Parent Experience with Genetic Testing for Pediatric Epileptic Encephalopathy: What Can We Do Better?

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Rhonda L. Feinbaum

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ABSTRACT

Parent Experience with Genetic Testing for Pediatric Epileptic Encephalopathy: What Can We Do Better?

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Rhonda L. Feinbaum

Epileptic encephalopathies are a group of severe childhood disorders characterized by seizures and cognitive and developmental deficits. Genetic testing is frequently used in the diagnosis of epileptic encephalopathy. The expanding list of epilepsy genes as well as the genetic and phenotypic heterogeneity of epileptic encephalopathy complicates the interpretation of genetic test results. The main goal of this study was to determine whether families of children with epileptic encephalopathy were provided with sufficient information to understand their child’s genetic test results and, if not, identify ways to improve their experience. This online survey asked parents and guardians of children with epileptic encephalopathy about their experience with genetic testing, the information provided pre-test and at the return of results, their understanding of the information, and what information would improve the genetic testing experience. In this retrospective study, respondents indicated a high level of perceived understanding of genetic test results (average 4.26 out of 5, $SD=0.79$, $n=64$). At the time genetic test results were returned, only 32.8% recalled receiving online information to better understand their child’s genetic test results and only 18.8% were provided with ways to contact other
families \((n=64)\). Respondents reported that these information sources were useful in learning about their child’s diagnosis. Thus, information that patient’s families need and want is frequently not given to them by health care professionals at the results return session. This study suggests that a concerted effort by health care professionals to offer comprehensive information and resources in the context of an ongoing dialogue with patient’s families would improve their understanding of genetic testing for epileptic encephalopathy.

Keywords: epileptic encephalopathy, pediatric epilepsy, genetic testing, family, parent, experience, understanding, genetic counselor
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INTRODUCTION

The epileptic encephalopathies are a group of severe childhood epilepsies characterized by seizures, abnormal electroencephalographic (EEG) findings and cognitive and developmental delays. In these disorders, uncontrolled epileptic activity contributes to cognitive impairment but many also have underlying neurodevelopmental abnormalities. (Hamdan et al., 2017; I. Helbig & Abou Tayoun, 2016). Seizures are often difficult to control with available therapeutic options in patients with epileptic encephalopathies. Epileptic encephalopathy syndromes are clinically defined by the type of seizures, age of onset, course of disease and the co-occurrence of other features. A subset of the earliest onset epileptic encephalopathies is described in Table 1.

Many genes linked to epilepsy have been identified in the last twenty years. The relatively recent ability to detect rare de novo variants using next generation sequencing technologies of family trios has greatly expanded the list of epilepsy genes (EpiPM Consortium, 2015; Hamdan et al., 2017; I. Helbig & Abou Tayoun, 2016; Mei, Parrini, Marini, & Guerrini, 2017). Epileptic encephalopathy genes are expressed in the brain and affect either development of brain structures or fundamental neurological processes including membrane excitability and synaptic function; some of these genes overlap with those of other neurodevelopmental conditions such as autism (Srivastava & Sahin, 2017). Genes encoding ion channels such as $SCN1A$, $SCN2A$, $SCN8A$, $KCNQ2$ and $KCNT1$ play a prominent role in the etiology of many epileptic encephalopathies. Epileptic encephalopathy genes exhibit both genetic heterogeneity, whereby variants in multiple genes result in the same clinical diagnosis, and phenotypic
heterogeneity, whereby variants in a single gene may cause epileptic encephalopathy as well as more benign or otherwise distinct forms of epilepsy (I. Helbig & Abou Tayoun, 2016).

Genetic testing is now routinely used in the diagnosis of children who present with seizures and cognitive or developmental delay (Poduri, 2017). A genetic diagnosis may aid in prognosis, guide treatment, reduce further testing, offer access to drug trials and provide parents with information for family planning as well as enable them to find a supportive community (Berkovic, 2015; Brunklaus et al., 2013; Poduri, 2017). The choice of genetic test depends on clinical evaluation as well as family history and for some patients the “diagnostic odyssey” involves multiple genetic tests. Genetic tests used in pediatric epilepsy include single gene sequencing, chromosome microarray to look for copy number variants (CNV), multi-gene panel, whole exome sequencing (WES) or whole genome sequencing (WGS) (K. Helbig, 2016). The most common test ordered today is a multi-gene panel of which there are over 100 commercially available that range in size from a handful to thousands of genes (Chambers, Jansen, & Dhamija, 2016; Ferraro, Pollard, & Helbig, 2016).

Realistic expectations about the likelihood of getting a positive result and the possibility of a negative result or a variant of uncertain significance (VUS) are important to establish with patients and families prior to genetic testing. Although the diagnostic yield varies with the patient cohort, it is approximately 5% for microarray and up to 45% for large multi-gene panels and whole exome sequencing (K. L. Helbig et al., 2016; Mercimek-Mahmutoglu et al., 2015; Poduri, 2017). Thus, a genetic cause may not be identified in about half the patients. In addition, the use of genome-wide tests means the potential for a secondary finding, a pathogenic variant in a clinically relevant gene unrelated to the primary indication of epilepsy, needs to be discussed with patient’s families (Ferraro et al., 2016; Poduri, Sheidley, Shostak, & Ottman, 2014).
<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Onset</th>
<th>Seizures</th>
<th>EEG</th>
<th>Other features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ohtahara Syndrome&lt;sup&gt;ab&lt;/sup&gt;</td>
<td>&lt;3 months (often by 10 days after birth)</td>
<td>primarily tonic * (may develop others including partial, myoclonic, tonic/clonic, infantile spasms)</td>
<td>burst suppression**</td>
<td>treatment refractory, often patients have underlying structural brain abnormality, infants are hypotonic and have difficulty feeding, may progress to other epileptic disorders, associated physical (frequently spasticity in limbs) &amp; cognitive disabilities, may have reduced life expectancy</td>
</tr>
<tr>
<td>West Syndrome&lt;sup&gt;cd, e&lt;/sup&gt;</td>
<td>4-8 months</td>
<td>infantile spasms† (evolve into other types as child gets older)</td>
<td>hypsarrhythmia†</td>
<td>treatment refractory, seizures infrequent at first then progress to runs or clusters, babies become irritable &amp; may behave as if they cannot see, associated developmental regression &amp; delay with mild to severe learning difficulties, may have abnormal skin structure, may have reduced life expectancy</td>
</tr>
<tr>
<td>Dravet Syndrome&lt;sup&gt;fg&lt;/sup&gt; (Severe myoclonic epilepsy of infancy)</td>
<td>&lt;1 year</td>
<td>febrile seizures, myoclonic, other seizure types, status epilepticus§</td>
<td>hypsarrhythmia</td>
<td>treatment refractory, early development often normal (&lt;2yrs), seizures escalate &amp; may be triggered by light, heat or emotion, associated developmental delay &amp; cognitive &amp; behavioral impairment, often language &amp; gait are affected, skin photosensitivity, may have reduced life expectancy</td>
</tr>
<tr>
<td>Epilepsy of infancy w/ migrating focal seizures&lt;sup&gt;h,i,j&lt;/sup&gt;</td>
<td>&lt;1 year</td>
<td>focal seizures, status epilepticus</td>
<td>shifting foci i.e. migrating spikes</td>
<td>microcephaly, hypotonia, feeding difficulties (often need feeding tube), severe neurological impairment, may have reduced life expectancy</td>
</tr>
</tbody>
</table>

Table 1. Examples of early onset epileptic encephalopathies

* tonic seizures- stiffening of arms and legs  
** burst suppression- high amplitude spike followed by little activity  
† infantile spasms- sudden bending of body forward with stiffening of arms and legs  
‡ hypsarrhythmia- chaotic, high amplitude interictal pattern  
§ status epilepticus- seizures lasting longer than 5 minutes or when seizures are so close together a person doesn’t recover between seizures  
The utility of a positive genetic test varies with each case. On the one hand, genetic testing can confirm a clinical diagnosis, for example 80% of children with Dravet Syndrome have a variant in SCN1A. In addition, there are more than a dozen genes for which identification of a pathogenic variant can impact treatment, notably the avoidance of antiepileptic sodium channel blockers for those with SCN1A variants and the prescription of a ketogenic diet for patients with pathogenic variants in the glucose transporter gene, SLC2A. (Akman, Yu, Alter, Engelstad, & De Vivo, 2016; Poduri et al., 2014). However, with use of large gene panels and genome-wide technologies there is a high likelihood of obtaining a variant in a newly discovered gene that provides little useful clinical information to the patient (Chambers et al., 2016; I. Helbig, 2017).

