

What I Wish I Knew Then...Reflections from Personal Experiences in Counseling about Down Syndrome

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Received: 17 April 2007 / Accepted: 2 July 2007
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Abstract Sharing the news about a newborn baby's diagnosis of Down syndrome with families is a scenario genetic counselors frequently face. Yet often we may feel uncomfortable or unsure how to best support families in this setting in a way that will foster competence and resilience. This commentary is a reflection of one genetic counselor's experiences in counseling about Down syndrome over the course of her career and how her thinking has transitioned from a medical based model of disability to a more individual and family-focused model. Ideas and suggestions are offered that genetic counselors can incorporate into their practice.

Keywords Down syndrome · Unexpected news · Genetic counseling

Case 1: Patient: I'm not really sure I know what Down syndrome is. Can you tell me a little bit more about it?

Genetic Counselor: *Down syndrome results from a chromosomal abnormality where a fetus has an extra copy of chromosome 21. It causes heart defects, gastrointestinal abnormalities, and mental retardation. Most children with Down syndrome have a typical facial appearance and have low muscle tone. Children with Down syndrome often need therapies and attend special classes in school. Many work in a sheltered workshop setting and most are unable to live independently as adults.*

Case 2: Patient: I've never known anyone with Down syndrome. Can you tell me more about it?

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Genetic Counselor: *Down syndrome is a genetic condition due to an extra copy of chromosome 21. The extra chromosome causes some differences in how babies with Down syndrome grow and develop. While Down syndrome is a lifelong condition, with good medical care and attention to areas where they may need some extra help, children with Down syndrome can thrive and grow into happy, healthy, and productive adults. Families who have a child with Down syndrome are typically no different from other families. Children often attend school in an inclusive classroom and participate in a variety of activities in the community with their typically developing peers. They are more like other children than different.*

Introduction

It is interesting how one's beliefs and perspectives change over time. When I first went into practice almost 20 years ago, I was the genetic counselor in Case 1. I thought as many health professionals did, that the birth of a child with Down syndrome was a devastating and mostly tragic event. I believed the theory that families experienced chronic sorrow over the perceived child they had "lost". I bought into the medical model of disability that with enough therapy and the right services provided by the 'system', we could "fix" the child and make them more "normal". My graduate training did not dissuade me from nor challenge me on these views. Even now I'm not really sure why, but just out of graduate school, I started attending meetings of our local Down syndrome support group. I remember the first meeting I went to... one parent came up to me and said accusingly, "What are you doing here? You're a genetic counselor!!" as if all we genetic counselors did was perform prenatal diagnosis and terminate pregnancies of babies with Down syndrome. But somehow I was drawn to the babies

and children with Down syndrome—their charm captivated me and I found I had to totally change what I thought I knew about people with Down syndrome and about their families.

There are articles written about the best ways to give unexpected news (Dent and Carey 2006; Skotko 2005; Krahn *et al.* 1993; Buckman 1992; Sharp *et al.* 1992; Cooley and Graham 1991). There are accounts written by families about the positives and the joys their child with Down syndrome has brought into their lives (Boehm 2007; Beck 1999; Berube 1996; Marsh and Boggins 1995; Meyer 1995). There are studies showing that marriages can and do withstand the challenges of having a child with a disability diagnosis (van Riper *et al.* 1992; Feingold 1996; Hirst 1991; Dyson 1991) and that brothers and sisters actually experience positive benefits from having a sibling with special needs (Skotko and Levine 2006; van Riper 2003). Yet still our society tends to cling to the old beliefs that the birth of a child with Down syndrome is a sad event, that families never really get over this experience, that their lives are now somehow less full, that marriages suffer, and siblings are neglected. There's a perception that adults with Down syndrome are perpetual children, that they can never live independently, that they will not work a real job, have real relationships, or experience quality of life. *In my experience, I have found none of these old beliefs to be universally true.* How can we, as genetic counselors, address these old ideas and beliefs? One way may be to start at the beginning... I truly believe that the way the diagnosis of Down syndrome is presented to families during those critical first days sets the stage for how the family will ultimately cope and adjust and that genetic counselors are in a unique position to help families move through those first fragile and shaky days and get on with their lives.

