J Genet Counsel (2012) 21:35–44
DOI 10.1007/s10897-011-9417-2

ORIGINAL RESEARCH

What is a "Balanced" Description? Insight from Parents of Individuals with Down Syndrome

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Received: 12 April 2011 / Accepted: 23 September 2011 / Published online: 20 December 2011

Abstract Genetic counselors and parents of individuals with Down syndrome (DS) agree that descriptions of DS in prenatal settings should be "balanced." However, there is no consensus regarding what constitutes a balanced description of DS. A survey was designed in collaboration with, and sent to the membership of, the British Columbia based Lower Mainland Down Syndrome Society (N=260). Respondents were asked how they would describe DS to a couple who have just received a prenatal diagnosis of the condition. We rated the descriptions provided for positivity/negativity. Completed surveys were returned by 101 members, the majority of whom were Caucasian (87%) and female (79%). Participants' descriptions of DS ranged from entirely positive (n=5; 10%) to entirely negative (n=4; 7%) in nature. Deriving a description of DS that would broadly be perceived as "balanced" may be impossible. Instead, it may be more important to explore the range of possibilities regarding the family experience of raising a child with DS using nonjudgmental terminology, and to help families evaluate these possibilities in the context of their own values, coping strategies, and support networks.

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Keywords Down syndrome · Balanced description · Balanced information · Parents · Opinions · Prenatal screening · Prenatal diagnosis · Genetic counseling

Introduction

> "Balance—when referring to something as variable and experientially related as 'life with a disability'—is an elusive commodity" Ahmed et al. (2007, p. 319).

Genetic counseling for Down syndrome (DS) was available prior to the advent of prenatal screening tests for DS (Weil 2003). But, as the availability of, and options for, prenatal testing for DS have increased, facilitating informed decision-making related to which testing option is optimal for an individual patient has been acknowledged as a critical goal of clinical encounters such as genetic counseling sessions (Hall et al. 2007). The importance of this issue is recognized in practice guidelines of professional bodies such as the American Congress of Obstetricians and Gynecologists (ACOG) (2007) and the Society of Obstetricians and Gynaecologists of Canada (SOGC) (2007). However, the SOGC guidelines do not specify the nature of the information that should be provided, and neither guideline mentions the importance of describing the conditions for which testing is offered, or the expectations for the quality of life of a family with a member with the condition in question. Additionally, there is no guidance provided as to how a condition such as DS should be described to prospective parents, nor is there elaboration about how to facilitate informed decision-making. It is perhaps unsurprising, then, that in a survey of 141 mothers who received a prenatal diagnosis of DS, a majority reported they had not received an explanation of DS at the time of screening (Skotko 2005).

Many studies have investigated the nature of the information that should be provided by health care professionals at the time of a diagnosis of DS. The majority have focused on recommendations for the delivery of a postnatal diagnosis of DS (Cooley 1993; Skotko 2005; Skotko et al. 2009), but three studies investigated parental opinions of what should be included in the context of a prenatal diagnosis of DS (Helm et al. 1998; Sheets et al. 2011a; Skotko 2005). A systematic review of 19 articles published between 1960 and 2008 relating to what parents felt should be included in a description of DS, found that a desire for balanced information was one of three major wishes expressed (Skotko et al. 2009). Provision of "balanced" information about DS has emerged consistently as a desirable goal, and providing balanced information has been described as a core element of genetic counseling practice (Weil 2003). Indeed, a recently published NSGC Practice Guideline regarding the provision of genetic counseling following a diagnosis of DS (Sheets et al. 2011b) states that it was written: "…to ensure that families are consistently given up-to-date and balanced information about the condition, delivered in a supportive and respectful manner". However, it is also frequently acknowledged that there is no clear consensus on what constitutes "balanced" information (Madeo et al. 2011; Sheets et al. 2011a; Williams et al. 2002). As Madeo et al. (2011) stated, "Standards of what constitute "unbiased" or "balanced" information do not exist" (p. 1779).

