

**Parental perception of pediatric clinical exome sequencing in a
Latino population**

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Abstract

Clinical exome sequencing (CES) is an established method for genetic diagnosis that is commonly used in clinical practices. Studies examining the CES experience for families have primarily been non-Latino white participants who speak English. To begin to address how these experiences may differ in other populations, we conducted in-depth semi-structured interviews. Interviews were completed with 22 Latino parents (14 in English; 8 in Spanish) of children who had pediatric CES at Columbia University Medical Center and received results within three to 20 months of the interview. We also measured acculturation with a standardized scale. Similar to prior studies, parents reported varied emotional reactions to their child's results. Parents largely understood their child's CES results though were not always able to correctly recall recurrence risk. The majority of participants reported feelings of anxiety while waiting for CES results. Some parents, particularly those not fluent in English with lower acculturation scores, reported language barriers to their understanding of the testing and their child's care. Faith appeared to be a positive coping mechanism that was more frequently reported by participants with lower acculturation scores. Our findings are consistent with many of the previous studies of parental experience of CES but highlight some key findings that warrant further study and potentially inform practice. Larger studies should explore potential associations between acculturation and parental understanding or emotional response to results. The minimal language barriers reported by Spanish-speaking patients reiterates the importance of a Spanish-speaking provider, when possible, and bilingual resources.

Key Words: clinical exome sequencing, Latinos, genetic counseling

Introduction

Clinical exome sequencing (CES) is an established method for genetic diagnosis. Implementation of CES as a first or second tier test provides an earlier diagnosis, eliminates the cost of traditional testing, and increases the diagnostic yield compared to the standard of care (Valencia et al. 2015; Kuperberg et al., 2016; Nolan & Carlson, 2016; Vissers et al., 2017).

Research to date has demonstrated mixed motivations for obtaining CES, including a sense of duty, lack of other options, hope for a diagnosis and the potential for it to guide care and treatment (Rosell et al., 2016; Wynn et al., 2018). Many parents report feeling that any information at all could improve their quality of life and sense of empowerment (Clift et al., 2015; Wynn et al., 2018), though previous failed attempts at diagnosis can reduce expectations of a diagnosis for parents of children getting CES (Rosell et al., 2016). While a hope for a diagnosis was often reported as motivation for testing, parents also feared receiving a diagnosis that indicated a shortened lifespan, and implications for their insurance (Rosell et al., 2016, Clift et al., 2015).

Similar to motivations for testing, reactions to test results are often mixed. Parents felt frustrated and sometimes depressed by their CES results (Rosell et al., 2016, Wynn et al., 2018). Parents who received a definitive diagnosis for their child experienced relief and appreciation, while others experienced a loss of hope by the lack of therapeutic options or isolation by the rarity of the diagnosis. Parents who received a possible diagnosis mostly experienced hopefulness as they continued to search for a diagnosis, or acceptance at not receiving a definitive diagnosis. However, those who received no diagnosis either experienced relief that nothing was found, or concern and disappointment as they still sought a reason for their child's symptoms (Krabbenborg et al 2016; Rosell et al., 2016).

Parental reaction may also have effects on comprehension of genetic information. In instances where the emotional impact is high, parents are less likely to process information which can result in a feeling of being overwhelmed. Parental experiences are perceived as positive when a dialogue is present between the geneticist and the parent without excessive jargon, emotional support is provided, and there is inclusion of medical management suggestions (Ashtiani, Makela, Carrion, & Austin, 2014; Browner, Preloran, Casado, Bass, & Walker, 2003; Walser et al., 2017).

With adequate pre-test counseling, parents have been able to accurately explain their results and explain the limitations of the CES (Rosell et al., 2016). Parents often request variants of uncertain significance (VUS) returned, although there may be confusion about their significance (Rosell et al., 2016, Wynn et al., 2018). In terms of secondary findings, parents may be willing to learn about them because they believe it may help them contribute to research and it may inform decisions about medical management (Clift et al., 2015).

A study by Wynn et al. found that most parents were able to correctly interpret their child's CES results, and most discrepancies were due to the clinician's reinterpretation of the laboratory report or the return of negative or uncertain results. Timing of testing also impacted parental perception, with a lower positive impact associated with increasing age of the child. The overall experience was perceived as positive in parents who interpreted the CES results as positive. However, one major limitation noted in this study was that most of the participants identified as white, non-Latino (2018).

The ethical principle of justice in research dictates that all groups of people are invited to participate in research. The US population of Latinos is about 17.8% and is projected to increase to 28.6% by 2060 (United State Census Bureau, 2017). Despite this, most research on genetic

disease susceptibility has been done on non-Latino white individuals (Kinney, Gammon, Coxworth, Simonsen, & Arce-Laretta, 2010; Wynn et al., 2018). Members of minority groups are also less aware of genetic testing as compared to Caucasians (Hann et al., 2017).

The Latino population remains underrepresented in genetic research. Language barriers, discrimination, low numeracy and literacy, higher rates of being uninsured, lower household incomes, and a higher rate of poverty are some of the barriers to implementing genetics into their healthcare (Kamara, Weil, Youngblom, Guerra, & Joseph, 2018).

Ponce et al. (2006) found that English language skills correlated with better health outcomes even when race, age, and income are controlled for. Less proficiency in English corresponds with lower health literacy and fewer regular visits to the doctors (Bruce et al., 2014; Kamara et al., 2018). In addition, physicians view non-English communication as a barrier to informed consent or interest in medical care (Bruce et al., 2014). Patients required to assess themselves in a language they are not fluent in report themselves as more unhappy, less confident, and even less intelligent in the subject matter (Wallin & Ahlström, 2006). Research into the attitudes and beliefs of Spanish-speaking people is important to developing public awareness among these people (Kinney et al., 2010).

Healthcare disparities can alter disease course in minorities. Breast cancer is detected at a later stage in Latina women, even while controlling for education, socioeconomic status, and detection method (Lantz et al., 2006). This discrepancy is reduced when controlling for detection method, but only in Caucasian and African American women and not Latinas, suggesting that cultural differences unique to Latinas may outweigh racial differences (Lantz et al., 2006).

When communicating genetic testing results, Latino patients request that the information be disclosed slowly and without technical jargon (Browner et al., 2003; Kamara et al., 2018).

Better outcomes have been demonstrated for patients who speak English when receiving a genetic diagnosis (Krabbenborg et al., 2016). In addition, people coming from a lower socioeconomic status (SES) have been shown to be accustomed to addressing immediate health concerns and less concerned with planning for future events (Kamara et al., 2018). Latinos are sometimes used to being given a much more paternalistic treatment by their doctors and may see non-directiveness as indication that something is not important (Browner et al., 2003; Kamara et al., 2018; Vadaparampil et al., 2010).

Not only can there be communication barriers between providers and the Latino population, but access to genetic testing may be limited as well. Fewer genetic tests are ordered and fewer referrals to genetic counseling are made by physicians who serve mostly minorities (Shields, Burke, & Levy, 2008). This discrepancy may be related to competing health priorities among minorities (who tend to present to care sicker and receive more emergency care), the lower level of board certification of minority-serving physicians, or simply minority patients being offered less genetic testing (Shields et al., 2008). Other barriers to genetic testing among Latinos are similar to reported barriers to general healthcare including logistical concerns - insurance status and competing life concerns, such as childcare and other life responsibilities (Sussner, Jandorf, Thompson, & Valdimarsdottir, 2013). The issue of “competing life concern” was reported by Sussner as the biggest barrier to Latinas pursuing testing, and made them less likely to undergo BRCA1/2 testing (2013).

In addition to considering how to tailor information to the Latino population, their perception of genetic testing could influence how information is received. Not much is known about the psychosocial and emotional reactions to genetic testing that are unique to Latinos (Hickey et al., 2014; Kinney et al., 2010). Male Latinos tend to experience anxiety around

personal peril and employment-related barriers, while females tend to be concerned with heredity implications of genetic diseases, such as how it would affect their children and pain associated with genetic testing (Hamilton et al., 2016, Hickey et al., 2014). Latinos perceive the risk of emotional harm from genetic testing to be higher than Caucasians do, though both ethnicities have relatively low levels of perceived risk of harm (Palmer et al., 2008). Fatalistic attitudes can prevent Latinos from recommending testing to other people (Vadaparampil et al., 2010).

