RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Hearing From Parents: The Impact of Receiving the Diagnosis of Williams Syndrome in Their Child

Jessica L. Waxler,[1]* Elizabeth M. Cherniske,[2] Kristen Dieter,[3] Pamela Herd,[4] and Barbara R. Pober[1,5]

[1]Massachusetts General Hospital, Boston, Massachusetts

[2]Yale University School of Medicine, New Haven, Connecticut

[3]Northwestern Reproductive Genetics, Chicago, Illinois

[4]University of Wisconsin, Madison, Wisconsin

[5]Department of Pediatrics, Harvard Medical School, Boston, Massachusetts

Manuscript Received: 9 May 2012; Manuscript Accepted: 11 October 2012

Healthcare providers often share difficult or life-altering news with their patients yet this challenging and delicate process is frequently met with dissatisfaction by those receiving this news. Articles and guidelines exist to aid providers in sharing diagnoses such as Down syndrome, but relatively few have focused on rare genetic conditions often diagnosed years after birth. For this reason, we sought to learn about the experience of receiving a diagnosis from parents of children with Williams syndrome. We asked members of the Williams Syndrome Association to complete an anonymous online survey about recollections related to the diagnostic process. Responses, both close-ended and open-ended, were received from 600 families across the United States. Analysis revealed a high proportion of families (59.91%) with at least some negative recollections about the experience (and nearly half of those with negative recollections denied recalling anything positive). Factors influencing a more positive overall perception of the experience included receiving written information about Williams syndrome and seeing a genetic counselor. Analysis of open-ended responses identified additional positive and negative themes; for example, nearly one quarter of respondents expressed a desire to be given hope when receiving the diagnosis. Based on these analyses, we offer several specific recommendations for improving the diagnostic process in the future. © 2013 Wiley Periodicals, Inc.

Key words: breaking bad news; Williams syndrome; Williams–Beuren syndrome; diagnostic process; parental recollections

How to Cite this Article:
Waxler JL, Cherniske EM, Dieter K, Herd P, Pober BR. 2013. Hearing from parents: The impact of receiving the diagnosis of Williams syndrome in their child.
Am J Med Genet Part A 161A:534–541.

INTRODUCTION

The Practice of Breaking Difficult News

Health care providers are frequently called upon to provide patients and family members with difficult or life-altering information. The task of delivering "bad news" (as it is often termed) arises with any diagnosis that negatively impacts a person's future or leaves a lasting emotional effect [Fallowfield and Jenkins, 2004; Harrison and Walling, 2010]. This near universal aspect of medical care is a component of most disciplines but is especially common in practices that care for patients with cancer, terminal illness, chronic disorders, birth defects, and genetic disorders. Unfortunately, many providers do not receive formal training or supervision in the acquisition or practice of this skill, making them feel inadequately prepared to deliver difficult news [Sharp et al., 1992; Dube et al., 2003; Fallowfield and Jenkins, 2004].

Several studies demonstrate that families express a high level of dissatisfaction after receiving difficult news. The focus of their dissatisfaction most often centers on the style and interpersonal skills of the provider and/or the content and quality of the information being provided [Barnett, 2002; Skotko, 2005; Skotko et al., 2009b; Gilbey, 2010]. Patients are, however, able to articulate preferences related to receiving potentially negative information. Suggestions for improving the process of breaking difficult news include being provided appropriate information, receiving the news in a private setting, and being connected to other families or individuals for support [McCluskey et al., 2004; Skotko et al., 2009b; Harrison and Walling, 2010].

Underscoring the importance of this topic, consensus guidelines and checklists are emerging to assist healthcare providers in per-

Additional supporting information may be found in the online version of this article.

*Correspondence to:
Jessica L. Waxler, 175 Cambridge Street, Room 502, Boston, MA 02114.
E-mail: jwaxler@partners.org

Article first published online in Wiley Online Library (wileyonlinelibrary.com): 7 February 2013
DOI 10.1002/ajmg.a.35789

forming the task of providing difficult news. For example, the National Society of Genetic Counselors (NSGC) recently published guidelines for sharing the diagnosis of Down syndrome with parents, both prenatally and at birth [Sheets et al., 2011]. Likewise, providers in other areas of medicine are crafting similar recommendations for delivering news and for training practitioners [Russ et al., 2004; Tluczek et al., 2006; Paul et al., 2009].

Diagnosing Williams Syndrome

Much of the current literature examining patient or parental responses to receiving difficult medical news pertains to conditions such as Down syndrome or cancer. In the case of Down syndrome, the diagnosis is typically established either before a child is born or immediately after birth, while most diagnoses of cancer are established later in life. Less information exists for the diagnosis of other genetic disorders, especially those where the diagnosis could be established in the newborn period but is generally not made until much later. In one study of cystic fibrosis, delay in diagnosis increased parental frustration; they felt this impeded delivery of optimal care to their child and also resulted in a loss of faith in medical professionals to identify disease [Merelle et al., 2003].

