



# Investigation of First Contact Regarding Out-of-Range Newborn Screening Results

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Consumer Task Force-Baby's First Test



# Conflict of Interest

- I have no financial conflicts of interest to disclose
- I am the mother of a child whose condition was found via newborn screening





# Purpose

- Evaluate first contact regarding out-of-range newborn screening
- Determine who is contacting parents
- Evaluate information given to parents
- Determine what additional information parents want



<https://unsplash.com/search/photos/newborn>



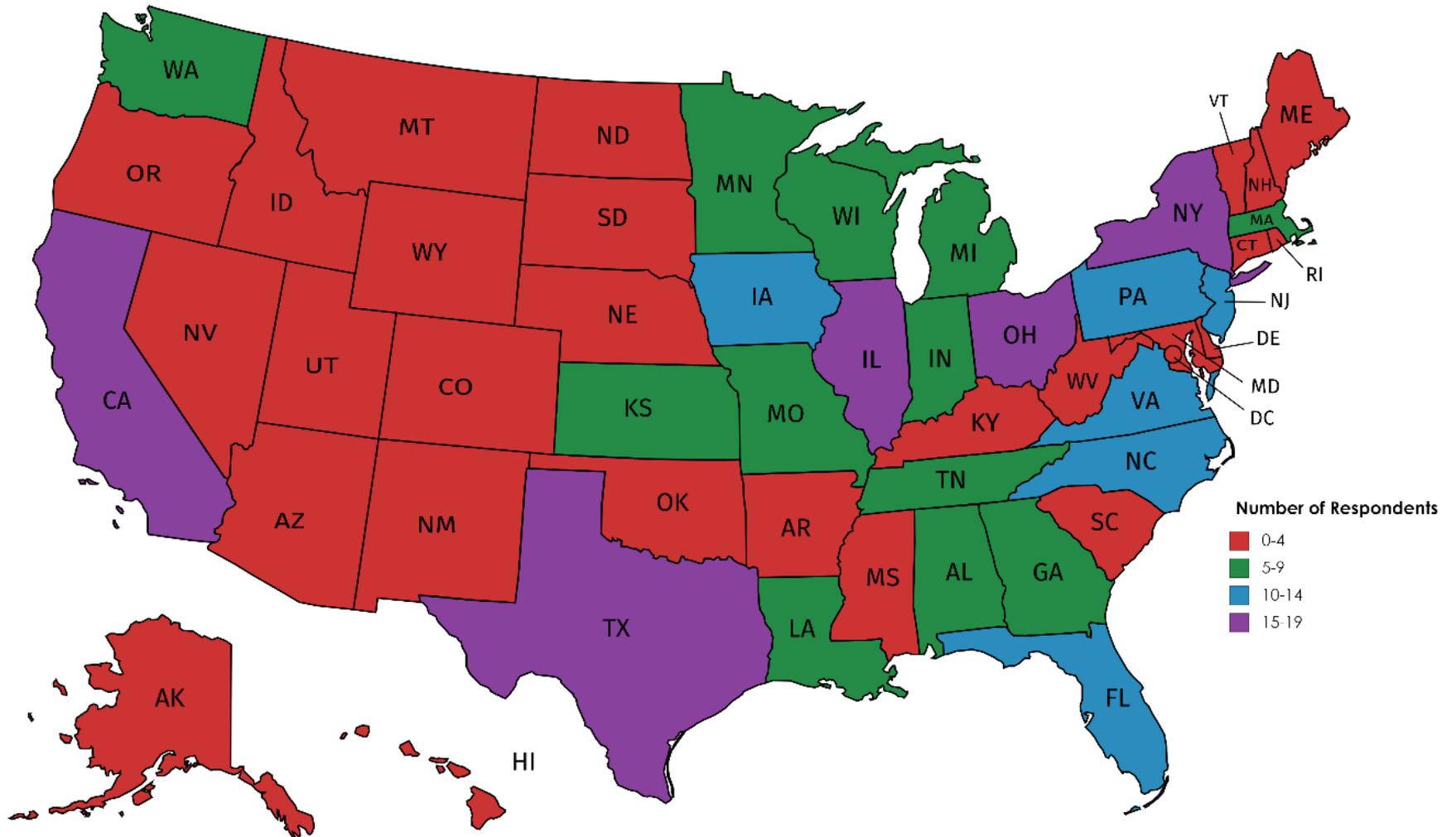