Latinos have been shown to want genetic testing for the purpose of helping affected individuals, their families, and other families, as well as to learn recurrence risk or to explain family history, whereas non-Latino Caucasians more frequently reported discovering the etiology of a disease as reasons to pursue testing (Palmer et al., 2008). However, a more recent study found that there was no significant differences between Latina and non-Latina women in perceived benefits or desired counseling topics in a hereditary cancer setting (Gammon et al., 2011). Studies show Latinos understand the basics of genetic testing and believe that it can provide valuable health information and may lead preventative measures; however, language barriers are listed as a hindrance to doctor-patient communication and the purpose of genetic testing, how it is performed, and the information it provides are less well understood (Hamilton et al. 2016).

Latina women have misconceptions about BRCA1/2 genetic testing, unawareness of familial breast and ovarian cancer, and ignorance about the availability of genetic risk assessment. Benefits of genetic testing reported by these women included information for their children and future generations (Kinney et al., 2010).

In addition, attitudes toward genetic testing in Latinos are often influenced by levels of acculturation, or the degree to which an individual from a minority culture has adopted the

mainstream culture. Latino women with lower acculturation tend to be diagnosed with breast cancer at a later stage than their more acculturated peers (Lanz et al., 2006). Knowledge of genetic testing is lower among Latinos than non-Latino whites, but this knowledge gap becomes smaller with increased acculturation (Sussner, Thompson, Valdimarsdottir, Redd, & Jandorf, 2009). As acculturation increases, Latinos report fewer barriers, more perceived benefits, and more favorable attitude towards genetic testing (Sussner et al., 2009).

One social barrier that cultural sensitivity can ease is the language barrier. Some medical terms in English may not have an equivalent in the patient's language, making it difficult for both translators and interpreters. In addition, when translating from one language to another, the concept or meaning of the sentence may be altered unintentionally which can skew results in a qualitative interview. Interpretation also depends on the skill level of an interpreter and their ability to interpret effectively.

Despite the recognized effects of a language barrier, this is infrequently addressed in qualitative studies. Qualitative interviews have been used in many studies to determine a patient's perception, but few studies have explored the effect of the language barrier. A review of 40 scientific cross-language qualitative studies found that researchers often do not acknowledge language barriers as a possible limitation of their study and do not explore the possibility of it affecting the analysis and interpretation of results. In addition, only seven studies pilot tested their interview guides in the native language of the participants, and half of the studies did not mention the credentials of the interpreter or at which point in the interview the interpreter was used (Squires, 2009).

This study aims to identify both common and unique themes pertaining to the Latino population and their experience of CES results. These themes include, but are not limited to, the

parental experience of education and consenting, initial reactions to the CES results, their decision to have testing, the experience of waiting for and receiving results, and the resources received and needed throughout the process. Barriers to their genetic healthcare will also be explored.

The projected increase in the Latino population within the United States highlights the need for minority populations to be significantly represented within healthcare research. This is the first study that aims to understand the experience of Latino parents who have had CES on their children. The findings from this study can be used to identify barriers to genetic testing in Latinos, and more specifically, the challenges associated with genetic testing when involving a child with a genetic condition. The results of this study have the potential to improve their healthcare in a positive manner by including genetics.

Methods & Materials

This study was carried out in the Division of Clinical Genetics at Columbia University Medical Center (CUMC) in New York City. Interview guides and participant contact materials were developed by the authors and translated by bilingual research assistants, and subsequently approved by the Columbia University Institutional Review Board. Interview guides assessed themes which were identified from the literature as having special relevance to the Latino experience with healthcare (access, language barriers, competing life concerns, cost, etc.) as well as recognized themes related to receiving genetic test results (impact of testing, sharing results, emotional reaction to and understanding of results).

Participants

The participants for this study were parents of Latino/a pediatric patients who had undergone CES at CUMC. Parents were seen in clinic and results session by various members of a team of seven genetic counselors, one of whom was Spanish-speaking, and one of five geneticists, two of whom were Spanish-speaking. A sample of participants were recruited from a database of patients who undergone CES at CUMC in the years 2017 and 2018. Latino participants (n=102) were selected for by filtering ethnicity of proband, parents, or grandparents as “Hispanic” or “not given,” “Country of Origin” for proband or parents as any Latin American country, and “Primary Language” as “English,” “Spanish,” or not listed. Subjects were further stratified by those receiving CES results between six to 18 months ago.

Procedure

The parents of each participant were contacted by invitation letter in both English and Spanish from their treating provider. Invitation letters were followed by up to three invitation phone calls made by English-speaking or bilingual RAs. Verbal consent was obtained, and interviews were scheduled. Consent forms were sent to enrolled subjects either electronically through email link to REDCap (Research Electronic Data Capture) or paper forms through the mail. Subjects’ contact history, consents, and interview schedules were tracked in REDCap.

In-depth, semi-structured interviews were conducted in either in English or Spanish based on participant preference. Interviews lasted between 30-60 minutes and were conducted by phone by one of two genetic counselors (JW, CK) and attended by genetic counseling interns (RS, DL) or a bilingual research assistant (KR). Data collection continued until saturation and all interviews were audiotaped. English interviews were transcribed verbatim by a private

transcription service. Spanish interviews were first transcribed and then translated by bilingual Latina research assistants KR or AE. To improve reliability, translation was completed first by one RA and then selected quotes were reverse translated (translated back into Spanish) by the second RA.

After each interview, subjects were sent an 8-question survey designed to assess acculturation. six questions were from Marin's Short Acculturation Scale for Hispanics (SASH), addressing language use and preferences, and was used as a measure of acculturation. Two additional questions were the authors' own and regarded language use with healthcare providers and at work. Responses were on a 5-point Likert scale of "only Spanish" to "only English," with higher scores indicating greater levels of acculturation. Subjects were sent a \$25 Amazon gift card to compensate them for their time.

Data Analysis

Transcripts were uploaded and coded in NVivo software (QSR International Inc., Burlington, MA, USA) in a three-stage process by three researchers. Participant narratives were reviewed using inductive thematic text analysis, a process of transcript review, interpretation, and consensus discussions. Transcripts were coded by researchers JW, RS, and DL on a rotating schedule. Primary coding was performed by the assigned researcher for the designated transcript. After completion, another researcher was assigned to secondary coding. After secondary coding, the final researcher performed tertiary coding. Methodological and analytical documentation included the identification of key phrases, similar experiences, common themes, and documentation of the subject reaction to the CES experience. All three researchers completed an analysis of each.

Results

Demographics

In total, 22 parents were interviewed. Of those parents, 4.5% (N=1) were male and 95.5% (N=21), were female, ranging in age from 24 to 53 years old at the time of the interview with an average age of 36. Most parents completed high school (86.4%, N=19) and some had a college degree (22.7%, N=5). Sixty-eight percent (n=15) of parents were employed full-time or part-time, while 32% (n=7) were unemployed. Sixty-four percent (n=14) of parents were married and 36% (n=8) were single. Fourteen parents elected to be interviewed in English and eight in Spanish. Besides one father, all of the participants were the mother of the child tested. All parents spoke some Spanish and the majority (86.4%) (n=19) identified Spanish at their primary language or both Spanish and English as their primary languages. Four (18.2%) parents reported they spoke no English. A minority (13.6%, n=3) of parents reported English as their single first language.

Parents had acculturation scores between 4.25 and 1.00. Ten (45.5%) of parents had a low acculturation score, classified by a score of 2.99 and below. Twelve (54.5%) parents had a high acculturation, classified by a score of 3.0 or above. Low-accultured parents had an average acculturation score of 1.72, while high-accultured parents had an average score of 3.41. Of the low-accultured parents, five held their genetics appointment in Spanish with a Spanish-speaking provider, two spoke Spanish and used a translator, and three spoke in English. Nine of the 10 low acculturated parents spoke Spanish in the interview for this study. Of the high-accultured parents, 10 spoke English during their genetics appointment and two spoke in Spanish with the providers. All 12 high acculturated parents spoke English in our interviews (Table 1).

The average age of the children was 8.4 years old, and 86.4% were insured through Medicaid/Medicare. Ten children were female and 12 were male, ranging in age from 1.5 to 18 years old at the time of the interview. The most common clinical indications were developmental delay (n=8), autism spectrum disorder (n=4), seizures (n=2), and hypotonia (n=2). Children were seen in the genetics clinic by one of seven genetic counselors and one of five geneticists. Thirteen were evaluated by a genetic counselor and/or geneticist who spoke Spanish, the other nine either conducted their appointments in English or with the use of a translator. One child had exome sequencing through her neurologist. The time from when parents received results to when the interview was conducted ranged from 3-21 months with an average time of 13 months (Table 2).

