

# SPEAKING GENETICS GARD DATA ANALYSIS SUMMARY

Prepared for the ISCC Speaking Genetics Working Group  
Made possible through funding provided by NHGRI  
Data generously provided by the Center for Genetic and Rare Diseases

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## BIRDS EYE VIEW: ABOUT THIS PROJECT

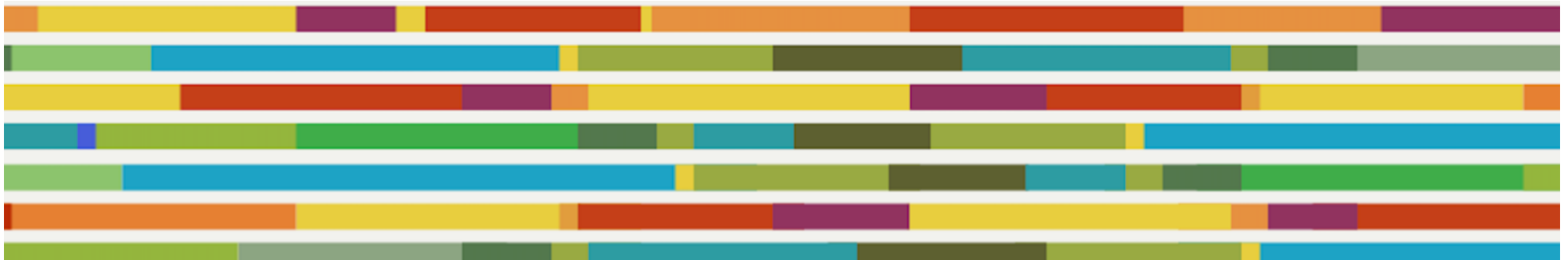
### What We're Trying To Do With Speaking Genetics

This project was designed to look at how people, actual consumers of information about genetic medicine, talk about and ask questions about genetic medicine using a data-driven approach.

We used the GARD data set as a proxy for patient-provider communications.

Through analyzing how people talk about and communicate about genetic medicine, we can help equip providers and others with insights and suggestions to communicate about genetic medicine more effectively and successfully.

This is a starting point in an effort to make genetic medicine more accessible to patients, non-specialist providers, caregivers and others.