It is challenging to predict the severity or course of epilepsy based on genotype because of phenotypic heterogeneity. In a study by Stamberger et al. (2016), variants in STXBPI encoding syntaxin binding protein 1 resulted in a diagnosis of early onset epilepsy and encephalopathy, Ohatahara, West, or Dravet Syndromes. One third of these patients became seizure free over time whereas another one third were refractive to therapy with no clear genotype/phenotype correlation (Stamberger et al., 2016). Further, STXBPI pathogenic variants can cause developmental delay and intellectual disability in the absence of seizures, which may be difficult for families who are constantly on edge waiting for the occurrence of a seizure that never comes (Gburek-Augustat et al., 2016). Even in the case of SCN1A where there is generally good genotype/phenotype correlation the outcome is not always predictable. Severe loss of function SCN1A variants are more likely to cause epilepsy with intellectual disability whereas certain missense variants result in milder Generalized Epilepsy with Febrile Seizures Plus, (I.
Helbig, 2017). However, there are reported cases of sibs with the same \textit{SCN1A} variant with discordant clinical outcomes (Depienne et al., 2010).

Parents expect genetic testing to be of personal use (Hayeems, Babul-Hirji, Hoang, Weksberg, & Shuman, 2016). In interviews with people with epilepsy and their family members, genetic information was perceived as having the potential to increase personal control and the ability to care for those affected by epilepsy (Shostak, Zarhin, & Ottman, 2011). However, parents of children that underwent genetic testing for other complex disorders, particularly in cases where genetic test results are ambiguous and have little effect on patient care, have expressed frustration at the lack of clarity in the communication of results and a poor understanding of the test results (Chen, Xu, Huang, & Dhar, 2013; Kiedrowski, Owens, Yashar, & Schuette, 2016). Preparing patient families for the type of test results that might be returned and conveying the possible intricacy of the genetic test results for epilepsy, including their impact (or lack thereof) on clinical management and reproductive risk is important. This often requires a concerted effort by a team of health care providers including neurologists and genetic counselors (Sheidley & Poduri, 2012).

Genetic counseling is a key component of a recent model proposed for advancing precision medicine in the treatment of pediatric epilepsy (Smith et al., 2016). Studies have demonstrated that genetic counseling improves knowledge for a variety of genetic conditions including cancer, deafness and congenital heart disease (Baldwin, Boudreault, Fox, Sinsheimer, & Palmer, 2012; Blue, Kasparian, Sholler, Kirk, & Winlaw, 2015; Meiser & Halliday, 2002). In addition, genetic counselors are trained to engage with patients, advocate for them and provide psychosocial support for patients (and families) living with a genetic condition (Veach, Bartels, & LeRoy, 2007).
Given the rapid and accelerating incorporation of genetic testing into the care of children with epilepsy, it is critical to understand the family’s experience. This is the first study to look at how genetic testing for epilepsy impacts patients and their families with a specific eye to whether the family’s informational needs are being met. The goals of this project are 1) to better understand how the process of genetic testing for pediatric epilepsy transpires and to what degree genetic counselors are involved, 2) determine whether the information provided is sufficient for parents to understand the health implications for their child, the impact of genetic results on their reproductive future, make necessary decisions for their child and find a supportive community and 3) identify information that families would like to have going forward in order to improve the experience of genetic testing for families with children with epilepsy.
METHODS

The Brandeis University Institutional Review Board deemed this research exempt from further review (http://www.brandeis.edu/ora/compliance/irb/about.html).

Participants

Parents or guardians of children with epilepsy and cognitive delay were recruited through a link posted on the following syndrome-based websites and Facebook pages: Aaron’s Ohtahara Foundation, The Cute Syndrome Foundation: SCN8A Families Support, The Dravet Syndrome Foundation, Infantile Spasms Community, and The Lennox-Gastaut Syndrome Foundation (Appendix A). The anonymous survey was accessible through the online Qualtrics survey platform for six weeks.

Participants provided consent by clicking through the link to begin the survey. Eligibility required that parents or guardians have a single child with epilepsy and cognitive delay or impairment who had genetic testing since January 1, 2014. Upon completion of the survey, participants were given the choice to enter a separate random drawing for a $75 Amazon.com gift card.

Instrumentation

The survey consisted of 30 multiple choice and Likert scale questions and three open response questions (excluding the final question about entry into the random drawing for the gift
certificate); the maximum number of questions for any single participant was 32 (Appendix B). The survey included questions about the following:

1) Demographics and background information
2) Logistics of genetic testing
3) Results of genetic testing
4) Communication by health care providers about genetic testing and results
5) How participants felt about the information provided regarding genetic testing and results
6) What information would improve the genetic testing experience for parents/guardians of children with epilepsy

Three open-ended questions allowed participants an opportunity to provide additional input including a) what information would have been helpful before their child’s genetic test was done b) what information would have been helpful upon receipt of results and c) general advice to health care professionals who will be counseling similar families in the future.

Data Analysis

SPSS software version 24 was used for analysis which included frequencies, descriptive statistics, chi-square analysis to determine the relationship between nominal variables and independent-samples t-tests to compare means. Open response questions were evaluated by thematic analysis.

Respondents rated a series of statements about their understanding of their child’s genetic test results on a Likert scale (strongly disagree, somewhat disagree, neither agree nor disagree, somewhat agree, strongly agree). Each answer was assigned a numerical value (strongly
disagree=1, strongly agree=5). Calculation of Cronbach’s alpha (0.806) demonstrated internal reliability and allowed for combination of values across Likert scale statements.

In order to generate one value that represented a respondents overall level of understanding about their child’s genetic test result, a “perceived understanding” combined mean score was computed from the following Likert statements: (1) I know who to contact if I have questions about my child’s genetic test result, 2) I am confident in my understanding of my child’s genetic test result, 3) I understand how this genetic information may or may not help my child, 4) I understand the risk of epilepsy to any other children or family members, 5) I have the necessary information to find the emotional support my family needs). Calculation of a respondent’s “perceived understanding” mean score required a response to at least four of the five statements.

Several dichotomous variables were created from the data for analysis.

1) Respondents were separated into two groups, +GC or -GC, for both the pre-test and results sessions based on whether a genetic counselor was included (either solo or as part of a team) in information delivery.

2) Two groups of respondents were defined with respect to whether all the five topics listed in Figure 2 were discussed at the initial results return session or not all (either none or a subset).

3) Likewise, respondents were separated into two groups based on whether they were given all four types of information in Figure 3 or not all (either none or a subset).
RESULTS

A total of 69 surveys were used for analysis: 107 people entered the survey, 10 did not proceed far enough to answer all required eligibility questions and 28 were ineligible. For those questions that all individuals saw, the total number of respondents ranged from 62-69 and is presented with each analysis.

Demographics of Sample

All respondents were a parent or guardian of a single child with epilepsy and developmental or cognitive delay who had genetic testing since January 1, 2014. Respondents reported the current age of their children to be from less than one year to 22 years ($M=6.3$ years, $SD=5.1$ years, $n=69$). In 89% of the 69 cases, seizures or developmental delay were first recognized by parents in their children at less than one year of age (4.3% 1-2 years, 4.3% 2-4 years, and 1.4% 4-6 years).

Of 64 respondents who provided information on their level of education, 14.1% completed high school, 28.1% 2-year college, 29.7% 4-year college and 28.1% post graduate education. Sixty-two respondents were involved in at least one of 17 foundations or support groups; the top three were the Dravet Syndrome Foundation (27/62), The Cute Syndrome Foundation (17/62) and Aaron’s Ohtahara Foundation (10/62) (Appendix C).
Overview of genetic testing

The genetic tests referred to in this study were ordered by health care providers across North America as well as in Europe and Australia (Table 2). Many children (60.8%) had more than one genetic test and had their most recent genetic test at four years or younger (63.7%) (Table 2). Sixty-nine percent of genetic tests were done in an outpatient setting and 30.9% during an inpatient stay in the hospital (Table 2). Of the 69 respondents, 84.1% reported a positive genetic test result that identified a genetic cause of their child’s epilepsy, 5.8% had a negative result with no genetic cause identified, 8.7% reported an uncertain test result and 1.4% (1 respondent) did not know or understand the result of their child’s genetic test (Table 2).

For the 58 respondents who reported that their child had received a positive genetic test result, presumed pathogenic variants were identified in 18 genes (Appendix D). Variants in *SCN1A* (26/58) or *SCN8A* (18/58) were reported in 75% of the individuals. Seven respondents reported that their child had variants in more than one gene, with one individual reporting pathogenic variants in three genes (*SCN1A, SCN3A, DHCR7*).