# Methodology

- Thirteen question survey through SurveyMonkey containing both qualitative and quantitative questions
- Study approved by Genetic Alliance IRB
  - No crossing of more than two answers to protect identity
  - Included only people whose child had had newborn screening after January 1<sup>st</sup>, 2014 to increase recall ability
- Sent out via advocacy organizations, Genetic Alliance and Baby's First Test

# Results

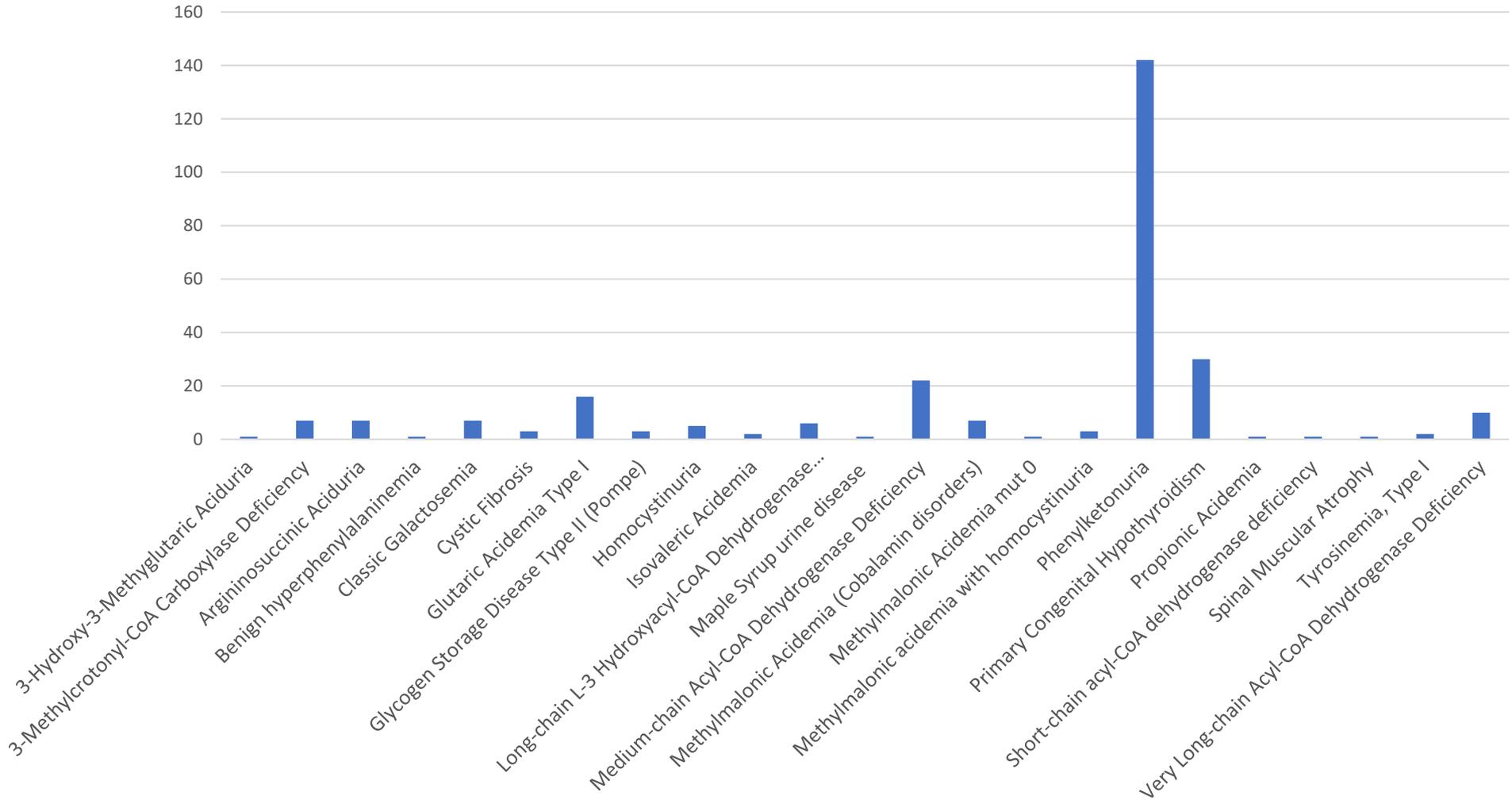
- 283 respondents
- Average time spent was five minutes
- 13% of the time the child was in the hospital at the time of the initial contact
- 98% of the respondents had a child who was diagnosed with a condition