## BIRD'S EYE VIEW: THE GARDA DATA, ITS SOURCES & WHAT WE ANALYZED

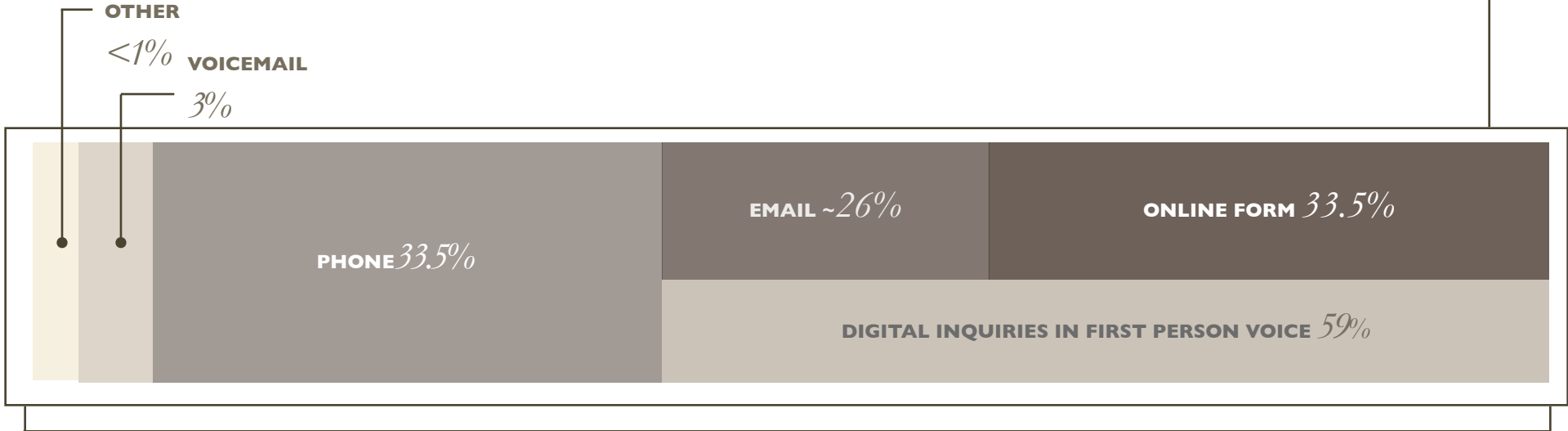
### The GARD Data & What We Analyzed

The GARD Data is a collection of twelve years of inquiries from the general public to the contact center alongside responses from the GARD staff and rich metadata about source of inquiry, type of user who made the inquiry, the reason for the inquiry etc..

The most meaningful analysis that we established came from mining the inquiries themselves and was where we put our focus. All the data we analyzed was in English.

**50,000** INQUIRIES

WHAT WE ANALYZED



**50,000** TOTAL RESPONSE-INQUIRY RECORDS BY TYPE OF CONTACT



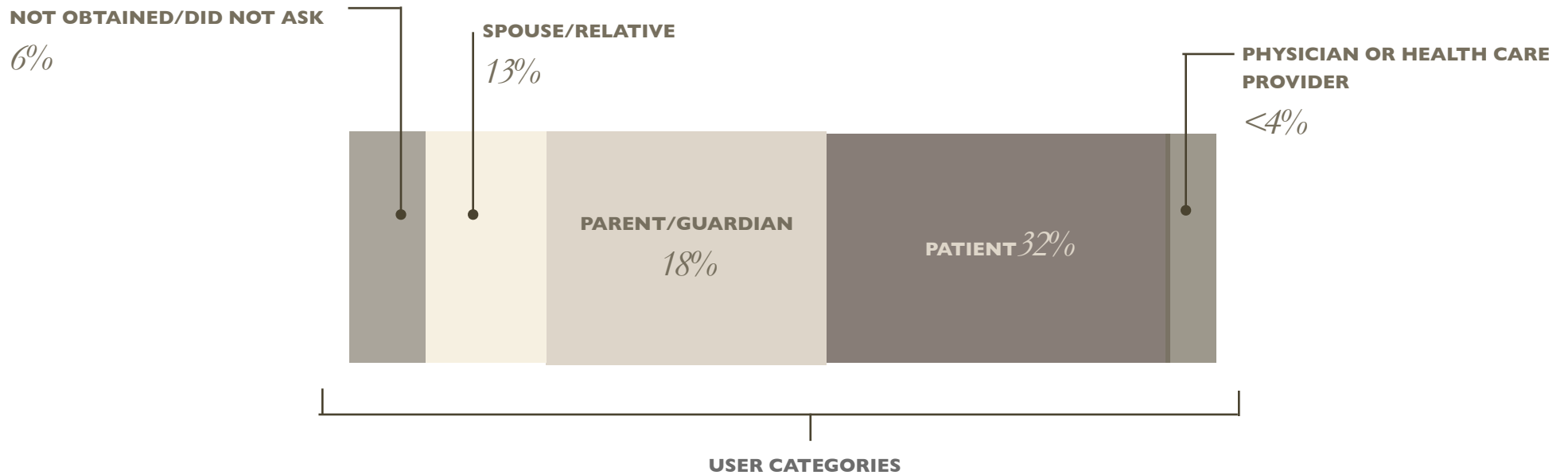
## BIRD'S EYE VIEW: SOME SPECIFICS

### More About What We Analyzed

In addition to looking at all 50,000 inquiries in aggregate, we also looked at specific sub-sets of the data.

Decisions about what specific sub-sets of the data to review were driven by stakeholder requests and/or data volume.

In particular, one sub-set we looked at were inquiries by specific user categories.