We sought to learn more from parents who have had a child diagnosed with Williams syndrome (WS) [OMIM 194050], a genetic syndrome which, unlike Down syndrome, is usually diagnosed months or even years after birth. WS is a micro-deletion disorder caused by the loss of 26–28 contiguous genes mapping to chromosome 7q11.23. A diagnosis of WS heralds the potential for multisystem medical concerns as well as the certainty of developmental delays and intellectual disability [Mervis et al., 2000; Pober, 2010]. At the same time, individuals with WS also display many positive personality features (e.g., empathy, good social and interpersonal skills) as well as certain relative cognitive strengths (e.g., strengths in selective language skills, good long-term memory) [Pober, 2010]. This duality is reflected in a message to parents from the parent support group, the Williams Syndrome Association (WSA)—"Williams syndrome: Extraordinary Gifts, Unique Challenges." Even though positive aspects of WS exist, its diagnosis has major and life-altering implications for the individual and the family.

Assessing the Experience

To gather perceptions from a large number of parents receiving the diagnosis of WS in their child, we used the internet to solicit anonymous feedback from WSA members. We anticipated this approach would allow for honest parental feedback and encourage responses from regions throughout the United States. We report here on the perceptions of remarks and actions, either positive or negative, by health care providers during the diagnostic process. We also expand on the details of these recollections that parents found either useful or unhelpful in learning about a new WS diagnosis. Using the responses collected from parents, we will make practical suggestions which healthcare providers can employ when telling families life-altering news in the hope of ultimately improving this experience for future families.

MATERIALS AND METHODS

Data Collection

Data were obtained from parents' responses to an online, anonymous survey hosted by Survey Monkey™. The survey, developed expressly for this study, contained 12 questions (in Supplemental Material A—See Supporting Information online). It prompted families to recall useful and supportive remarks, as well as inappropriate or unhelpful remarks, made during the WS diagnostic process. Eleven of the questions were closed-ended with "yes/no" response frames; 5 of these 11 questions were followed by text boxes in which parents could choose to insert open-ended and free form remarks. For example, one question asked:

> "Was there any information that you did not receive, but in hindsight would have found useful in helping you find support and/or learn more about the diagnosis of Williams syndrome? If so, please describe this in the text box below."

The 12th and final question was entirely open-ended, allowing respondents to provide additional information on any topic of their choice.

The survey questions and electronic method of distribution were approved by the Massachusetts General Hospital Institutional Review Board.

Respondents

Participants were recruited for this study through the non-profit parent support group, the WSA. On the authors' behalf, the WSA sent out two email announcements to its membership providing them with a link to the survey. At the time the survey was initially distributed in June 2008, the WSA membership consisted of 1,500 families. Individuals were asked to participate if they had a family member diagnosed with WS. The respondent had to have been present when the diagnosis was given and, therefore, was almost exclusively a parent. A total of 600 responses to the survey were received, resulting in a 40% response rate. Twenty-six responses were excluded, for either answering fewer than half the questions or disclosing their child was not diagnosed with WS, leaving a pool of 574 responses. While, ideally, the sample would be representative of the entire population of parents with Williams syndrome, a complete registry from which such a sample could be drawn does not currently exist. That said, we think the WSA membership is closer to representative than a single provider-based sample because it includes families from across the United States. Furthermore, recruiting through providers has the potential to bias a sample toward those making more frequent visits to the doctor (i.e., increasing the likelihood a person would learn about the study) and consequently could disproportionately result in the recruitment of children who had more serious health problems associated with the syndrome.

Surveys were completed by caregivers living in the United States and residing in 48 different states. The age of individuals with WS for whom the survey was successfully completed ranged from 1 month to 55 years (mean age of 14.42 years; SD $\pm$ 11.34). In 91% of cases, the diagnosis of WS was established by age 5 years and

nearly 1/3 of the individuals with WS were between the ages of 6 and 15 years old at the time of survey completion.

Deriving Perception of the Diagnostic Experience

Analyses focused on capturing respondents' perception of their diagnostic experience (our dependent variable) and the factors that influenced this perception (our covariates). The diagnostic experience was categorized using responses to two survey questions, numbers 6 and 7, which solicited positive and negative recollections, respectively (Fig. 1A). We used specific answer combinations to classify the respondent's overall feelings of the experience as either positive, negative, or mixed. If a parent reported *only* positive recollections and denied negative recollections, we assumed his or her overall experience was primarily positive. Conversely, we assumed an experience was primarily negative when *only* negative recollections were reported. Parents reporting both positive and negative recollections were classified as having a mixed experience. This algorithm allowed us to classify parental perception of the diagnostic experience in 439 of the 574 respondents; the diagnostic experience could not be classified for the remaining 135 respondents as they failed to comment on either positive or negative recollections (Fig. 1B).