Giving Unexpected News

The literature often talks about the process of giving “bad” or “difficult” news. I'm not fond of the term “bad” news because that implies a value judgment on the information given. And difficult for whom? The family... or the giver of the news? As genetic counselors, I'm sure many of us have had families whose initial reaction to their new baby with Down syndrome has been one of instant love and acceptance thus those descriptors are not universally relevant. However, I do think it is fair to say that the news is often “unexpected” and I feel this may be a more appropriate descriptor. Many of the articles in the literature share helpful and practical information from families' experiences that genetic counselors have incorporated into their practice. In addition to those recommendations, I would like to add a few more from my own experience:

- 1) When you first walk in the room, *tell the family “congratulations!”* on the birth of their new baby. They will hear “I'm sorry” often enough from others but let them remember you were one of the few who actually congratulated them. “I'm sorry” tells them the birth of their child is indeed a tragedy whereas “congratulations” reminds them this is their new baby after all, whose birth is to be celebrated.
- 2) Through your discussions with the family, help them *put the diagnosis in proper perspective.* This child will be more LIKE other children than different. The features we see of Down syndrome are only a small part of the whole person who has their own unique personality, capabilities, and gifts. Help families focus on what their child CAN do, not what they cannot. Sure, there are some things that will need to be done differently over the years (Van Cleve and Cohen 2006; Van Cleve *et al.* 2006; American Academy of Pediatrics 2001) but help families relegate the diagnosis to the level of importance it deserves, not overblown out of proportion. Families will soon realize their experience is not so different.
- 3) *Reassure families they have the skills to raise this child.* Remind families that their baby is a baby first... he needs the same things as any other baby—love, food, sleep, lots of cuddles. You can often see the relief in their faces when they hear they don't have to learn a whole new set of parenting skills, that what they know already is sufficient and indeed valuable. There is no handbook or manual on how to raise a child with Down syndrome. You just do it, just like with any other child. As parents get to know their precious new baby, they will instinctively know what their baby needs, just like other moms and dads do. As with most children, parents may make some mistakes along the way but, for the most part, all will come out OK in the end. Let them know you will be there for them, if needed, for questions or guidance or if they just need to talk.
- 4) Remind them to *have expectations for this child.* If you had a child who was capable of making straight A's, you wouldn't be satisfied if they came home with C's. Same for the child with Down syndrome, don't settle for “average” if they are truly capable of more. You will be surprised how many children meet those expectations and more. Let them know this child will follow the same developmental progression as any other child... they will walk, talk, run, play, get in trouble, annoy their siblings—all the typical kid things!
- 5) Emphasize the *importance of a balanced family life*—each family member is important and has something to contribute—don't put all the focus, finances, and energies on the one member who has a label. Families often feel overwhelmed by the obligations of weekly therapy

appointments, doctor's visits, and case workers. It can seem to families like everyone else knows what is best for their child and that their input and insights are unwelcome. But you can reassure them that **THEY** are the real experts on their child and to trust their gut feelings about what is right. Give them permission to say "no" when such obligations intrude on the family's well-being. Now I don't mean for families to not follow through on important and necessary medical follow-up. The Down Syndrome Health Care Guidelines have been written to ensure optimal health and developmental potential (American Academy of Pediatrics 2001; Cohen 1999). But as an example, early intervention doesn't have to be structured therapy or provided through the "system". You can encourage families to be creative and look for other inclusive opportunities in the community that can address motor, language and social skills—Mommy & Me, Gymboree, Kindermusik, infant swim classes at the YMCA, or dance. Sports participation can be in the community leagues, separate activities such as Special Olympics are not the only option. In addition, encourage families to be in collaborative relationships with the professionals in their child's life. Our goal is a "resilient family" (Rolland and Walsh 2006; King *et al.* 2006; Walsh 2003).