Defining a "Balanced" Description[1]

The first attempt to define "balanced" information about DS (of which we are aware) came from Bryant et al. (2001) who used the term in their content analysis of serum screening leaflets offered to women prior to prenatal screening in the UK. Examples of information the authors classified as "negative" included either content they judged to be negative (e.g., the prevalence of heart defects in DS) or the manner in which the information was presented [such as "emphasizing infant mortality rates rather than survival rates" (p. 1058)]. Examples of information the authors classified as "positive" included information related to treatments or educational strategies that have led to improved outcomes or the increasing life expectancy among individuals with DS. The authors concluded that information provided about DS should be "balanced"; by containing statements that are positive, negative, and neutral in content as well as tone. Soon thereafter, Williams et al. (2002) found that 70 healthcare professionals who regularly communicate about DS reported minimal personal experience/contact with individuals with DS in either their personal life or their professional training, and their lack of experience resulted in a tendency to follow a "checklist" approach when providing a description of DS, rather than a more subtle approach incorporating experience or stories. It follows from their study that a "balanced" description of DS should include both potential medical issues and information related to quality of life.

More recently, Ahmed et al. (2007) developed and evaluated a patient-friendly web resource: The Antenatal Screening Web Resource (AnSWeR). The stated goal of AnSWeR was to provide a counter-balance against the surplus of available "medico-clinical" information [perceived to be negative by authors such as Bryant et al. (2001) and Williams et al. (2002)] with case stories of families with members who have DS, and incorporating feedback from parents of individuals with DS. In an evaluation of whether users of the resource felt it to be "balanced," some participants (mostly health care professionals) felt that because there was less information about pregnancy termination as compared to information about raising a child with DS, the resource implied judgment that continuing a pregnancy was preferable to terminating. Some participants (mostly health care professionals) felt that AnSWeR painted a picture of DS that was too positive, while others (mostly mothers of children with DS) did not share this opinion. There was also a discrepancy between the health care professionals and parents when it came to opinions about including photographs of people with DS. Health care professionals tended to view them as "cute" and positive, while the parents tended to see them as "scary" and negative. The authors suggested one potential method for operationalizing "balance" when providing information would be to define it as "an equal proportion of 'negative' and 'positive' facts about the condition" (p. 319). They concluded, however, that the task of conveying balanced information about DS is very challenging given the wide diversity in participant perceptions about which information on AnSWeR was positive versus negative. Because of the divergent perspectives of different groups on the nature of positive and negative information about DS, it is critical to document the perceptions of "balanced" information of those to whom healthcare professionals' descriptions of DS are most important—parents of individuals with DS.

There have been recent efforts to solicit the input of parents of individuals with DS about how the condition should be described—once in the development of interactive educational material about DS for medical residents

[1] Previous research studies have not always distinguished between "balanced information" and a "balanced description," and have used the two terms interchangeably. In order to allow a more nuanced discussion of the nature of "balance," we conceptualized the term "information" as relating to the facts that are provided. While facts may seem to be neutral in and of themselves, the context and manner in which facts are communicated or articulated, in other words, the description, can be perceived to fundamentally alter this neutrality.

(Kleinert et al. 2009), and later in the collaborative development of a patient resource "Understanding a Down Syndrome Diagnosis" (Down Syndrome Consensus Group 2011). The booklet developed by the Down Syndrome Consensus Group took into consideration input from the organizations ACOG, the National Society of Genetic Counselors (NSGC), the American College of Medical Genetics (ACMG), the National Down Syndrome Congress (NDSC), and the National Down Syndrome Society (NDSS). This booklet includes information about the range of potential medical complications, as well as what families can expect in terms of educational outcomes and the impact of raising a child with DS on the family. In spite of these valuable advances, there is still uncertainty as to the definition of a "balanced" description, and, to our knowledge, parents of individuals with DS have not previously been asked to provide a "balanced" description of DS in their own words.

Purpose of the Present Study

The purpose of this study was to explore how parents of individuals with DS would describe DS in a "balanced" manner. The ultimate goal of this study was to generate insights to allow genetic counselors to better serve individuals whose children/pregnancies have received a diagnosis of DS.

Methods

Sample and Procedures

The survey was distributed to all members of the LMDSS (N=260) by mail at two time points: December 2008, and January 2009, and a reminder letter was sent in follow up. Members were considered eligible if they were a parent or step-parent of an individual with DS. Respondents completed the survey anonymously; consent was implied by return of a completed survey. One hundred one members returned completed surveys. Four surveys were returned to sender due to incorrect information, four surveys were returned incomplete from ineligible members (social worker, infant development program worker, grandparents, community connections program worker), and six members called to say they were not eligible. It is unknown how many of the remaining 246 potentially eligible LMDSS members were actually eligible, but a conservative assumption that all 246 were eligible, yields an estimated response rate of 41% [a typical response rate for surveys of this kind (Kaplowitz et al. 2004)]. The Institutional Review Boards (IRBs) of the University of British Columbia and BC Women's Hospital approved this study (IRB #H08-01118; CW08-0178).