# Number of Respondents by State



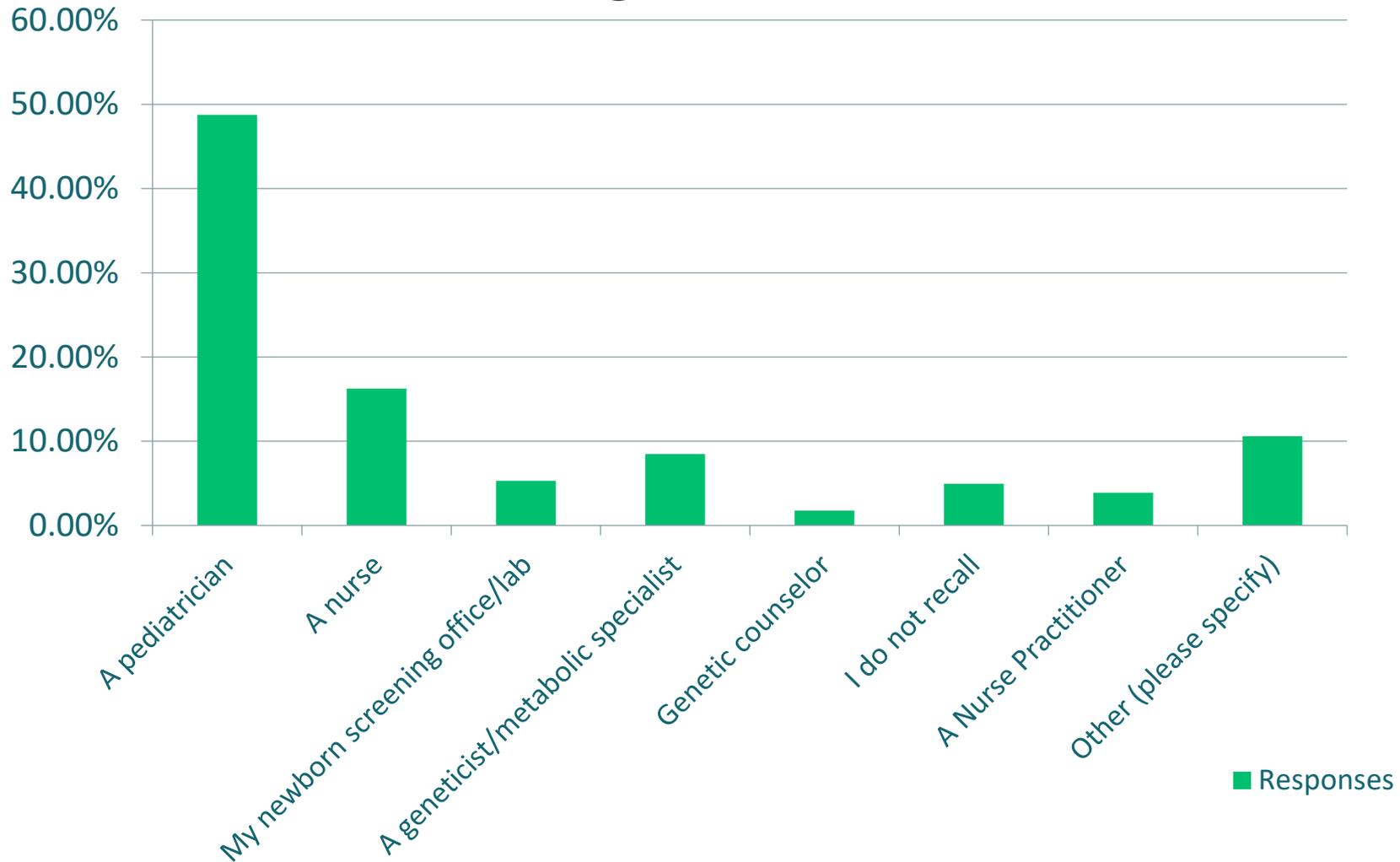
# Number of Responses by Condition

Responses



