Communicating the postnatal diagnosis of Down’s syndrome: an international call for change

Cambiare il modo di comunicare la diagnosi postnatale di sindrome di Down: una esigenza internazionale

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Summary

Objectives. To analyze the reflections of mothers in the United States and Spain about how they received a postnatal diagnosis of Down’s syndrome (DS) for their children.

Methods. An 11-page survey was mailed to 2,945 mothers in the United States and to 6,125 mothers in Spain.

Results. The majority of mothers reported that their physicians were insufficient and, at times, offensive when delivering a diagnosis of DS. Mothers in both countries claimed that their physicians rarely talked about the positive aspects of DS and did not provide enough up-to-date printed materials or referrals to other parents.

Conclusions. Mothers have now called upon the global community of physicians to improve the way in which postnatal diagnoses are delivered. Specific recommendations are offered herein.

Introduction

Despite the increased usage of prenatal testing, the majority of mothers who have children with Down’s syndrome (DS) do not learn of the diagnosis until after their child is born. Delivering that diagnosis well can challenge even the most practiced physicians, and receiving the news typically alarms the otherwise happy new parents. Until recently, research into this staged drama was limited to mere reflections or very small sample sizes. Now, two large comprehensive studies have been carried out in the United States and Spain, analyzing the reflections of mothers who had received diagnoses of Down’s syndrome for their children. The majority of mothers in the United States reported being frightened or anxious upon learning that their child had Down’s syndrome. They claimed that their physicians talked little about the positive aspects of DS and rarely provided a sufficient amount of up-to-date printed materials or referrals to other parents. Mothers in Spain also reported feelings of anger, guilt, fear, and anxiety upon receiving the diagnosis for their child. They, too, claimed...
that they had received poor information and were not given a sufficient number of phone numbers to other parents who had children with DS. The purpose of this research article is to ask: what similarities and differences exist between the way in which a postnatal diagnosis is delivered in the United States and Spain? And, by extension, what common recommendations can be extracted for the international community of physicians?

Materials and Methods

SAMPLE
For this study, surveys were distributed exclusively to mothers of children with DS (as opposed to other family members) in order to standardize the perspectives of our respondents and to record the sentiments of the person most intimately involved with the pregnancy. This study was implemented in the United States and Spain for multiple reasons: (1) the evolution of the disability parent movement in Spain has mirrored, in many ways, that of the United States; (2) Spain has a national Down’s syndrome parent organization, allowing for access to a large sample size, and (3) the Institute on Community Integration (INICO) in Salamanca, specializes in disability research, could provide technical support for a widespread survey distribution.

In Spain, 6,125 surveys were distributed to mothers on the mailing lists of (1) the Down’s syndrome parent groups associated with national Down’s syndrome organization, Federación Española del Síndrome de Down, (2) the Fundación Síndrome de Down de Cantabria, and (3) the Fundación Síndrome de Down de Madrid. Because there is no national database of families who have children with DS in the United States, 2,945 surveys were distributed through 5 DS parent support groups, chosen for their large membership size and geographic distribution: the Mile High Down’s syndrome Association (Colorado), Triangle Down’s syndrome Network (North Carolina), Massachusetts Down’s syndrome Congress (Massachusetts), Down’s syndrome Association of Los Angeles (California), and the Down’s syndrome Association of Rhode Island (Rhode Island).

SURVEY INSTRUMENT
An 11-page survey instrument, previously described \(^1\) \(^2\) \(^25\) \(^26\), was translated into Spanish and received approval from the Committee on Human Studies at Harvard Medical School. The survey gathered both quantitative and qualitative data with yes/no questions, open-ended questions, and a series of statements for which the mothers were asked to rate their level of agreement on a Likert scale of 1 to 7 (with “7” indicating “strongly agree”, “4” being “neutral”, and “1” meaning “strongly disagree”).

DATA ANALYSES
For this current study, the Kruskal-Wallis test was used to measure the significance between the means of mothers’ responses in Spain and those from the United States. Reported here are the p values. The means and SDs used to calculate this significance were taken from previous papers \(^1\) \(^25\) \(^26\).

Results

RESPONDENTS
Of the 2,945 surveys mailed in the United States, 1,250 (42.4%) were returned. Of these, 43 were excluded because they were completed by fathers, and 81 were excluded because they were returned with an indication that the respondent did not want to or could not complete the questionnaire. An additional 141 were completed by mothers who had received a prenatal diagnosis based on amniocentesis results, and 103 underwent some form of prenatal screening with no confirmatory chronic villus sampling (CVS) or amniocentesis testing. These results are reported elsewhere \(^1\) \(^2\); analyzed here are the remaining 882 responses. Of the 6,125 surveys mailed in Spain, 501 (8.2%) were received. Of these, 29 were excluded because they were completed by fathers or other family members. An additional 5 were completed by mothers who had received a prenatal diagnosis based on amniocentesis results, and 45 were from mothers who had triple screen analyses without further prenatal testing. These results are reported elsewhere \(^25\) \(^26\); analyzed here are the remaining 422 responses.