Measures

In collaboration with board members of the British Columbia based Lower Mainland Down Syndrome Society (LMDSS—the largest provincial non-profit organization that provides support, resources and information for individuals with DS and their families), we developed a self-report survey composed of 26 questions (both open and closed ended) designed to address the study objective and to obtain demographic information, including information about the degree to which a respondent's child with DS was affected (e.g., number and nature of medical problems reported, and whether the child worked or continued on to further education). We piloted a draft of the survey by asking LMDSS board members (n=11) to complete it and incorporated their feedback by changing wording, including more open-ended questions and space for responses, and providing an example scenario (written in collaboration with LMDSS board members) to provide more context for the survey.

We included 13 demographic questions to obtain information regarding both the participant and the participant's child with DS: age, sex, ethnicity, religious affiliation, and the degree to which children with DS were affected (as detailed above). Rather than simply asking parents how they would describe DS, we first provided them with the following scenario and example of how DS might be described:

Scenario

A 32 year old pregnant woman decides to have prenatal screening for Down syndrome. After discussing it with her doctor and her partner, she chooses the triple marker screen blood test. The result of her test is a "screen positive for Down syndrome." The chance that the child has Down syndrome is 1/258. She and her partner are referred for Genetic Counselling to find out more. The Genetic Counsellor provides the couple with information about their screening test results and about their options for diagnostic testing, and explains that the decision about whether or not to have a diagnostic test is theirs. The couple decides to have a diagnostic test (amniocentesis) to find out for sure whether or not their child has Down syndrome because they are very anxious about not knowing. The diagnostic test shows that their baby has Down syndrome. They return to talk to the Genetic Counsellor, who tries to provide a balanced view of Down syndrome. She explains that individuals with Down syndrome can be healthy, but do have some degree of intellectual disability, and sometimes there are health concerns, like heart problems, but that individuals with Down syndrome can often work in a supported environment. The Counsellor provides them

with an information package from the LMDSS, and asks whether they might like to speak with parents of a child with Down syndrome. She also explains that it is the couples' decision whether they choose to continue with, or end the pregnancy, but that she will do her best to make every effort to support them in their decision making.

This scenario was followed by an open-ended question: "What do you think should be included in an appropriate, balanced, description of Down syndrome? (For example: How would you describe Down syndrome to the couple in the scenario who have just found out that their unborn child has Down syndrome?)".

Data Analysis

First, descriptions of DS that were provided by parents were reviewed by the authors for their degree of positivity and negativity. Specifically, guided by Ahmed et al. (2007)'s possible definition of "balance" as an equal proportion of positive and negative facts about the condition, we devised a 5-point scale (1 = entirely negative, 2 = mostly negative, 3 = balanced, 4 = mostly positive, 5 = entirely positive) to rate participants' descriptions. Following independent ratings by all three authors, discrepancies were discussed and consensus was reached. Discrepancies between author ratings were largely due to initial differences in approach regarding which participant responses (or portions thereof) were eligible for inclusion in the analysis. Specifically, some of the text entered by participants clearly did not directly address the question on the survey, but other responses were more ambiguous.

Subsequently, in order to explore apparent thematic differences between parents' perceptions of a "balanced" description of DS and the "severity" of their child's DS, the authors also rated "severity" of DS in the participants' children by categorizing the child's experience on a 3-point scale (1 = mild, 2 = moderate, 3 = severe), taking into consideration the age of the child, the number and nature of medical problems reported, whether the child participated in the standard curriculum in school (if applicable), whether the child worked or continued on to further education (if applicable), and whether the child lived independently (if applicable). All three authors independently rated the degree to which they judged the participants' children to be affected by DS. Following independent ratings, discrepancies were discussed and consensus was reached. In this case, discrepancies between author ratings were mostly due to rater error (e.g. failing to observe a relevant data point).

Finally, many participants took advantage of the space provided to make suggestions for how health care professionals should communicate about DS. Since this feedback contained valuable incidental findings, consistent with the overall goal of the study (to empower genetic counselors to better serve individuals whose children/pregnancies have received a diagnosis of DS), the authors systematically reviewed all participant responses to extract suggestions for healthcare professionals regarding communication about DS.

Data analysis was descriptive in nature, and participant verbatim quotations were selected that illustrate key points.