To gain additional insight and collect granular detail on parental perceptions of specific aspects of the diagnostic process (such as opinions of the healthcare provider, quality of educational information received about WS, etc.), three of the authors (each a qualified genetics service provider) read the open-ended responses to all free-text questions. Each response was scored using criteria formalized in a codebook (See Supplemental Material B in Supporting Information online for more details). This read of open-ended text also allowed us to generate and tally a list of events spontaneously reported by families (such as their reaction to being shown a picture of a person with WS); several of these events are shown in Table IA. Examples of quotes that particularly illustrate the general findings or that provided suggestions for improving the diagnostic process in the future are provided in the Results and Discussion Sections.

The open-ended reading also provided a second and independent method for categorizing the parents' diagnostic experience. This approach yielded a distribution of positive, negative, and mixed experience categories that did not significantly differ from that generated by analyzing responses to survey questions #6 and 7 (data not shown). Accordingly, all analyses relied on the data generated from questions #6 and 7, using the algorithm depicted in Figure 1.

Covariates

To assess factors influencing the diagnostic experience, we examined several variables including the age of the individual with WS at the time the parent completed the survey, the age of the individual with WS at the time of diagnosis, and whether or not the family saw a genetic counselor during the process. We also explored the effect of receiving information versus not receiving information, specifically "written information, online resources, or other resources" from a "doctor, nurse, or genetic counselor," when the diagnosis was first established. These covariates were selected following a literature review searching for aspects of the diagnostic process that were commonly mentioned as impacting the experience [Sharp et al., 1992; Girgis and Sanson-Fisher, 1995; Chisholm et al., 1997; Skotko et al., 2009a].

As some covariates could be related to each other, we performed multifactorial analyses. For example, failure to meet with a genetic counselor could reflect an older aged child who was diagnosed many years ago prior to the widespread incorporation of counselors into genetics clinics. Likewise, seeing a genetic counselor could increase the likelihood resources were received and impact the overall perception indirectly.

Analysis

The relationship between the covariates and the respondent's perception of the diagnostic experience was examined using multinomial regression models (Stata 11.0), which takes into consideration the predominately categorical nature of the outcomes. We developed three different models, adding additional variables in succession. Each variable was examined not only for its independent impact on parental overall diagnostic perception but also for any mediating impact on the other covariates. The first model regressed age of diagnosis and the current age of the child. The second model, in addition to the variables from model 1, added the covariate of genetic counselor involvement. The final model further incorporated whether or not parents reported receiving written information or other resources about WS at the time of diagnosis.

RESULTS

Perception of the Experience

Based on responses to survey questions #6 and 7, we classified the parental perception of diagnostic experience for 439 respondents as follows: 176 cases (40.09%)—a primarily positive experience; 112 cases (25.51%)—a primarily mixed experience; and 151 cases (34.40%)—a primarily negative experience (Fig. 1B). The final regression models included 385 respondents due to missing information on other covariates.

Age of Diagnosis and Current Age

Multinomial regression model 1, including age of diagnosis and current age of the individual with WS, indicated that parents with an older child at the time of diagnosis were more likely to report a primarily positive rather than a negative experience (RRR 1.47; $P < 0.002$). In contrast, the older the current age of the child with WS (e.g., the age of the child at the time of survey completion), the parents' perception of the diagnostic experience was less likely to be classified as positive versus negative (RRR 0.97; $P < 0.01$). Neither of these differences remained statistically significant when comparing those with a mixed experience to those with a negative experience.

Genetic Counselor

The second model added a measure to capture whether or not the parent receiving the diagnosis had seen a genetic counselor during the process. While the inclusion of this variable had no meaningful impact on the age of diagnosis or current age measures, families

