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## LATINAS AND THE TRADITIONAL GENETIC COUNSELING MODEL: A QUALITATIVE STUDY

Stephanie Thompson

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LATINAS AND THE TRADITIONAL GENETIC COUNSELING MODEL:

A QUALITATIVE STUDY

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The traditional genetic counseling model reflects an individualized counseling session that includes the presentation of information about genes, chromosomes, personalized risk assessment, and genetic testing and screening options. Counselors are challenged to balance providing educational information with discussion of implications of this information in an allotted amount of time. The aim of this study was to explore the perceptions of pregnant Latinas on the benefits and limitations of the traditional prenatal genetic counseling model and to determine the specific preferences for receiving prenatal genetic counseling. Data were collected through focus groups and one-on-one, semi-structured interviews of twenty-five Spanish speaking Latinas who received genetic counseling during their current pregnancy. Their responses were evaluated using thematic analysis to identify major themes in participant responses by utilizing a grounded theory approach. Several themes were identified including an overall satisfaction with their prenatal genetic counseling appointment, desire for a healthy baby, peace of mind following their appointment, no desire for invasive testing, and faith in God. Several participants also stated a preference for group genetic counseling over the traditional individual genetic counseling model. Our data indicate that Latinas value the information presented at prenatal genetic counseling appointments despite disinterest in pursuing genetic testing or screening and suggest that group prenatal genetic counseling may be an effective alternative to the traditional genetic counseling model in the Latina population.

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## **Introduction**

The traditional genetic counseling model comprises an individualized counseling session that includes the presentation of information about genes, chromosomes, personalized risk assessment, and genetic testing and screening options, facilitation of decision making about genetic testing and/or screening, and provision of support to the patient (National Society of Genetic Counselors 2005). A key role of genetic counselors is to educate their patients about genetic conditions, genetic testing, risk assessment, and treatment options (O'Shea et al. 2011). During an initial genetic counseling visit, a large amount of information about genes, chromosomes, inheritance, and screening and testing options is provided; however, most patients may be able to recall less than half of that information (Aldaba et al. 2012). Counselors are challenged to balance the provision of enough basic genetic information to ensure clients' understanding of the genetic condition in question with a personalized discussion of what this information means to them.

While much of an initial genetic counseling visit focuses on providing patients with medical and genetic information about a particular condition, many patients are primarily concerned with understanding their personal risk of acquiring or having a genetic condition (Salemink et al. 2013). The comprehension of information provided during a genetic counseling session may be limited by the knowledge, understanding, and perception of genetics that individuals have prior to the session. Since time is limited in genetic counseling sessions, increasing patient knowledge prior to the session could result in more focused attention devoted to discussing patient concerns. Using computer-based information programs and informational websites can increase knowledge and recall of basic genetic information including inheritance and genetic testing as well as understanding of

what to expect in the counseling session (Green et al. 2004; Albada et al. 2011). However, a limitation of web-based information delivery is the potential lack of access to computers and the internet, particularly among underserved patient populations. While offering less opportunity for interactivity, informational brochures have been shown to increase the knowledge level of genetic testing and screening options of patients referred for prenatal genetic counseling (Dahl et al. 2006b). Since the majority of patients at a prenatal genetic counseling visit are knowledgeable about the procedural aspects of screening and testing, but fewer are knowledgeable about the interpretation or clinical utility of the results of these tests, these brochures have been most effective when they present information on procedures, testing, and interpretation of results (Dahl et al. 2006a).

Beyond web-based interventions and informational brochures, the use of a one-on-one educational genetics tutorial individually tailored to patients of low-socioeconomic background was helpful in increasing the knowledge level of these individuals (Sim et al. 2011). In this study, non-English-speaking participants who were scheduled for prenatal genetic counseling received a one-on-one tutorial session presented by a doctor in the participant's native language which included information on basic genetics, what to expect at the genetic counseling appointment, and testing options preceding their genetic counseling appointment. While an individualized genetics tutorial may not be feasible to implement at many clinics, presentations of genetic information to a group of individuals in their preferred language might be a more practical option.

