants (parents and siblings) indicated that although there were ongoing challenges associated with having a family member with DS, the positive consequences far outweighed the negative.<sup>13–14,39–44</sup>

Another possible explanation for why parents of children with DS may not receive accurate, up-to-date information about the family experience of living with DS at the time of diagnosis is that the healthcare providers who deliver the diagnosis of DS are typically not the same healthcare providers who provide ongoing care to children with DS and their families. Because of this, their understanding of the family experience of living with DS may be very limited; it may be based primarily on the interactions healthcare providers have had with expectant or new parents shortly after a diagnosis of DS has been confirmed. This is problematic because in most families of children with DS, views of the situation change dramatically overtime. More specifically, although many parents may initially view the birth of a child with DS as a tragedy or the worst thing that could have happened, most parents eventually develop a much more positive view. One parent noted,

*"It is really not quite the tragedy. . . At the time you really feel that this is the biggest tragedy that ever happened. If we could have known what it would be like to have M., we wouldn't have been nearly so sad. No one really mentioned the positive side."<sup>14</sup>*

Another parent discussed the positive consequences of having a child with DS,

*"Our entire family and marriage is stronger. It has changed our view of the world, our view of ourselves, and others. It has made use more giving and less selfish. It has drawn us closer to God. It has caused us to be more concerned about others who are different. It has shown us what we value in life—relationships—not power and wealth. It has made us more content to just be!"<sup>39</sup>*

As far as the 284 parents who completed the online survey, current views of the situation were much more positive than initial views. Also, mothers tended to have more positive views than fathers, both initially and at the time they completed the survey. In response to the question, "In general, which of the following best describes how you currently think about the fact that your child has Down syndrome?" 60% chose Blessing in disguise (64% mothers and 46% fathers), 18% chose Challenge to be overcome (15% mothers and 25% fathers), 22% chose Just something to accept (21% mothers and 27% fathers), and one father chose The worst thing that could have happened. For the question, "Think back to what you thought when you first learned of your child's diagnosis of Down syndrome," 7% of the parents chose Blessing in disguise (9% mothers and 0% fathers), 28% chose Challenge to be overcome (28% mothers and 29% fathers), 21% chose Just something to accept (18% mothers and 31% fathers), 24% chose A Tragedy (24% mothers and 25% fathers), and 20% chose The worst thing that could have happened (21% mothers and 15% fathers).

In conclusion, preliminary findings from an ongoing study about resilience and adaptation in families of children with DS suggest that parental preferences concerning the informing pro-

cess match fairly well with current guidelines concerning the best way to inform parents of a DS diagnosis.<sup>47</sup> However, these findings also suggest that many healthcare providers are not following the recommended guidelines. In fact, some seem to be using an approach similar to the approach used by healthcare providers decades ago, when negative consequences were thought to be inevitable after the birth of a child with DS. There is currently a critical need for ongoing education with healthcare providers regarding the best approach to use when informing expectant and new parents of a DS diagnosis. In addition, existing guidelines need to be expanded to include accurate, up-to-date information about the family experience of living with DS. Not only will these efforts help to increase parental satisfaction with the informing process to a more acceptable level, these efforts should help to decrease parental uncertainty. More importantly, these efforts are likely to give parents of children with DS a renewed sense of hope, something that may serve parents well as they deal with the ongoing challenges associated with raising a child with DS.

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