- 6) *Families need us to be a real person* in conversation with them about what it means to have a child with Down syndrome. We don't have to sugarcoat what the experience will bring but we also don't have to gloss over or ignore the real positives. Be a sounding board and a real source of support as parents work through the myriad of feelings and emotions about their new baby. Resilient families are able to "make meaning out of adversity" (Walsh 2003). They also maintain connectedness with their families and community as well as seek connections with families in similar situations. Often when you initially offer a connection with a local family after the baby is born, that offer is declined, but later that same family will say they wish they had had the opportunity to talk with a veteran family in those early days. They forget they were so wrapped up in their own feelings and reactions at the time that they just could not see how important talking with another family and sharing experiences would be. But it's just those personal connections that make all the difference for many families in negotiating this new territory. Encourage them to allow you to have another family visit them in the hospital or give them a call. If they decline initially, keep offering at later conversations. When the time is right, they will agree. Have the experienced parent be the one to make that first call... they know how to begin the conversation in a way that will put the new parent at ease, rather than the

awkwardness of the new parent having to dial up a complete stranger. And finally, make sure families have current written information about Down syndrome on hand when they are ready to start learning more. Many Down syndrome parent groups will provide such welcome packets at no charge.

- 7) If a family leads the discussion into a more spiritual direction, often I find the conversation turns toward the theme of *the miracle you ask for may not necessarily be the miracle you receive*. As one perinatologist states, "each birth is a miracle and the uniqueness of each of us is what makes us perfect. The gift of life can be 'perfect' even in the presence of serious problems" (Boehm 2007). As a new genetic counselor, I was often unsure how to proceed when families turned the discussion towards religion and spiritual issues. I'm still not sure I'm very accomplished in this realm, but I've learned to be more comfortable with these discussions and I've found that this theme works well for me. I've had the good fortune of having one of our hospital chaplains (who also happens to be the mother of a young boy with Down syndrome) partner with me in discussions with families who were having a difficult time resolving their spiritual beliefs with the birth of their baby with Down syndrome. Your hospital chaplains are an underutilized resource! It is interesting that it is in these discussions with families, where we are talking about how to find meaning in the birth of this child with Down syndrome, where I feel I am making the most difference with my families. This is the heart of genetic counseling to me, not the review of chromosomes and recurrence risks and features, but the relationship between human beings and the discussion of what it means to be human.

The reality is that families can and do thrive with a child with Down syndrome. Many families describe having a child with Down syndrome as a life changing event, but it is *not a life they would change!* They tell me later they feel guilty that they were so sad at the birth of their child; that their child is a joy to them and they would not change him or her now. They have come to learn what is really important in life, lessons they probably would not have learned if not for their child with Down syndrome: the strength of love, the importance of family, an appreciation for what is inside a person, that IQ or the acquisition of material things are not the only measures of success or happiness.

Down Syndrome Now

From getting to know many people with Down syndrome, I've learned they are not so different at all, they want what

we all want: love, acceptance, respect, dignity. Adults with Down syndrome are achieving more than ever before. With a lifespan now reaching into the 60's, people with Down syndrome are dreaming big dreams. More and more are finishing high school and moving on to secondary education. They are moving out of the sheltered workshops and working real jobs for real pay. People with Down syndrome take real pride in their work and in their paycheck! They are moving out of group homes where they have no real choice about who they live with or what they do with their days and moving into supervised apartments or even purchasing homes. They are participating in the decisions that affect their lives and voicing opinions on what they want and don't want. They have friends, boyfriends, and girlfriends and even get married. They enjoy music, movies, reading, sports, hobbies, and church. They enjoy volunteering and having the opportunity to give back to others. They want and enjoy the same things as you and I. *What make their lives difficult are the attitudes of others, not Down syndrome!*

My graduate training taught me to maintain a "professional distance". But the most rewarding and meaningful aspect of my work has been the relationships I've developed working *together* with families and people with Down syndrome over the years. Through them sharing their lives and experiences with me, I have come to understand a very different picture of what Down syndrome is than what I was initially taught. Now I am the genetic counselor in Case 2. I am more aware of the true joys AND the real challenges of having a child with Down syndrome and of their inherent capabilities and possibilities. I am thankful they were willing to teach me as well. It is because of them I hope I am a better genetic counselor.

Acknowledgments Appreciation goes to Kathy Peters, MS, CGC, for title suggestions and to Allyn McConkie-Rosell, PhD, CGC, for thoughtful discussions on this topic and encouragement to write up these experiences. Many thanks also to Ann Martin, MS, CGC and Anne Bandholz, MS, CGC for critical reading of the manuscript. Reviewer comments on a previous version of this manuscript were most helpful.

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