

# Comparison Of Rare Disease Patient And Caregiver Perceptions Regarding Access To Disease Information And Interactions With Their Healthcare Providers: Results From An International Survey

PCR147

Siva Narayanan PhD<sup>1</sup>, Wes Michael MBA<sup>2</sup>.

<sup>1</sup>Avant Health LLC, Bethesda, USA; <sup>2</sup>Rare Patient Voice, Towson, USA.

## BACKGROUND

- Rare diseases (RD) are often hereditary and begin in early childhood. About 30% of children with a rare disease die before the age of 5 due to delayed diagnosis.
- The disease journey for RD patients is challenging due to the lack of awareness around it, the lack of specialists, and lack of adequate diagnostic treatments and tools.
- Healthcare Providers (HCPs) are not often trained to recognize RD but rather common diseases, and they are not well informed about the latest RD research.
- Due to the lack of training in RD, educating patients and communicating the life changing news in a sensible manner are fields that need improvement for HCPs.
- In addition, due to the social lack of awareness, patients and their caregivers often have to educate others and self-educate.

## OBJECTIVE

- To assess perceptions of RD patients and their caregivers regarding access to RD information and interactions with their healthcare providers (HCPs).

## METHODS

- An online survey. The quantitative data was analyzed using SPSS, international survey with 800 participants was conducted across Germany, United Kingdom (UK), Spain and the United States (U.S), using Rare Patient Voice online patient panels.
- RD patient/caregiver opinions about their (or their family member's) experience living with RD, including information about interactions with their HCPs were gathered using an online survey.
- All participants were sourced from the Rare Patient Voice panels and outreach program. Participants were eligible to participate if they had a RD, were over 18y and from one of the 4 countries mentioned above. Adult caregivers of patients with a RD condition were included as well.
- A combination of qualitative and quantitative data were collected from participants; the quantitative data was analyzed using SPSS software.

## CONCLUSION

- Majority of the RD patients and caregivers reported difficulties attaining information regarding their RD or new treatment options and reported rarely receiving RD information from their medical team and that the received information was not easy to understand. Perceptions varied between patients and caregivers. Better strategies to improve knowledge and awareness of RD among HCPs, and better communication of RD information to patients/caregivers is warranted.

## REFERENCES

Alvarez, R. (2022) Patient perspective: My rare disease journey, Cold Spring Harbor molecular case studies. Available at: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8958917>

Guilbert, M. et al. (2021) Results of a patient reported experience measure (PREM) to measure the rare disease patients and caregivers experience: A Spanish cross-sectional study, Orphanet journal of rare diseases. Available at: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7866674>

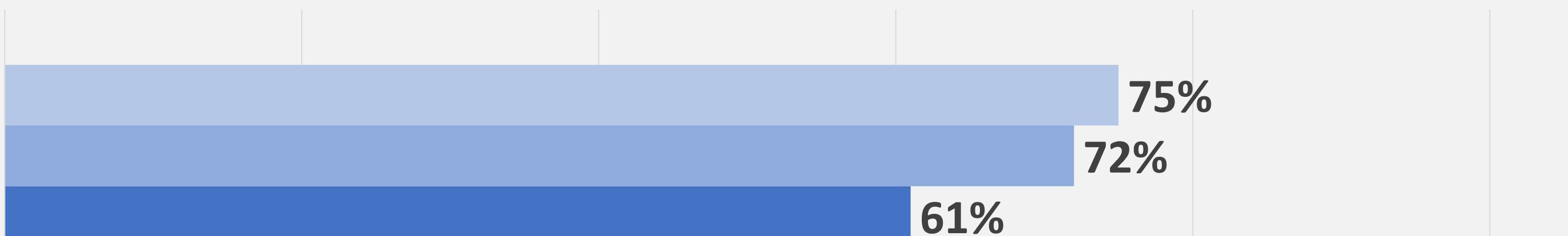
Baumbusch, J. et al. (2018) Alone in a crowd? parents of children with rare diseases' experiences of navigating the healthcare system, Journal of genetic counseling. Available at: <https://pubmed.ncbi.nlm.nih.gov/30128673>