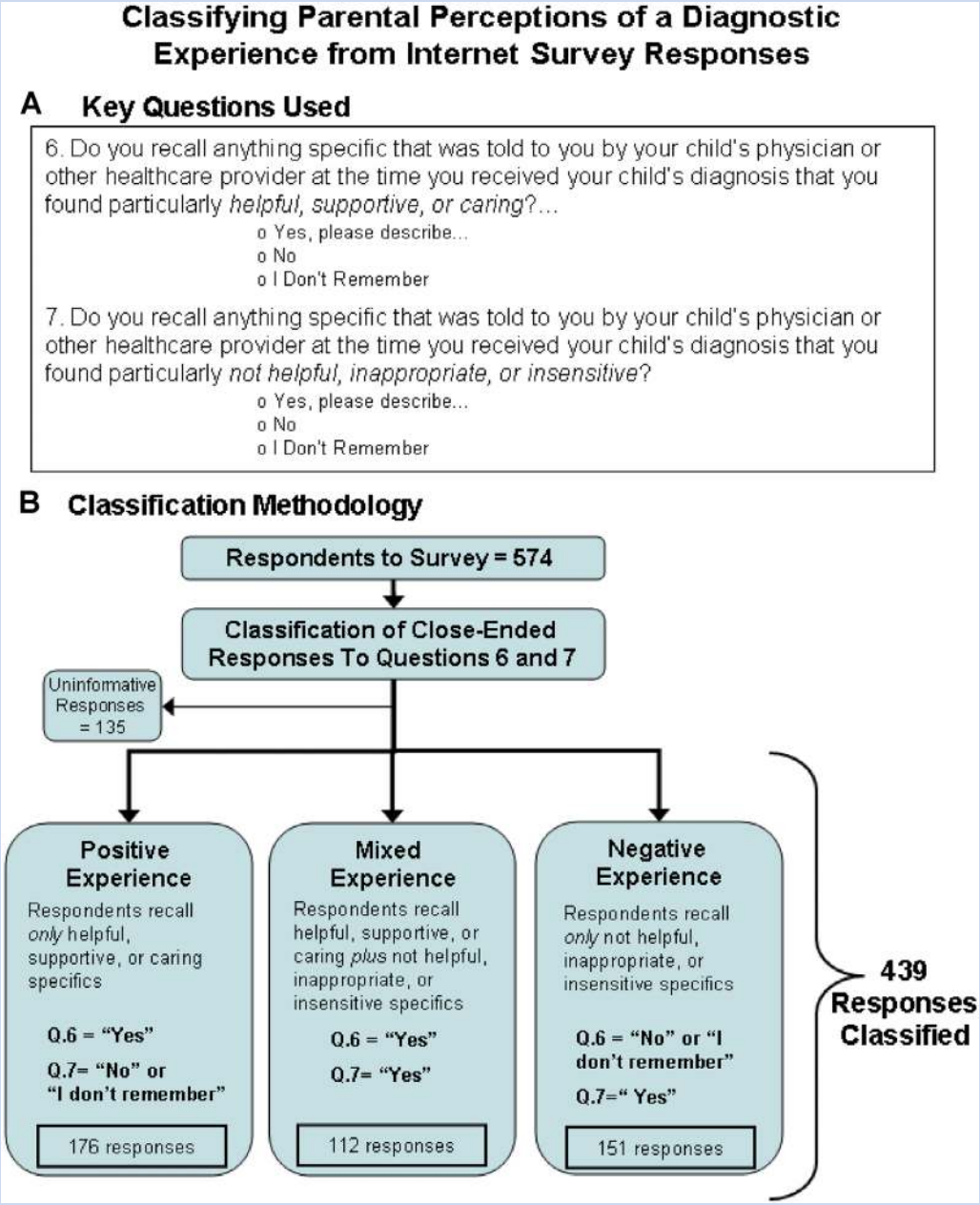

FIG. 1. Parental perceptions (of receiving a diagnosis of WS in their child) were scored based on answers to two internet survey questions shown in (A). The answers were classified as positive, mixed, or negative providing a categorical value for the respondent's perception of the diagnostic experience (N = 439). [B] [Color figure can be seen in the online version of this article, available at http://wileyonlinelibrary.com/journal/ajmga]

who saw a genetic counselor (n = 368, 81.60%) as part of their diagnostic process were more likely to have a positive (RRR 1.95; $P < 0.05$) or a mixed experience (RRR 2.76; $P < 0.001$) than a negative experience.

Information

The third model factored in whether parents received written information or access to other information regarding WS at the time of diagnosis. Families who reported they did not receive written information were less likely to be classified as having a positive experience (RRR 0.16; $P < 0.0001$). Inclusion of this variable mostly explained the association between seeing a genetic counselor and reporting a primarily positive experience, reducing the size and significance of the genetic counselor coefficient. Similarly, those who reported not receiving information were less likely to report even a mixed experience versus a negative experience (RRR 0.33; $P < 0.0001$). The genetic counselor variable, however, remained statistically significant, though was slightly reduced in the magnitude of the association (RRR 2.29; $P < 0.01$).

TABLE I. Events and Emotions from Open-Ended Responses

(A) Specific events noted in open-ended responses potentially impacting family perception of the diagnostic experience

Event	Families commenting positively	Families commenting negatively
Provider showed me a picture of WS	9	23
Received the diagnosis outside the office (i.e., phone)	9	30
Being told WS was a "better diagnosis"	38	7
Being used as a "teaching example"	2	17

(B) Specific emotions detected by genetics providers in open-ended responses according to criteria in codebook[a]

Emotion	Families rated as expressing that emotion	% of 439 respondents
Hope	112	23.38
Externalizing emotion (i.e., anger)	89	18.58
Internalizing emotion (i.e., sadness)	173	36.12
Relief/validation	87	18.16
Removal of guilt	16	3.34

[a]See Supplemental Material B.