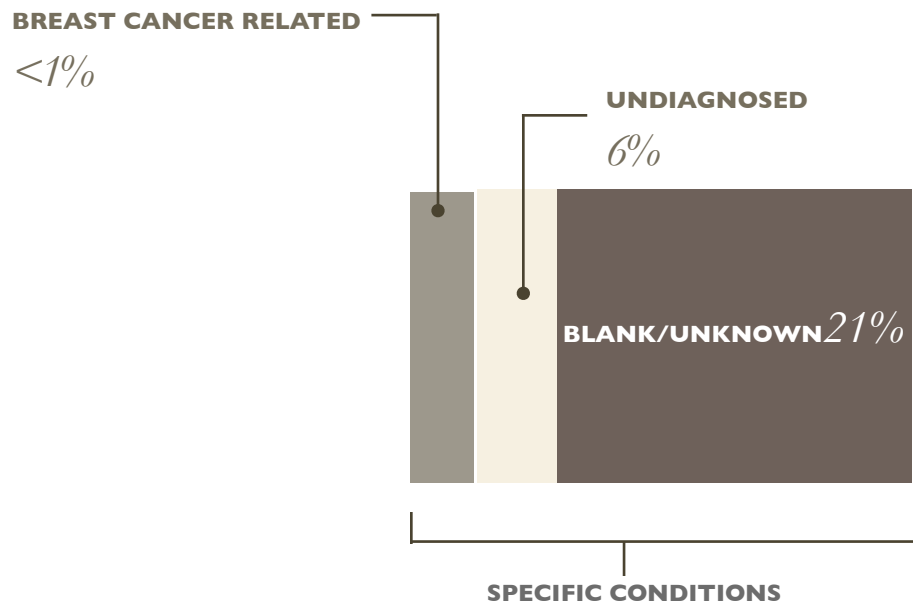




## BIRD'S EYE VIEW: SOME MORE SPECIFICS

### And A Little More About What We Analyzed

We also looked at inquiries made about specific conditions.

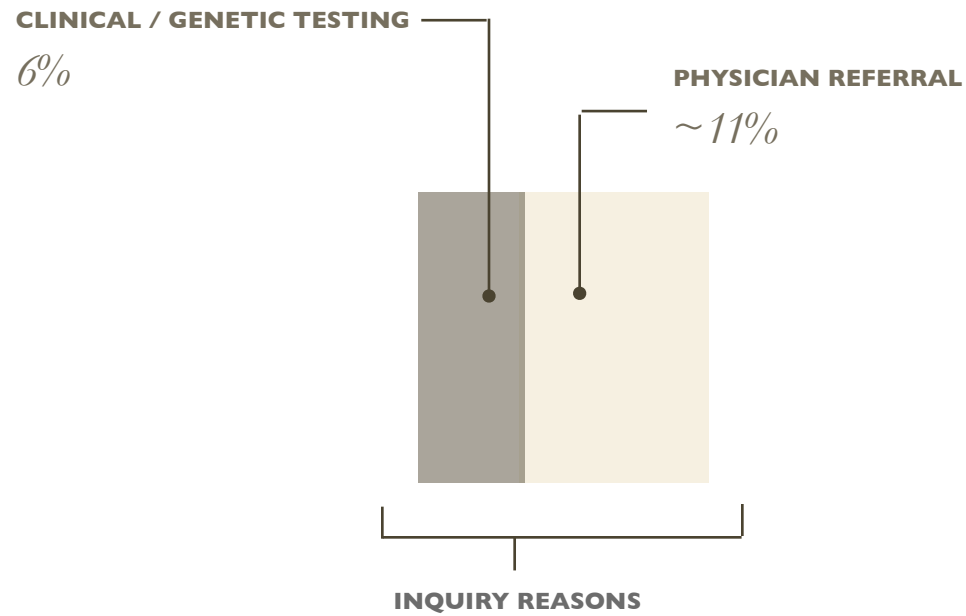




## BIRD'S EYE VIEW: SOME MORE SPECIFICS

### And Even A Little More About What We Analyzed

Another sub-set of the data that we examined were specific reasons for inquiry.



## BIRD'S EYE VIEW: HIGH LEVEL THEMES

*Here's what we found*

### 4 THEMES

Looking across all 50,000 inquiries in aggregate, four primary themes emerged from the data that present areas of difficulty, frustration, concern or confusion for consumers of genetic medicine.

These are not the only themes, but they seemed to be the most meaningful to surface. You may have already suspected that these themes existed. And, you may have suspected what some of the specific challenges for people are. Now, you know.