All children received CES. Ten children (45.5%) had CES results which did not identify any variants, 9 (40.9%) had pathogenic variants identified, and three (13.6%) had VUS identified. Two children had negative CES results with a VOUS or pathogenic mutation on previous panel or microarray. Of the pathogenic results, four (18.2%) were de novo, three (13.6%) could not be determined because one or both parents had not been tested, one (4.5%) was an X-linked disease maternally inherited, and one (5%) was an autosomal recessive condition with bi-parental inheritance. Nine children (41%) had prior genetic testing before the CES was ordered, eight (36%) had CES as a reflex test following an initial negative work up, and seven children (32%) had CES concurrently with other testing. In addition to CES, 19 children (86%) had a karyotype and 21 children (95%) had a microarray, either before or at the same time as CES. One child had CES ordered by a neurologist and it is unknown if a karyotype or microarray were completed (Table 3).

Understanding of CES

In general, when asked to recall CES, parents provided a limited description and often stated they were not familiar with the terms of exome sequencing. Most parents did not distinguish CES from other genetic tests their child had concurrently or in an earlier work up, though some recalled a discussion of the comprehensive nature of the testing or the occurrence of parental sample collection. Though often unable to provide a detailed description of CES, most parents felt comfortable with the information provided to them.

Motivations for CES

Families expressed an obligation to pursue testing, often expressing that they “had to find out” what caused their child’s condition, regardless of treatment options (n=18). However, many parents communicated that they had hoped that CES would provide a diagnosis for which there was a treatment or cure for their child’s condition (n=6). While many parents expressed hope that a genetic cause could be determined, several also hoped that a cause would not be found. Some had ambivalent feelings about what they were hoping for.

A few also indicated the potential to provide information about the risk for other children or to guide reproductive risk for themselves or their children as a motivation to have testing (n=3). Several participants (n=3) were interested in the potential to learn about secondary results. Many parents (n=14) also explained the testing with regards to how it would inform them of inheritance, such as which side of the family the condition came from. Eleven parents described the purpose of testing as partially to learn which parent had passed on the genes causing the condition. Sometimes the question of inheritance was tied to feelings of guilt or fault.

Waiting for Results

Waiting for the results was frequently considered to be the most difficult part of the entire CES process. When commenting about the amount of time expected for the results to arrive, most patients remembered being told to wait three to four months. The only mother who repeatedly contacted the hospital asking for her results did so at one and a half months, far earlier than most others expected their results, and may have not understood the expected turnaround time.

Experience with genetics and how results were received

Parents generally felt their experience in genetics was positive. With few exceptions, they felt comfortable with their providers and felt their questions were answered. Five parents reported feeling nervous or overwhelmed at various points in the testing process.

Some (n=11) recalled receiving the results first by phone, some recalled receiving them through an in-person visit, and a few thought they learned the results in a letter. One could not recall how they first learned the results. Three parents stated they thought receiving results by phone was the best way, and three thought the results would have been better delivered in person. Most were satisfied with how they received the results or agreed that there was no perfect way. Two parents did not recall ever receiving the full results.

Understanding of and Reaction to Results

Parents had various reactions to the results, reacting both negatively (sadness, surprise, mistrust, disappointment) or positively (hopeful, satisfied, or relieved). There was no noticeable difference between parental emotional reaction and their acculturation levels. While some parents expressed an initial disbelief in results, most expressed a trust in the results and the

prognosis. Some parents expressed the permanence of the results, though others, both with negative and positive results, felt the results may change and may warrant testing in the future as technology advances.

Positive Results (N=9)

Interpretation

Almost all parents of children who had a pathogenic variant understood this to be the cause of their child's condition (n=8). Two parents with *de novo* results were able to explain the genetic cause did not come from them but were unsure about recurrence risk and had decided to forgo having additional children due to the perceived risk of recurrence. Three of the parents were able to name the condition of the diagnosis provided by the test. Most parents had already been convinced there was a condition present in their child before the CES, and that the genetics confirmed the condition's presence.

Reaction

Emotional reactions to positive results were mixed. Parents of children who received positive results often expressed strong negative emotions, such as being shocked, devastated, unsatisfied, in disbelief, or sad (n=5, 62%). One parent reported that his wife doubted the genetic etiology and believed vaccines were responsible for the child's condition. A mother had initially been told the results were negative and was in denial when she learned about the diagnosis.

One parent who reported only a modest emotional reaction felt this was related to already being aware that her child had a serious condition, and the results did not change this. Three parents with positive results expressed hope that the results could lead to some treatment, while

two parents with de novo positive results expressed relief to learn they had not caused the condition. One mother of a child with a de novo muscular dystrophy shared how the results helped her to stop blaming herself.

VUS Results (N=3)

Interpretation

One parent had a child with a de novo VUS but interpreted the result as negative. She reported wanting more information about why CES was performed. Another mother had a child with a paternally inherited VUS and understood the uncertainty of the result, the inheritance, and the recurrence risk. The mother of the child with the maternally inherited mitochondrial VUS was confused about the result and thought further genetic studies were needed.

Reaction

Patients with VUS results reacted in a variety of ways, from confusion to unconcern. The parent of the child with the de novo VUS reported being happy that there was nothing found but was confused about why CES was performed and what clinicians expected the result to be. Feelings of relief were expressed by the parent of the child with the paternally inherited VUS. The mother of the child with the maternally inherited mitochondrial VUS was concerned about inheritance and was fearful of a hereditary condition.

Negative Results (N=10)

Interpretation

Some parents (n=3) who received a negative CES result for their child seemed to understand there was still the potential for a genetic diagnosis despite negative results. Two participants expressed a wish for future treatments or testing options. Many parents (n=4) saw

CES as definitive and believed failure to find a variant meant that the child's condition was not genetic and understood negative results to have eliminated a potential genetic diagnosis. Two parents had negative CES results but VUS on an epilepsy panel and did not distinguish between the two tests. The parent whose child had a negative CES result but a de novo pathogenic deletion on microarray also did not differentiate between the two tests.

Reaction

Parents who received negative results and felt the results ruled out a diagnosis expressed happiness and relief (n=5). Two parents reported being unconcerned about the results due to the distractions of caring for their families, one of whom never received the results. Other parents with negative results were disappointed to not have an answer (n=3), though two of these parents also felt glad the process had been done. One parent with a negative result was unsure if doing the test was the right thing for her.

Timing of CES

Most parents (69%) would have liked to have had the testing done sooner, and no parents wished for testing to be done later. Of the parents with pathogenic results who were asked if they desired to have testing done sooner, all agreed. For parents who had negative or uncertain results many were content with the timing of the test (67%), though some (n=2) would have liked to have the testing done earlier.

Impact of CES

Regardless of the emotional impact of learning the results, almost all parents stated that they did not regret doing the test or learning the results, and would do the test again. Relatively

few sought advice from others before undergoing CES, and none had doubts about the testing process.

Most parents reported no significant impact on their lives regarding insurance, jobs, or relationships, but genetic testing has made them more cautious for their other children, has caused minor lifestyle changes, and has improved some of their medical interventions. Six parents mentioned that there were no negative effects of the testing (2 negative, three VUS, one positive). Eight parents mentioned that the tests did not affect their child's care in any way, five of them from the positive result group. Eight parents mentioned the results affected their reproductive decisions or feelings about recurrence risk, most (n=5) had positive results. Five parents were reassured there was little chance of recurrence, and one said she was considering her previous decision to forgo reproduction. Another parent with a positive result said the result allowed them to be prepared for a second affected child. A parent with a negative result was uncertain of the implications of the test.

Three parents with positive results and one with a VUS mentioned access to a trial treatment or some improvement in the care as a result of the genetic test. Three parents (one VUS, two positive) mentioned considering a trial but not seeing improvements or not joining the trial. One parent with a positive result explained they had undergone much personal growth since the diagnosis. Another discussed being more cautious with her current child.

Sharing CES Results

All parents chose to share their child's CES results with some family members, such as their other children, their parents, grandparents, or their siblings. Some parents stated they had a family meeting, while others reported they had family members who constantly checked in with them about their child's health. One mother of a child with a de novo muscular dystrophy

expressed difficulty explaining the condition because of limited information available and the misconception that genetic conditions always run in families.

Others shared that members of their family were not as accepting of the results. The father who was interviewed for the study stated that his wife did not accept the test results. She believed vaccines were responsible for their child's autism and planned to pursue direct-to-consumer testing (DCT) as a second opinion. Another mother of a child with seizures noted that the father had unrealistic expectations that their child would recover or catch up.