Qualitative Themes

The scoring of free-text responses identified several events or emotions in the diagnostic process which were spontaneously mentioned by a subset of families (see Table I). Specifically, nearly one quarter of respondents (23.38%) stressed the importance of receiving "hope" in the sense of being provided reassuring or optimistic information about their child's potential abilities. Many noted the importance of being connected to resources like a WS specialist or another WS family, while a handful of respondents relayed that it was comforting to learn that having a child with WS "wasn't their fault." Families also seemed to find it comforting to hear they would be able to handle the diagnosis of WS. One family wrote: *The genetic counselor was particularly compassionate and provided handouts on local and national support organizations. We were so blindsighted [sic] that there was anything wrong with our baby that being given a direction to go in was appreciated.*

In contrast, respondents spontaneously expressed negativity about other events during to the diagnostic process. For example, 17 families reported receiving out-dated information and in most cases were very upset by this. Likewise, a similar number of families disliked when their child was used as a teaching-example. One family wrote: *After my son was operated on for SVAS a group of doctors came through the hospital and came into our room. They looked athim and said 'Yes, that looks like a Williams kid'. Iwas very offended because they talked as if I were not present at all.* Receiving a diagnosis by phone also received a high level of negative feedback; one family wrote: *It was a 2 minute conversation where I was told the results . . . at the time, I was home alone with my 3 month old. Needless to say I was devastated. Looking back now, I am appalled that I was given those results over the telephone.* Although not specifically tallied, events such as a healthcare provider presenting only a "problem list" or spending what the family felt was an inadequate amount of time with them each received criticism from several families.

Overall, families had comments of both praise and frustration regarding their care providers; some families strongly liked or disliked all their care providers while others rated each provider differently. Genetics healthcare providers and pediatricians were most frequently mentioned as the provider involved in the diagnostic process but others, such as cardiologists and developmental pediatricians, were also reported as having a role. Of those 204 respondents who commented about a genetics healthcare provider, 73 (35.78%) had a primarily negative opinion. Of those commenting on a pediatrician or other provider, 51/95 (53.68%) and 116/223 (52.02%), respectively, expressed an opinion that was primarily negative.

Lastly, using criteria established in the codebook, the authors detected emotional reactions in many of the responses despite the fact some families went through the diagnostic experience a decade or more earlier. Specifically, the raters scored the presence of emotions such as anger (18.58%) or sadness (36.12%) in the open ended responses received from our final cohort of 439 respondents. For example, one mother remembered getting the diagnosis when her was son was very young and wrote, . . .*I have never forgotten it. How I felt like a knife stabbing me in my gut. My son is now 29 years old and I remember that day like it was yesterday . . . today I still cry when I talk about it.* Some families expressed feelings of both anger and sadness.

DISCUSSION

Room for Improvement

Sharing difficult news with patients is one of the greatest challenges healthcare providers face. Our study indicates a high rate of parental dissatisfaction with the process of learning their child has WS. Nearly 60% of the parents in our survey relayed they were told something inappropriate, not helpful, or insensitive at the time their child was given a diagnosis. In fact, over one third (34.4%) of these individuals denied recalling anything positive about their diagnostic experience with most of their negative recollections being directed at one or more specific care providers. Though this negativity showed certain patterns (described further below), its pervasive presence highlights the ongoing need for improvement in how providers deliver difficult news.

A respondent was more likely to be classified as having a negative diagnostic experience if she or he reported receiving no resources or contact information for reaching out to another parent. Respondents also voiced frustration (in their open-ended responses) about receiving outdated information: *Not much was said, except that the pediatrician brought out an ancient text book from the 1950s and showed me a picture of a person w/Williams that was terrifying!* When asked what they wished they had been given at the time of diagnosis, several respondents expressed the desire to hear a range of abilities and receive a list of resources. One family wrote: *I would have liked to have been handed a packet of info regarding Williams syndrome at the time of diagnosis, instead of going home devastated and turning to the very disturbing Internet. That shows some truthful, but very scary, information, with no doctor present. . .*

This desire to receive information at the time of diagnosis is consistent with other reports in the literature. Specifically, several studies have also found that families would like to be given high-quality verbal information and written resources to take home after receiving a diagnosis [Boyd, 2001; Oshea et al., 2007]. Other studies demonstrate that patients have difficulty processing verbal information at the time difficult news is given, further strengthening the importance of providing appropriate written information that can be referred to later [Skotko, 2005].

Another predictor of an overall negative experience was having an older aged child with WS at the time the survey was completed. As mentioned above, these parents were more likely to recall a primarily negative experience compared to parents with a younger WS child at the time of survey completion. Several explanations may account for this observation including that health care providers' abilities to share the diagnosis of WS have gradually improved or that, with time, parents can better define the gap between how they wished the diagnostic experience had been versus the actual process and events that transpired.

Qualities of a More Positive Experience

Other aspects of our study shed light on ways parents suggest the diagnostic experience could be made more positive. As already discussed, being provided information (that is up to date, as well as appropriate in message, content, and tone), and having ready access to information, favorably impacts the parental perception of diagnostic experience. Those families who saw a genetic counselor were more likely to have a positive diagnostic experience, though the impact of seeing a genetic counselor could be partially explained by receiving information. This is not surprising as genetic counselors receive extensive training on how to deliver difficult news and provide appropriate support as well as resources [American Board of Genetic Counseling, Inc., 1996].