Each theme is presented in more detail later in this document and illustrated with verbatim examples from the data alongside insights and suggestions.



## CORNERSTONE CONCEPTS: OVERVIEW

5-15+%

This theme brings to light how people struggle with basic understanding of foundational concepts in genetic medicine, including terms like dominant, recessive, x-linked and basic concepts of heredity. Their lack of comprehension snowballs into their understanding of other areas of genetic medicine, such as genetic testing and disease/condition specific concepts.

### In Their Own Words

“If your a carrier of the <disease/condition> are there any paths you can take to try not pass it on to your children????”

“Caller is a carrier for <disease/condition>. She was confused by the words carrier and recessive. She wondered if I could help her understand these words.”

“Hello, I'm trying to get phenotype information for xxxxx -I zqzzzz with a "PM" phenotype. I'm familiar with the M allele, the null and the z, but am clueless about the "P" allele.”

“What causes the genetic mutation in the mitochondrial DNA or nuclear DNA? Is it idiopathic?”

“looking for information about how haploid cells and diploid cells work (i.e. mitosis versus meiosis and how daughter cells come about, are they exactly alike, etc.)”

“when the doctor said that if both of are carriers then there's a 1 in 4 chance of the baby having the disorder. Does that mean that the baby is one of the 3 who didn't get the disorder?”



CORNERSTONE CONCEPTS: INSIGHTS & SUGGESTIONS

**Insight #1**

Math, probability and risk are challenging for people to understand!

We use phrases like “a 1 in 2 chance”, “on average half {their/your} children will be affected and half {their/your} children will not be affected”, “50/50 percent chance per child (of being affected)”. These are not easy concepts for people to grasp and can lead to real issues with literacy and comprehension.

**Suggestions**

Developing canonical explanations for these concepts that leverage real-world analogies that draw on very familiar concepts could be helpful.

Drawing pictures can also be effective.

Using interactive aids, such as a special set of dice and supporting materials could be used to better illustrate risk on a per-child basis.

**Insight #2**

People like to feel like they have some sense of control, which genetic and other inherited conditions don’t give them. There are a lot of unknowns and variables in genetic medicine that are challenging for people to cope with.

People want to feel that they can somehow avoid passing on an inherited disease to their children or that they can do something to prevent the spread of the disease to others.

**Suggestions**

Acknowledging the many variables and the lack of control may be an effective starting place.

Explaining as best as possible the “knowns” and how to account for them could help in many cases.

Using examples that are close to home and that seem more mundane to explain heredity may be useful. For example, the color of a child’s eyes or hair can’t be “prevented” or “avoided”.

GENETIC TESTING: OVERVIEW

5-10%

Questions about and understanding of genetic testing represent a high level theme that can be further broken down into more specific sub-themes, including: who should be tested and why, what to test for, what tests are available, testing logistics, difficulty understanding and interpreting test results.

**In Their Own Words**

<p>“Female inquirer who's pregnant, was given diagnosis of carrying the CF gene and is scheduled for amnio. Would like to find out whether her husband should be tested”</p>	<p>“Caller's paternal half-uncle diagnosed with &lt;disease/condition&gt;. Caller wanted to know how to go about getting carrier testing.”</p>	<p>“I recently participated in a genetic research study for &lt;disease/condition&gt; through 23andme. ... They have provided the raw data, which I obviously cannot interpret. If I were to e-mail the raw data, could someone there interpret it for me?”</p>
<p>“My DNA test tells me where the mutations are but is there any testing to indicate which mutations are present and to what symptoms these mutations are related to?”</p>	<p>“My husband and i are currently trying to conceive and are wondering if there is test to see if we are carriers, and what the tests involve.”</p>	<p>“genetic testing report that she requested help with interpreting.”</p>

## GENETIC TESTING: INSIGHTS & SUGGESTIONS

### Insight #1

As with other tests and diagnostics, such as imaging, consumers are not best equipped to interpret results on their own.

Genetic testing results contain very detailed information that is often not black or white in its interpretation.

### Insight #2

Testing logistics, including where to get testing and what can be tested for, are not straightforward for consumers to find.

People like to feel some sense of control and would like to be empowered to take proactive measures in their lives.