One model for group prenatal care is the Centering Pregnancy Model (CPM), in which patients are grouped by gestational age and are given educational information about different aspects of pregnancy. This model enables group education as well as the



opportunity to build a support network with other pregnant women (Rising 1998). Women who participated in CPM reported higher levels of pregnancy knowledge compared to those who received traditional prenatal care (Baldwin 2006). Hispanic women who participated in CPM also reported high levels of satisfaction with this model (Robertson et al. 2009). These findings suggest that integrating a genetic counseling presentation into CPM may be a feasible option to increase patient knowledge.

The aim of this study was to explore the perceptions of pregnant Latinas on the benefits and limitations of the traditional prenatal genetic counseling model and to determine the specific preferences for receiving prenatal genetic counseling in the Latina population. Specifically, we assessed the preferred way to present educational information about genes, chromosomes, age related risks for a pregnancy to have chromosome abnormalities, and testing and screening options as well as the preferred structure to use in a counseling appointment.

## **Methods**

### Study Design

We conducted focus groups and semi-structured interviews with pregnant Latinas who received prenatal genetic counseling during their current pregnancy and for whom Spanish was their primary or preferred language. The study protocol was approved by the Committee for the Protection of Human Subjects at the University of Texas Health Science Center at Houston and at the Harris Health System (HSC-MS-13-0485).

## Study Population

Eligible women included those who were age 18 or older, who were seen at Lyndon B. Johnson General Hospital in Houston, Texas, for prenatal care, and who were referred for genetic counseling due to a primary indication of advanced maternal age and/or abnormal maternal serum screen (MSS) indicating an increased risk for Down syndrome or Trisomy 18. Exclusion criteria included having a positive MSS indicating an increased risk for an open neural tube defect (ONTD), having a high resolution ultrasound that detected structural abnormalities or soft marker signs, and all other indications for genetic counseling, such as a family history of a birth defect or genetic condition.

Eligible women were identified from a password-protected electronic database of prenatal patients seen for genetic counseling between August 2013 and February 2014 and were invited to participate in the study via a telephone call from AH, SN, or ST.

Demographic and personal information including age, gestational age, number of children, previous experience(s) with genetic counseling, education level, religion, marital status, annual income, health insurance information, and length of residence in the United States was obtained from participants' medical records, when possible, and was supplemented by patient report.

## Data Collection

Data were collected through focus groups and one-on-one, semi-structured interviews. Participants who could not attend a focus group were invited to complete a phone interview. The focus groups and interviews were all conducted by the same facilitator (ST) with the assistance of a trained Spanish interpreter using an interview guide

that encouraged discussion of participants' prenatal genetic counseling experiences. The interview guide included several yes/no and other descriptive, open-ended questions to assess knowledge of genetic counseling prior to the appointment, expectations for the genetic counseling appointment, satisfaction with the appointment itself and with the way information was presented in the appointment, and preferences for alternative models of counseling including the possibility of group counseling sessions. Focus group sessions and interviews were audio-taped and transcribed by the interviewer, and analyzed by ST and JL to identify common themes in member responses.

Thematic analysis was used to identify major themes in participant responses, using a grounded theory approach (Braun & Clark 2006; Grubs & Piantanida 2010). In this approach, transcripts are coded into nodes and grouped into categories to reflect similar themes. ST and JL independently reviewed three transcripts and compared identified themes in order to determine consistency in coding and to limit bias. ST coded the remainder of the transcripts and the analyses were reviewed by JL, SP, and AH.

## **Results**

Of 111 eligible women who were invited to participate in the study, 60 were reached by phone and 39 provided verbal consent. Ten women participated in three focus groups held between November 2013 and February 2014. One woman completed a one-on-one interview in person. Fourteen women participated in phone interviews, and indicated that problems with transportation, work schedule, or child care precluded their participation in scheduled focus group sessions. The remaining fourteen individuals who provided consent did not attend their scheduled focus group or could not be reached for a phone interview.

## Demographics

Demographic and clinical characteristics of participants are summarized in Table 1. Most were multigravida and had between 1-5 children. None of the participants had higher than a secondary education, and average length of residence in the U.S. was 12 years. The majority of participants (76%) had Children's Health Insurance Program (CHIP) perinatal coverage while twenty percent received another form of government funded financial aid.

Most participants (88%) received individual prenatal care, while 12% (3/25) participated in CPM. Only one participant received genetic counseling prior to her current pregnancy. Of note, two participants had a child with Down syndrome.