Some parents (n=4) mentioned sharing the results with their friends, while others (n=4) reported sharing their child's diagnosis with the child's school. Ten parents shared results with healthcare providers. There were no overt patterns of sharing based on type of result. Parents listed reasons not to share with others due to privacy concerns or their own judgement about who they felt needed to know. Some stated that other people who knew about their child's symptoms before CES either did not believe the child's symptoms were real, thought the symptoms were due to the child's upbringing, or believed in alternative remedies, which is why they chose not to share the diagnosis.

Barriers to CES

The majority of parents did not report physical barriers to their genetics care such as time off work, child care, and transportation. However, some parents reflected on the difficulty of multiple appointments and taking time off work or finding child care. One mother who received car service expressed exasperation with it and eventually purchased a car to attend appointments.

One parent who reported never learning the results of her child's testing and not following up to inquire about it expressed that it was not her priority given her child's other

medical needs. Another parent experienced the death of her grandmother when the results became available and she elected to delay the receipt of the results because of this.

Only two parents had an out of pocket cost of \$7,000 and \$200, respectively. The majority of other parents (n=19) reported no cost of testing. Few recalled having a discussion about potential cost at the time the testing was ordered. When asked about whether or not they would have pursued the test had there been a cost, two indicated an upper limit of \$1,000 and \$300 respectively, two mentioned they could not have afforded anything, and others (n=3) felt they would have found a way regardless of the cost.

Language as a barrier

Only one participant used a phone translator during their genetics appointment. All other parents who conducted the interview in Spanish had Spanish-speaking providers who either spoke Spanish with them, chose to speak in English, or used a staff member to translate in-person. The Spanish-speaking parent who did not have a provider who spoke Spanish recalled that a phone interpreter was used and reported problems with understanding. This parent reported wanting more information and not understanding the reason for the test. Three of the parents were able to communicate with Spanish-speaking doctors in the session. These parents reported no problems with the communication. When asked directly if they were satisfied with the information provided in session, only one parent who communicated with the doctor directly in Spanish reported being unsatisfied.

All of our participants spoke at least some Spanish, but several said they preferred to speak to providers in English. Three parents had concerns about not knowing as much medical terminology in Spanish, and one expressed the preference for English based on the concern that Spanish language encounters with providers could cause delay accessing records in case of an

emergency. Though each of these four parents said they were comfortable in Spanish, they scored high on the acculturation scale (average of 3.5). One parent with an acculturation score of 4.25 reported being perfectly bilingual, and preferred Spanish for the benefit of the people who attended the session with her. No parents with a low acculturation score preferred English with their providers, and those who voiced a preference for Spanish had an average acculturation score of 1.9 (n=5).

Additional Resources

Additional resources accessed by parents were the use of the internet, their job experience in the healthcare field, or parents of other children with the same condition. Overall, Spanish-speaking, low acculturated parents had the most barriers regarding use of support groups. Only one parent, noted to have high acculturation, was actively enrolled in a support group. Three parents, all Spanish-speaking with low acculturation, expressed interest in attending a support group, and two of these parents stated they were able to find resources in Spanish. One Spanish-speaking parent stated that she wanted to find resources but was discouraged because of the language barrier, while another Spanish-speaking parent stated she would have liked therapy with psychologists. However, one parent whose child has Neimann-Pick and stated that the online support group provided to her by her child's neurologist had a representative who could communicate with her in Spanish.

Latino experience

Parents were asked how they felt their ethnicity or culture impacted their experience. Many felt that it had little impact and often reflected on how they may differ from other Latinos. Others expressed that the larger Latino community may have a different experience because of

cultural approaches to about health, such as belief in alternative treatments, lower education, language barriers, and differences in how information and decisions about health are approached. Some used the examples of specific family members and friends and hypothesized that they may have relied more on their provider to make the decision to pursue testing, or have more difficulty accepting a diagnosis or a prognosis. In terms of acculturation level, very little difference was found in the experience of parents based on their acculturation levels.

Religion & Faith

Some parents occasionally used language relating to “God,” in vernacular such as “thank God” or “oh my God” (n=3). Two mothers expressed faith that God could change something in relation to their child’s genetic diagnosis while the other mother used her faith to come to terms with her son’s diagnosis. All parents who made explicit mentions of faith were slightly above the average age of parents in our study, had acculturation scores less than 1.5, and spoke Spanish during the CES results session. However, one chose to speak English during their interview. All three had children with pathogenic CES results, two of which were de novo and one where inheritance was unknown.

Discussion

This study provides insight into the experiences and perceptions of Latina mothers and one Latino father of children who have had CES. We examined how these experiences were influenced by degree of acculturation. Overall parents reported the experience of having CES performed on their children to be positive with few reporting negative experiences. Most negative experiences were related to distress upon receiving a diagnosis, waiting for results, and difficulties with language.

Most parents were unable to recall the term “exome sequencing” or describe the test in any detail but were satisfied with their understanding and the information provided to them regardless. The primary reasons for pursuing testing was a desire to learn everything possible about their child’s condition, to obtain advice from providers regarding their child’s diagnosis, and a hope for new therapies or treatments.

Reactions to results were influenced by type of result. Parents receiving positive results often expressed feelings of shock or sadness and occasionally hope. Parents who received negative results or VUSs often felt relief, confusion, and occasionally disappointment. Most participants had good recall of their child’s CES results and an appropriate understanding of the results.

We did not observe a difference in the understanding and interpretation of CES results between high acculturated parents and low acculturated parents. This is somewhat inconsistent with a previous quantitative study (N=120) of Latina women, which found increased acculturation correlating with increased knowledge of genetic testing (Sussner et al., 2009). However, we did not specifically ask about prior knowledge of genetic testing.

Few differences across acculturation levels were observed for barriers to testing, regrets around testing experience, or satisfaction with the process of genetic testing. Spanish speaking parents were more likely to express difficulty in communicating or understanding the providers, and also in finding additional resources during and after the testing process. Similar to a study by Rajpal, Munoz, Peshkin, & Graves, few participants felt that their cultural background personally influenced their experience but agreed that for other Latino parents it might (2017).

Our findings of parents’ feelings of guilt about potentially passing down genetic conditions contrasts with findings in a recent study of parents in South Africa. A study by

Gardiner et al (2018) showed that parents of children with hearing loss reported that discovering a genetic etiology for the hearing loss would decrease their personal feelings of guilt. Only one parent in our study also expressed the idea that discovering a genetic cause would remove their feelings of guilt; many more reported apprehensions that the genetic testing would indicate that one of the parents were at “fault” genetically for their child’s condition. Gardiner’s study had findings which were concordant with ours, such as the perception that a genetic etiology was associated with a positive family history.

Lack of diagnostic odyssey and its impact on parental reactions

The majority of the children in the study had CES as part of their initial genetic evaluation or as reflex testing following negative first tier testing. This timing is in contrast with other studies of parents of children with CES, where frequently, the child had CES after years of evaluation (average of 78 months in the case of Valencia et al. 2015, and 91 months in Rosell et al. 2016). This difference may account for some of the differences we observed in our study. Many of the parents did not describe the CES testing experience as different than prior or other genetic testing their child had or was having. In contrast, other studies have reported parents to understand CES accurately as distinct from previous tests, with characteristics such as being untargeted (Rosell et al. 2016).

While waiting for results has been reported as a source of frustration and disappointment (Rosell et al. 2016), in our study, parents recalled the waiting period as a source of anxiety and anticipation. As CES testing becomes part of second and even first tier testing, this will affect the patient/parent experience. Their understanding of the testing may differ as they have had only one or two appointments in genetics rather than many over a span of several years. The CES

process is likely one of the first times where parents have had to wait over a month for a test result.

Faith as a coping mechanism

One theme that emerged was the use of faith and spirituality as a coping mechanism, although we did not specifically ask about this during our interviews. Faith has been previously explored as having a significant impact on the genetic testing decision-making process in individuals who consider themselves religious. For these individuals, there is an added step in the decision-making process. Some Latinos may believe genetic testing is punishable by God, while others process their understanding of the diagnosis and treatment within the context of their faith (Hamilton, et al. 2016; Scully, Banks, Song, & Haq, 2017). Clinicians are less likely to address faith and are viewed as disengaged when patients mention their religion (Scully et al., 2017). Faith has also been observed as a method of adaptation and resilience in Latinos with cancer. This belief system aids in their acceptance of disease and they respond to illness to overcome disease (Jurkowski et al. 2010; Hunter-Hernández et al. 2015).