Pre-test information session

Prior to genetic testing for their child, 86.8% of the 68 respondents met with a health care professional to discuss the test whereas 8.8% did not and 4.4% were unsure. The 59 respondents who met with a health care professional before testing reported that they discussed the test with the following: 1) neurologist (*n*=36), 2) genetic counselor (*n*=30), 3) geneticist (*n*=26) 4) epileptologist (*n*=17), 5) primary care physician (*n*=7) and 6) nurse/nurse practitioner (*n*=4). Forty-two percent met with a single provider while the rest interacted with a team of two or more. Fifty-one percent met with a genetic counselor and in all but one case the genetic counselor was part of a team.
When asked whether they were provided with written or online material describing the genetic test to take home, 50% of 68 respondents replied “Yes”, 36.8% “No” and 13.2% didn’t remember. For those who were not provided with material to take with them, 80% said that they would have wanted this information.

| Region where sample for child's most recent genetic test was collected | CT, MA, ME, NH, RI, VT, Prince Edward Island | 8 (12.5%) |
| DC, DE, MD, NJ, NY, PA, VA, WV, Quebec | 7 (10.9%) |
| AL, FL, GA, KY, LA, MS, NC, SC, TN, Puerto Rico, Virgin Islands | 4 (6.3%) |
| AR, IA, IL, IN, KS, MI, MN, MO, MS, NE, ND, OH, OK, SD, WI, Ontario | 15 (23.4%) |
| AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan | 9 (14.1%) |
| AK, CA, HI, ID, NV, OR, WA, British Columbia | 9 (14.1%) |
| Europe | 10 (15.6%) |
| Australia | 2 (3.1%) |
| Total | 64 |

| Number of genetic tests child has had in his/her lifetime | 1 | 26 (37.7%) |
| 2 | 13 (18.8%) |
| 3 | 6 (8.7%) |
| 4 or more | 12 (17.4%) |
| > 1, unsure of exact number | 11 (15.9%) |
| Don't know/understand | 1 (1.4%) |
| Total | 69 |

| Age of child when his/her most recent genetic test was performed | 0-6 months | 9 (13.0%) |
| 7-12 months | 12 (17.4%) |
| 1 year old up to 2 years | 9 (13.0%) |
| 2 years old up to 4 years | 14 (20.3%) |
| 4 years old up to 6 years | 6 (8.7%) |
| 6 years old up to 10 years | 10 (14.5%) |
| 10 years and older | 8 (11.6%) |
| I can't remember | 1 (1.4%) |
| Total | 69 |

| Sample taken for genetic test | Inpatient in hospital | 21 (30.9%) |
| Outpatient | 47 (69.1%) |
| Total | 68 |

| Result of child's most recent genetic test | Positive | 58 (84.1%) |
| Negative | 4 (5.8%) |
| Uncertain | 6 (8.7%) |
| Don't know or understand | 1 (1.4%) |
| Total | 69 |

Table 2. Overview of genetic testing
The majority of the 68 respondents (79.4%) reported that they were given enough information prior to their child’s genetic test to understand what information the test would provide. However, 13.2% said that they were not given the information they needed and 7.4% did not remember. Of the nine respondents who were not satisfied with the information provided before the test, two did not discuss the test with a health care provider prior to sample collection.

Respondents who reported dissatisfaction with the information provided prior to administration of the genetic test were given the opportunity to comment on what would have been helpful (Appendix E). Eight out of nine offered suggestions, with the most common theme, expressed by four respondents, being a desire for general information about what the test looked for and what the possible outcomes of the test might be. One parent explained what he/she would have liked to have known:

“What mutations they were looking for and what possible conditions these results would show.”

Two of the eight respondents expressed confusion as to what a negative test result might mean for the future and two respondents commented that they agreed to the test without a full understanding of what it might reveal.

“We agreed to the test because the neurologist indicated their (sic) may be an underlying food allergy or environmental allergy. The genetic test was part of a complete epilepsy panel. Never did we imagine that the results would come back as a genetic mutation.”

Delivery of results

Genetic test results were initially delivered most frequently over the phone (50%, n=68) followed by in person (38.2%), in writing (10.3%) and 1 respondent did not remember (1.5%).
Approximately 66% of those who received the results over the phone or in writing were offered an in-person follow-up appointment.

The health care professionals with whom genetic test results were first discussed are shown in Figure 1A. Twenty-eight of the 68 respondents initially discussed results with a genetic counselor. Two reported that they did not discuss the genetic test results with a health care provider and one was not sure/did not remember. Of the 65 respondents who spoke with a provider, 58.5% discussed genetic test results with a single health care professional while the remaining interacted with teams of two or more (Figure 1B). In 20 of the 28 cases where respondents discussed genetic test results with a genetic counselor they were part of a team.

![Figure 1. Health care professionals involved in discussion of genetic test results](image)

**A**) Health care professionals with whom respondents initially discussed genetic test results (*n*=68). **B**) The number of health care providers with whom respondents initially discussed genetic test results (*n*=67).

Next, we asked respondents about the topics they recalled being discussed at the initial results disclosure (Figure 2). Genetic test results were discussed 96.9% of the time and 78.1% of the time the discussion included how the results would impact their child’s future health.

Respondents recalled discussing the three remaining topics less frequently; the impact of genetic
test results on their child’s treatment were discussed 60% of the time, what the results might mean for other members of the family (61.5%), or to future pregnancies (59.4%) of the time. Of the 65 respondents who provided feedback on at least four out of five queries, 42% indicated that they recalled discussing all five topics listed in Figure 2 in the initial conversation about their child’s genetic test results.

Figure 2. Topics discussed with respondents at the time genetic test results were returned

Respondents were also asked about whether they recalled being given specific resource material at the time that genetic test results were returned (Figure 3). Respondents reported being given a copy of their child’s genetic test report 78.5% of the time and 69.2% were provided with a written explanation of the genetic test results. Online resources to help them understand genetic test results (32.8%) or information on how to connect with other families (18.8%) were less frequently offered.
Figure 3. Information given to respondents at the time genetic test results were returned

We subsequently asked respondents if there was any information that they were not given that would have been helpful to understand their child’s genetic test results. Thirty-one respondents (46.3%) answered “yes” and 27 provided feedback about what information they would have liked to receive (see Appendix F for all responses). The most repeated theme (7/27) was a need for information about how to connect with other families with the same genetic condition and/or find support groups. One respondent wrote

“It took me almost a year to find the Facebook group that connected me to other families.”

Another theme expressed by two respondents quoted below was a desire for more information on the genetic condition and/or mutation, particularly information about prognosis.

“Online places to go to learn specifics about our disease.”
“It would have been helpful to have been told (even a ballpark) what percentage of SCN1A mutations go on to a Dravet syndrome diagnosis.”

**Reported confidence in understanding of genetic test results**

Respondents evaluated their experience with genetic testing for their children by rating a series of five statements focused on their understanding of the genetic test results. The majority of respondents indicated that they were comfortable with and understood how the genetic test result might affect their child and family (Table 3). The respondents who either somewhat or strongly agreed with each statement ranged from 71.4%-87.3% (Table 3). The respondents’ overall “perceived understanding” computed as mean of the first five statements in Table 3 was 4.26 (out of 5) with a standard deviation of 0.79.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly disagree</th>
<th>Somewhat disagree</th>
<th>Neither agree nor disagree</th>
<th>Somewhat agree</th>
<th>Strongly agree</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>I know who to contact if I have questions about my child's genetic test result</td>
<td>1 (1.6%)</td>
<td>4 (6.3%)</td>
<td>5 (7.8%)</td>
<td>13 (20.3%)</td>
<td>41 (64.1%)</td>
<td>64</td>
</tr>
<tr>
<td>I am confident in my understanding of my child's genetic test result</td>
<td>2 (3.2%)</td>
<td>1 (1.6%)</td>
<td>5 (7.9%)</td>
<td>25 (39.7%)</td>
<td>30 (47.6%)</td>
<td>63</td>
</tr>
<tr>
<td>I understand how this genetic information may or may not help my child</td>
<td>0 (0.0%)</td>
<td>3 (4.8%)</td>
<td>7 (11.1%)</td>
<td>16 (25.4%)</td>
<td>37 (58.7%)</td>
<td>63</td>
</tr>
<tr>
<td>I understand the risk of epilepsy to any other children or family members</td>
<td>4 (6.3%)</td>
<td>3 (4.7%)</td>
<td>4 (6.3%)</td>
<td>11 (17.2%)</td>
<td>42 (65.6%)</td>
<td>64</td>
</tr>
<tr>
<td>I have the necessary information to find the emotional support my family needs</td>
<td>3 (4.8%)</td>
<td>9 (14.3%)</td>
<td>6 (9.5%)</td>
<td>16 (25.4%)</td>
<td>29 (46.0%)</td>
<td>63</td>
</tr>
<tr>
<td>I am clear on what the next steps are to find a cause for my child's epilepsy*</td>
<td>0 (0.0%)</td>
<td>1 (1.6%)</td>
<td>0 (0.0%)</td>
<td>2 (3.2%)</td>
<td>2 (3.2%)</td>
<td>9</td>
</tr>
<tr>
<td>I have a follow-up plan to obtain updated genetic information**</td>
<td>1 (1.6%)</td>
<td>1 (1.6%)</td>
<td>1 (1.6%)</td>
<td>1 (1.6%)</td>
<td>1 (1.6%)</td>
<td>6</td>
</tr>
</tbody>
</table>

Table 3. Reported understanding of genetic test results

*Only respondents who indicated that their child had received either negative or uncertain genetic test results saw this statement.
**Only respondents who received an uncertain genetic test result viewed this statement.
The nine respondents who reported receiving an uncertain or negative genetic test result were asked about their understanding of the next steps for their children (Table 3). Four strongly or somewhat agreed with the statement “I am clear on what the next steps are to find a genetic cause for my child’s epilepsy” whereas five strongly or somewhat disagreed. Likewise, for the six respondents with uncertain genetic test results, two strongly or somewhat agreed with the statement “I have a follow-up plan to obtain updated genetic information” and three somewhat or strongly disagreed.

