Lessons Learned: Bleeding Disorders Community Response on Gene Therapy Research

Elaine Chan[1], Meg Bradbury[1], Mabel Cresiconi[1], and Tessa Field[2]

[1]Hemophilia Federation of America, Washington, DC, USA, [2]Spark Therapeutics, Philadelphia, PA, USA

Objective/Purpose of the Project

Recent medical advancements have pointed to gene therapy as an emerging treatment intervention for individuals with hemophilia. To better understand knowledge and perception of gene therapy, patients with hemophilia and their caregivers were surveyed to:

- assess patients and caregivers' knowledge of gene therapy;
- understand how and from whom they prefer to learn about their bleeding disorder and the potential of gene therapy; and
- identify knowledge gaps

Methods

- An instrument was developed (IRB approval #AAAA001) using Likert-type scales, multiple choice, and open-ended questions. Prior to enrollment, construct and face validity of the instrument was obtained by piloting to community members; adaptations were made as necessary.
- Recruitment and access to the survey was done via a Web based Community Research Portal, email list serve and social media postings with enrollment occurring between July-October, 2018. Target audience was the bleeding disorders community (BDC).
- Using Microsoft Excel 2016 and NVivo 12 descriptive statistics and qualitative data analysis was conducted (n=136).
- Participants were given the option to leave the survey at any time (attrition) as well as opt of out answering particular questions throughout the survey. Various survey questions were formated for multiple responses as well to allow for participants to select all options that were applicable.

Results

Demographics

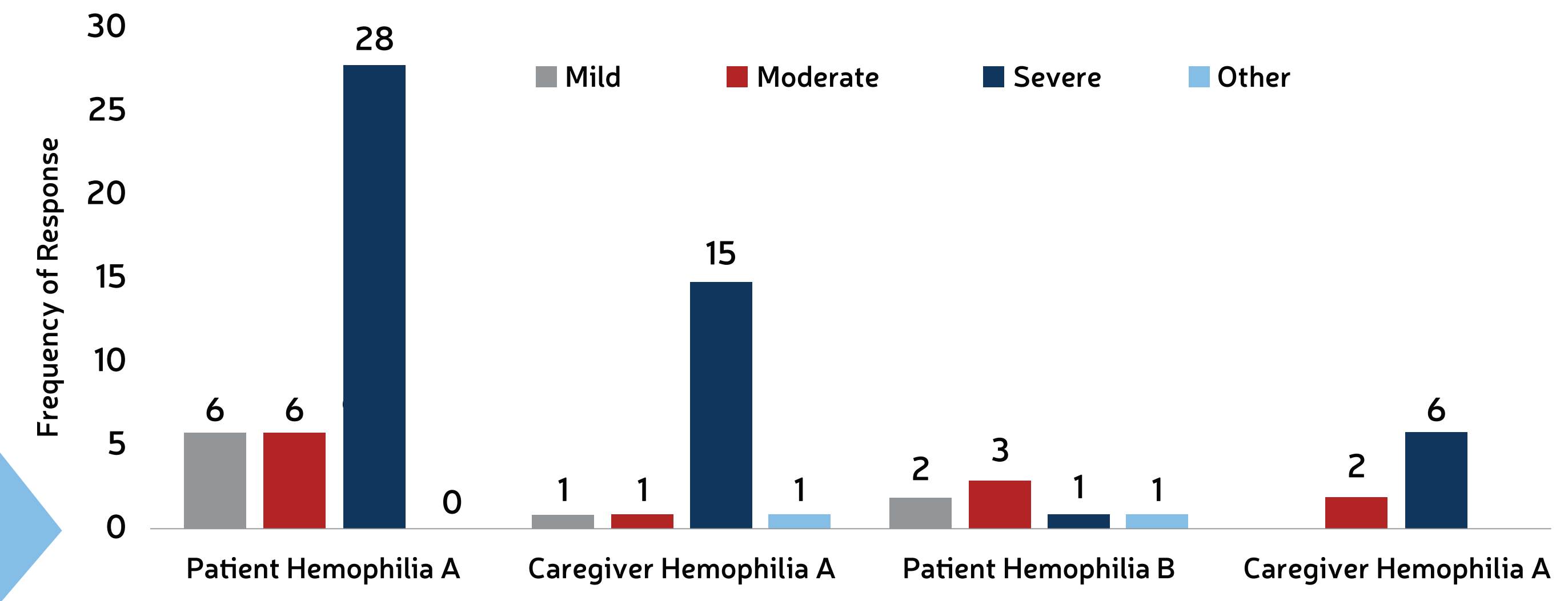
Gender	
Male	44% (n=44)
Female	56% (n=57)

Patient or Caregiver	
Patient	47% (n=64)
Caregiver	29% (n=39)
Other respondents*	24% (n=33)

*Other participants included, but not limited to, individuals who work within the bleeding disorder community and representatives of bleeding disorders organizations.