The majority of the mothers from the United States and Spain were white, Catholic, and college graduates, although the religious identification of women in Spain tended to be more uniform (Tab. I). When her child with DS was born, the average mother was 32.3 years of age in the United States (SD = 5.6) and 33.7 in Spain (SD = 6.61). Approximately 29% of the mothers from the United States and 39% of those from Spain were over the age of 35 when they had their children with Down’s syndrome. Mothers from the United States provided perspective on postnatal medical support from 1964 to 2003 (Fig. 1); mothers from Spain provided commentary from 1978-2003 (Fig. 2). Changes in these opinions have slowly improved over time in both of these countries; the evolution is described in previous reports \(^1\) \(^25\) \(^26\).

REACTIONS TO RECEIVING THE DIAGNOSIS
The response upon learning that a child has DS is similar between countries: Mothers consistently reported being frightened and anxious (Tab. II). While those in the United States tended to have more anxiety (United States: M = 5.8, SD = 1.7; Spain: M = 5.14, SD = 2.33; p < 0.01), the fear level between mothers were statistically similar. In neither country was suicidal ideation common. About half of the mothers in Spain and the
United States had some prior knowledge about DS prior to receiving the diagnosis, whereas the other half did not. In sum, neither group of mothers felt positive upon receiving the diagnosis, with mothers from Spain feeling even more gloom (United States: $M = 2.9$, $SD = 1.9$; Spain: $M = 2.27$, $SD = 1.94$; $p < 0.001$).

**PHYSICIAN BEHAVIORS**

When their physicians explained DS for the first time, mothers in both the United States and Spain reported that these doctors neglected to talk about the positive aspects of the condition (Tab. II) (United States: $M = 3.0$, $SD = 2.1$; Spain: $M = 2.87$, $SD = 2.26$; $p = 0.06$).

About half of the mothers in the United States suggested that their physicians talked about negative aspects of DS (such as, medical complications and cognitive difficulties), and some reported that their pediatricians emphasized these aspects. The same responses could be found in Spain, but the majority of these mothers reported that their physicians did not talk about the positive aspects either, suggesting that they received little to no information at all (Tab. II).

Mothers from both countries expressed a wish that they had learned of the diagnosis as soon as their physicians suspected it. They took note of the “silence” that seemed to surround their child’s birth, sensing that their physicians were trying to avoid telling them something. Mothers reported that this hesitancy by physicians only contributed to mounting fear and anxiety. Many of the mothers also reported, with anger, that they were informed without their partner present and often in a public setting. By contrast, mothers who had their partners present appreciated the added support, and those who had their own rooms were grateful for the privacy to express their emotions, as needed.

In both countries, mothers reported that some physicians are still (even today) using offensive language. In some cases, both in Spain and the United States, physicians intimated that the birth of a child with DS was regrettable, and in other circumstances, pediatricians used antiquated and derogatory vocabulary, such as the “M word.”

Mothers in both countries also felt that they were not provided with enough telephone numbers of other parents who have a child with DS. While the feelings
in Spain were more grim (Tab. II) (United States: M = 2.4, SD = 2.1; Spain: M = 1.84, SD = 1.78; p < 0.001), the mothers in the United States rarely reported that they had received a referral. Those who did, valued the educational materials, networking, and social opportunities provided by these parent groups.

**Printed Materials**

Nearly all mothers in both the United States and Spain reported that their physicians did not provide them with enough up-to-date printed materials on DS, with the mothers in Spain reporting even less (Tab. II) (United States: M = 2.4, SD = 2.0; Spain: M = 1.79, SD = 1.66; p < 0.001). While mothers in the United States reported that the little information that they did receive was easy to read and comprehend, the literature did not provide a balanced approach of the positive and negative aspects of DS (Tab. II).