Results

Sample Demographics

The majority of participants were Caucasian (87%) and female (79%) (see Table 1). There was an almost even split between participants who reported being "religious" and those who reported that they were not "religious."

Participants' children with DS were diverse in terms of age, sex, medical concerns, intellectual and social ability (See Table 2). Three participants volunteered that their children with DS had completed college courses.

Response to the Scenario Provided

Of the 101 respondents, 79 provided an answer to the question assessing their opinion of how to describe DS. Participants were not asked specifically for their opinion of the scenario that was provided, but 12 parents spontaneously reported that they felt the approach in the scenario was appropriate/balanced. For example, one mother of a 3 year old son with DS wrote, "The above scenario was very balanced in my opinion."

Table 1 Demographic information for sample of parents of individuals with DS (N=101)

	Number (n)	Percent (%)
Mothers	88	87.1
Fathers	13	12.9
Religious[a]	44	44.4
Non-religious	55	55.6
European	79	79
Asian	6	6
Mixed	8	8
Other (Canadian)[b]	7	7

Percentages are based on non-missing data

[a] Participants were asked: "Would you say that you are religious?" and given the option to reply "Yes" or "No."

[b] The only response given when participants selected "other" was Canadian. The options "African" and "Aboriginal" were also given, but no respondents selected these options

Table 2 Demographic information for participants' children with DS (N=102)

	Number (n)	Percent (%)
Females	53	52
Males	49	48
Medical problems[a]		
Has had surgery (for any indication)	60	58.8
Vision loss	49	48.0
Heart defect	42	41.2
Hypotonia	41	40.2
Thyroid dysregulation	32	31.4
Dental problems	30	29.4
Hearing loss	29	28.4
Gastrointestinal problems	17	16.7
Level of Independence		
Under 5 years old	16	16
Between Age 5–19	53	52
Standard curriculum at school	4	7.5
Modified curriculum at school	47	88.7
Left school	2	3.8
Over Age 19	33	32
Living with parent(s)—full time	19	57.6
Living with parent(s)—part time	4	12.1
Living with parent(s)—amount of time unspecified	4	12.1
Not living with parents	6	18.2
Work/Volunteer—independent	5	15.2
Work/Volunteer—supported	17	51.5
Not working	11	33.3

One respondent had twins with DS

[a] Participants could select as many medical problems as were applicable

Positivity Vs. Negativity

Participants' descriptions of DS ranged from entirely positive to entirely negative. By consensus, all three authors rated five descriptions as entirely positive (10%), 25 as mostly positive (49%), 12 as "balanced" (24%), five as mostly negative (10%) and four as entirely negative (7%). See Table 3 for examples of descriptions in each category.

Some participants whose descriptions were "mostly negative," were of the opinion that some of the differences that exist in caring for a child with DS mean that not everyone would find the experience fulfilling, as this mother of an 18 year old daughter with DS put it: "Sometimes people just talk about "all the blessings of these special people" but the reality is very difficult and not for everyone. To help these children thrive you need very dedicated and caring parents so children can't be resented or neglected." The extra dedication required for parenting a child with DS came up often for this group, for example, this father of a 22 year old daughter stated that: "Having a child with DS is more of a lifetime commitment than for a "normal" child."

By consensus ratings of the three authors, 33 children were rated as mildly affected, 48 as moderately affected, and 21 as severely affected. There were no apparent thematic differences in the nature of respondents' descriptions of DS when the descriptions were compared by severity rating. Parents of individuals with DS who were mildly affected provided the full range of descriptions from totally positive to totally negative. Similarly, parents of individuals with DS who were severely affected provided the full range of descriptions from totally positive to totally negative.

Suggestions Regarding Communicating a Diagnosis of DS

Participants spontaneously volunteered many constructive suggestions for genetic counseling at the time of a prenatal diagnosis of DS (see Table 4).

While contact with families of individuals with DS came up repeatedly as a suggestion for helping families who have received a prenatal diagnosis of DS, this experience can be difficult for the families who are contacted. A mother of a 15 year old daughter with DS raised this point by saying: "I would introduce them to my daughter BUT I have done that over the course of the years and I have still had parents who have still decided to abort. (That hurt me a lot. Although, I tried not to take it personally, it did still hurt)."

Discussion

> "It is unrealistic to expect one person's account of their life to be 'balanced' from anyone else's perspective" Ahmed et al. (2007, p. 319).