Similar to other studies in a prenatal setting, we found that several parents, all with lower acculturation levels, referenced back to their faith as a coping mechanism, expressing having faith in God, waiting for a change from God, or believing their child's condition was a result of God (Thompson et al., 2015; Seth et al., 2011). We also noted that there may be generational differences due to the Latinas referencing faith being above the average age of our participants. Our cohort did not use their faith as a form of denial about their child's condition, but rather as a coping mechanism or form of acceptance. Recognizing that this need may be more frequent in the Latino community could build additional rapport with parents and extract positive reactions toward genetic testing in their children.

Language and Cost Barriers in Latinos

Our study found few language barriers encountered during the genetics appointment and only occasional language barriers during the subsequent search for additional information or resources. In fact, of the nine parents who we identified as having expressed misconceptions or confusion about the implications of their results (incorrect recurrence understanding, dismissal of positive results, believing that negative results signified no possible genetic etiology, confuse CES and microarray results, or report not knowing the purpose of the CES), seven of those nine were English speakers with high acculturation.

In past studies language barriers were a reported concern affecting the ability of Latino patients to access genetic testing (Kamara et al., 2018; Kinney et al., 2010). Compared to these two other studies, our cohort was less foreign born, more English speaking, younger, and more educated. The lack of reported language difficulties in our study was most likely due to the availability of Spanish-speaking receptionists, genetic counselors, and geneticists in the clinic. The solitary Spanish-speaking patient who used a phone interpreter in the counseling session and had no Spanish-speaking providers was one of the only parents to report confusion and low satisfaction with the information provided. This study illuminates the increased patient comprehension that can be provided by a bilingual staff and suggests that always having a Spanish-speaking provider on staff and simply asking a patient if they would prefer that Spanish-speaking provider allows greater understanding by Spanish speaking patients.

Several of our participants reported difficulties finding additional resources in Spanish, such as focus groups or literature on the internet. A similar need for culturally-appropriate materials was found among New York Latinos in the cancer genetics setting by Sussner et al. (2009), and in a group of Salt Lake area Latinos in Kinney et al. (2010). Our participants

described using Spanish language resources for providing themselves with helpful information but also as being used as a resource for educating extended family members who do not speak English.

Although a minority of bilingual parents preferred using Spanish in the sessions with providers for the benefit of Spanish speaking family members also present, almost all of the parents who spoke some English expressed a preference to speak with their providers in English. This occurred even when the provider spoke Spanish and was attributed to ignorance of medical terminology in Spanish or concern that Spanish language encounters with providers could become an impediment to medical care. This indicates that although Spanish may be their first language and the language that they are most comfortable using, for those who are bilingual, there is a level of discomfort when using Spanish with providers.

Another barrier that has been explored is the cost of genetic testing. Cost and lack of insurance have previously been found to be the most frequent barriers for BRCA1/2 testing in Latinos in New York City (Kinney et al., 2010). Our study found almost no concern or even discussion of financial costs when testing was ordered among our parents, though our cohort is refined for those who already received testing. Five parents either did, or indicated that they would have, paid at least \$300 for the cost of testing. Only two parents claimed to have been unable to afford any cost whatsoever.

Sharing of results

A study by Velicer et al. found that women who intended to pursue genetic testing were twice as likely to have spoken to relatives about the testing (2001). This is consistent with our study, where participants who already had testing were generally willing to share their test results with providers, family, and friends, with only one parent stating they only shared the

results with their child's other parent but no other family members. Other studies have found Latinos to be more likely to share medical information or involve family members in the decision-making process with a cancer diagnosis (Maly et al., 2006), and family history of cancer risk (Corona et al., 2012).

Study Limitations

There are several limitations to our study. All parents received care at a single institution. Parents were cared for by seven GCs and five geneticists, and differences in how each individual provider practices may have affected the experiences of the participants. While it is a strength of our study that we conducted interviews in both Spanish and English, the Spanish and English interviews were conducted by separate individuals and there is potential that information is lost in the translation of the Spanish interviews. All quotes from the Spanish language interviews were translated back into Spanish by a native speaker to confirm the original translation was correct, though there is debate in the literature about whether it is appropriate to analyze interviews in two different languages in a single study. Additionally, the Spanish-language interviews were conducted by a genetic counselor fluent in Spanish but not a native speaker, however a native speaker was present and assisted in any linguistic difficulties. All interviews were conducted by phone, and the lack of nonverbal cues may have influenced the interviews and therefore our results. The timing of the interview from when the parent had received the results varied from three months to one year and nine months. We specifically elected not to complete an interview less than three months from receiving results to limit the immediate and sometimes transient reactions to results. The timing likely resulted in some recall bias.

We attempted to capture a diverse sample of Latino/a parents of child with diverse types of CES results. This diversity may have limited our ability to capture more subtle themes

experienced within parents of a certain acculturation or with specific types of results. As all of the participants had already undergone CES, our participants are representative of only this subsample. In addition, the term Latino refers to a heterogeneous group of peoples. The Latino population in our study are those living in New York, who are primarily from the Dominican Republic and Puerto Rico by descent. The experiences we observed may differ from experiences of other Latinos with different ancestries and cultures.

Practice Implications

Many of the parents in our study understood CES as an investigation of many genes and were confused or relieved when no genetic cause was discovered after testing. This finding suggests that a more thorough explanation of the limitations and negative predictive value of testing needs to be conveyed to patients. Several patients seemed to be unaware of the possibility of an undetected genetic cause. Likewise, some parents misunderstood the implication of results which were de novo and had decided to forgo having additional children due to the risk of recurrence. Additional time is needed in sessions to convey the recurrence risk of de novo conditions and explain why there may be no family history.

To address the anxiety of the waiting process, placing emphasis on the average turnaround time multiple times during the consult may be helpful. In addition, psychosocial counseling and support should be provided throughout the process, from pre-results to post-results, to alleviate and explore any concerns parents may have. Providing a visual timeline that illustrates the process of CES that highlights the average wait time for return of results may also be supportive in generating realistic expectations. Some parents may find it helpful to connect with other parents who have had CES for their child. These parents may be a source of comfort

or a more accurate resource during the wait time. If possible, Latino parents should be connected to each other, as the cultural and language aspects may provide an extra level of comfort.

The Latino parents in our study did not differentiate between CES testing and previous genetic testing performed, suggesting that extensive information about the specific genetic tests may not be needed. Instead, it may be helpful to focus on the differences between a positive, negative, and VUS result, and what these results could mean for their family, their child, and their child's medical management. This information may be further useful as in the form of written documentation, available to Latino parents in both Spanish and English regardless of what language they used in the results session. To support parents who are more comfortable speaking Spanish but lack confidence in speaking Spanish with providers, better educational materials can be developed to help them understand medical terminology in Spanish. In addition, acknowledging the role of faith and God in the parental experience of CES for their child may also be conducive to the establishing a stronger bond between the provider and parent.

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Conflict of Interest

Author Daniel Luksic and Author Radha Sukhu declare that they have no conflict of interest.

Informed Consent

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients included in the study.

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Appendix I. Study Participants