**Discussion**

Mothers in both the United States and Spain rarely reported that the birth of their child with DS was a positive experience. They were frightened and anxious upon receiving the diagnosis and frustrated that their physicians failed to explain the positive aspects of DS, provide up-to-date printed materials, or offer the contact information for local parent support groups. While statistical differences emerged between Spain and United States, the differences ranged between terrible and just bad (Tab. II). Mothers in Spain tended to respond with more dissatisfaction for their physicians. This could be explained by (1) cultural differences between the two countries, (2) differences in physician practices in Spain and the United States, or (3) both. To the extent that national laws reflect the consent of the people, Spain and the United States have different cultural respects for individuals with Down’s syndrome. Spanish laws assume that mothers would want to terminate a child with Down’s syndrome if diagnosed prenatally. According to Articles 144 and 145 of the Spanish penal code (Organic Law 10/1995, November 23, 1995), a woman is legally allowed to terminate a pregnancy only if (1) her life is at risk by continuing the pregnancy, (2) the pregnancy is the result of rape, or (3) the fetus would be born with grave physical or psychological defects. The reasoning for the third case, as outlined in the law, is to “avoid the birth of persons with high probability to be unhappy and with a subminimal quality of life”. The purpose is also to “avoid disturbance of the parents and the family atmosphere”. Article 2 of the Ministerial Notice of Health and Consumption (July 31, 1985) outlines which “physical and psychological defects” qualify; among those included

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**Tab. II. Comparison Between Mothers’ Responses in the United States and Spain to Postnatal Diagnosis.**

<table>
<thead>
<tr>
<th>Responses</th>
<th>United States</th>
<th>Spain</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>When I learned that my child had DS</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I had no prior knowledge about DS</td>
<td>4.1</td>
<td>4.40</td>
<td>0.08</td>
</tr>
<tr>
<td>I felt positive</td>
<td>2.9</td>
<td>2.27</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>I had suicidal thoughts</td>
<td>1.6</td>
<td>1.73</td>
<td>0.51</td>
</tr>
<tr>
<td>I felt anxious</td>
<td>5.8</td>
<td>5.14</td>
<td>&lt; 0.01</td>
</tr>
<tr>
<td>I felt frightened</td>
<td>5.5</td>
<td>5.41</td>
<td>0.30</td>
</tr>
<tr>
<td><strong>Physician Behavior</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>My physician provided me with enough telephone numbers of parents of a child with DS</td>
<td>2.4</td>
<td>1.84</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>My physician provided me with enough up-to-date printed material on DS</td>
<td>2.4</td>
<td>1.79</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>My physician pitied me</td>
<td>3.7</td>
<td>3.43</td>
<td>0.03</td>
</tr>
<tr>
<td>My physician emphasized the negative aspects of DS</td>
<td>3.7</td>
<td>2.71</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>My physician told me about the negative aspects of DS</td>
<td>4.1</td>
<td>2.95</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>My physician emphasized the positive aspects of DS</td>
<td>3.0</td>
<td>2.57</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>My physician told me about the positive aspects of DS</td>
<td>3.0</td>
<td>2.87</td>
<td>0.06</td>
</tr>
<tr>
<td>The printed materials that I received from my physician</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>were easy to read and comprehend</td>
<td>4.0</td>
<td>2.81</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>were helpful for understanding DS</td>
<td>3.7</td>
<td>2.62</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>emphasized the positive aspects of DS</td>
<td>2.9</td>
<td>1.96</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>emphasized the negative aspects of DS</td>
<td>3.1</td>
<td>1.76</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>provided equal mixture about the positive and negative aspects</td>
<td>3.0</td>
<td>1.99</td>
<td>&lt; 0.001</td>
</tr>
</tbody>
</table>

* N = 882. † N = 422. ‡ Mothers were asked to rate their level of agreement with the statements on a Likert scale of 1 to 7 (with 1 indicating strongly disagree, 4 neutral, and 7 strongly agree).
The person to deliver the diagnosis should be a physician. When a mother learns about the diagnosis of DS for her child, she is certain to have questions and concerns. Mothers who received the news from nurses, lactation specialists, and, in some cases, volunteers did not feel that they received sufficient information or support;

2) **The physician should deliver the diagnosis with both parents together, whenever possible.** In many cases, pediatricians or neonatologists inform the father about the diagnosis of DS, obligating him to share the news with the mother. Mothers have insisted that it is both unfair and inappropriate to ask fathers to fulfill such a role since they are often just as shocked and unfamiliar with DS as the mothers. More commonly, mothers are informed alone without the comforting support of their partner. Frightened and anxious, they report intense difficulty in repeating to the fathers what a physician had explained to them. In short, the physicians should wait, within reason, for the father to be present when delivering the diagnosis. If the father of the child is not readily available, the physician should explain the news to the mother and offer to review everything again when the father returns;

3) **The physician should deliver the diagnosis in a private setting.** Receiving the diagnosis for the first time can be emotionally taxing for some, if not most, mothers. Those mothers who received the unsuspecting diagnosis with other family members present or with a hospital roommate nearby reported intense anger for their physician. Mothers ask that physicians bring the mother and father into a private room to explain DS. If possible, a private room should also be offered after the diagnosis is conveyed so that the couple has space to explore their deep emotions;