When asked for a "balanced description" of DS, the nature of the narratives provided by participants in this study varied widely. Our results highlight the importance of the observations of Williams et al. (2002) and Ahmed et al. (2007), who suggested there is enormous complexity in attempting to convey "balanced" information. If we assume that participants' descriptions were informed by their unique perceptions and experience, then this variance probably reflects extreme variation in the lived experience of families in which an individual has DS. Considering the descriptions that were entirely positive and those that were entirely negative could inform genetic counseling practice in the following way. Clinicians often spend time talking with clients about the variability of the DS phenotype. The data presented herein suggest that adopting the same

Table 3 Examples of descriptions of DS categorized by their rating on the 5-point scale used to assess degree of positivity or negativity

Rating of positivity or negativity	Example description
Entirely positive	"Your child will grow up, move out, have relationships, friends, a job, with/without support :)."—*mother of a 6 year old daughter with DS*
	"I could not imagine life without my daughter. She has brought so much joy to our family and has taught us to be more accepting of others. We don't know what the future will be for any of our children. Think positive and take things day by day."—*mother of a 10 year old daughter with DS*
Mostly positive	"Very loving and forgiving—no pretenses or mind games. Varying stages of intellectual disability which you cannot tell until they mature. Generally happy-go-lucky, somewhat stubborn. Physical disability varies too."—*mother of a 17 year old daughter with DS*
Balanced	"Varying degrees of mental abilities. Some Down syndrome are much lower functioning than others. Happy most of the time. As adults many live at home with aging parents. Some are capable of semi-independent living. Always learning throughout life."—*mother of a 24 year old daughter with DS*
Mostly negative	"Your life will be changed forever but not always in a bad way. You will be challenged daily, the demands physically and emotionally will be huge. If you are dedicated to improving the quality of life for your child and are persistent, the rewards will be huge. It is so exhausting but so gratifying as well."—*mother of a 9 year old son with DS*
Entirely negative	"I would want to tell them that the journey is filled with grief, is never-ending and the suffering for the whole family is insurmountable and my suggestion would be to terminate the pregnancy if they were given the choice, **without a doubt**." (Emphasis made by participant.)—*mother of a 21 year old son with DS*
	"It needs to be clear that this child is going to take all of their attention, time, and care, and that financially it will make it tough plus it is very necessary to have extended family support. There is a lot of personal care (e.g. changing diapers for 11 years); not a lot of free time to pursue any personal goals; health issues cropping up gradually, lack of speech abilities, a lot of false hope given by professionals, problems in the school system, not as easy as they say."—*mother of a 16 year old son with DS*

approach towards discussing the impact of having a child with DS on individuals, couples and families might be a strategy that parents would appreciate. It could be helpful to parents for a clinician to explicitly articulate their understanding that the family may want to know not just about the potential severity of the child's DS, but also about how this might impact their family. This is consistent with one of the recommendations included in the recently published Practice Guidelines (Sheets et al. 2011b). Clinicians could help their clients to understand the full range of experiences that parents have had as a result of having a child with DS (i.e., some experience having a child with DS as wonderful and life enriching, while others find it a struggle full of hardship). It might be appropriate to help parents understand that, in the same way that severity of DS cannot be predicted, it is hard to predict how the family would respond to having a member with DS. Moreover, patients could benefit from attempting to imagine what life might be like for their family if they had a child with DS. Genetic counselors could ask parents to speculate on how their family might react to the addition of a child with DS, and then use insights gained from either the genetic counselors' experience with individuals with DS or, for example, the experiences of participants in this study and those reported elsewhere in the literature, to encourage patients to elaborate on particular topics or to consider an element of raising a child with DS from an angle they hadn't considered.

With respect to specific suggestions from participants for genetic counselors who engage in disclosing a diagnosis of DS (see Table 4), there were many recommendations that reflected those that have emerged in previous studies. Brasington (2007) also emphasized the importance of conveying the similarities between individuals with DS and individuals without DS: "This child will be more LIKE other children than different" (p. 732). Over 96% of 687 parent respondents in another study felt that discussing how DS children are more like other children than different was essential to include in the initial conversation following the diagnosis of DS (Sheets et al. 2011a). The only item in that study that was rated by more parents as being essential was a discussion of available early intervention centers. Participants in the present study felt that it was important to mention adoption as an option, and this was also recommended in the recently published Practice Guidelines (Sheets et al. 2011b). One participant suggested limiting the amount of information that is provided at the time of diagnosis, which was also a finding of Skotko et al. (2009), and is consistent with the Practice Guidelines recommendation to tailor the amount of information provided to the needs of the individuals. The present participants also suggested counselors avoid using inappropriate language or terminology; this too is in line with the Practice Guideline recommendation that information provided should be nonjudgmental and unbiased, and corroborates the findings of Helm et al. (1998) and Skotko et al. (2009).