Many of the positive recollections that parents shared focused on events or information which they found helpful for coping with the diagnosis and continuing to move forward. For example, families appreciated being given "hope" or the sense that raising a child with WS was not a solely negative task but one that will also be accompanied by certain joys. Similarly, many parents reported they were happy to be (or in some cases wished they had been) connected with other healthcare providers or families who could help with the "next steps." One parent clearly expressed this need: *[sharing that] you aren't the only one going through this and that there is help out there, is probably the biggest help you can give a parent when telling them what is wrong with their child.* These sentiments were echoed by many respondents who were clearly seeking a "game plan" and not just a list of problems associated with the diagnosis. Even those who didn't receive this level of support voiced their wishes for it: *It would have been helpful to have resources or someone explain how to navigate all the different doctors/therapist [sic] and develop a plan with us for our son.*

Applying Lessons Learned

The opportunity for parents to write in open-ended remarks allowed us to discover several events that can contribute to the positive or negative perception of the diagnostic experience. Though the actual numbers are small, we chose to list several of them because they were spontaneously remarked on by at least 15 families and also because they represent specific, but potentially modifiable, actions taken by healthcare providers (see Table I). For example, 38 families found it helpful to hear that if their child had to have a genetic syndrome, WS was a "better diagnosis" to have than other syndromes; however, 7 families were not all comforted by this. One person wrote, I was told *something along the lines of 'if you have to have something wrong with your child. . . WS is a good one'. Oddly enough it was helpful.* But another wrote, the doctor *did apologize but made the comment that of the two syndromes she suspected, WS was the best to have. I found this insensitive, because obviously it would have been best to not have any genetic syndrome!* Another individual preference was the reaction to seeing a medical textbook picture of someone with WS. Although common in the practice of genetics, it was appreciated by only 9 of the 32 families who mentioned it. This was particularly troubling for one mother who wrote the doctor *put a medical textbook in front of my husband and me. He pointed to a picture of a very facially deformed looking individual (presumably with WS), and said 'Your baby looks like this'. It was a shocking picture and in all the years of going to WSA conferences, I've never seen a person with WS that looked like that. I thought my baby was adorable and thought the doctor was insulting. . .*

Our findings also raise awareness of the need to consider the family's current situation when choosing the words to convey the diagnosis of a genetic disorder. Specifically, parents of a newly diagnosed adult with WS are very familiar with their child's abilities as well as the areas where their child needs additional help. For these families, a sense of relief may influence their perception of the diagnostic process. For example, one family wrote *We always knew our daughter had developmental problems but when she was 2 we first had genetic testing and they came up with no diagnosis, so we treated the symptoms and went on not knowing. . .* The mother goes on to write that after establishing a diagnosis of WS *I was relieved to know that we had a diagnosis and that there were experts who knew something about this.* This phenomenon of relief may offer an explanation for the possibly counterintuitive finding that the older the individual with WS was at the time of diagnosis, the more likely the diagnostic experience was recalled as being positive. Russ et al. also explored the parental reactions to receiving a diagnosis and suggested that some expectation of the diagnosis lessens the emotional reaction [Russ et al., 2004].

The strengths of this study are several-fold. They include responses from a large sampling of parents living in 48 of the 50 states, anonymous responses (which we believe promotes honesty), and careful review and classification of open-ended remarks which provided a unique view into the mindset of parents who have had a child diagnosed with a life-long genetic disorder. Given the rarity of WS, our recruitment methodology provided the most powerful and timely approach possible to collect important feedback.

There are, however, several limitations of this study. Foremost among them is the fact that those responding likely constituted a self-selected population, not necessarily representative of all parents who have a child with WS. Specifically, we did not collect information on socioeconomic status or ethnic background and, clearly, participation was limited to those with access to a computer. Additionally, we could have garnered further insight on pre-existing respondent perspective had we used a tool that assesses "backgrounds, needs, and expectations" [Peters and Petrill, 2011]. Their work relies heavily on an idea well established in the genetic counseling literature: that past experiences influence perceptions of new diagnosis and the genetic counseling process. Accordingly, an assessment of these experiences will be important for future studies. Another study limitation is our response rate of 40%, even though this is comparable to that observed in a previous similar study based on a survey [Skotko, 2005]. Even if we classify all those who failed to complete the survey as having a positive recollection of the diagnostic process, the percent of families recalling partially or entirely negative experiences is not trivial, but rather equals 17.5% (263/1,500). Finally, our survey could have also benefited from variation in question order. Specifically, we asked about positive recollections before negative recollections and this may also bias responses. Several responses from parents suggest ways to revise the questionnaire in future studies. For example, findings presented in Table I derived from information volunteered spontaneously by parents rather than being elicited in response to specific questions, such as seeing a picture or receiving a diagnosis outside the office. Given the repeated yet unprompted mention of these events, we feel confident that they can impact the diagnostic experience and would be a fruitful source of information in future surveys.