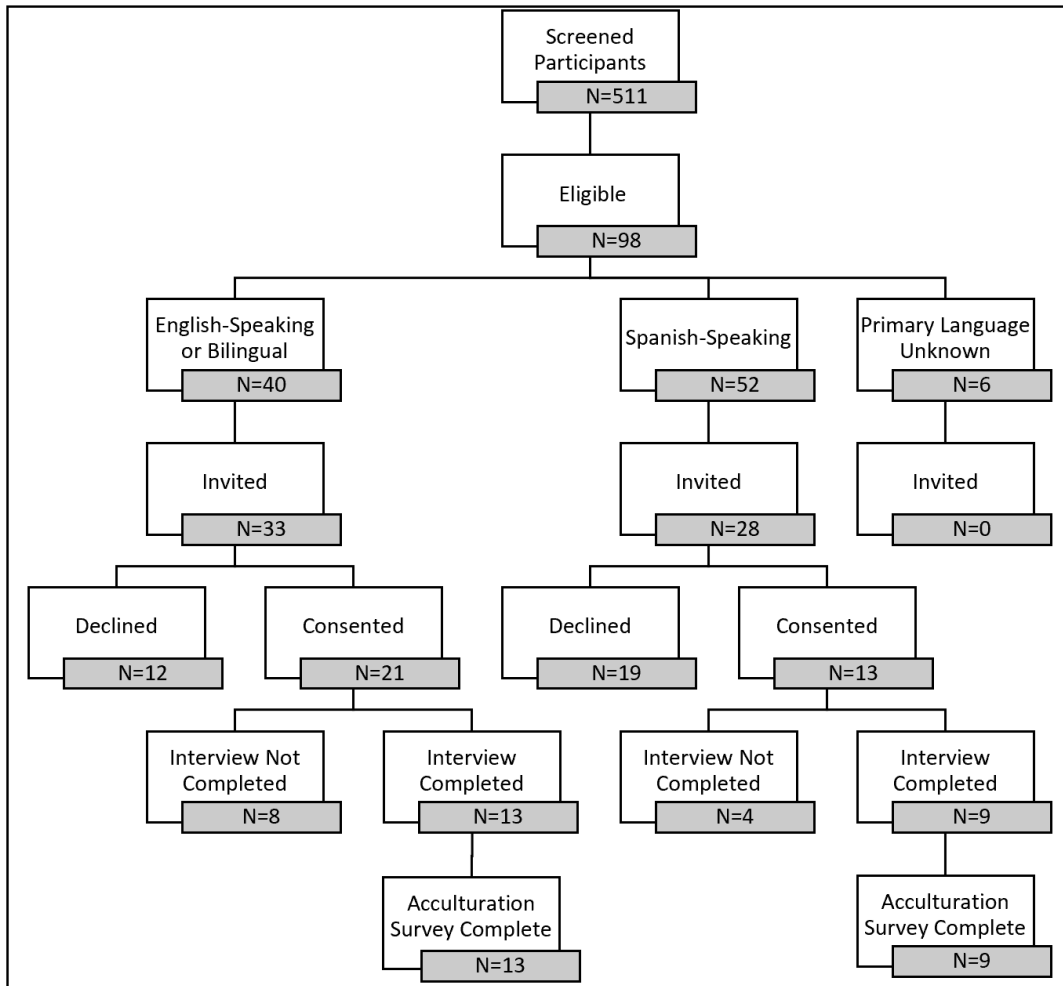


Figure I. Flow diagram of study participants. Parents of patients who were of Latino ancestry and had CES for their children were eligible. Some eligible participants were not invited because they could not be reached, or the study period ended before they were contacted. Participants who consented to the study were sent a consent form by email or paper, according to their preference. After consent was received, participants were contacted by phone for the interview on a scheduled date. Some participants chose not to complete the interview after consent or cited scheduling conflicts as a barrier. Participants who completed the interview were invited to complete the acculturation survey after the interview.

Appendix II. Parent Demographics

Table I. Parent Demographics

Study ID	Parent's Gender	Parent's Age	Country of Birth	First Language	Second Language	Education	Employment ^a	Marital Status	Interview Language	Acculturation Score
1504 ^b	Female	33	US	English	Spanish	less than HS	not employed	married	English	3.8
1509	Female	40	US	English	Spanish	HS Graduate	employed	married	English	3
1514	Female	32	not US	Spanish	n/a	Some College	employed	not married	Spanish	1.25
1523	Female	53	not US	Spanish	English	Associate Degree	employed	not married	English	2.8
1524	Female	37	US	English	n/a	Associate Degree	employed	not married	English	3.6
1526	Female	40	US	English & Spanish simultaneously		Associate Degree	employed	not married	English	3.6
1528	Female	35	US	English	Spanish	Associate Degree	employed	married	English	4.2
1532 ^c	Female	31	US	Spanish	English	less than HS	not employed	not married	English	3.4
1533	Female	24	not US	English & Spanish simultaneously		HS Graduate	not employed	married	English	3.4
1534	Male	35	US	Spanish	English	HS Graduate	employed	married	English	2.8
1536	Female	47	not US	Spanish	English	HS Graduate	employed	married	English	3
1539	Female	25	US	Spanish	English	HS Graduate	not employed	married	English	3.8
1549	Female	26	US	English	Spanish	HS Graduate	employed	married	English	3.8
1550	Female	29	not US	Spanish	English	HS Graduate	employed	married	English	2
1551	Female	41	not US	Spanish	English	less than HS	employed, part time	not married	Spanish	1.6
1554	Female	38	not US	Spanish	English	HS Graduate	employed	not married	Spanish	1
1556	Female	36	not US	Spanish	n/a	HS Graduate	employed, part time	married	Spanish	1.4
1559	Female	30	not US	Spanish	English	College Degree	employed	married	Spanish	1.6
1562	Female	40	not US	Spanish	English	HS Graduate	not employed	not married	Spanish	1.3
1563	Female	40	not US	Spanish	English	HS Graduate	employed	married	Spanish	1.4
1590	Female	39	not US	Spanish	n/a	HS Graduate	not employed	married	Spanish	1.2
1599	Female	31	not US	Spanish	n/a	HS Graduate	not employed	married	Spanish	1.6

Abbreviations: HS, high school; US, United States

^a employed full time unless otherwise noted

^b = did not receive results of CES

^c = interview not recorded

Appendix III. Child Demographics

Table II. Child Demographics

Study ID	Child's Gender	Child's Age ^a	Child's Insurance	Child's Diagnosis	CES Result	Prior Testing
1504	male	12	Medicare or Medicaid	developmental delay and fhx of brother with learning disabilities	Negative	microarray at the same time as CES, chromosomes ordered after birth, not by genetics
1509	male	3	Private	developmental delay, ophthalmoplegia, encephalopathy	MT-ND5, pathogenic, de novo; Diagnosis of Leigh syndrome	mt genome sequencing concurrent with CES
1514	male	2	Medicare or Medicaid	hypotonia, bronchiolitis	EIFG3, VUS, de novo; clinical implications uncertain	microarray with reflex to CES
1523 ^b	female	17	Medicare or Medicaid	seizures	Negative	chromosomes, microarray, epilepsy panel (PLCB1 VUS) w/ reflex to CES
1524	male	15	Private	connective tissue disorder	Negative	connective tissue panel, chromosomes, microarray with reflex to CES
1526	female	18	Private	immunodeficiency	Negative	microarray and chromosomes concurrent with CES
1528 ^b	female	15	Medicare or Medicaid	seizures	Negative	microarray, chromosomes, seizure panel (DEPDC5 VUS maternally inherited) before CES
1532 ^c	female	1.5	Medicare or Medicaid	Chiari II brain malformation and encephalocele	Negative	chromosomes, microarray before CES
1533	female	3	Medicare or Medicaid	autism, optic nerve hypoplasia	Negative	microarray (regions of homozygosity) concurrent with CES
1534	male	3	Medicare or Medicaid	autism	NRX1, pathogenic, maternally inherited; Diagnosis of NRX1 X-linked intellectual disability	chromosomes, microarray, fragile X with reflex to CES
1536	female	20	Medicare or Medicaid	fibromatosis and a fhx of hemochromatosis	Negative	chromosomes, NF1 previously, microarray (regions of homozygosity) with reflex to CES
1539	female	1.5	Medicare or Medicaid	vomiting, FTT, developmental delay, cerebral volume loss	PDHA1, pathogenic, de novo, Diagnosis of x-linked pyruvate dehydrogenase E1 alpha deficiency	chromosomes and microarray with reflex to CES
1549 ^d	male	2	Medicare or Medicaid	hypotonia, developmental delay, nystagmus	Negative	microarray (Xq22.2-q22.3 de novo pathogenic deletion), fragile X, chromosomes concurrent with CES
1550	male	2	Medicare or Medicaid	congenital heart defect, cardiomyopathy	TPM1, VUS, paternally inherited; clinical implications uncertain	metabolic testing, chromosomes, microarray, cardiomyopathy panel concurrent with CES
1551	male	18	Medicare or Medicaid	short stature, obesity, & developmental delay	SETD5, pathogenic, inheritance unknown; Diagnosis of SETD5 syndrome	chromosomes, microarray, fragile X before CES
1554	male	5	Medicare or Medicaid	autism, developmental disorder, macrocephaly, food allergies	SETD2, pathogenic, inheritance unknown; Diagnosis of STED2 related autism, intellectual disability, overgrowth syndrome	microarray, chromosomes, autism panel, fragile X before CES
1556	female	4	Medicare or Medicaid	motor delay, regression	Homozygous NPC1, pathogenic, inherited; Diagnosis of Niemann Pick type C	chromosomes, microarray (maternally inherited X dup), fragile X with reflex to CES
1559	male	3	Medicare or Medicaid	autism	Negative	chromosomes, microarray, fragile X, NSD1 before CSE
1562	male	6	Medicare or Medicaid	growth retardation, congenital heart defect, hearing loss, dysmorphic	CHD7, pathogenic, not maternally inherited; Diagnosis of AD CHARGE syndrome	chromosomes, microarray before CES
1563	female	10	Medicare or Medicaid	retinitis pigmentosa	PDE6A, de novo, pathogenic; Diagnosis of retinitis pigmentosa	microarray and chromosomes at the same time as CES
1590	male	14	Medicare or Medicaid	developmental delay	GRIN2B, pathogenic, de novo; Diagnosis of GRIN2B-related neurodevelopmental disorder	chromosomes, fragile X, Angelman, microarray before CES
1599	female	2	Medicare or Medicaid	hypoglycemia, developmental delay and spasticity	MT-TC, VUS, mother homoplasmic; counseled not associated	metabolic testing, chromosomes, microarray before CES