Sixty-eight percent received genetic counseling from a Spanish-speaking genetic counselor, and the remainder received genetic counseling with the assistance of a professional medical interpreter. Nearly all were offered amniocentesis, ultrasound, and Non-Invasive Prenatal Testing (NIPT) at their genetic counseling appointment, and most elected to proceed with ultrasound only.

## Prior Knowledge and Expectations of Genetic Counseling Appointment

Sixty-eight percent of participants (n=17) stated that they were aware that they were having genetic counseling prior to their appointment. Twenty percent (n=5) stated age would be discussed, thirty-six percent (n=9) expected they would discuss the risk for a problem with the pregnancy, including Down syndrome, and sixteen percent (n=4) cited genetic testing as the reason for the appointment.

Table 1: Demographic Characteristics of Study Participants					
Characteristic	N=25				
	Mean	SD	Range	<i>n</i>	%
Age at delivery	38	2	35-44		
Gestational age (weeks) at time of genetic counseling	20	3	14-25		
Gestational age (weeks) at time of focus group/interview	30	4	22-38		
Time (weeks) between genetic counseling appointment and focus group/interview	10	3	5-15		
Years lived in US	12	7	0.5-23		
Approximate annual income	22,600	9,900	9,600-41,600		
<b>Education</b>					
No formal education				1	4
Grades 1-6				7	28
Grades 7-9				8	32
Grades 10-12				9	36
<b>Marital Status</b>					
Single				5	20
Partner				1	4
Married				19	76
<b>Religion</b>					
Catholic				24	96
None				1	4

Sixty-eight percent of study participants (n=17) were aware of prenatal testing and screening prior to their genetic counseling appointment. Thirteen of these individuals received information about these options from a medical professional, while others learned about testing and screening from friends and family members. Most were aware that these tests and screens are available and that they are used to evaluate the pregnancy for conditions such as Down syndrome.

When asked what concerns or questions participants had prior to their genetic counseling appointment over half of the respondents stated they had no concerns or questions prior to their appointment. Some women stated they were worried about their age and the risks associated with pregnancy for a woman over the age of thirty-five, and others reported concerns about pregnancy-specific problems or conditions.

#### Evaluation of Prenatal Genetic Counseling Experience

Overall, participants reported being satisfied with their genetic counseling appointment. All respondents reported that all of their questions were answered during their session. All reported understanding the information presented during the session and stated they had enough information to make a decision about testing and screening options available to them. One participant expressed:

“I had several questions and at that point they helped me, they answered my questions and told me what could happen. They offered if I wanted to do the studies. That it was my decision whether I wanted to do it or not, and I have informed myself, and I know people that have babies that have problems, so I’m not scared of it, and I told them that I did not want to do the rest of the studies.”

Sixteen participants stated that they would have found it helpful to have information written down to take with them after their genetic counseling appointment, and twenty respondents would have preferred to have information before their appointment about what to expect at the appointment and/or the risks and testing/screening to be discussed. As one participant stated:

“I wish this appointment had happened sooner...until now I did not know that we could go to counseling. If I had known I would have gone earlier...because I already have four babies and had I known I would have gone to more teachings to know more about babies, how do they come and all that.”

### *Faith in God*

About one third of participants referenced their religious faith in regard to decisions about prenatal testing or perceptions of testing outcomes. Several stated that they declined invasive testing because of a desire to leave the outcome in God’s hands. Two participants challenged the veracity of age-related risk for a chromosome abnormality by stating that genetic conditions are a result of God’s will. Several other women who participated also stated they had faith that everything would be fine with their baby.

### *No desire for invasive testing*

No participants elected to proceed with any invasive testing and six women (24%) elected NIPT. Of note, participants who elected to proceed with NIPT at the study site had to travel to another laboratory to have their blood drawn. One individual mentioned this as the least helpful part of her genetic counseling appointment. However, most expressed appreciation for the information received at the genetic counseling appointment. Several women referred to the amniocentesis procedure as dangerous, risky, or scary. One woman

declined amniocentesis, but proceeded with NIPT because she perceived that NIPT was less risky yet informative regarding the health of her baby. Some stated that they had decided not to have any prenatal testing or screening before the genetic counseling appointment, but followed through with the appointment nonetheless.