Results from our study, in spite of the limitations just presented, highly parallel the increasing body of literature offering recommendations for breaking difficult news. Several acronyms that are commonly employed to remind providers of the protocols for breaking news include very similar recommendations. In the ABCDE method, those sharing difficult news are reminded to have "advance preparation," "build therapeutic relationship," "communicate well," "deal with reactions," and "encourage/validate emotions" [Rabow and McPhee, 1999]. Likewise the SPIKES protocol makes suggestions related to the "setting," "perception," "invitation," "knowledge," "exploration," and "summary/strategy" [Baile et al., 2000].

In the setting of Down syndrome, several of the recommendations offered by Skotko et al. are reinforced by findings from this work, including the importance of conveying accurate information that includes positive elements at the time of diagnosis [Skotko et al., 2009a], and connecting newly diagnosed families with other families [Skotko, 2005]. The recently published practice guidelines from NSGC and other researchers also make similar recommendations including being aware of the parents' current state of mind, emphasizing a balanced perspective, and providing appropriate resources for families [Dent and Carey, 2006; Sheets et al., 2011].

Take Home Message

No matter how the message is given, sharing a significant diagnosis has a long lasting effect on families. Research suggests that the moment a healthcare provider breaks difficult news will be remembered indefinitely [Russ et al., 2004]. This is certainly the case for many of the respondents in this survey who freely shared their experience with palpable emotions. As one person wrote, *I will always remember the day we found out the diagnosis... There were definitely things that were said that still cause me pain...* Comments such as this were frequent even when the experience was positive.

In light of the potentially life-changing impact of the message, healthcare providers need to strive to share information as thoughtfully and sensitively as possible. The respondents to this survey had many different experiences but, even so, several common recommendations emerge on ways to improve the diagnostic process. As discussed, our results confirmed some previously published recommendations such as having up-to-date information available at the time of the diagnosis but also the need to engage families in a dialogue about the diagnostic process. For certain elements of the diagnostic process, such seeing a picture, families voiced the need to have their preferences assessed. And for these more choice-driven events, the experience may have been more positive had the family been asked their preference.

Accordingly, healthcare providers who share news should continually check in with the family throughout the process. The existing acronyms (i.e., ABCDE and SPIKES mentioned above) may include assessment or validation of the patient but they do not emphasize the need to periodically re-engage. In the practice of genetic counseling, establishing a mutually agreed upon agenda (e.g., "contracting") is a well described technique but primarily focuses on the beginning of the session [Uhlmann et al., 2009]. Responses from the families in this study indicate that the process needs to be more iterative and that the "contracting" process needs to be revisited as the dialogue ensues.

Among the many lasting messages, the families in this study felt strongly that when delivering difficult news healthcare providers should:

- Above all, engage the family in a dialogue to help guide the diagnostic process, particularly for choice-driven events.
- Deliver the message accurately and compassionately.
- Have up to date information readily available.
- Listen to the family and answer their questions.
- Stop and think before giving a diagnosis over the phone, showing a picture, or bringing a trainee into the room.
- Involve knowledgeable professionals like genetic counselors who are trained to share difficult information and find appropriate resources.
- Connect families with support groups, other parents, and appropriate care providers.
- Assist with developing a game plan (e.g., action plan), rather than simply provide a "laundry list" of potential problems.
- Remember your words will have a lasting impact on the family.

Though many of these suggestions echo recommendations previously identified in the breaking "bad" news literature, there are several points that are special to the practice of clinical genetics. These include showing medical photographs, and the lengthy and highly variable time interval for availability of test results. These too need to be given some consideration when sharing a diagnosis; for instance, families should be asked in advance how they wish to be informed of results (e.g., on the phone and if so, on a home, mobile, or work phone).

Even if the recommendations shared above improve the diagnostic process, it is important for providers to remain aware of the enduring impact a diagnosis such as Williams syndrome can have on the family. As one respondent eloquently wrote:

When any child is diagnosed with any kind of disorder or syndrome health care providers need to understand that at that moment you may be smashing all the dreams the parents have for that child-birthday parties, sleepovers, first love, prom dates etc. . . In that split second their lives are changed forever and they are standing on the edge of the line that divides us from the rest. There are certainly other syndromes out there that are more traumatic to be diagnosed with—either way we are overwhelmed about where to go, who to see and what to do. Having the person who gives that diagnosis to the parents understand that they are changing that family's world with the words that are coming out of their mouth may soften the blow. Being prepared to provide information is so important.

ACKNOWLEDGMENTS

We would like to thank the many families who shared their experiences receiving the diagnosis of Williams syndrome in their child as well as the Williams Syndrome Association for distributing our survey to their membership.

REFERENCES

American Board of Genetic Counseling, Inc. Adopted 1996 and revised periodically. ABGC accreditation—Standards for program. http://www.abgc.net/Training_Program_Accreditation/Program_Standards.asp

Baile WF, Buckman R, Lenzi R, Glober G, Beale EA, Kudelka AP. 2000. SPIKES-A six-step protocol for delivering bad news: Application to the patient with cancer. Oncologist 5:302–311.

Barnett M. 2002. Effect of breaking bad news on patient's perceptions of doctors. J R Soc Med 95:343–347.