Sources of information

Respondents \((n=65)\) were asked to recall all sources of information that helped them understand their child’s genetic test results (Figure 4). The five sources most often reported to be utilized by respondents were 1) Syndrome support groups and Facebook forums 2) neurologist or epileptologist 3) internet search 4) genetic counselor and 5) geneticist.

Figure 4. Sources of information respondents reported using to understand genetic test results \((n=65)\).
Suggestions for health care professionals who will be counseling other families

We asked respondents to provide advice for health care professionals who will be working with families undergoing genetic testing for epilepsy in the future and 27 provided feedback (Appendix G). As in the previous open response questions, the most common themes were the importance of providing families with information on how to make connections with others with similar diagnoses (30%) and the desire for specific information regarding their child’s genetic finding (15%).

Although not addressed directly in our survey questions, several noteworthy themes were revealed about how genetic testing information is delivered. First, parents reported a desire to be treated with respect and as an equal contributor to their child’s care.

“Try to be compassionate and understanding. Listen to the parents and respect the fact that although you may be an expert in your field, we are experts in our children.”

Second, as reflected in the statements by the following two respondents, it is critical that providers be well-informed and, for newly defined genetic epilepsies, communicate honestly with patients about the current limitations and gaps in knowledge.

“I think that health care professionals often overstate what they can know about test results in any given moment.”

“That they (health care providers) educate themselves prior to meeting with a parent.”

Third, patients need to hear information multiple times and want an opportunity to reconnect with health care professionals after the return of genetic test results so that they can ask follow-up questions.

“Listen carefully, explain things as basically as you can, offer another follow up appointment within the month to answer questions that arise after the diagnosis sinks in.”
DISCUSSION

The main goal of this study was to determine whether families of children with epilepsy were provided with sufficient information to understand their child’s genetic test results and, if not, identify ways to improve their experience. We found that the information discussed at the results return session and resources provided to respondents impacted their perceived understanding of genetic test results. The most striking result of this work was the need respondents expressed for information on ways to connect with other families with similar diagnoses and for online resources that provide information specific to their child’s genetic finding. Inclusion of a genetic counselor in the pre-test information session and the return of results may positively impact information delivery, but our data set is not adequate to demonstrate this definitively. Moving forward, special attention should be paid to making sure patients and their families get the information they need, when they need it, and we offer suggestions on how to do so.

Information provided is related to understanding of genetic test results

The relation between information discussed with or provided to respondents at the return of genetic test results and their perceived understanding was assessed using independent-samples t-tests. Respondents were grouped based on whether they recalled that all five topics (what the results were, what they could tell about a child’s future and how the results would impact treatment, other family members and future pregnancies) were discussed at the time genetic test results were returned. There was a significant difference in the perceived understanding scores
for respondents who reported that all topics were discussed ($M=4.61, SD=0.58$) and those who reported four or less topics discussed ($M=4.01, SD=0.83$); $t(61.94)=3.39, p=0.001$. Respondents who discussed all five topics with their health care providers exhibited greater understanding of the genetic test results.

In addition, respondents were similarly grouped based on whether they received all the following information: a copy of their child’s genetic test report, written information explaining the results, online resources to help understand genetic test results and information on how to connect with other families with similar diagnoses. Respondents who were given all four information sources ($M=4.78, SD=0.44$) had higher perceived understanding of their child’s genetic test results than those who were given three or less ($M=4.16, SD=0.80$); $t(22.33)=3.50, p=0.002$.

One potentially confounding factor is that the results of genetic testing may influence reported understanding; a positive test result provides clarity and may enable access to specific information relevant to the patient. We conducted an independent-samples t-test to compare perceived understanding of respondents whose children had positive genetic test results with those who had negative or uncertain test results. Respondents who indicated that a genetic cause for their child’s epilepsy was identified reported greater understanding of the test results ($M=4.34, SD=0.77$) than those whose child had negative or uncertain genetic test results ($M=3.74, SD=0.73$); $t(62)=2.20, p=0.031$.

To further address the potential effect of test outcome on perceived understanding, we re-analyzed the data from the subset of respondents whose children had positive genetic test results for a relation between topics discussed or information provided and perceived understanding (see Appendix H). These data were similar to that reported above for the whole cohort and supported
the idea that a thorough discussion of the genetic test results as well as providing patients’ families with multiple resources that they can access at home leads to greater understanding.

Our observation that only 32.8% of respondents recalled being given online information to better understand their child’s genetic test results and only 18.8% were provided with ways to connect with other families at the results return session could potentially be explained by a lack of information available. Indeed, of the respondents whose child had a negative or uncertain genetic test result, in which case specific relevant information might be unlikely to exist, 0/9 received any online information resources and 1/9 said that they had been given ways to connect with other families. However, a lack of information provided (38.2% given online information and 20% provided ways to connect with other families) was still observed in the subset of respondents (n=55) for whom a genetic cause for their child’s epilepsy was identified (Appendix I).

Further, when we looked only at the responses from the families who reported that their child had an SCN1A-related epilepsy, which can include a spectrum of epilepsy phenotypes ranging from febrile seizures to Dravet Syndrome, we also observed an information deficit; 28% of 25 respondents recalled being provided with online resources and 25% of 24 respondents said they were given information on how to connect with other families. Resources that provide information on SCN1A and ways to connect with other families with similar diagnoses are available and have been for some time (for example the Dravet Syndrome Foundation was formed in 2009). This suggests that lack of available information is unlikely to be the sole reason that these resources were not given to families.
The role of the genetic counselor in providing information

Although this study was not explicitly designed to test the effect of a genetic counselor’s presence on information delivery, we were curious to see if the presence of a genetic counselor was related to respondents’ report of confidence in pre-test information and/or topics discussed and information sources provided during the results return session. The role of a genetic counselor could not be isolated because in 29/30 pre-test and 20/28 results return sessions genetic counselors worked as members of a team with other health professionals. However, we compared the responses of those who consulted a genetic counselor, either solo or as part of a team, to those who did not.

The relation between the inclusion of a genetic counselor in the pre-test consultation and whether respondents reported that enough information was provided to understand the genetic test was examined by chi-square test. There was a significant relationship between these variables, $\chi^2 (2, n=59) =8.97, p=0.011$. The 7/59 respondents who met with a health care professional but reported that they did not get sufficient information to understand the test were less likely to have interacted with a genetic counselor prior to testing (0/7). In comparison, these seven respondents all met with a neurologist in the absence of a genetic professional.

A chi-square test for independence was also performed to examine the relation between the presence of a genetic counselor and the breadth of topics discussed during the results return session. The relation between inclusion of a genetic counselor and whether all five topics (what the results were, what they could tell about a child’s future, how the results would impact treatment, other family members and future pregnancies) were discussed was significant $\chi^2 (1, n=65) =7.45, p=0.006$. The discussion of genetic test results in sessions that included a genetic counselor was more likely to cover all five topics. In contrast, when a similar analysis was done
comparing those respondents who saw a neurologist and whether all five topics were discussed there was no significant relationship.

The relationship between the presence of a genetic counselor at the results return session and respondents receiving access to further information was also explored using chi-square analysis (Appendix J). With regards to whether a copy of the genetic test report, written information explaining test results, online resources or information to connect with other families was provided at the results return appointment, the presence or absence of a genetic counselor was found to be related in just one case, the provision of online resources. Respondents who met with a genetic counselor were more likely to be given online resources, $X^2 (2, n=64)=13.139$, $p=0.001$.

However, the 18.8% of respondents who reported being given information on how to connect with other families with similar diagnoses were not more likely to have met with a genetic counselor. Existing genetic counseling practice guidelines, for example for families with children with a new diagnosis of Fabry Disease or Down Syndrome, explicitly include information on support groups as an essential resource (Laney et al., 2013; Sheets et al., 2011).

Taken together these data indicate that while the inclusion of a genetic counselor is related to a more thorough explanation of genetic tests and results, genetic counselors have room to improve when counseling families of patients with pediatric epilepsy.

**Limitations of the study**

The greatest limitations to this study were the small sample size, lack of sample diversity and ascertainment and recall bias. A small sample may not accurately represent the target population and limits statistical power. Further, this group of respondents was heavily biased
towards families whose children had received a positive genetic test result; in this cohort 84% reported that a genetic cause for their child’s epilepsy had been identified, well above the expected 25-50% (K. L. Helbig et al., 2016; Mercimek-Mahmutoglu et al., 2015; Poduri, 2017).