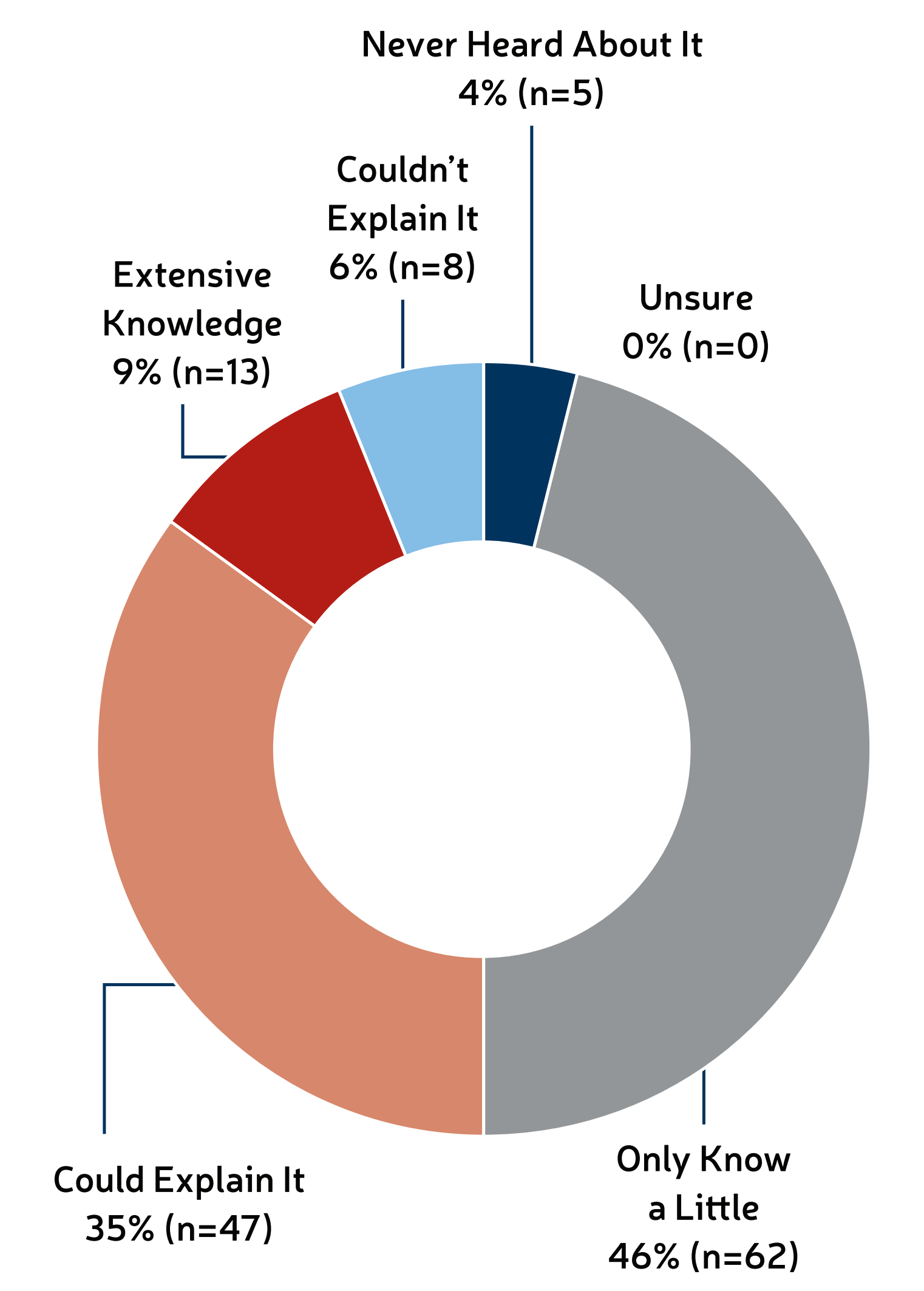
Majority of survey participants self-identified as patients with hemophilia A.

Patient vs. Caregiver Stratified by Disease Type and Severity (N=128)