Boyd JR. 2001. A process for delivering bad news: Supporting families when a child is diagnosed. J Neurosci Nurs 33:14–20.

Chisholm CA, Pappas DJ, Sharp MC. 1997. Communicating bad news. Obstet Gynecol 90:637–639.

Dent KM, Carey JC. 2006. Breaking difficult news in a newborn setting: Down syndrome as a paradigm. Am J Med Genet Part C Semin Med Genet 142C:173–179.

Dube CE, LaMonica A, Boyle W, Fuller B, Burkholder GJ. 2003. Self-assessment of communication skills preparedness: Adult versus pediatric skills. Ambul Pediatr 3:137–141.

Fallowfield L, Jenkins V. 2004. Communicating sad, bad, and difficult news in medicine. Lancet 363:312–319.

Gilbey P. 2010. Qualitative analysis of parents' experience with receiving the news of the detection of their child's hearing loss. Int J Pediatr Otorhinolaryngol 74:265–270.

Girgis A, Sanson-Fisher RW. 1995. Breaking bad news: Consensus guidelines for medical practitioners. J Clin Oncol 13:2449–2456.

Harrison ME, Walling A. 2010. What do we know about giving bad news? A review. Clin Pediatr (Phila) 49:619–626.

MacQueen KM, McLellan E, Kay K, Milstein B. 1998. Codebook development for team-based qualitative analysis. Cult Anthropol Methods 10: 31–36.

McCluskey L, Casarett D, Siderowf A. 2004. Breaking the news: A survey of ALS patients and their caregivers. Amyotroph Lateral Scler Other Motor Neuron Disord 5:131–135.

Merelle ME, Huisman J, Alderden-van der Vecht A, Taat F, Bezemer D, Griffioen RW, Brinkhorst G, Dankert-Roelse JE. 2003. Early versus late diagnosis: Psychological impact on parents of children with cystic fibrosis. Pediatrics 111:346–350.

Mervis CB, Robinson BF, Bertrand J, Morris CA, Klein-Tasman BP, Armstrong SC. 2000. The williams syndrome cognitive profile. Brain Cogn 44:604–628.

Miles MB, Huberman AM. 1994. Qualitative data analysis, 2nd edition. California: Sage Publications. 338 p.

Oshea J, Smith O, O'Marcaigh A, McMahon C, Geoghegan R, Cotter M. 2007. Breaking bad news—Parents' experience of learning that their child has leukaemia. Ir Med J 100:588–590.

Paul CL, Clinton-McHarg T, Sanson-Fisher RW, Douglas H, Webb G. 2009. Are we there yet? The state of the evidence base for guidelines on breaking bad news to cancer patients. Eur J Cancer 45:2960–2966.

Peters KF, Petrill SA. 2011. Development of a scale to assess the background, needs, and expectations of genetic counseling clients. Am J Med Genet Part A 155A:673–683.

Pober BR. 2010. Williams–Beuren syndrome. N Engl J Med 362: 239–252.

Rabow MW, McPhee SJ. 1999. Beyond breaking bad news: How to help patients who suffer. West J Med 171:260–263.

Russ SA, Kuo AA, Poulakis Z, Barker M, Rickards F, Saunders K, Jarman FC, Wake M, Oberklaid F. 2004. Qualitative analysis of parents' experience with early detection of hearing loss. Arch Dis Child 89:353–358.

Sharp MC, Strauss RP, Lorch SC. 1992. Communicating medical bad news: Parents' experiences and preferences. J Pediatr 121:539–546.

Sheets KB, Crissman BG, Feist CD, Sell SL, Johnson LR, Donahue KC, Masser-Frye D, Brookshire GS, Carre AM, Lagrave D, Brasington CK. 2011. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: Recommendations of the National Society of Genetic Counselors. J Genet Couns 20:432–441.

Skotko B. 2005. Mothers of children with Down syndrome reflect on their postnatal support. Pediatrics 115:64–77.

Skotko BG, Capone GT, Kishnani PS, Down Syndrome Diagnosis Study Group. 2009a. Postnatal diagnosis of Down syndrome: Synthesis of the evidence on how best to deliver the news. Pediatrics 124: e751–e758.

Skotko BG, Kishnani PS, Capone GT, Down Syndrome Diagnosis Study Group. 2009b. Prenatal diagnosis of down syndrome: How best to deliver the news. Am J Med Genet Part A 149A:2361–2367.

Tluczek A, Koscik RL, Modaff P, Pfeil D, Rock MJ, Farrell PM, Lifchez C, Freeman ME, Gershan W, Zaleski C, Sullivan B. 2006. Newborn screening for cystic fibrosis: Parents' preferences regarding counseling at the time of infants' sweat test. J Genet Couns 15:277–291.

Uhlmann WR, Schuette JL, Yashar BM, editors. 2009. A guide to genetic counseling, 2nd edition. New Jersey: John Wiley & Sons, Inc. pp. 78–80.