The respondents were ascertained through online syndrome-based support groups and therefore the value placed on this resource may not be generalizable to all families with children with epileptic encephalopathy. Although it is difficult to predict the population size that would meet the study criteria, the number of Facebook “likes” for the syndrome-based groups was cumulatively in the tens of thousands, several orders of magnitude greater than our number of respondents. Further, the rapid and short-lived response immediately after posting of the link to the survey indicated that the respondents in this study were highly pro-active with a significant online presence.

Recall bias is a potential confounder in any retrospective study. Respondents were eligible for this study if their child had had genetic testing any time since January 1, 2014 and therefore the event in question may have been several years in the past. In addition, recall may have been impacted by the stress of ongoing medical challenges for their child and the genetic test result itself.

What can we do better?

This study shows that genetic testing for pediatric epilepsy is provided by diverse health care professionals. Genetic counselors are not always included in either the pre-test information session or the return of genetic test results; 44% of all respondents met with a genetic counselor before their child’s test was done and 41% discussed the results with a genetic counselor (n=68 for both). Although more frequent inclusion of genetic counselors might increase the information
and support given to patient families, this may be impractical. Therefore, moving forward, any information sources identified that would be helpful to patient families should be available to a variety of health care professionals.

At the time testing is offered, providing patient’s families with written or online information that explains the genetic test and its potential results would help those who don’t receive pre-test counseling as well as serve as a resource for those who need to revisit the information at a later time. A few genetic testing companies provide patient friendly literature that explains genetic testing for epilepsy and there is general information about genetic testing on sites like the Epilepsy Foundation (https://www.epilepsy.com/learn/diagnosis/genetic-testing). However, there is the room for further development of this literature and genetic counselors could encourage genetic testing companies to provide this information in a form that can be given to families.

Maintaining an ongoing discussion with patient’s families is important. Only about half of the respondents who reported that their child had received a negative or uncertain genetic test result were confident in what the plan was to find a genetic cause or update their child’s genetic test information. Further, of the 41 respondents who received their child’s genetic test results over the phone or in writing, 34% (14) were not offered a follow-up in person appointment to discuss the results. Comments from respondents indicated that a follow-up appointment after the initial results disclosure would be helpful so that they could hear the information again and ask questions once they had time to process the test results. Although some health care professionals already do so, providing a clear plan and an avenue for re-contact may help patients feel supported.
Referrals to syndrome-based support groups were both the most asked for information and the reported number one source of information for this group of respondents. Parent volunteers often run these groups and the groups may evolve over time. The challenge is how to keep track of this information and get it to patient families in diverse settings. Information on support groups for some of the more well-characterized encephalopathies, like Dravet syndrome, is collected on the NIH Genetic and Rare Diseases website (https://rarediseases.info.nih.gov). EpiGC (a group of epilepsy-focused genetic counselors) is collating information on gene-specific as well as syndrome based epilepsy support groups (https://www.epigc.net). Maintaining an up-to-date centralized patient resource for pediatric epilepsy in this rapidly growing field will require significant ongoing effort. Moving forward, health care professionals can support their patients by directing them to these and other resources but also by listening carefully and discussing genetic tests and results thoroughly.
REFERENCES


Poduri, A. (2017). When should genetic testing be performed in epilepsy patients? *Epilepsy Currents, 17*(1), 16-22. doi:10.5698/1535-7511-17.1.16


Appendix A. Recruitment Link

Has your child had genetic testing to find the cause of his/her epilepsy? If so, we invite you to help us help families! Share your experiences with the genetic testing process to help clinicians better serve families in the future – and enter to win a $75 Amazon gift card. Click here to learn more!
Questions?? please contact Rhonda Feinbaum at rfeinbaum@brandeis.edu
Appendix B. Survey Instrument

Q1 Thank you for your interest in our research study. My name is Rhonda Feinbaum, and I am a genetic counseling student at Brandeis University. For my master’s thesis, I am conducting a research study to understand how genetic testing for pediatric epilepsy takes place and whether parents feel that the information provided by health care professionals about genetic testing meets their needs. The aim of this research is to help genetic counselors and other healthcare providers understand parents’ experiences with genetic testing for their children with epilepsy and to identify ways in which they can improve this experience.

Who can participate? You can participate if you are a parent or legal guardian of a child who (1) has epilepsy, (2) has cognitive delay or impairment, and (3) who has had genetic testing since January 2014. We encourage you to participate in this study regardless of whether or not your child received a genetic diagnosis for his or her epilepsy. Participants must be at least 18 years old, and we ask that only one parent or guardian for each affected child respond to this survey.

What does participation involve? The survey will take approximately 15 minutes to complete. It asks questions about your experiences throughout the genetic testing process, including questions about the information (or lack of information) you received from healthcare providers about genetic testing, how results were communicated to you and how you felt throughout the process. It also asks you to identify ways in which the process might have been improved.

How will the results be used? We hope to use the results of this survey to help genetic counselors and other healthcare providers improve the genetic testing experiences for families in the future.

What should I know about this study? The survey is anonymous (we will not ask you for your name) and voluntary. You may exit the survey at any time, and you may choose to skip any question(s) that you prefer not to answer. Participants completing the survey will have the option to enter a raffle to receive a $75 Amazon gift card at the end of the survey. This research study has been approved by the Brandeis University IRB (Institutional Review Board), Waltham, MA.

Who should I contact if I have question about this study? If you have any questions about this study feel free to contact me by email at: rfeinbaum@brandeis.edu or my master’s thesis supervisor, Dr. Judith Tsipis at tsipis@brandeis.edu.

How do I begin? To begin, click [>>] below. By clicking the link below, you are agreeing to participate in this survey.

Thank you again for your interest!
Q2 Are you the parent or legal guardian of a child who has both epilepsy and cognitive delay or impairment?
   - Yes (1)
   - No (2)

Skip To: End of Block If Are you the parent or legal guardian of a child who has both epilepsy and cognitive delay or impa... = No

Q3 Do you have more than one child with epilepsy?
   - Yes (1)
   - No (2)

Skip To: End of Block If Do you have more than one child with epilepsy? = Yes

Q4 In what year was your child with epilepsy born?
   ▼ 1990 (1) ... 2017 (28)

Q5 How old was your child when you first became aware of his/her developmental delay or seizures?
   - 0-6 months (1)
   - 7-12 months (2)
   - 1 year old up to 2 years (3)
   - 2 years old up to 4 years (4)
   - 4 years old up to 6 years (5)
   - 6 years old up to 10 years (6)
   - 10 years and older (7)
   - I can't remember specifically (8)

Q6 Has your child had genetic testing for epilepsy at any time since January 1, 2014?
   - Yes (1)
   - No (2)

Skip To: End of Block If Has your child had genetic testing for epilepsy at any time since January 1, 2014? = No
Q7 How many genetic tests has your child had in his/her lifetime?
   o 1 (1)
   o 2 (2)
   o 3 (3)
   o 4 or more (4)
   o More than one, but unsure of exact number (5)
   o I don't know (6)

Q8 How old was your child when his/her most recent genetic test was performed?
   o 0-6 months (1)
   o 7-12 months (2)
   o 1 year old up to 2 years (3)
   o 2 years old up to 4 years (4)
   o 4 years old up to 6 years (5)
   o 6 years old up to 10 yrs (6)
   o 10 years and older (7)
   o I can't remember specifically (8)

Q9 What was the result of your child's most recent genetic test?
   o POSITIVE: The test identified a genetic change that likely is the cause of my child's epilepsy. (1)
   o NEGATIVE: The test did NOT find a genetic cause for my child's epilepsy. (2)
   o UNCERTAIN: The test found some genetic changes but it is not clear if they caused my child's epilepsy. (3)
   o I don't know or understand the result of my child's genetic test. (4)

Display This Question:
If What was the result of your child's most recent genetic test? = POSITIVE: The test identified a genetic change that likely is the cause of my child's epilepsy.

Q10 If you know the genetic cause of your child's epilepsy, please indicate the responsible gene or genes listed below. (select all that apply)
   □ ARX (1)
   □ CDKL5 (2)
☐ CHD2 (3)
☐ DNM1 (4)
☐ GRIN2A (5)
☐ GABRB2 (6)
☐ GABRG2 (7)
☐ IQSEC2 (8)
☐ KCNQ2 (9)
☐ KCNT1 (10)
☐ PCHD19 (11)
☐ SCN1A (12)
☐ SCN1B (13)
☐ SCN2A (14)
☐ SCN8A (15)
☐ SEPSECS (16)
☐ SLC2A1 (17)
☐ SLC6A1 (18)
☐ STXBP1 (19)
☐ TBC1D4 (20)
☐ TSC1 or TSC2 (21)
☐ Copy number variant (deletion or duplication of multiple genes) (22)
☐ Other. Please explain (23) ________________________________________________
Q11 When answering ALL of the following questions please consider only your child's most recent genetic test, whether or not it resulted in a diagnosis.