## RESULTS

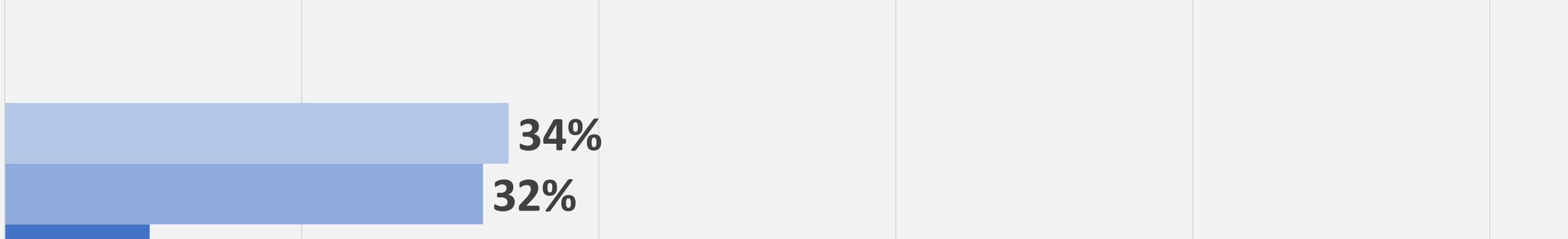
- Survey participants included 100 from Germany, 168 from Spain, 252 from the UK and 280 from the U.S. Majority (78%) were RD patients, 15% were caregivers of a RD patient, and 7% represented "both" groups.
- Majority of patients (72%), caregivers (61%) and both (75%) reported it was 'difficult/very difficult/extremely difficult' attaining information regarding their RD or new treatment options under development for their condition.
- Between 10%-34% of respondents reported "often/always" that their needs were heard by their medical team.
- Between one-fourth and one-third of respondents (patients: 27%, caregivers: 33%, both: 25%) reported that the information received from various sources is 'often/always' easy to understand.
- Only a small proportion of patients (12%), caregivers (22%), both (13%) reported 'often/always' receiving education about their RD from their medical team.
- Between one-third and half of the respondents (patients: 48%, caregivers: 33%, both: 48%) reported that their medical team is 'never/rarely' educated about their RD.

### Experience of patients, caregivers, and patients who are caregivers (both) with their medical team and rare disease

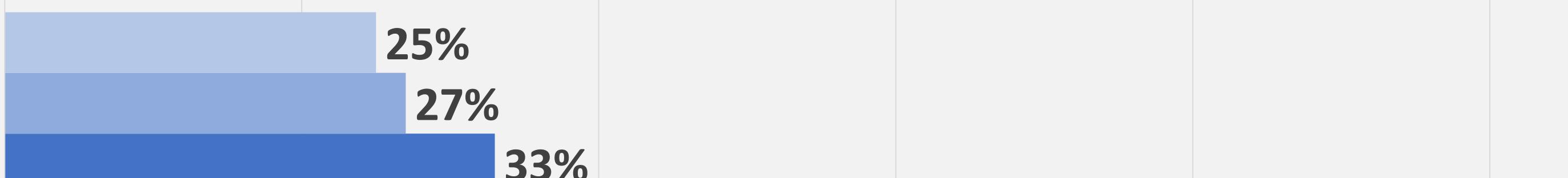
Find it 'difficult/very difficult/extremely difficult' to attain information regarding their RD or new treatment options under development for their condition



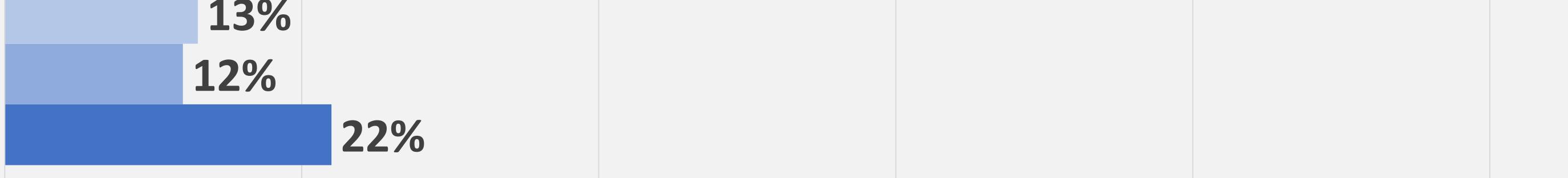
Feel that their needs are 'never/rarely' heard by their or their family member's clinical team (doctor and nurses)



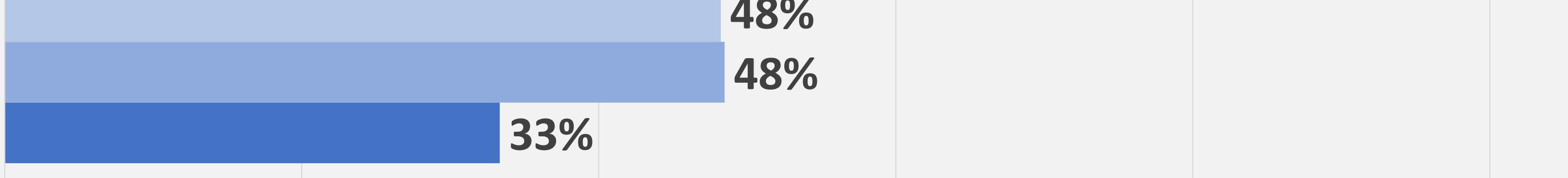
Information about the rare disease condition being presented (from sources such as pharmaceutical studies, patient leaflets, etc.) is 'often/always; easy to understand'



When visiting the doctor, 'often/always' receives education about the rare disease condition



Doctors or nurses who are taking care of them or their family member is 'never/rarely' educated about the rare disease condition



■ Both ■ Patient ■ Caregiver

0% 20% 40% 60% 80% 100%

For more information: snarayanan@avant-health.com

Presented at ISPOR 2025, Montreal, QC, Canada