Table 4 Recommendations from parents for genetic counseling for DS

Parental suggestion	Frequency of suggestion	Quote(s)
Emphasize similarities between people with DS and people without DS, as well as between parenting a child with DS and a child without DS	34	"D.S. people have different skills + abilities just like 'ordinary' people."—*mother of a 21 year old daughter with DS*
		"I am a believer that all children have some level of disability my child's just has a name."—*mother of a 15 year old daughter with DS*
		"To me D.S. is not the issue. The extreme health issues are. A 'NORMAL' child can be born with major health issues, including cancers, brain defects, hearing loss that cannot be detected through screening."—*mother of a 17 year old son with DS*
		"As I read once, the news that your child has DS is like the death of your child because all the dreams and plans you had are over. That is only partially true, yes your "typical" child is dead but your dreams and plans are the same, (health, happiness, friends, success…) You just have to let go of the plans you had for your 'typical' child and have them for your DS child."—*father of a 4 year old daughter with DS*
		"Like the Poem—Something like "Holland instead of Italy" It's not the trip you planned but it is great just the same."—*father of an 8 year old son with DS*
Use a variety of methods for describing children with DS, including suggesting contact with families with a member who has DS	22	"Balanced, honest representation of possibilities and issues: medical issues, stats, success stories, pictures."—*mother of a 3 year old son with DS*
		"it might be good to have a video of different levels of what D.S. looks like. There is a great deal of variety in the amount an individual is affected by D.S. and parents who have little knowledge deserve to have a clearer picture than what words can convey."—*mother of a 5 year old daughter with DS (diagnosis made prenatally)*
		"A video could be made showing **many different** DS people at different age levels, in different activities, with family and in the community. Included in the video could be a scientific description and the possible health issues that can sometimes go along with D.S. Also several parent statements of the **challenges** and the **rewards**."—*mother of an 18 year old daughter with DS*
		"They should get to know families who have a D.S. member to get a more realistic view of life in general."—*mother of a 17 year old son with DS*
Emphasize the normal curve and describe the range of severity of DS, rather than focusing only on extreme examples	18	"That those with DS IQ fall into the normal bell curve with some quite mentally handicapped and some almost of our IQ, but most are mildly mentally handicapped—thus can learn, go to regular school, learn skills for daily living, work, have a social life and marry if they choose."—*mother of a 29 year old son with DS*
		"Provide range of probabilities of the severity of different medical issues & intellectual disabilities **if** asked by the couple."—*mother of a 5 year old son with DS*
Ensure that information provided is up-to-date	9	**"**A short, simple booklet that is current and has pictures would be helpful. We were given an outdated book when our son was born"—*mother of a 28 year old son with DS*
		"If the information is accurate and not negative. I hear from lots of parents news or info they get is all so negative. There is a lot to be positive about and parents need accurate information and lots of support."—*mother of a 6 year old daughter with DS*
Mention that adoption is an option	6	"I think that you should also make them aware that adoption is an option."—*mother of a 21 year old daughter with DS*
		"I think we must keep in mind that this is not for everyone but there are options like adoption there."—*mother of a 6 year old daughter with DS*
Do not use inappropriate language or terminology	4	"Do not use words that attach judgment or inappropriate language."—*father of a 7 year old daughter with DS*
Acknowledge that grieving is normal	4	"Be made aware early that it is a legitimate grieving process to go through and not to feel guilty."—*mother of a 5 month old son with DS (diagnosis made prenatally)*
		"I would start by reading to them and giving them a copy of "Welcome to Holland" by Emily Kingsley.—*mother of a 23 year old daughter with DS*
Ask parents up front for questions	1	"Ask first "What questions do you have?" You may be surprised what **their concerns** are."—*mother of a 6 year old daughter with DS*

Table 4 (continued)

Parental suggestion	Frequency of suggestion	Quote(s)
Limit amount of information provided at the time of diagnosis	1	"**do not** send a package home with a new parent (as I received) that outlines everything under the sun that will likely go wrong with your child. Give the package that discusses pros and cons and keep it limited. You don't need to discuss health issues at 50 years of age."—*mother of a 7 year old son with DS*