### Suggestions

Reviewing genetic test results either in person or by some virtual means with a patient would provide opportunities for Q&A.

Examples of how to interpret common test results may be effective.

### Suggestions

Providing clear lists of available genetic tests labeled as “Available Genetic Tests” possibly with some coding of “reliability” or similar would empower consumers.

Outlining a basic “how to” for obtaining the most common genetic tests could be helpful.

A map or flow of common or possible steps in obtaining tests for less common genetic tests could be useful.

## FINDING & UNDERSTANDING INFORMATION: OVERVIEW

# 10 - 80+%

As we looked across the GARD data, this theme was really recurrent. People were either looking for information in more accessible terms or just looking for information about specific diseases, conditions and similar because they were unable to obtain such information from other sources, including their physicians in some cases. In other cases, physicians themselves were looking for more accessible information to share with patients.

### In Their Own Words

“I have tried to locate info on this and with all the medical jargon it is really hard for me to understand all the terminology.”

“I thought I'd contact you because my primary doctor isn't helping me and I'm not sure where to start on my quest”

“When I tried pressing further to better understand what this means for any future childbearing, the doctor indicated that I would need to talk to a specialist.”

“Do you know someone who could put the information below into lay terms?”

“Are there any publications or information available that would be suitable for a family to read that would enable them to gain a better understanding of the condition?”

“I am trying to educate myself to help the family better understand this syndrome. Your website is vast with information but I need information that can be understood by the general public. Is it possible to receive that kind of information”

FINDING & UNDERSTANDING INFORMATION: INSIGHTS & SUGGESTIONS

**Insight #1**

People want to understand the conditions that they and their families are being affected by and often encounter speed bumps because the information they encounter is too technical, dense or difficult to understand.

Additionally, information about genetic medicine is not always presented in user-friendly ways that consumers have become increasingly accustomed to.

**Suggestions**

Drawing on concepts, tools and practices in health literacy to present at least core and common concepts in genetic medicine could be effective.

Assessing written content via a reading level calculator and adjusting content could be useful.

Breaking down concepts to their core and explaining fundamentals in basic terms could be helpful.

**Insight #2**

People often simply don't know where to look for any information at all about specific conditions. They want some help and to be "pointed in the right direction".

**Suggestions**

Highlighting "Additional Resources" or "Looking For Information About" on websites and other collateral could be very useful to consumers.

Readily directing consumers to support groups, peer groups and online communities for those in similar circumstances or with similar conditions may also be helpful.

Equipping providers with a list of "first step" could be very useful

## COSTS & INSURANCE COVERAGE: OVERVIEW

24%

This theme really showcases the anxiety and lack of clarity that people have around how they can pay for not only genetic testing but also genetic treatments and/or diagnosis.

### In Their Own Words

“cannot find a more affordable genetic lab”

“they are hurting financially from all the medical bills as much of her treatment is not covered by insurance. We need resources ... We need help! please. ”

““I am trying to find help for my sister ... they r havin a hard time getting control of it she was diagnosed in 2004 she just had a stroke. the ml in her brain is blocked she has had left carotid angioplasty with patch a double renal bilateral bypass. her dr. ... has submitted remicaid to the insurance company and they wont approve it they have submitted it 3 to 4 times and they r tryin again. im tryin to find some help for her. ”

“I am the parent of a 3 year old that was recently diagnosed with <condition/disease>. My employee changed insurance providers and our new plan no longer covers genetic testing. She was in the approval process for her next set of testing when this change occurred. Is there anyone that is conducting microarray testing on <condition/disease> patients that could help us?”



## COSTS & INSURANCE COVERAGE: INSIGHTS & SUGGESTIONS

### Insight #1

Medical care is expensive and those expenses are crippling to many people.

The costs of dealing with genetic conditions, diseases, testing and treatment are possibly more expensive than other conditions and testing

### Suggestions

Highlighting resources for financial assistance prominently in locations that are highly visible on public websites and collateral could be very helpful.

### Insight #2

People often don't know what their insurance carriers will or won't cover, which leaves them unsure about the best course of action.

This situation can be further complicated when tests or conditions go by multiple names and similar.

### Suggestions

Encourage transparency among payers about their coverage.