^a = child's current age

^b = negative result on CES, VUS on epilepsy panel

^c = interview not recorded

^d = negative result on CES, pathogenic deletion on microarray

Appendix IV. Information about Genetics Evaluation and Language Usage

Table III. History of Genetics Visits

Study ID	Date of Initial Genetics Evaluation	Date of Results Disclosure	GC	MD	Language Spoken at Visit	Interpreter Usage ^b	Interview Language
1504	5/11/2017*	7/13/2017	LT	LP ^a	English	No	English
1509	3/1/2017*	11/1/2017	IL	WC	English	No	English
1514	3/30/2017*	8/16/2017	PA	KY	Spanish	Yes	Spanish
1523	8/10/2017*	12/18/2017	LT	AI ^a	English	No	English
1524	5/18/2017*	8/29/2017	JS	LP ^a	English	No	English
1526	9/27/2017*	1/19/2018	MR	KY	English	No	English
1528	4/5/2017	11/16/2017	JS	LP ^a	English	No	English
1532	8/24/2017	11/9/2017	AW	KY	English	No	English
1533	3/5/2018*	6/1/2018	AW	WC	English	No	English
1534	2/28/2018*	4/18/2018	AW	KY	English	No	English
1536	2000	8/3/2018	JS	EP	English	No	English
1539	10/25/2018*	3/5/2018	MR	EP/AI ^a	English	No	English
1549	12/8/2017*	8/28/2017	JS	LP ^a	English	No	English
1550	2/1/2017*	6/1/2017	JS	AI ^a	Both	No	English
1551	8/13/2013	2/22/2018	MR	KY	English	No	Spanish
1554	6/1/2015	6/1/2017	EG ^a	AI ^a	Spanish	No	Spanish
1556	2/14/2017*	6/1/2017	EG ^a	AI ^a	Spanish	No	Spanish
1559	9/1/2016	5/1/2017	JS	LP ^a	Spanish	No	Spanish
1562	8/1/2012	7/1/2017	EG ^a	AI ^a	Spanish	No	Spanish
1563	5/3/2017*	8/1/2017	LT	LP ^a	Spanish	No	Spanish
1590	2013	2/6/2018	EG ^a	KY	Spanish	No	Spanish
1599	10/10/2016	1/18/2018	JS	AI ^a	Spanish	No	Spanish

* CES ordered at first visit or as reflex

^a Spanish speaker

^b indicates if an interpreter was used during the CES results session

Appendix V. Quotations from Parents

Table IV. Quotes obtained from Qualitative Interviews from Latino Parents of Children who have had CES

Theme	Sub-theme	Participant	Quotations from parents
Understanding of CES	Basic understanding of CES	1533	"I don't remember the names; I just know that they told me there were going to be two different tests done, and that one was going to be more simple and faster, and it was going to be like overview, and if they didn't find anything, they told me they might send his genetics testing to be more -- further explored, but that that would take more time."
		1549 ^b	"I could have eaten salad my whole pregnancy, or I could have eaten vegetables my whole pregnancy, and you know, wouldn't have changed anything... they did tell us that ... the X-chromosome is the mom, so of course I felt like 100 percent guilt... after that, they told me OK, you know, thankfully there's nothing wrong -- you know, it's not something that you carry... So, you know, that definitely was like a reliever."
Motivations for CES	Explanation for Condition	1549 ^b	"I really just wanted to know what was wrong and what he had."
		1533	"Half of me wanted there to be a certain reason, explanation in those results for why [name] is the way he is. Then the other half of me didn't want there to be anything because I didn't want to think that it was something in his genetics."
	Assuaging Guilt or Shame	1509	"...only the mother carries it, and the mother gives -- so I felt really bad."
		1533	"Because I don't want to think there's a reason, like, that could be my fault, or his father's fault that he was diagnosed with autism and all these other things."
Waiting for Results		1514	"After a month and a half we called to learn about the results. We called after a month and a half because no one would give us any information. We even went to the hospital in person asking about the genetics results and no one ever gave us an answer. At the two month mark the woman [GC] gave me a call. She told us that the result was negative."
Experience with Genetics	Suggestions for emotional preparation	1551	"...for example, if their appointment is at two pm, ask them to arrive 30 minutes earlier to prepare the mother or father for what they're about to go through with their children."
How Results are received	Preferences	1526	"...but especially for tests that are done like these, I believe that it's better to be, you know, to meet in person."
		1539	"...it was good that they give me the news over the phone, because that gave me -- immediately after I hung up, I went and did my research online."
Interpretation of Positive Results	Implications of Result	1539	"...this doesn't come from me or her dad; this - this is all on her own. So it's [already] genetic because she has it, so there is a 25 percent chance if we have another child that this might happen again... So as of right now we are not trying to have children. And maybe later on in the future, maybe adopt..."
		1509	"But at the same time, she explained it to me that it's not that I have something... when she said that to me, and the father, I was like, okay, that's it, no more kids for me."
Reaction to Positive Results	Shock, devastation, dissatisfaction, disbelief	1509	"And obviously, that day I was devastated; I couldn't ask -- they were talk -- once they told me about the syndrome, from there, I couldn't hear; I couldn't speak; I couldn't do anything else, because obviously, I was done, you know?"
		1556	"Very bad. I almost didn't believe it. In that moment, I thought that it couldn't be that. And it was when I entered into a doubt that it couldn't be that, that at best they were wrong or that it shouldn't be there."
		1540	"I completely understand it, but it's just, it's very -- like, it's very broad type of diagnosis where -- you know, like I said, it's kind of, you don't know what's going to happen, and you're kind of out there, and you don't know, oh, well if this is this."
		1509	"I was completely in denial; I was like no, it cannot be happening to my son, you know what I mean? So yes, I definitely told them, I think you guys are wrong, because doctor told me something different."
		1534	"And she still believes it's a vaccine, and I see it, OK, the result says it's one of us, and so it's from a genetic level."

Understanding of VUS Results	Minimal understanding	1599	"...but what I thought is that she could come out with the genes like her father's family and stuff."
		1550	"They found like a genetic in the dad, that has to do like something with the dad, but they said it doesn't necessarily mean that our kids are all going to have the genetic."
Reaction to VUS Results	Skepticism, unconcerned, relief, fear	1514	"Of why the test was being done, of what they were looking for in my child, that is what I had doubts about. Why did they want to the testing and what were they expecting the results to be?" - 1514
		1550	"I was calm because it didn't seem like they were -- like it was a dangerous, you know, genetic result, so I was not really scared or anything like that."
		1599	"Well, we were scared because we didn't know if it was from my part or his part."
Understanding of Negative Result		1523 ^a	"Explained to me that it was not -- 100 percent all the gene is investigated, but the most common that could cause this problem."
Reaction to Negative Results	Relief, happiness	1524	"I mean, I felt great, yes. [laughter] It was great news."
		1526	"I feel more safer knowing that it's nothing, you know, concerned. And like I said, I was very relieved when I actually got the results."
		1523 ^a	"...because when you have something and your kid has the same thing, what you can think, that I pass it to her. So now I feel, you know relieved that there's nothing like that in this family."
		1563	"I got up to thinking the worst for my girl...And for one moment I was thinking that since I had diabetes during her pregnancy, that probably it could be that, something that passes, but it reality it wasn't that."
	Devastation	1528 ^a	"Kind of upset and devastated at the same time"
	Unconcern	1536	"But it would sound better if it had more -- that if something show up."
	Confusion	1504	"And have you tried to reach out to them to find out about the results? A: I'm a little occupied." - 1504
1528 ^a	"Yeah. I was the one that had it. So I know that -- something I -- I guess, something that I had, it -- she has that could be a possibility of why she would have had a seizures, but they really didn't determine that was the cause. Like you know, that can -- it was weird to understand -- like I understood it, but not at the same time; it was weird. You know, that gene there could me a possibility that could have caused it, but it's not 100 percent sure,"		
Timing of CES	Earlier testing	1539	"I probably would have wanted -- would have wanted -- done it sooner, so we could have found out sooner. But I feel the timing was perfectly fine, because by now she -- she's doing so much that -- I'm -- I'm grateful for whatever I can get."
Impact of Testing	Reproductive Choices	1509	"...because you know, if I knew that I had something that I could damage -- you know, I wouldn't ever get pregnant like you know, I did."
		1533	"And maybe the next time I have a child, everything will be fine and different. Then again, we don't know the cause of autism, so it could just be it wasn't the hospital; maybe it's just, that's just what happens."
	Enrollment in trials	1549 ^b	"so you know, we just put in the back of our heads, and we're like, OK, well maybe with time if we get to it, we will. But, that was it with that."
	Personal Growth	1509	"Like, the person I was when I found out to the person I am now -- it was completely different, because I -- I've learned to live for her and around her and her condition. So it's easier. Back then, I -- it was all so new to me that I just thought the worst of the worst."
	Caution	1524	"Definitely. Even myself, even myself. I have a three-year-old, and any little bump or any little thing that I see that wasn't there that is forming, taking him to the doctor. Or googling it, or whatever, whichever."