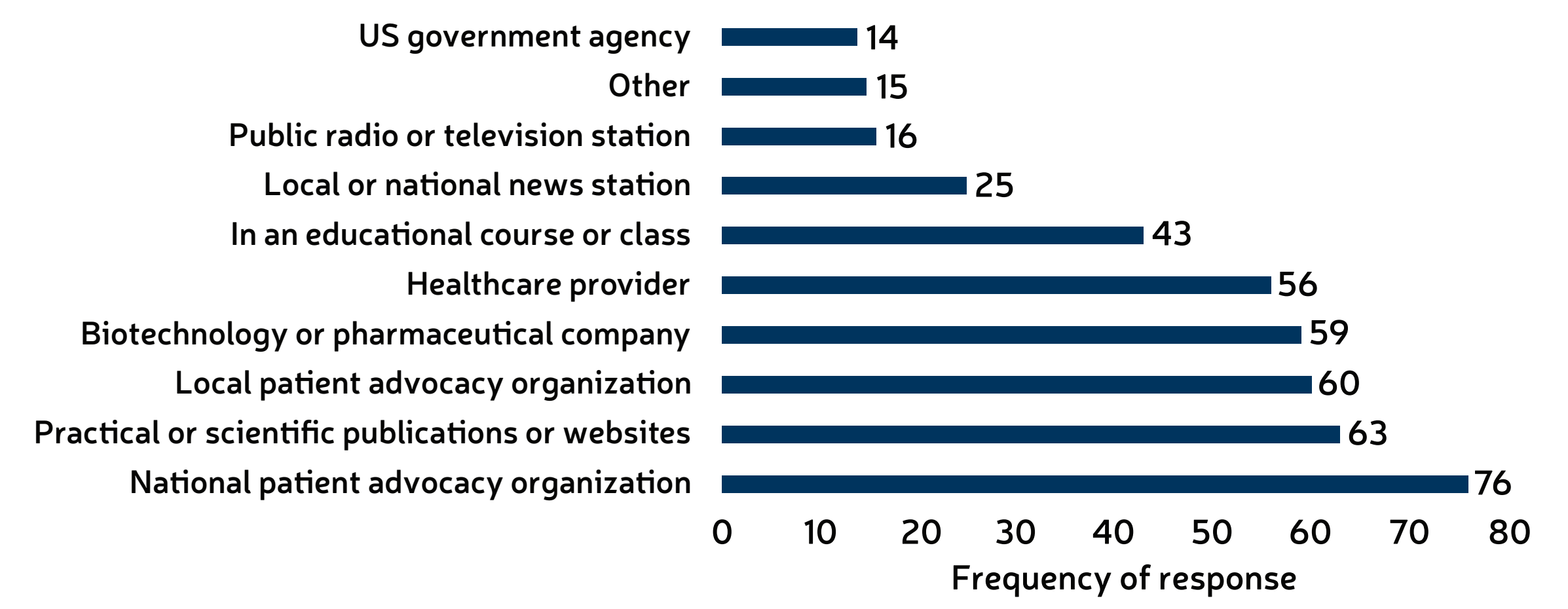
Community Awareness of Gene Therapy

Gene Therapy Awareness (N=135)



Most participants have heard of 'gene therapy' but the extent of knowledge varied. When stratifying by patient vs. caregivers, the patients self-reported a higher level of knowledge (49%) while 33% of caregivers reported having at least enough knowledge to explain gene therapy.

Source of Gene Therapy Awareness (N=130)



How Would You Describe Gene Therapy?

Through thematic analysis, there was a wide range of themes that emerged from written responses:

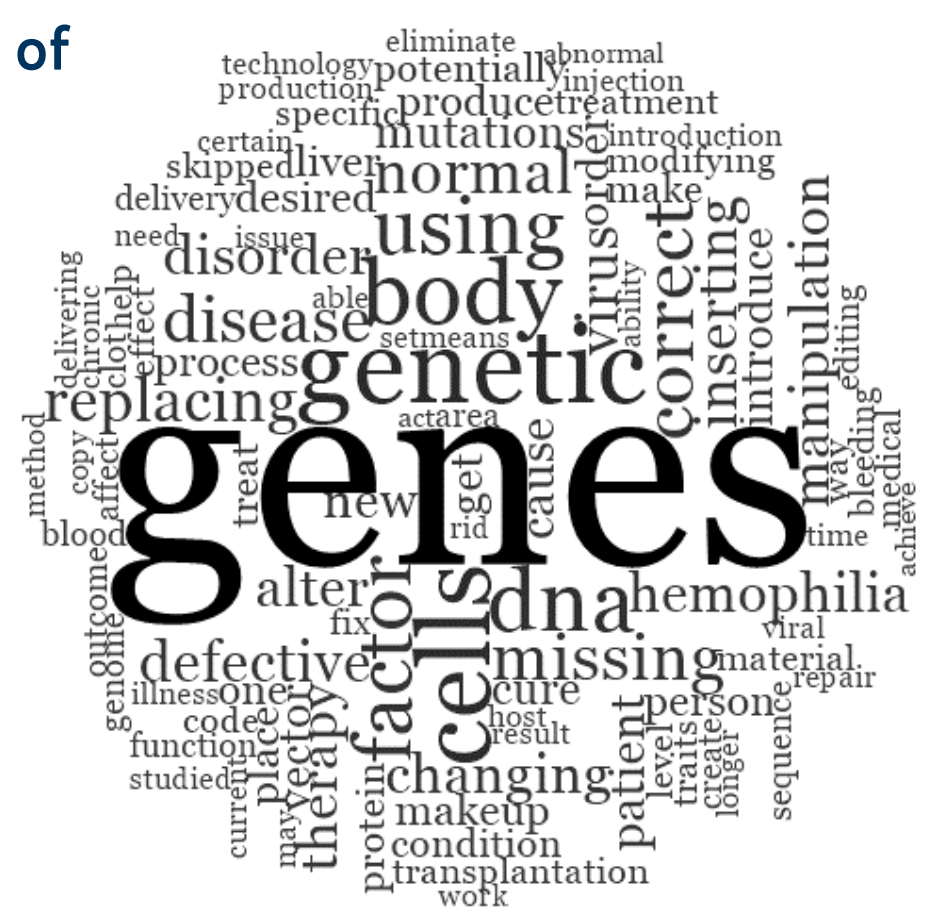
Risks, Emotions, Expectations, & Concerns
"Gene therapy is something that could potentially help hemophilia patients in the long term and eliminate the need for weekly infusions."

Future Therapy or Long-term Outcomes
"It's the future of our care."