Q12 Was your child admitted for an inpatient stay in the hospital when he/she had the genetic test?
   o Yes (1)
   o No (2)

Q13 Before your child's sample was taken for genetic testing, did you discuss the test with a health care professional?
   o Yes (1)
   o No (2)
   o Unsure (3)

Display This Question:
   If Before your child's sample was taken for genetic testing, did you discuss the test with a health care professional... = Yes

Q14 Which of the following health care professional(s) met with you to discuss the genetic test at that time? (select all that apply)
   □ Genetic counselor (1)
   □ Neurologist (2)
   □ Epileptologist (3)
   □ Nurse/Nurse practitioner (4)
   □ Primary care physician (5)
   □ Geneticist (doctor who specializes in genetics) (6)
   □ Not sure/Don't remember (7)
   □ Other health care provider. Please explain (8)

Q15 Did you feel you were given enough information before the genetic test was done to understand what information the test would provide?
   o Yes (1)
   o No (2)
   o Don't remember (3)
Q41 Please explain what information would have been helpful before your child's genetic test was done.

________________________________________________________________
________________________________________________________________

Q16 Were you provided with material describing the genetic test that you could access at home? (written or online)
   - Yes (1)
   - No (2)
   - Don't remember (3)

Q17 Would you have liked to have received such information?
   - Yes (1)
   - No (2)

Q18 Were you told how long it might take to receive the genetic test results?
   - Yes (1)
   - No (2)
   - Don't remember (3)
Q19 Do you feel that information would have been helpful?
   - Yes (1)
   - No (2)

Display This Question:
   If Were you told how long it might take to receive the genetic test results? = Yes

Q37 Was the amount of time that it took to receive your child's genetic test results about what you were told to expect?
   - Yes (1)
   - No, the results came back faster than I was told to expect (2)
   - No, the results took longer to come back than I was told to expect (3)

Q39 REMINDER. Please continue to consider only your child's most recent genetic test results when answering the following questions, whether or not it resulted in a diagnosis.

Q20 How were you first told about the results of your child's genetic test?
   - In person (1)
   - Over the phone (2)
   - In writing (by letter, email or other) (3)
   - Don't remember (4)

Display This Question:
   If How were you first told about the results of your child's genetic test? = Over the phone
   Or How were you first told about the results of your child's genetic test? = In writing (by letter, email or other)

Q21 Were you offered a follow-up appointment to discuss the results in person with a health care professional?
   - Yes (1)
   - No (2)
Q22 When your child's genetic test results first became available who did you discuss them with? (select all that apply)

- Genetic Counselor (1)
- Neurologist (2)
- Epileptologist (3)
- Nurse/Nurse practioner (4)
- Primary care physician (5)
- Geneticist (doctor who specializes in genetics) (6)
- Not sure/don't remember who I discussed my child's results with (7)
- Did not discuss my child's test results with a health care provider (8)
- Other health care provider. Please explain (9)

Skip To: Q25 If When your child's genetic test results first became available who did you discuss them with? (sel... = Did not discuss my child's test results with a health care provider
Q23 In thinking back to the initial conversation you had with your health care provider(s) about your child's genetic test results, please indicate which (if any) of the following topics were discussed.

<table>
<thead>
<tr>
<th></th>
<th>Yes (1)</th>
<th>No (2)</th>
<th>Don't recall (3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>What the results of genetic testing were</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(1)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>What the genetic test results could or could not tell you about your child's future health</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(2)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>How the genetic test results would or would not impact your child's treatment</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(3)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>What the genetic test results might mean for other members of your family</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(4)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>How the genetic test results for your child with epilepsy might impact decision-making regarding future pregnancies</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(5)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Q24 At the time that you discussed your child's genetic test results with your health care provider were you given any of the following?

<table>
<thead>
<tr>
<th></th>
<th>Yes (1)</th>
<th>No (2)</th>
<th>Don't recall (3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A copy of your child's genetic test report (1)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Written information that explained your child's genetic test results (2)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Online resources to help you understand your child's genetic test results (3)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Information on how to connect with other families with children with epilepsy or a similar diagnosis (4)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Q25 Is there any information that you were not given about your child's genetic test result that would have been helpful to receive?

- Yes (1)
- No (2)

Display This Question:

If Is there any information that you were not given about your child's genetic test result that woul... = Yes

Q42 Please explain what information would have been helpful to receive about your child's genetic test results.

________________________________________________________________________________________

________________________________________________________________________________________

________________________________________________________________________________________

End of Block: Default Question Block
Q32 Please rate the following statements based on your experience with genetic testing for your child. When answering the questions please consider only your child's most recent genetic test whether it gave a diagnosis or not.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly agree (1)</th>
<th>Somewhat agree (2)</th>
<th>Neither agree nor disagree (3)</th>
<th>Somewhat disagree (4)</th>
<th>Strongly disagree (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I know who to contact if I have questions about my child's genetic test result (1)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I am confident in my understanding of my child's genetic test result (2)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I understand how this genetic information may or may not help my child (3)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I understand the risk of epilepsy to any other children or family members (4)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I have the necessary information to find the emotional support my family needs (5)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I am clear on what the next steps are to find a cause for my child's epilepsy (6)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I have a follow up plan to obtain updated genetic information (7)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>The health care provider gave me enough information to understand my child's genetic test results (8)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>I had to search the internet and/or consult other sources to understand my child's genetic test results (9)</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
<td>o</td>
</tr>
</tbody>
</table>

End of Block: Uncertain test results
Start of Block: Option D genetic test results

Q36 Please rate the following statement based on your experience with genetic testing for your child. When answering the question please consider only your child's most recent genetic test whether it gave a diagnosis or not.

I know who to contact if I have questions about my child's genetic test result.

- Strongly agree (1)
- Somewhat agree (2)
- Neither agree nor disagree (3)
- Somewhat disagree (4)
- Strongly disagree (5)

End of Block: Option D genetic test results
Q38 Please rate the following statements based on your experience with genetic testing for your child. When answering the questions please consider only your child's most recent genetic test whether it gave a diagnosis or not.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly agree (1)</th>
<th>Somewhat agree (2)</th>
<th>Neither agree nor disagree (3)</th>
<th>Somewhat disagree (4)</th>
<th>Strongly disagree (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I know who to contact if I have questions about my child's genetic test result (1)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I am confident in my understanding of my child's genetic test result (2)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I understand how this genetic information may or may not help my child (3)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I understand the risk of epilepsy to any other children or family members (4)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I have the necessary information to find the emotional support my family needs (5)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I am clear on what the next steps are to find a cause for my child's epilepsy (6)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>I had to search the internet and/or consult other sources to understand my child's genetic test results (8)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

End of Block: Negative
Q30 Please rate the following statements based on your experience with genetic testing for your child. When answering the questions please consider only your child's most recent genetic test whether it gave a diagnosis or not.

<table>
<thead>
<tr>
<th></th>
<th>Strongly agree (1)</th>
<th>Somewhat agree (2)</th>
<th>Neither agree nor disagree (3)</th>
<th>Somewhat disagree (4)</th>
<th>Strongly disagree (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I know who to contact if I have questions about my child's genetic test result (1)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I am confident in my understanding of my child's genetic test result (2)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I understand how this genetic information may or may not help my child (3)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I understand the risk of epilepsy to any other children or family members (4)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I have the necessary information to find the emotional support my family needs (5)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>The health care provider gave me enough information to understand my child's genetic test results (6)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I had to search the internet and/or consult other sources to understand my child's genetic test results (7)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Start of Block: Block 3
Q33 Which of the following have helped you understand your child's genetic test results? (select all that apply)

- Family members (1)
- Genetic counselor (2)
- Neurologist or Epileptologist (3)
- Pediatrician (4)
- Geneticist (12)
- Internet search (5)
- Syndrome based support groups and Facebook forums (6)
- Child's medical record (7)
- Personal friends (8)
- Scientific literature (9)
- Handout explaining diagnosis (10)
- Other. Please explain (11) _______________________________________

Carry Forward Selected Choices - Entered Text from "Which of the following have helped you understand your child's genetic test results? (select all that apply)"

Q34 Out of the information sources that you used to understand your child's genetic test results, please rank them starting (#1) with the most valuable to you.

______ Family members (1)
______ Genetic counselor (2)
______ Neurologist or Epileptologist (3)
______ Pediatrician (4)
______ Geneticist (5)
______ Internet search (6)
______ Syndrome based support groups and Facebook forums (7)
______ Child's medical record (8)
______ Personal friends (9)
______ Scientific literature (10)
______ Handout explaining diagnosis (11)
______ Other. Please explain (12)

End of Block: Block 3

Start of Block: Block 4
Q35 Please let us know of any suggestions or advice that you have for health care professionals who will be counseling other families undergoing genetic testing for epilepsy.

________________________________________________________________________
________________________________________________________________________

Q38 Are you involved with any of the following foundations/support groups? (select all that apply)

☐ The Dravet Syndrome Foundation  (1)
☐ PCHD19 Alliance  (2)
☐ Cute Syndrome Foundation  (3)
☐ Wishes for Elliot  (4)
☐ Families SCN2A Foundation  (5)
☐ Aaron's Ohtahara Foundation  (6)
☐ Lennox-Gastaut Syndrome Foundation  (7)
☐ Doose Syndrome Epilepsy Alliance  (8)
☐ Infantile Spasms Community  (9)
☐ Other. Please list  (10) _____________________________________________

Q39 Please indicate the geographic region in which your child most recently underwent genetic testing for epilepsy.