While the Practice Guidelines recommend providing "a range of possible outcomes to illustrate what life is like for individuals with Down syndrome and their families", which is similar to sentiments expressed in this study, participants in the current study went further to suggest ways in which this information could be conveyed. It has previously been recommended that written information about DS should be provided in addition to an oral description (Helm et al. 1998; Sheets et al. 2011a, b), but some participants in this study felt that it is important to show parents up-to-date videos of what life is like for individuals with DS. It was also suggested that providing stories from parents might be helpful. One difficulty with providing stories of families with an individual with DS, whether the stories are part of videos or in written format, is the potential for bias in those who offer to share their stories. As Ahmed et al. (2007) said, "Finding people willing to talk openly about 'negative'-life experiences may be more difficult than finding those whose experience is more positive" (p. 319–320). Indeed, one participant in the present study called the authors out of concern that the responses to the survey might be too positive because, in her words, it is "taboo" in the DS community to say anything negative about life with a child with DS. Another way of illustrating what life is like for families with an individual with DS is for patients to meet such families. This suggestion emerged in the present study, as it has repeatedly in prior work (Sheets et al. 2011a, b; Helm et al. 1998; Skotko et al. 2009). A novel aspect of the present study, however, was that we obtained some insight into the experience of the family with a member with DS to whom prospective parents are referred. Based on this information, we suggest that it is very important for counselors who facilitate such connections to consider and discuss with the family of the individual with DS how they might feel if after meeting, the prospective parents choose to end their pregnancy with DS.

One participant specifically suggested asking parents what their questions are prior to providing information about DS. Genetic counselors could consider incorporating this emphasis into contracting at the beginning of the session and integrating this emphasis on the parents' questions throughout sessions; thus, really using parents' questions to tailor the description. This may seem obvious, but genetic counselors may feel as if there is so much information to cover in a session with limited time, that there is a danger of dominating the conversation (Roter et al. 2006).

An additional replication of previous findings (Brasington 2007; Bryant et al. 2001; Helm et al. 1998; Skotko et al. 2009) was the reiteration that parents desire up-to-date information about DS. While many of the participants who made this request had children with DS who were over 20 years old, there were still parents of children as young as 3 years old requesting that information be accurate and up-to-date. The results suggest an improvement as far as providing materials, but perhaps more work could be done to ensure the information provided is representative of the latest anticipated outcomes and experiences of individuals with DS. The demographic information in Table 2 paints a picture of current expectations for the lives of individuals with DS; however, it is far from comprehensive. The collection of further data in this direction is an important endeavor for continuing research.

Practice Implications and Research Recommendations

In order to stay in touch with the latest outcomes and experiences of individuals with DS, perhaps genetic counselors in a given geographic region could conduct environmental scans of their local DS community (Graham et al. 2008) at regular intervals (e.g., every 5 years), and/or focus on building and/or maintaining strong links and communication with local DS support organizations. This suggestion echoes the calls of Skotko et al. (2009) to collaborate with leaders in the DS community during genetic counseling training; Madeo et al. (2011) who suggest, "those who practice genetic counseling must individually and collectively strive to more firmly establish and maintain relationships with members of the disability community" (p. 1778); and Wertz and Gregg (2000) who argue that "Education for professionals should always include experience with the lives of people with disabilities, outside a clinical setting" (p. 263). Parental suggestions for describing DS in a variety of ways could inspire or be used to strengthen collaboration with local DS communities; in particular, collaboratively creating videos which include

different family stories and individuals with DS at a range of ages. Additionally, conducting research into the opinions of individuals with DS themselves would be a very worthwhile avenue to pursue.

Specific recommendations for clinical practice include:

- Directly state that it is not possible to predict how a family will react to raising a child with DS, and explore coping mechanisms that have worked for the family in the past.
- Become involved with your local Down syndrome support group and explore your own biases with respect to disability. Develop a thorough understanding of positive outcomes as well as negative outcomes experienced by families with a member with DS. Increasing your own knowledge about the range of possibilities when it comes to life with DS will enhance your ability to provide a more nuanced description of what life with DS can be like.
- Ensure that communication regarding a diagnosis of DS and life with DS are parent-driven and responsive to parental emotions and informational needs, partly by continuing to ask parents for their questions.
- Ask parents to imagine what their life might be like with a child with DS, and use your knowledge of other families' experiences of life with DS to encourage parents to elaborate on different aspects of their anticipated life with and/or without their child with DS. What experiences are common to all parents? What would be different about having a child with DS specifically?