#### *Peace of mind following the appointment*

Responses regarding what women perceived as the most helpful part of their genetic counseling appointment varied and included hearing explanations of chromosomes and Down syndrome plus age-related risks for a chromosome abnormality in pregnancy, and answering questions about family history. Others noted that receiving good news, such as learning the risk for a chromosome abnormality was lower than expected, was also one of the most helpful parts of the session, and some reported feeling calmer or more secure at the end of their session. Four respondents reported feeling worried, nervous, or sad at the beginning of the session, but were more reassured at the end.

#### Alternative Models for Prenatal Genetic Counseling

About half of participants stated a preference for an individual genetic counseling appointment and the remainder preferred a group session or stated no preference for the structure of a genetic counseling appointment. Respondents cited privacy and individually focused attention of the genetic counselor toward patients as advantages of individual sessions; stated advantages of group sessions included social support and gaining knowledge from others in similar situations. The majority reported being comfortable talking about genetic counseling in front of other women.



Table 2: Summary of Themes in Participant Responses

<u>Theme</u>	<u>Selected Responses from Participants</u>
Desire for a healthy baby	<p>“More than anything, I needed to know, I wanted to know...if my baby is fine or if something is happening... There’s just the questions.”</p> <p>“I would like to go and be told that my baby’s going to be born 100% well.”</p>
Faith in God	<p>“They offered me all types of testing and...I declined everything because I’m just going to leave it in the hands of God and I really don’t want anything done.”</p> <p>“She did explain to me all the different types of testing that there is and if I wanted to get any, and I told her that I didn’t want any and if I do have, whoever has a child like that, it’s because that’s the way that God wanted it. It’s not a matter of the age or anything and there’s people who even have had children like that who don’t have a history in the family of having something like that.”</p>
No desire for invasive testing	<p>“It’s not that it was less helpful, it’s just that it makes you a little bit scared when they tell you about the study when they use the needle. They say that the baby could be at risk or the mom. Yes, that was a little scary when they tell you about the needle. It’s just more scary that something might happen to the baby.”</p> <p>“I already knew that I was not going to have any testing done, but...there’s people that don’t know and they want to hear. It didn’t cost me anything to stay and listen to what they were saying.”</p>
Peace of mind following appointment	<p>“She [the counselor] actually left me with a peace of mind... basically she told me that there are risks, but they are small risks and as a mother that helped me get more calm.”</p> <p>“That day I went to the meeting with my husband and both of us came out of there with a better peace of mind feeling more comfortable. We were both conscious that having a baby at this age there was a risk, but after they asked for everything... we were a little more calm afterwards when we left.”</p>

Table 3: Selected responses about structure of genetic counseling appointment

Individual Counseling

“Well because...when you’re in one-on-one with the genetic counselor you feel more comfortable asking all the questions that you have and...you can feel shy or...maybe you’re afraid to ask something because you don’t know what the other ones are gonna say because...it’s...personal and...I would feel better...if it’s something more private.”

Group Counseling

“In group...it’s better because I can hear the other cases of the other people and it seems less frightening.”

“With the other women I can relate to them and kind of feel like we cohabitate in the same situation and...ask each other how we feel, how’s our pregnancy going, things like that.”

## **Discussion**

Overall, participants were satisfied and had very positive responses regarding their one-on-one prenatal genetic counseling appointment. Participant responses indicated that the objectives of the genetic counseling process were accomplished. Several themes were identified as contributing to participant satisfaction with the genetic counseling appointment, including the desire for a healthy baby, peace of mind following the genetic counseling appointment, faith in God regarding pregnancy and prenatal testing outcomes, and lack of preference for invasive testing. Participants seemed to be well informed of their personal risks for having a baby with a chromosome abnormality and of the available testing and screening options, and their responses indicated that they felt autonomy in decision-making about testing and screening. Although none proceeded with invasive genetic testing, participants expressed appreciation for the information they received, and responses indicated that they perceived value in having the information. Overall, participants expressed feeling calm following their genetic counseling appointment based on the information they received. These findings suggest that a traditional prenatal genetic counseling model may be effective from our participants' perspectives.