☐ Region 1 (CT, MA, ME, NH, RI, VT, Prince Edward Island)  (1)
☐ Region 2 (DC, DE, MD, NJ, NY, PA, VA, WV, Quebec)  (2)
☐ Region 3 (AL, FL, GA, KY, LA, MS, NC, SC, TN, Puerto Rico, Virgin Islands)  (3)
☐ Region 4 (AR, IA, IL, IN, KS, MI, MO, MN, NE, ND, OH, OK, SD, WI, Ontario)  (4)
☐ Region 5 (AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan)  (5)
☐ Region 6 (AK, CA, HI, ID, NV, OR, WA, British Columbia)  (6)
☐ Other. Please list  (7) _____________________________________________
Q40 Please indicate your highest completed level of education.
   o Grade School (1)
   o High School (2)
   o 2-year college (3)
   o 4-year college (4)
   o Post graduate (5)

Q40 Thank you for your time. Are you interested in enrolling in the raffle for a $75 Amazon gift card?
   o Yes (1)
   o No (2)

End of Block: Block 4
Appendix C. Syndrome based groups

Figure 5. Syndrome based groups in which respondents reported being involved. 
\((n=62)\).
Appendix D. Positive genetic test results

Figure 6. Genes with pathogenic variants reported for 58 individuals with positive genetic test results.

Seven respondents reported that their child had variants in more than one gene.
Appendix E. Information that would have been helpful before child’s genetic test

<table>
<thead>
<tr>
<th>Information that would have been helpful before child’s genetic test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possibly out comes that it would show Maybe to talk to someone in before results to help a parent cope if negative outcome</td>
</tr>
<tr>
<td>They just told me since seizure was so long they wanted to rule out genetic cause but didn't think she fit into any genetic diagnosis. I think the ordering provider should have explained a few mutations that they tested for</td>
</tr>
<tr>
<td>There was long delays and difficulty getting insurance to cover it. Our child suffered but testing was hard to get.</td>
</tr>
<tr>
<td>I sometimes feel like doctors feel like I am not intelligent enough or with the rime to explain to. She has great doctors but sometimes feel talked down to. I would have liked to know some of the possibilities of genetic causes. And since it was negative what will that mean for her future. But neurology is so subjective and almost an art form.</td>
</tr>
<tr>
<td>We agreed to the test because the neurologist indicated their may be an underlying food allergy or environmental allergy. The genetic test was part of a complete epilepsy panel. Never did we imagine that the results would come back as a genetic mutation.</td>
</tr>
<tr>
<td>La ricerca del bene, sapere cosa pensassero i dottori</td>
</tr>
<tr>
<td>What mutations they were looking for and what possible conditions these results would show</td>
</tr>
<tr>
<td>I had to pay out of pocket for the test to understand what was going on with my child. I feel I was just expected to accept all of the varying things going on with my child as opposed to finding the cause.</td>
</tr>
</tbody>
</table>

Note: only those who said that they did not receive enough information prior to their child’s genetic test saw this open response question.
Appendix F. Information that would have been helpful to receive about genetic test results

| Information on the genetic mutation; how results could impact child in treatment, future issues |
| Things like typical body appearances |
| Ways to contact other families. I was the one demanding testing, i was told many times nothing would be found. Only when i said i would go to a different hospital for testing was i given the test. |
| We were told by our neurologist’s nurse that there was no new information that resulted from the test, but when we requested a copy of the test result we found that two additional genetic concerns were present. Months later the neurologist advised us to be more cautious because of those additional genetic concerns and also advised us about potential sibling risks, but initially we were told that everything was fine. |
| Support groups Online places to go to learn specifics about our disease |
| Specific doctors specializing in the genetic condition |
| More information on the other variants found besides scn1a nonsense. |
| Would have liked to have been given a written handout with a basic outline of the findings, and possibly a list of resources for finding other information. |
| Information on meds that are contraindicated to persons with this genetic mutation. |
| More about what it means for her prognosis |
| Understand her conditions and places to go for support. |
| More information on parents effects on mutation. |
| It would have been helpful to have been told (even a ballpark) what percentage of SCN1A mutations go on to a Dravet syndrome diagnosis. I felt I was blindsided by the diagnosis because the way the genetic results were explained to us, we were on the mild end of the spectrum and he might even outgrow the seizures. Then almost overnight, he leapt to the severe end simply for having two or three non febrile seizures. I also was told later (we didn’t get into much at the results appointment because I had no plans to have more children) that my chances of having another child with the same mutation, because they initially thought it was de novo because I tested negative, were akin to winning the Powerball. I went on to have another child with a different father and that child (born in 2016) has the same mutation. He hasn’t yet tracked the same. It was then I was told the mutation was likely carried in my eggs. |
| More information. I feel like there is not enough out there to find that can help with anything. No one seems to know what to expect or look for or really do. |
| I would like to know what all was tested for. |
| Links to research studies or anything that has been studied or written on the gene |
| I would have liked more information on why the genetic test result cannot predict the prognosis of the disease and why there exists such wide variability in the outcome. We did get this information when we had the genetic counselling. But it was a few months before we could get a genetic counseling appointment and so we were left with little knowledge until that time. I would have also liked information about how to connect with other families with children of the same condition. It took me almost a year to find the facebook group that connected me to other families. |
| I found the SCN8A parent group, which was a huge blessing. Although, our genetist did give us some online resources, I had already found much more before we had our appointment. how to follow up on what future research might tell about the variations of unknown significance. |
ANY information aside from “she could die,” would have been helpful. Support Group, Registries, Doctors that have treated, etc

It would have been helpful to receive information regarding support groups or other families with similar test results. But at the time of our diagnosis, there were less than 150 people affected world-wide, so there was not much information available. My independent searches and subsequent contacts have educated our neurologist.

Our neurologist had never heard of SCN8A prior to our child's diagnosis. So, there was very little information explained to us. We have since sought further information and made connections to supports groups and physicians, who study this disorder, on our own.

How to connect with other families and any studies.

Information on next steps or where to go might have helped make the news received less heavy. Maybe a database that the geneticist uses to look up the mutations. Also a little courtesy from the geneticist. A phone call to drop the bomb, state very rare oh and your child is the only one with the variant and good luck with that goodbye was rather a shock. The geneticist only relayed basic information and got off the phone as quick as possible. I just stood there left in the dark. Some extra info, any would have helped with the blow.

Resources to help connect with other families/groups/foundations

Words that a parent could understand without being a doctor or scientist

Information about other families or online resources
**Appendix G. Suggestions or advice for health care professionals who will be counseling other families undergoing genetic testing for epilepsy**

Have genetic results and potential impact information ready for family; if there are any organizations or support groups available; contact information for future questions or other testing

We were seen at Nationwide Children’s Hospital. Excellent genetic counselor. Very thorough in the entire process. Unfortunately, there is very little information available about our condition. UBA5 variant -Early Infantile Epileptic Encephalopathy Type 44.

Offer testing for children with epilepsy. Offer support groups. Knowledge is power.

Thank you for dealing with this issue.

Be prepared to help emotional support a parent if it's a terrible diagnosis Help them get into a geneticist as fast as possible

Always schedule an in person appointment to discuss a positive genetic test result. Give written handouts. Offer referrals to geneticist or other professionals who may be helpful. Offer information on groups or organizations who can provide support the the patient and family.

Listen carefully, explain things as basically as you can, offer another follow up appointment within the month to answer questions that arise after the diagnosis sinks in.

I wish it happened much sooner. His dr. and I discussed the most likely outcome of the testing. We both felt strongly it would come back positive, and it did. I wish I knew more about the possibility of my older son passing this on. He is 12 years old, but has a different father.

Provide more people resources — specifically groups or other families diagnosed with epilepsy.

I feel that the ARX testing should be in the first group of tests run. I feel like it took too long to get a diagnosis for him.

I think after intitial results, being able to connect with other parents of children with the same diagnosis as your child is valuable in letting you know you are not alone. I feel comfort in knowing that even though my daughter’s mutation is rare, our geneticist is still reaching out to researchers and asking them to study it more.

It helps to know specifics. Otherwise you worry even more or at least I do.

Research studies, scientific studies , or any information so they don't run home searching google for misinformation

In my daughter's case, genetic testing was performed only after I insisted. She was 3 years at that time. We had been battling seizures since she was 10 months. In this time, we had noticed certain triggers like excitement which our neurologist thought wouldn't cause seizures. We started searching the internet and came across Dravet syndrome and how excitement was one of the triggers. After learning this, we asked for genetic testing. Her neurologist didn't feel genetic testing was necessary since most epilepsies are idiopathic. While the test result did not alter the medication my daughter was taking, we were lucky to have not been on medication that is contraindicated for her syndrome.