DNA and Hepatic Cells
"The process of splicing genes into the patient's DNA to either permanently or temporarily alter the gene code of the patient."

Cure
"... gene therapy is the ability to splice into a gene some of what is missing in our DNA makeup to have our body start to reproduce the missing coagulation needs of our blood so that we can lead more normal lives. Also, for someone like myself, who is mild, that it could cure my hemophilia completely, which would be a massive step forward."

Temporary Fix
"Manipulating genes to solve a problem/disease/disorder. It does not alter genes for future generations."

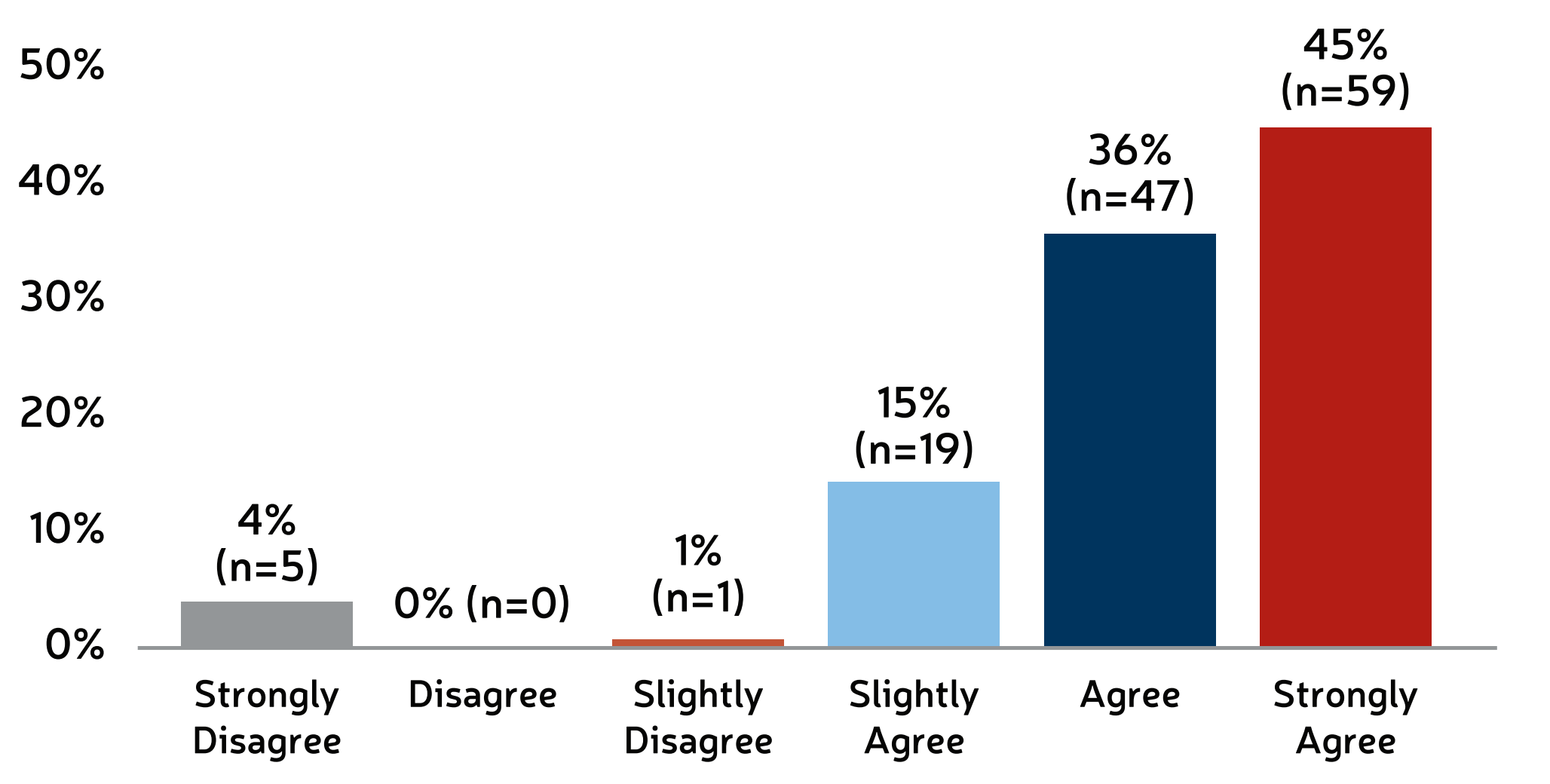


The word cloud summarizes list of words used by participants when asked, 'What is gene therapy?' with size of each word illustrating the frequency by participants.