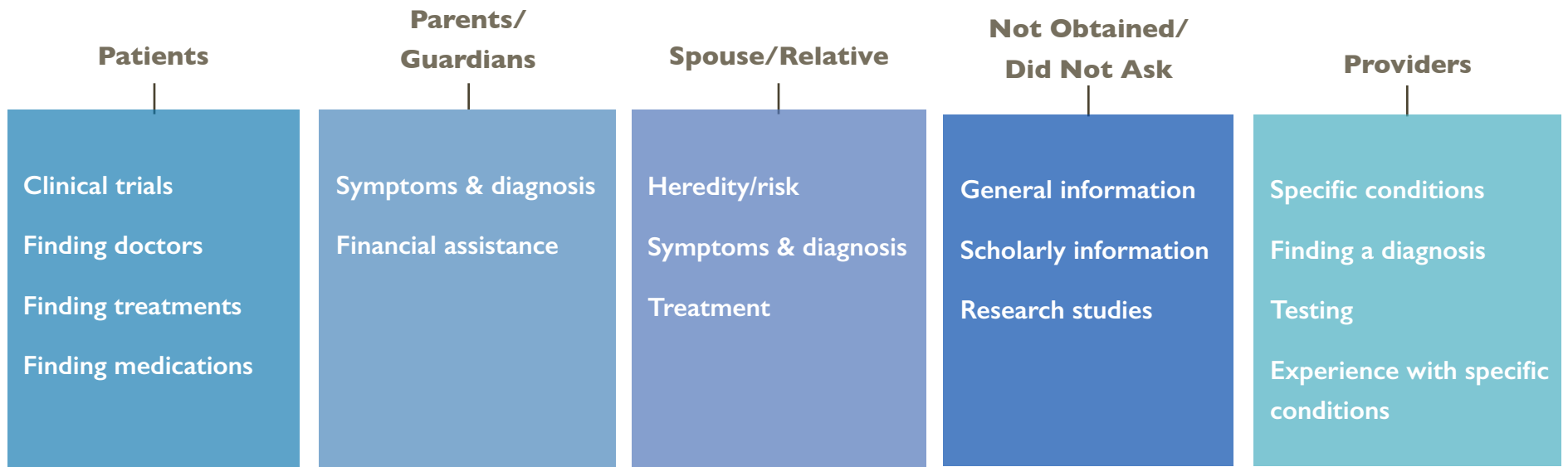
Develop consistent naming and / or easy-to-use thesauri/look-up tools so that consumers can verify coverage and costs on an apples-to-apples basis.



## DEEPER DIVES: THEMES FROM DIFFERENT USER CATEGORIES

*Here's what we found*

We looked at several user categories to see if different themes emerged from different types of inquirers.