Sharing CES Results	Support from family	1523 ^a	“Well, we have a close family in - since they are very healthy, the four of them, but this problem with [name], it worries everybody, the family. So, all the time, my sister and my brother, they are calling me, oh, how is [name] doing...”
	Difficulty sharing with others	1549 ^b	“I feel like explaining it to people is very difficult as well, because you know, like I kind of don’t really know much about it, so when people ask me, oh, what does he have, I just say it’s a genetic disorder. And then they’ll ask me, oh, who else has it? And I -- and you know, the results actually came back that it just started with him, so you know, nobody -- nobody in our family has it...”
	Unacceptance by others	1539	“He blocked it out ...you know, treat her like a baby -- like a normal baby, and he makes plans for the future for her. So that’s pretty much all he can handle.”
	Suggestions by others	1549 ^b	“You know, she’s kind of like - she raised - [laughter] she’s raised like all my brothers and sister, and she, you know, starts Googling stuff and saying, oh, well if you tried this, this can cure him...”
		1534	“...nobody outside the family because you know, they’re like, some people do not believe he has autism, and just him, us raising him in the wrong way. That’s why we keep some things to ourselves.”
Barriers to CES	Job	1509	“...it’s been extremely hard with my job, extremely hard with his father’s job. It’s been really crazy. It’s not an easy situation. The appointment, the therapies, me going here or going there is like, oh my gosh.”
	Transportation	1539	“No, because I was receiving the transportation through her insurance. It was more, like, mentally draining having to wait for - for a car service for almost an hour. And, you know - and the child is hungry, you’re hungry, you have all these [bags?], and you’re at a hospital.”
	Language	1554	“Sometimes when those that work with Dr. Iglesias would come, they would put them on, but I would end up speaking in English because I can also speak it, but I like to speak in my language, in Spanish.”
		1563	“Yes, because I don’t speak English and I also don’t have anyone to help me.”
Additional Resources	Resources in Spanish	1556	“...no one speaks Spanish there and I’ve tried to communicate with them by email in Spanish and they respond in Spanish, that is they translate it and ... but I communicate very little because it’s difficult...”
		1556	“Yes, there is very little information in Spanish. Up until now, I still have very little information in Spanish, so I would like to know how are the symptoms or what happens next or how do treatments react and almost all of that isn’t in Spanish. There is very little.”
Religion and Faith		1590	“Well, I wait in God. To change, to be something...”
		1563	“Yes, I have faith in god that it could change...”
		1563	“...only a miracle from God that that did happen.”
		1554	"I didn't want my son to be like this, but that's how God gave him to me and I accept it."
		1554	"Again, I tell you, God over all things because God knows why he gave him to me like this."

^a Negative result on CES, VUS on epilepsy panel

^b = Negative result on CES, pathogenic deletion on microarray

Appendix VI. Interview Script

Introduction

- Tell me a little bit about why your child was referred to genetics or why you sought a genetics evaluation.
- How and when did you first hear about exome sequencing? (Probe to find out when they first heard about it, who told them about it, or where they heard/read about it)
- What were your first feelings or thoughts about whether this was something you wanted to consider for your child? What did you think this could add to your child's care? What was your attitude towards genetic testing prior to getting the test done?
- When making the decision about whether to do the genetic testing or not, did you seek input from other doctors or medical professionals beyond your genetics team? Apart from medical professionals, did you ask anybody else their opinion about what you should do? (If yes) Who was that and why did you ask them for advice? What were the most important reasons why you decided to have the testing done?

Pre-test/Consent Visit

- After hearing about the test, what resources did you use to learn more?
- Is there information you wish you had been given or understood before your child had the test? (If yes) What information is that? (probe about if they understood what positive, negative, VUS, and secondary finding results meant)
- What were your hopes and expectations about the results for this test?
- What were your reservations and doubts about this test?
- Could we have done a better job discussing the testing during your visit? How so?

Language

- What language did you use to speak with the doctor about the test? With the genetic counselor?
- Was this the language you felt most comfortable speaking in about this topic? If no, what would you have preferred?
- Did you use a translator/interpreter? If yes, who? Was the translator/interpreter on the phone, in person, a relative/friend at the appointment? How was this experience?
- Did you have any difficulty finding other resources in your preferred language?

Cost/Logistics

- What made you decide to go to the genetics appointment? Did you think about not going or cancelling the appointment? Why?
- Did you take a day off work to attend the appointment? Did you need childcare for your other children to attend the appointment?
- What other arrangements, if any, did you make in order to attend the appointment?

- How did you travel to the appointment? How did you pay for this transportation?
- Do you remember if the entire cost of the testing was covered by your insurance or if you had to pay any part of the cost? (If had to pay some part of cost) How much did you have to pay?
- Did you consider the cost of testing when deciding whether or not to have it?
- Did you receive any assistance (from friends or family members) to help you to pay for the test?
- How much would you have been able and willing to pay for the test if your insurance had not covered it?

Understanding of results

- Who told you the results for the first time? The second time?
- How did they communicate the results for each discussion (e.g., in face-to-face meeting with physician, by mail, by telephone)?
- What language did the provider speak to tell you about the results? Was this your preference? Was an interpreter used? Who?
- Were you satisfied with how you learned the results? If not, how could this experience have been better? (by phone, in person, with someone else present?)
- Did you feel the results were communicated in a way that enabled you to understand what they meant for your child? What would you have liked to have been done differently?
- How do you feel about the results and why? What have the positives and negatives been about having this result?
- Are your feelings about the results what you expected they would be? If not, how are they different from what you expected?
- Did you have any misunderstandings about the test results that you understand better now?
- Did you look for any additional information about your child's results online or somewhere else? Where did you find information? Did you find that information helpful? Not helpful?

Social Impact

- Did you share the results with your child's other parent? What was his/her response? Was it the same or different to your reaction? Was his/her reaction helpful or harmful? In what way?
- Who did you share your child's results with? Did you share your test results with any other family members, friends, your child's teachers?
- Did you share your test results with your child's therapists/doctors? Which doctors? What was their reaction?

- Have the results changed their perceptions/feelings about your child? Have the test results changed your interactions with these people? Brought you closer? Weakened your relationship? Caused conflict?
- Have any of your family members had genetic testing after learning about your child's test results? (inherited conditions only)
- Are there family members you decided not to share the results with? (If yes) Why did you decide not to share the results with them?

Life Impact

- Have the results allowed you to join any specific support groups, research studies or meet other individuals with the same condition? If so, what impact did that have? If not, what prevented you from finding/joining support groups? Were you unaware that these groups were available to you? Uninterested? Could not find a group in your preferred language?
- Did anything negative happen to you in your job, insurance, or other areas of your life because of the results you received?

Decision Satisfaction/Regret

- Overall, how do you feel about your decision to have testing for your child?
- Currently, what do you think has been the greatest benefit from the test?
- If you were able to go back and choose the most optimal time for your child to have this test, when would you have liked to have it done? (probe to find out if they wanted it before a certain age or symptom onset)
- What would you have changed about this experience?

In Conclusion

- What do you think has been the most difficult part of the entire process from when you first began thinking about having the test through getting the result?
- Is there any kind of additional information or support that would have been helpful for you to have at any point in the process? (If yes) What kinds?
- What advice would you give other parents contemplating this test for their child?
- We are specifically trying to learn more about the experience of having this kind of testing in Latino families. How do you think your experience may have been different from people from other cultures or ethnicities?
- Before we end, is there anything else you would like to say about the whole exome sequencing experience?

Introduce that we will be sending them an electronic link/ paper survey with 8 questions \$25 gift card.