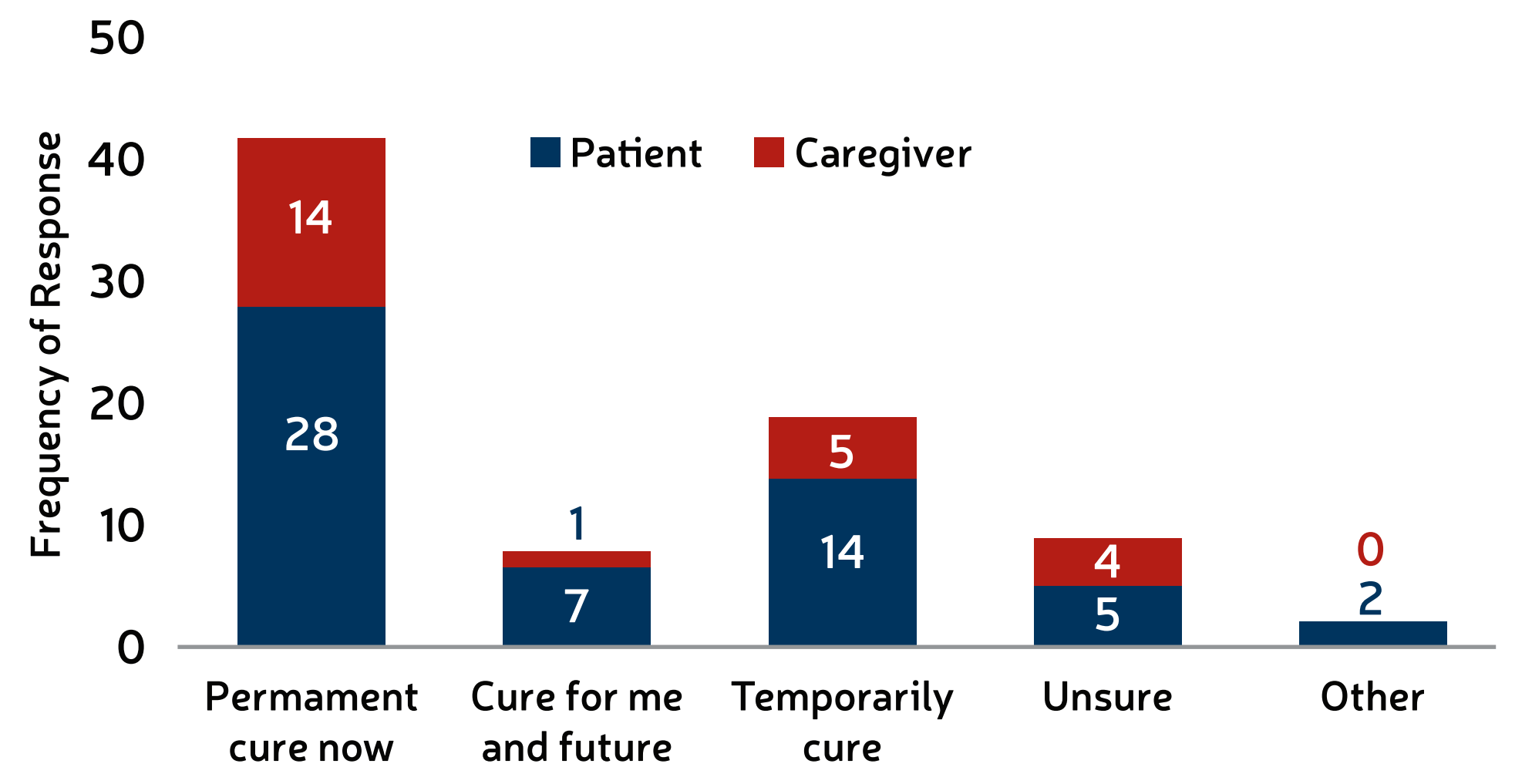
*The selected quotes above are participants perceived knowledge of what is gene therapy. This does not represent actual gene therapy treatment for bleeding disorders. This is not an exhaustive list of all the themes identified but a sample for discussion/presentation purposes.

Expectations of a Potential Gene Therapy

I am excited about the potential of gene therapy to improve life for people with chronic diseases.