There were no apparent thematic differences between the "balanced" descriptions of DS that parents provided in this study as compared with the "severity" of the manifestation of DS in their child. It would be interesting for future research to explore what factors do contribute to parental response to having a child with DS and quality of life of the family (e.g., the child's age, family socioeconomic status, etc.). Identification of risk factors and protective factors could help to tailor support provided to a given family. It would also be interesting to further explore perceptions of "balanced" descriptions from the viewpoint of individuals who have terminated a pregnancy affected by DS and, in another direction, from the viewpoint of individuals who have children affected by other genetic conditions.

Study Strengths and Limitations

A significant strength of this study is the anonymous nature of the survey, which allowed participants to share their perspectives candidly. One participant expressed the opinion that it is "taboo" within the DS community to say anything negative about your child with DS. Another wrote "*I had amnio done with my 3rd child and would have terminated the pregnancy if the baby had Down syndrome*." (Mother of a 28 year old son with DS). This participant may not have been comfortable with this disclosure if the questionnaire was not anonymous. It does remain possible, however, that a social desirability bias may have affected the responses we received, and so is a potential limitation of the study.

The use of a scenario preceding the question asking parents how they would describe DS provided a specific focus for participants, and makes the findings more relevant for use in the prenatal genetic counseling setting. However, had we asked the parents to provide a balanced description of DS without this context, it might have been possible to access a greater diversity of spontaneous responses and avoid any bias in their responses.

Restricting the survey to members of a Down syndrome support group could have biased the results of the study given that there are potentially systematic differences between individuals who join support groups and those who do not. For example, it is possible that those who join support groups in an effort to advocate for individuals with DS were more likely to complete the survey and to report the positive aspects of life with DS, and/or that individuals who join support groups may feel more positively about their child with DS as a result of this support. However, although more descriptions were rated as positive (30) than negative (9), we still received a wide variety of opinions and descriptions ranging from entirely positive to entirely negative.

Distributing the survey only in English restricted the responses to individuals who were English speaking, and likely contributed to the majority of respondents being Caucasian. It is thus not possible to generalize the results to individuals of different ethnicities. It is very possible that individuals of different ethnicities might have very different perspectives on both what constitutes a "balanced" description of DS and how it is best to describe DS to a couple who receive a diagnosis of DS prenatally.

Conclusion

Given that it seems difficult to reach consensus on what constitutes balanced *information* (facts), we argue that it is not feasible for any single *description* to be perceived by everyone as "balanced." In a genetic counseling context, information cannot be provided without becoming a description; thus, we argue that "balance" is an unattainable goal. Instead, it is important to ensure the information provided is up-to-date (which includes information that many consider to be "negative" about medical outcomes and that many consider to be "positive" about treatments and educational interventions that lead to improved out-

comes), and that the provision of information, or the description, is non-judgmental and responsive to patient needs.

Further, just as there is a range of severity of medical concerns and intellectual disability for individuals with DS, there is diversity in how families will react to raising a child with DS. In the same way that we cannot predict the medical concerns a child with DS will have, we cannot predict how a family will cope with raising a child with DS. As one participant (a mother of a 22 year old daughter with DS) succinctly stated, "[it's not possible to] tell by a test what life will be like having a DS child. The test only shows the condition." We suggest that, when discussing a prenatal diagnosis of DS, genetic counselors should explore a spectrum of experiences with parents; not only in terms of the degree to which a child can be affected by DS, but also in terms of the diversity in life experiences of families that include an individual with DS. Exploring the potential reactions of a family to a member with DS could include both the provision of stories from other families' perspectives and/or encouraging patients to imagine what their life might be like with a member with DS. In the process of exploring a family's anticipated reactions to either having a member with DS or ending a pregnancy affected by DS, it would be natural also to explore coping strategies and support networks. We also recommend genetic counselors strengthen relationships with local DS community groups and increase awareness and knowledge of current expectations with respect to the lives of individuals with DS and their families.

Acknowledgements This research was made possible by funding from the National Society of Genetic Counsellors Prenatal Special Interest Group, and a Rare Disease Foundation Microgrant. A. I. was a student of the University of British Columbia (UBC) MSc in Genetic Counselling program when this research was conducted. Special thanks to Rachelle Blais, the board members of the LMDSS, Theresa Preston, and the participants for taking the time to elaborate on their answers to our many questions. J. A. was supported by CIHR, the Michael Smith Foundation for Health Research, and BC Mental Health and Addictions Research Institute.

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