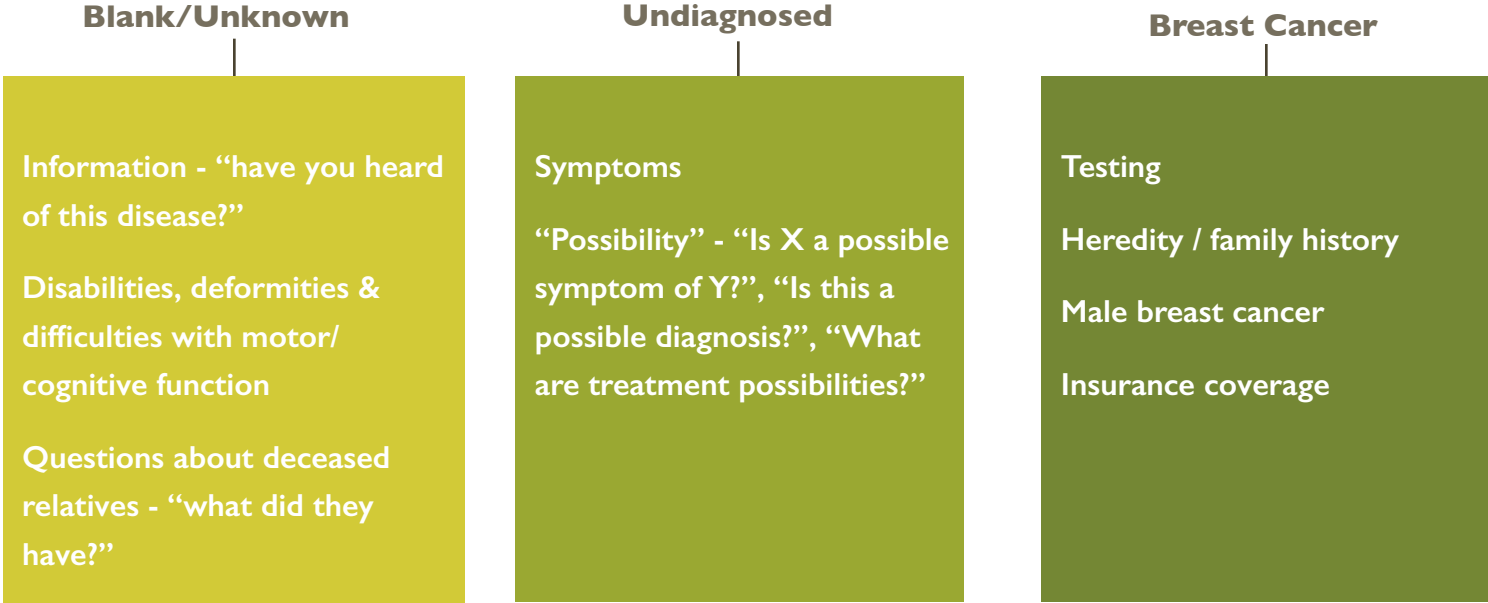




## DEEPER DIVES: THEMES FROM SPECIFIC CONDITIONS

*Here's what we found*

We looked at several specific conditions to see if different themes emerged from.





## DEEPER DIVES: THEMES FROM INQUIRY REASONS

*Here's what we found*

We looked at several inquiry reasons to see if different themes emerged.

### Physician Referral

- Searching for specialists based on a diagnosis
- Clinical trials
- Treatment options

### Genetic Testing

See earlier section

## INTERESTING ANGLES & FOOD FOR THOUGHT

*Here are some other interesting things that we found that we wanted to share*

We turned these themes up while looking at the data for other reasons.

### **“Is X related to Y?”**

We see a lot of inquiries that relate to the possible connections between things. “Is this symptom related to this one?”, “Is this disease related to this one?” and “Are these outcomes connected to one another.”

Possibly important for explaining concepts.

### **“Skip a generation”**

Yes, people still say this. There is still this notion of conditions/ diseases/disorders/traits by-passing one or more generations.

It would be great if the provider community could find an effective way to leverage this folk-explanation to their advantage to promote better understanding of heredity.

### **“Frustration & desperation”**

We see a lot of inquiries that suggest a deep level of frustration with either a lack of information/resources, frustration with a disease/ condition/disorder and/or depression related to same.

Keeping this top of mind when dealing with patient-centered interactions in this arena could be very effective.

### **“Translocations, deletions, trisomy (including 13 and 18)”**

Translocations, deletions - including micro-deletions - and trisomy 13 and 18 are some of the more difficult concepts for laypeople to understand and also among the most unique. They can be difficult to find information about and understand how to deal with them.

A sense of empathy and acknowledgement of difficult concepts, emotional distress and unknowns could be helpful.

## DIRECTIONS FOR FUTURE WORK

*Here are some ideas about where to go from here*

We turned these themes up while looking at the data for other reasons.

### Review provider education materials for core concepts

Our analysis was a linguistic analysis of the GARD data. We did not review provider education materials for terms used, method for teaching about core concepts or similar. Reviewing these materials vis-a-vis the observations here would be a solid next step.

### Languages other than English

This data set included English-only data and the analysis was English first. Moving to languages other than English would be a solid next step in this work.

### Analyze additional data sources

Moving to different kinds of data would make sense. This analysis was a first step and leveraged data that was available to NHGRI. There are lots of other data sources out there, many of which have already been identified by the working group.

### Move to actual constructions & phrases

All this linguistic research is supposed to lead us to a place where we have a good understanding of what actual linguistic constructions will resonate with people. Analyzing the GARD response data will be very valuable for this effort.

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