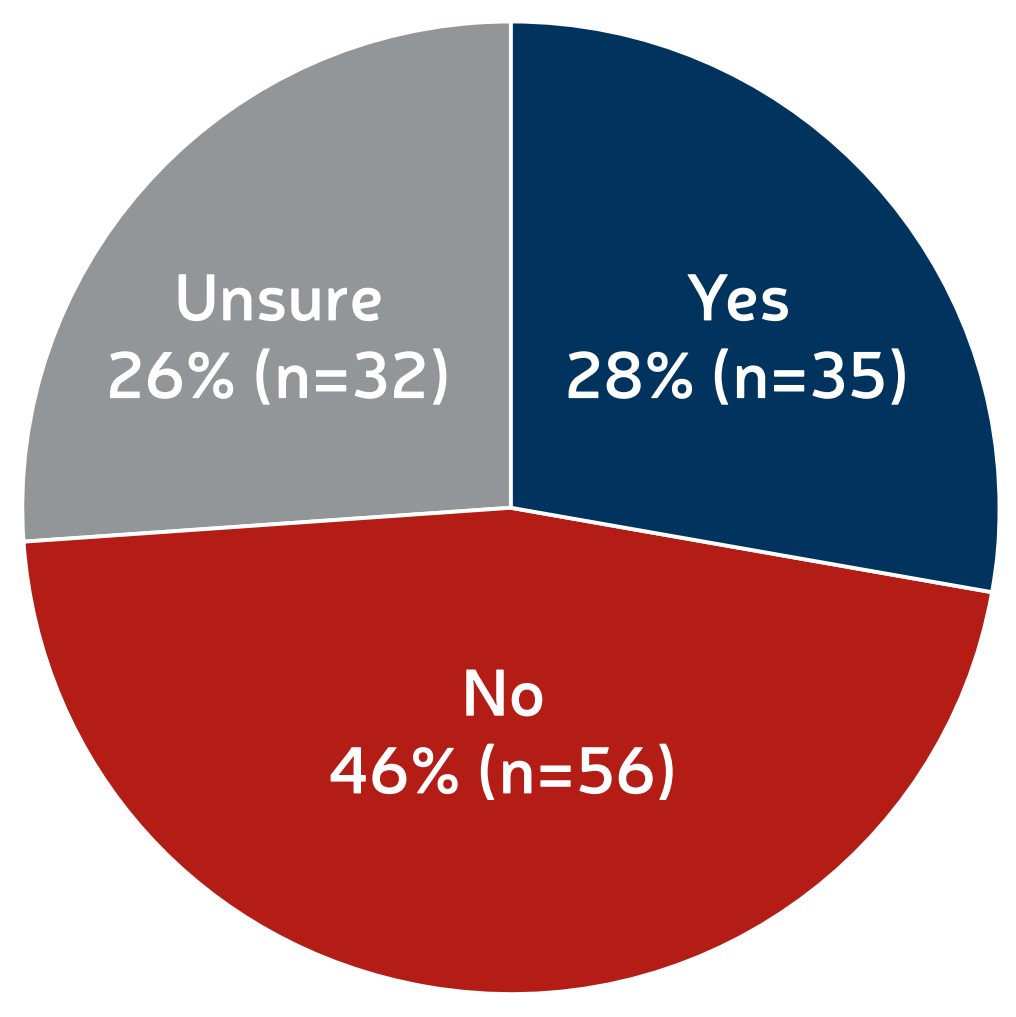
Expectations for a Potential Gene Therapy (N=57)



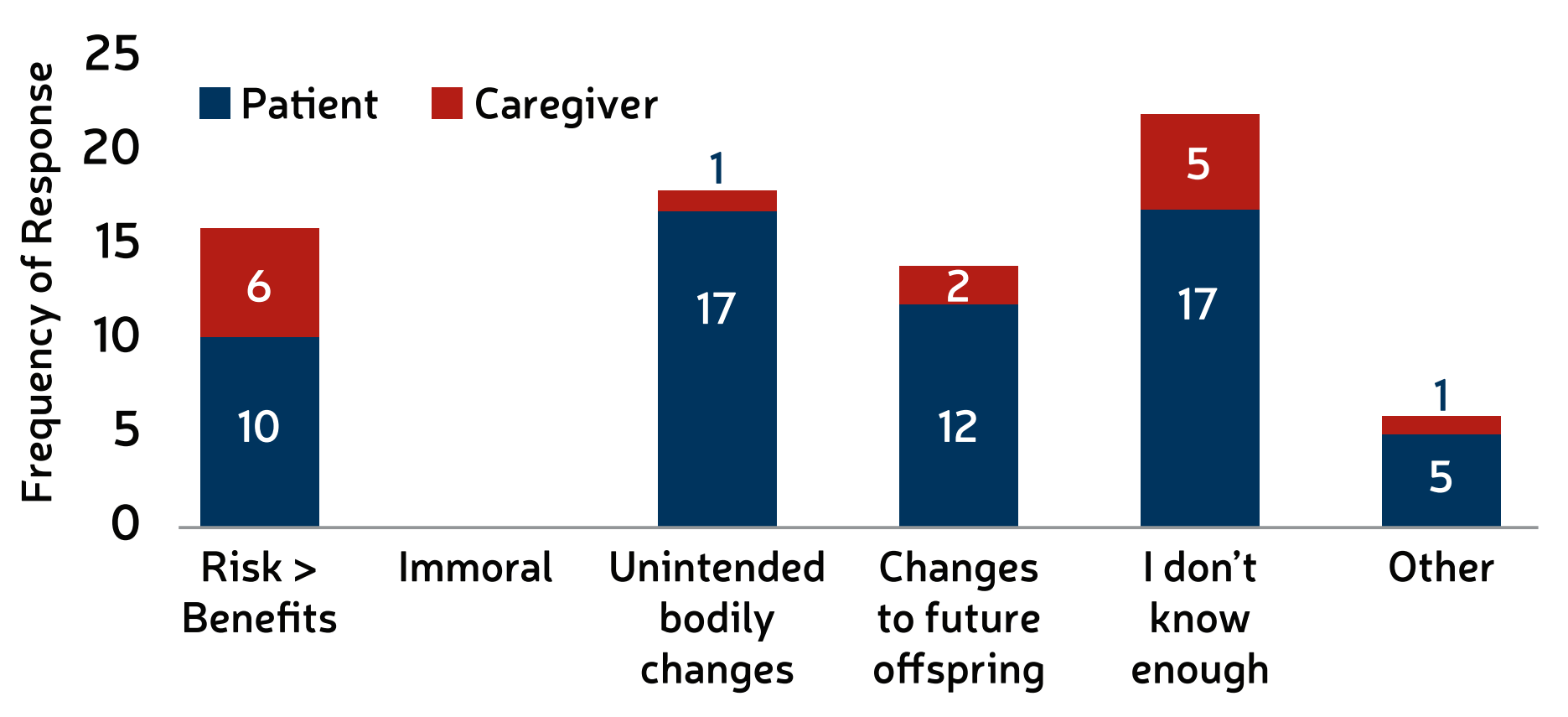
A majority of participants noted expecting gene therapy to provide a "permanent cure". However, cure was not defined within this study and it is acknowledged this phrase may be define differently depending on the individual. While it is clear and noteworthy that community expectations are high, it is also important to note that gene therapy for BD is currently in the research stage and these long-term data are currently being collected.