Although no questions in the interview guide referenced specifically faith or spirituality, a number of women discussed their faith in God as an important factor in decision making about prenatal testing in regard to their pregnancy in general. This suggestion is consistent with previous studies reporting that Latinas may decline invasive prenatal testing procedures in favor of leaving the outcome to God when termination is not being considered (Hunt & Voogd 2005). While faith in God was one reason some of the women we interviewed declined invasive testing, others cited different reasons. Other

research exploring the role of religion and spirituality in decision making for prenatal testing in Latinas suggests risks associated with the amniocentesis procedure itself contribute to declining that test (Seth et al. 2011). Concern about the potential risks of amniocentesis was also represented in our participants' responses.

Part of the decision making process for prenatal testing and screening considers whether or not termination is an option a woman would consider if her pregnancy had a chromosome abnormality. None of the women participating in our study expressed an interest in pursuing termination if a chromosome abnormality was detected prenatally. It is important to consider that the majority of the women in our study were Catholic, as that might influence this decision. Nonetheless, most of the respondents indicated that the information presented on this topic was of value to hear. This suggests that prenatal genetic counseling may still be beneficial for individuals who are in the third trimester of pregnancy when testing and screening options are limited.

In our study, none of the participants requested their genetic counseling appointment and all were referred by their physicians. However, as genetic counseling continues to gain popularity, the demand for genetic counseling will likely increase.

To inform future efforts to maximize efficiency of prenatal genetic counseling services, we found that about half of the women in our study expressed a preference for group counseling rather than individual counseling, or stated no preference. Group prenatal genetic counseling sessions for routine indications such as advanced maternal age could provide an environment that normalizes the process. Advantages to group counseling voiced by our participants included the value of having the perspectives of other individuals

in a similar situation to their own and the opportunity to learn from others. Many also expressed comfort with discussing genetic counseling information in front of other women. While utilizing a group counseling model may introduce greater efficiency in the provision of prenatal genetic counseling services, it is important to note that about half of our sample indicated a preference for individual counseling due to privacy concerns and a desire for one-on-one attention from a genetic counselor. Future efforts to explore the provision of group versus individual prenatal genetic counseling for Latinas may need to take into consideration personal preferences for appointment formats.

### Study Limitations

Although a strength of this study is its focus on Latinas whose preferred or primary spoken language was Spanish, one limitation was the use of a translator to directly interpret participants' responses; thus we were unable to analyze responses in participants' spoken language. Focus groups and one-on-one interviews were conducted with the assistance of a professional Spanish interpreter and analysis was based on the English translation of participant responses. Because we excluded individuals with ultrasound abnormalities, soft markers on ultrasound, and other indications for genetic counseling such as a family history of a genetic condition, our study did not include the perspectives of this group of women, which may limit the generalizability of our findings. All participants were over the age of 35, which also may limit our ability to generalize our findings to younger women.

### Research Recommendations

Additional research could also include a comparison analysis of satisfaction and knowledge between women who received prenatal counseling in a group setting and those

who received traditional genetic counseling. Another consideration would be the cost effectiveness of group counseling versus individual counseling.

### Practice Implications

Several suggestions from participants could be implemented into a genetic counseling session with minimal effort. The majority of respondents would have liked to have information on what to expect at their appointment before arriving, and more than half of our participants stated they would have preferred to have some information on testing and screening prior to their genetic counseling appointment. A patient-friendly brochure could be created for referring providers to provide to their patients when referring them for genetic counseling.

Many patients also reported a desire to have information that was discussed during the genetic counseling appointment in writing to take home with them. Some would want this information for their reference, while others would plan to share it with family members. Creating a patient-friendly education document in the patient's first language could enhance the prenatal genetic counseling appointment.

Based on participant responses, obtaining information about personalized risks for the pregnancy seems to be more important to patients than uptake of testing. Therefore, it might be beneficial for women to receive genetic counseling even in the third trimester when testing options are limited. This could be especially helpful for women who plan to have more children after their current pregnancy. This would likely increase the number of patients scheduled for genetic counseling, which could provide an opportunity to utilize group genetic counseling.

Given the number of women in the study who expressed an interest in group prenatal genetic counseling, it could be advantageous to explore group counseling as an option, especially for Latinas. In order to address the issue of privacy, patients who elect group genetic counseling could be given the option to schedule an individual appointment following the initial session. Adding group genetic counseling to a clinic schedule could potentially be more time and cost effective for routine indications such as advanced maternal age.

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## VITA

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