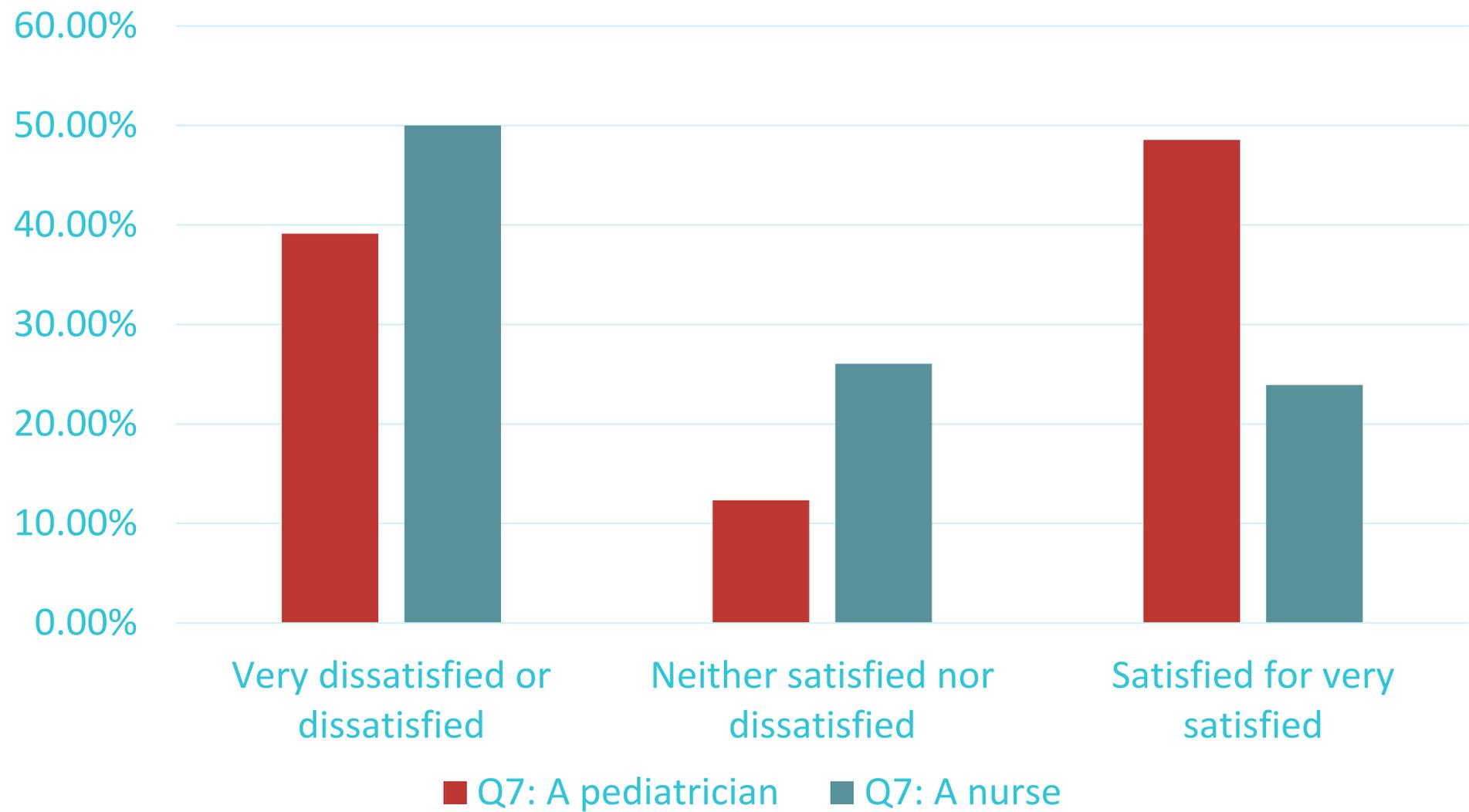


# Who contacted you about an out-of-range result?



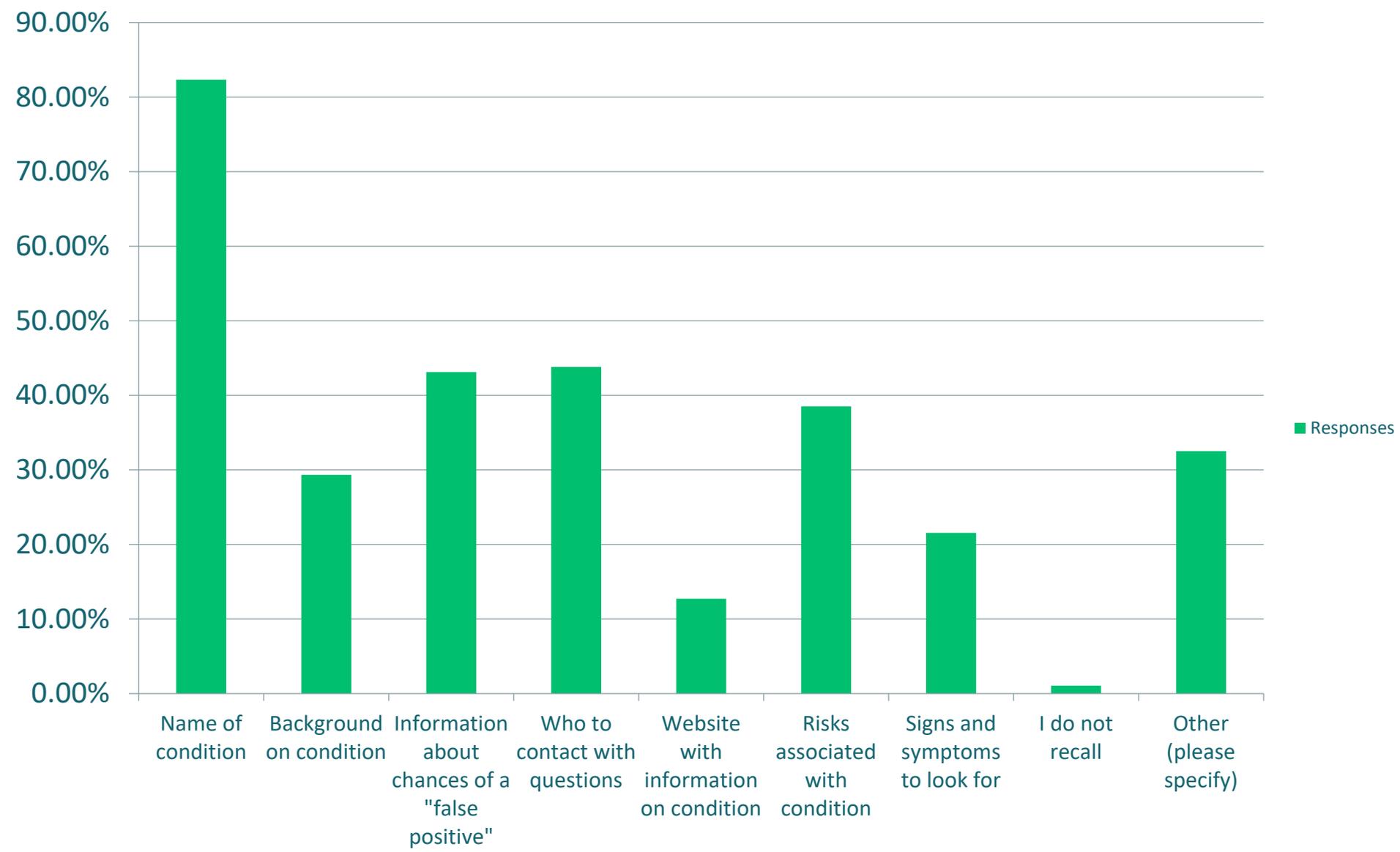


# Overall, how satisfied were you with the information you received during the initial contact with five being very satisfied