Awareness of gene therapy vs. gene editing (N=123)

Our analysis reveals that for some of our participants, these two technologies are indistinct. The implications of and differences between these two technologies are important for the community to understand. These results suggest clarification between these terms is warranted.



Concerns for a Potential Gene Therapy Treatment (N=43)



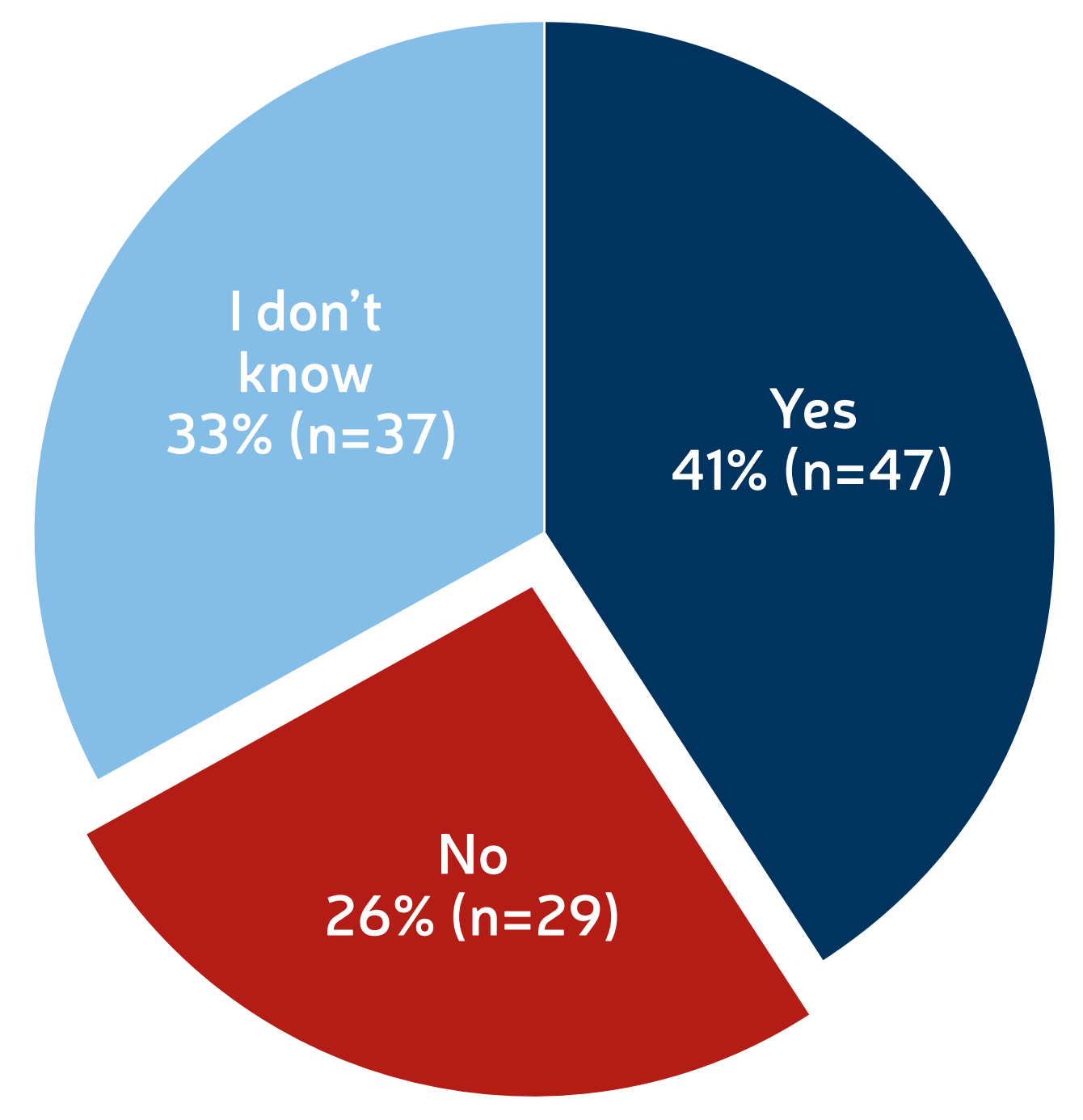
Those participants who selected 'other' mentioned, cost, insurance coverage, happy with current treatment, prior adverse outcomes in gene therapy research.