Make completing the process is simple as possible. Coordinate with the parents and do an online meeting or a phone meeting to complete the paperwork and answer questions. Refresh the parents memory often. Ask parents to ask their other family members if they have any questions. Remember they at they are very busy taking care of their disabled loved one and me understand things once but will still need it to be explained again at a later date.
It’s worth every penny. It’s helped me as a parent knowing the root cause of my daughter’s epilepsy and it’s changed our process of which drugs to try first.

If you are aware of support groups, it is REALLY important you make families aware. Same with registries or potential research opportunities in which our child can participate. Try to be compassionate and understanding. Listen to the parents and respect the fact that although you may be an expert in your field, we are experts in our children.

I think that health care professionals often overstate what they can know about test results in any given moment. My daughter has moved from having one variant of uncertain significance, to two, to two others mutations that my relationships with genetic researchers uncovered, to two confirmed likely pathogenic/pathogenic variants, one uncertain significance, and one that is no longer considered pathogenic (although there is not full agreement from researchers). And none of the two confirmed pathogenic/pathogenic variants are associated with epilepsy like my daughter's...

Understanding what the genetic mutation caused -- is it a dysfunctional channel, a wrongly coded protein, etc -- would have helped me understand why symptoms present. In addition, it would be immensely helpful to know where in the body this gene impacts would be helpful in order to understand the breadth of symptoms we might see (for example it's expressed in GI tract, respiratory tract, heart, neurons of the brain, neurons of the periphery). Sadly, as in our case, this is not fully understood, but it would be helpful to have access to whatever info is available.

Dare più informazioni e supporto, per aiutare la famiglia

Genetic testing was not covered by insurance. We went two years on a medicine that intensifies seizures in scn8a Epilepsy. We finally were offered a grant in exchange for our info to be used to prove genetic testing should be covered by insurance companies. We had whole exome sequence done. If the insurance had paid for the cheap epilepsy panel. Thousands would have been saved. My child also would have been treated with correct seizure meds. In my experience my fb support group is the most beneficial.

Here were under 100 known cases at the time of her diagnosis. I have learned so much from the SCN8A support group.

Maybe some research on what that gene mutation means. Please don't just state very rare look it up on the internet. Even after dropping the bomb schedule one more appointment for questions that will arise after the person digests the information. One time phone call most people need time to take it all in then the questions will start. Please just don't state if you have questions look it up...ugh [could've been we just had a geneticist with poor "bedside manner" :-] ]
Please be realistic when providing families a time line of when results will become available. We were told 3-6months give or take a month. However at the 5 month mark we were disheartened to here that the blood test had not even gone over seas onto the machine yet. We had to continuously ask for updates after this point. It ended up taking 1 year to get the results. To people not directly affected they see as a further 6months not too bad. However for a young couple coming to terms with there child disablity, ongoing medical needs and unknown life expectancy this is simply not acceptable. Along with wanting to find appropriate treatments and wanting to provide her with a healthy sibling this all was put on hold until the results came back. We were prepared for the worst and had a plan B in place sourcing a donor egg and sperm in the USA. Luckily for us they were able to isolate our daughters gene and we are able to have children naturly as our daughter carries a de novo mutation. We will be forever greatful for all the hard work that went into discovering our daughters mutatuon. However I believe better communication and more frequent updates would have been emenasly benificial in managing our expectations on the time line. If they had of said 6months to 1 year we would have paid for this test privately. Thanks

| To really have the doctors breakdown what the mutation mean for your child. |
| That they educate themselves prior to meeting with a parent. |
| Provide non-medical support information: Facebook support group info, relevant non-profit info, etc. |
Appendix H. Perceived understanding in the subset of respondents whose child had a positive genetic test result

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
<th>95% CI for Mean</th>
<th>t</th>
<th>df</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>All Topics Discussed</td>
<td>M 4.67, SD 0.50, n 26</td>
<td>M 4.06, SD 0.86, n 29</td>
<td>0.24, 0.99</td>
<td>3.27**</td>
<td>45.60</td>
<td>0.002</td>
</tr>
<tr>
<td>All Information Provided</td>
<td>M 4.78, SD 0.44, n 10</td>
<td>M 4.25, SD 0.80, n 45</td>
<td>0.16, 0.91</td>
<td>2.92**</td>
<td>24.53</td>
<td>0.007</td>
</tr>
</tbody>
</table>

Table 4. Independent-samples t-test comparing mean perceived understanding in cases where 1) all five topics were or were not discussed (see Figure 2 for topics) and 2) where all four types of information was or was not provided at the time results were returned (see Figure 3 for information resources).

*p<0.05
† Note: Satterthwaite approximation employed due to unequal group variances.
Appendix I. Topics discussed and information provided to respondents whose children had positive genetic test results

<table>
<thead>
<tr>
<th>Topic</th>
<th>Yes %</th>
<th>No %</th>
<th>Don't recall</th>
</tr>
</thead>
<tbody>
<tr>
<td>What results of genetic tests were (n=55)</td>
<td>96.4%</td>
<td>3.6%</td>
<td></td>
</tr>
<tr>
<td>What genetic test results could or could not tell you about your child's future health (n=55)</td>
<td>80.0%</td>
<td>14.5%</td>
<td>5.5%</td>
</tr>
<tr>
<td>How the genetic test results would or would not impact child's treatment (n=56)</td>
<td>62.5%</td>
<td>25.0%</td>
<td>12.5%</td>
</tr>
<tr>
<td>What the genetic test results might mean for other members of family (n=56)</td>
<td>64.3%</td>
<td>30.4%</td>
<td>5.4%</td>
</tr>
<tr>
<td>How the genetic test results might impact decision-making regarding future pregnancies (n=55)</td>
<td>63.6%</td>
<td>29.1%</td>
<td>7.3%</td>
</tr>
</tbody>
</table>

Figure 6a. Topics discussed with the subset of respondents whose children had positive genetic test result.

<table>
<thead>
<tr>
<th>Information provided</th>
<th>Yes %</th>
<th>No %</th>
<th>Don't recall</th>
</tr>
</thead>
<tbody>
<tr>
<td>Copy of genetic test report (n=56)</td>
<td>80.4%</td>
<td>19.6%</td>
<td></td>
</tr>
<tr>
<td>Written information explaining test results (n=56)</td>
<td>69.6%</td>
<td>30.4%</td>
<td></td>
</tr>
<tr>
<td>Online resources to understand results (n=55)</td>
<td>38.2%</td>
<td>60.0%</td>
<td>1.8%</td>
</tr>
<tr>
<td>Information to connect with other families (n=55)</td>
<td>20.0%</td>
<td>74.5%</td>
<td>5.5%</td>
</tr>
</tbody>
</table>

Figure 6b. Information provided to the subset of respondents whose children had a positive genetic test result.
Appendix J. Comparison of the information provided to all respondents who met with a genetic counselor with those who did not

<table>
<thead>
<tr>
<th>Information provided</th>
<th>No GC</th>
<th>Yes GC</th>
<th>Chi-sq</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Copy of child's genetic test report</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>27</td>
<td>52.9%</td>
<td>24</td>
<td>47.1%</td>
</tr>
<tr>
<td>No</td>
<td>10</td>
<td>71.4%</td>
<td>4</td>
<td>28.6%</td>
</tr>
<tr>
<td>Don’t recall</td>
<td>0.0%</td>
<td>0</td>
<td>0.0%</td>
<td></td>
</tr>
<tr>
<td>Written information explaining test result</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>25</td>
<td>55.6%</td>
<td>20</td>
<td>44.4%</td>
</tr>
<tr>
<td>No</td>
<td>12</td>
<td>60.0%</td>
<td>8</td>
<td>40.0%</td>
</tr>
<tr>
<td>Don’t recall</td>
<td>0.0%</td>
<td>0</td>
<td>0.0%</td>
<td></td>
</tr>
<tr>
<td>Online resources to help understand test result</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>6</td>
<td>28.6%</td>
<td>15</td>
<td>71.4%</td>
</tr>
<tr>
<td>No</td>
<td>31</td>
<td>73.8%</td>
<td>11</td>
<td>26.2%</td>
</tr>
<tr>
<td>Don’t recall</td>
<td>0.0%</td>
<td>0</td>
<td>100.0%</td>
<td></td>
</tr>
<tr>
<td>Information on how to connect with other families</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>6</td>
<td>50.0%</td>
<td>6</td>
<td>50.0%</td>
</tr>
<tr>
<td>No</td>
<td>30</td>
<td>61.2%</td>
<td>19</td>
<td>38.8%</td>
</tr>
<tr>
<td>Don’t recall</td>
<td>1</td>
<td>33.3%</td>
<td>2</td>
<td>66.7%</td>
</tr>
</tbody>
</table>

Table 5. Chi-square analysis comparing the information resources given to all respondents who met with a genetic counselor (either one-on-one as part of a clinical care team) and those who did not meet with a genetic counselor at the time genetic test results were initially returned.