4) **The physician should convey the diagnosis as soon as he or she suspects it.** One of the most common sources of frustration – and anger – among mothers was a period of “silence” that seemed to loom around the birth of their child. Mothers felt avoided, as though they could not get any answers from physicians or the medical staff. The hesitancy from doctors to convey their suspicions contributed immensely to the anxiety surrounding the day. Rather than waiting for the diagnosis to be confirmed with a genetic karyotype, a physician should share with parents his or her suspicions about DS and what steps should be taken to confirm the diagnosis. Mothers said they valued transparency in the decision-making process. Of course, a physician can also inappropriately inform a mother too soon. Some mothers were informed while episiotomies were being sutured. In sum, a physician should convey the diagnosis when he or she suspects it, but not until a mother has settled from the birthing process itself;

5) **The physician should use sensitive language.** The tone of the conversation is frequently established by the first words used by the physician. Instead of saying “I’m sorry” or “I have some bad news to share,” physicians should first congratulate parents on the birth of their child. Mothers emphasized that having a child with DS is not a tragedy and should not be introduced as one. Also, the “M word” is wholly derogatory and no longer deserves a place in medical nomenclature;

6) **The physician should include the positive aspects of DS when explaining the diagnosis.** Mothers would like to receive a balanced and accurate portrait of DS. Many reported that their physicians failed to
mention any of the positive possibilities and realities for people living with DS today. Mothers considered it the responsibility of physicians to stay informed of the educational and social potentials of children with DS. Knowing the scientific literature alone is not sufficient. Physicians can find useful information about the achievements of people with Down’s syndrome in Common Threads: Celebrating Life with Down Syndrome 27, Down Syndrome: Visions for the 21st Century 28, and Down Syndrome: A Promising Future, Together 29.

7) The physician should not share his or her personal opinion. Mothers requested that physicians offer sound medical advice based on the most up-to-date information. Unless specifically requested by the mother, personal opinions are not warranted. Some mothers felt that their decision to have a child with DS was being questioned by their physicians;

8) Parents should be provided with up-to-date printed material. When the diagnosis is being shared, physicians should provide up-to-date printed material on DS. Many mothers suggested the books: Babies with Down Syndrome: A New Parent’s Guide 30, Common Threads: Celebrating Life with Down Syndrome 27; Life as We Know It: A Father, a Family, and an Exceptional Child 31; Count Us In: Growing Up with Down Syndrome 32; and A Parent’s Guide to Down Syndrome: Toward a Brighter Future 33. When printed materials cannot be provided for financial reasons, a reference list of the most current literature should be substituted. One such list can be found on the web page for the National Down Syndrome Congress in the United States (http://www.ndsccenter.org/resources/print.htm);

9) The physician should offer the contact information of a local support group. Mothers repeatedly stated that their local DS parent support groups were of invaluable help, especially during the first few years of a child’s life. Every physician should know the contact information of the local DS support group in their area. Oftentimes, a national DS parent organization, like the National Down Syndrome Society in the United States, maintains a directory of these groups online for easy and ready access.

As with all retrospective studies, this research is subject to recall bias. The mothers answered the survey with an average of 11 years of hindsight in the United States and 10 years in Spain. Because of the clarity in which they responded, however, receiving an unexpected diagnosis might represent a true flashbulb memory – complete, accurate, and immune to forgetfulness 34-36. This study is also subject to selection bias. Only mothers who were members of DS support groups were sampled. It is possible that only mothers who had the most difficult experiences coping with the birth of their children with DS would enroll in a DS support group and only those who had particularly stressful birthing experiences would choose to respond to our survey. Experience with DS support groups suggests that this is not the case in either country, and since there are no national databases of families who have children with DS, using support groups remains the most robust distribution mechanism. The response rate from mothers in Spain was also low, compared to that of the United States. Possible explanations include: (1) mothers in Spain are not as accustomed to receiving research questionnaires in the mail as are the families in the United States, (2) mothers from the United States were more apt to respond since the researcher was their citizen while mothers from Spain were reluctant to respond to someone from another country, or (3) a combination thereof.

Between Spain and the United States, however, more than 1,300 mothers have now called for a reform in the way physicians deliver a diagnosis of Down’s syndrome. Previous research in England, Scotland, Ireland, Sweden, and Australia also suggested that women are strongly dissatisfied with the way in which the diagnosis of DS is conveyed 34-6 7 11 12 14 17 19 21 23. Clearly, the worldwide community of pediatricians and neonatologists have now received a mandate for improvement. Mothers are asking that physicians engage in an international dialogue on how best to implement their recommendations. While the communication of such sensitive diagnoses will continue to remain challenging, one thing remains clear: the time for change is now. Let us begin.

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Communicating Diagnosis of Down Syndrome

References