# What information were you given during the initial contact? Please check all that apply.





# Positive responses

- Themes
  - Previous relationship with physician
  - Older child who already has the condition
- ***“... She wanted to be the first to tell us because we had a long standing relationship. While she didn't have a lot of answers she did say that she will be in close contact with the genetic care team and we will all learn together how to best move forward. I am very glad my first contact was not with a person I had never met.”***



# Negative responses

- Themes
  - False sense of security
  - Lack of knowledge
- ***“The pediatrician laughed and said he would be “retarded” if he had PKU...”***
- ***“Go to the ER immediately. If you don’t your child will be mentally retarded. If you have questions call Dr ....” That was the message left on my voicemail.”***
- ***“It’s just a bad read, your odds are better that you win the lottery twice than this being a thing”***



# Given the wrong condition

- *“Given the wrong name. **We were told of a similar condition but not the condition our child had.** The pediatrician messed up.”*
- *“I was actually **told the incorrect name** but was able to figure it out on my own.”*
- *“We were told he’d **failed the “PKU test”**, but they didn’t actually know which condition he’d tested positive for since it tested for a variety of conditions.”*
- *“**Name of condition was given but wrong.**”*

# Was there any information you wish you had been given during the initial contact that you did not receive?

- No or positive response (~30%)
  - *“Our doctor was extremely sensitive and supportive. In hard times like that, it is impossible to have all questions answered right away. Our doctors did the best they could with the limited knowledge they had.”*

# Theme

- Explanation lacked needed relevant information (~25%)
  - Accurate websites/resources (~9%)
  - Availability of treatment (~7%)
  - Support groups (~5%)
  - Who to contact with questions (~5%)

*“I would have liked an informed explanation. **Our Dr. knew nothing about the condition and just said we needed to go to a metabolic and genetics hospital out of town and they could answer our questions. That led to a lot of online research and no sleep for my husband and I for 3 days. Worry unimaginable and broken hearts. He is a perfectly healthy boy thriving greatly.**”*

# Theme

Feeling like there was a lack of knowledge (~8%)

*“The initial contact I had was a phone call from a nurse at the pediatricians office. **She told me my daughter was positive for PKU (she was pos on NBS for MCADD). She could give me no information aside from scheduling with the pediatrician that afternoon and to take my daughter to the ER if I became concerned for her. She wasn’t very compassionate and had zero information to provide to us aside from the fact there were abnormalities seen.”***

# Theme

Lack of empathy(~7%)

***“Yes, I wish the person contacting had been understanding of the severity of the news I was being given, had given some information about what was happening and what his possible outcomes were.”***





# Conclusion

- Major themes
  - General feeling that more information or a place to find accurate information is needed
  - Once families were connected with the specialist people felt they were given the answers they needed



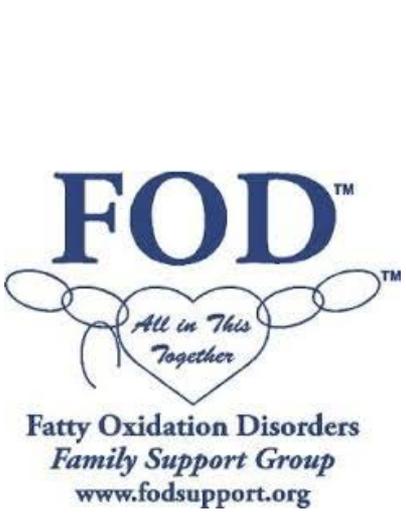
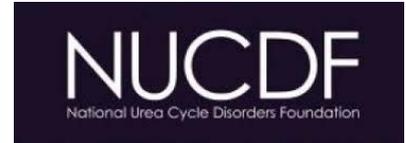


# Next Steps

- Deeper analysis of the data
- Should there be protocols on who from the PCP office contacts families?
- What other ways can families receive the information they need?
  - Video series?



Thank you for your help!





# Acknowledgments:

- Genetic Alliance
- Baby's First Test
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- Bay Path University

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