What do you want to know about a potential gene therapy for hemophilia? (N=122)

Top 5 Responses

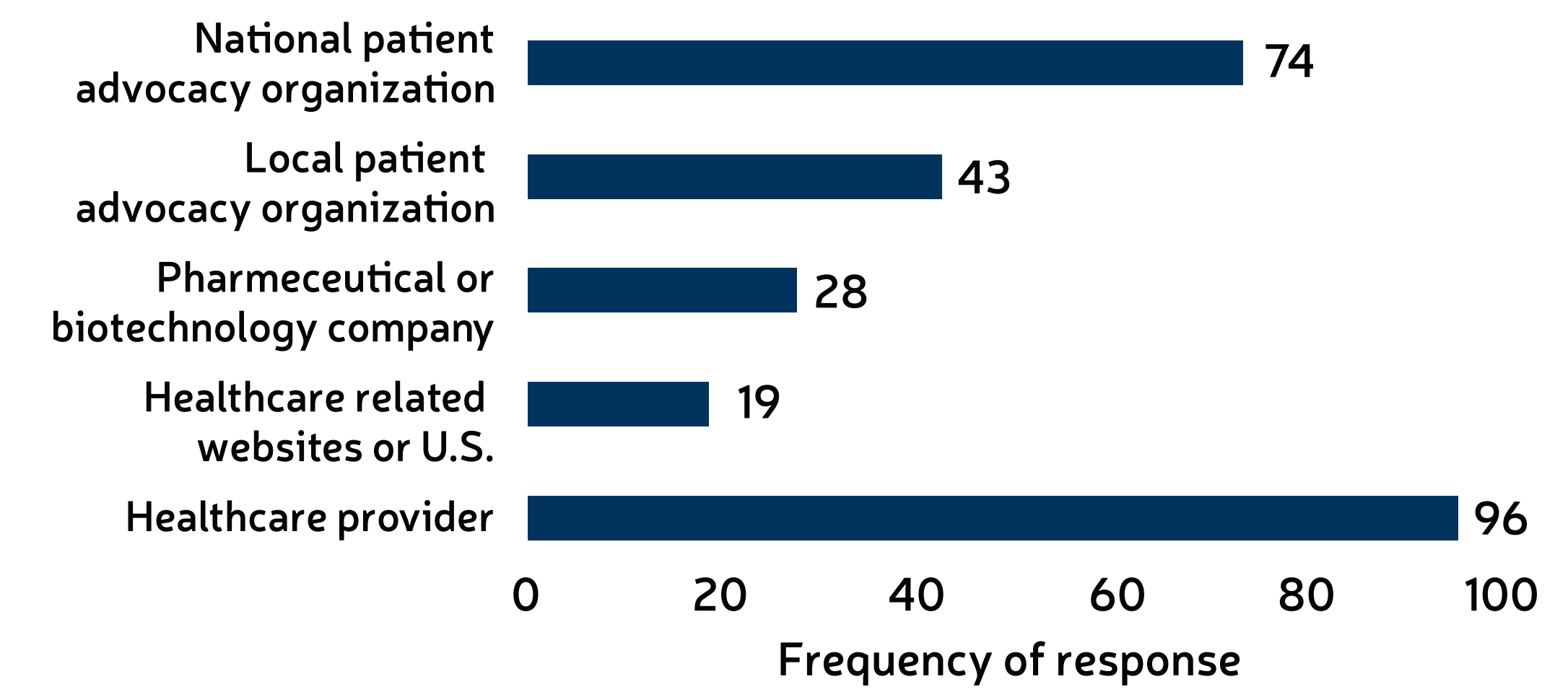
- What might be the side effects of a potential gene therapy? (n=102)
- Is gene therapy safe? (n=96)
- Will insurance pay for it? (n=90)
- How long could a potential gene therapy last? (n=89)
- How much could a potential gene therapy cost? (n=86)

Perceived Healthcare Provider Gene Therapy Research Knowledge (N=113)



Do you think your healthcare providers know enough about gene therapy research for hemophilia to discuss it with you and answer the questions you have about it?

Which of the following sources of information on gene therapy would you trust to learn more about it? (N=113)



Conclusion

While expectation and excitement around the potential of gene therapy is high, the study demonstrates participants' perception of access to limited information around the topic as a whole, which may lead to an increase the increase in frequency of the choice " I don't know enough" as a concern for a potential gene therapy. Participants express a desire to learn more about gene therapy research, specifically from patient advocacy organizations and in-person interactions with healthcare providers. Patient advocacy organizations are encourage to offer educational resources on gene therapy research to address participants' needs. While participants reported healthcare providers as being their most trusted source of information regarding gene therapy; less than half thought their healthcare provider knew enough about gene therapy research to answer their questions. Further assessment of healthcare provider knowledge and their resources for understanding gene therapy may provide insight into how to best equip HCPs to facilitate the BDC in understanding this emerging field.

Limitations: Our survey title, "Gene Therapy and You" may have a response bias. The title may have attracted individuals with more optimistic views of, or interest in, gene therapy, or perhaps implied a need to have experience with gene therapy. While strategies were employed to mitigate this, the participant sample may not representative of the BDC with an over representation of individuals who were interested and in favor of gene therapy. Based on the research team's interactions with the bleeding disorders community, this may have led to an under representation of individuals with less optimistic views or interest in gene therapy.