



# Perceptions of Rare Disease Patients and their Caregivers Regarding Access to Disease Information and Interactions with their Healthcare Providers: Results from an International Survey

Siva Narayanan PhD, Avant Health LLC, Bethesda, USA. Wes Michael MBA, 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 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, using an online survey. The quantitative data was analyzed using SPSS, while the qualitative data (verbatim from respondents) was analyzed using the MAXQDA 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. 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; over 300 unique RDs were represented within the study cohort.
- Only 14% of respondents reported 'often/always' receiving education about their RD from their medical team and only 28% reported that the information received is 'often/always' easy to understand.
- Approximately 46% reported that their medical team is never/rarely educated about their RD. (Fig 1)
- Majority (70%) of participants reported they 'often/always' had to educate others (including their medical team) related to their RD. (Fig 2)
- Majority (71%) of participants reported having difficulties attaining information regarding their RD or new treatment options under development for their condition. (Fig 3)
- Furthermore, only 29% reported "often/always" that their needs were heard by their medical team. The results varied across the countries. (Fig 4)

Figure 1. Are the HCPs educated about the patient's RD condition?

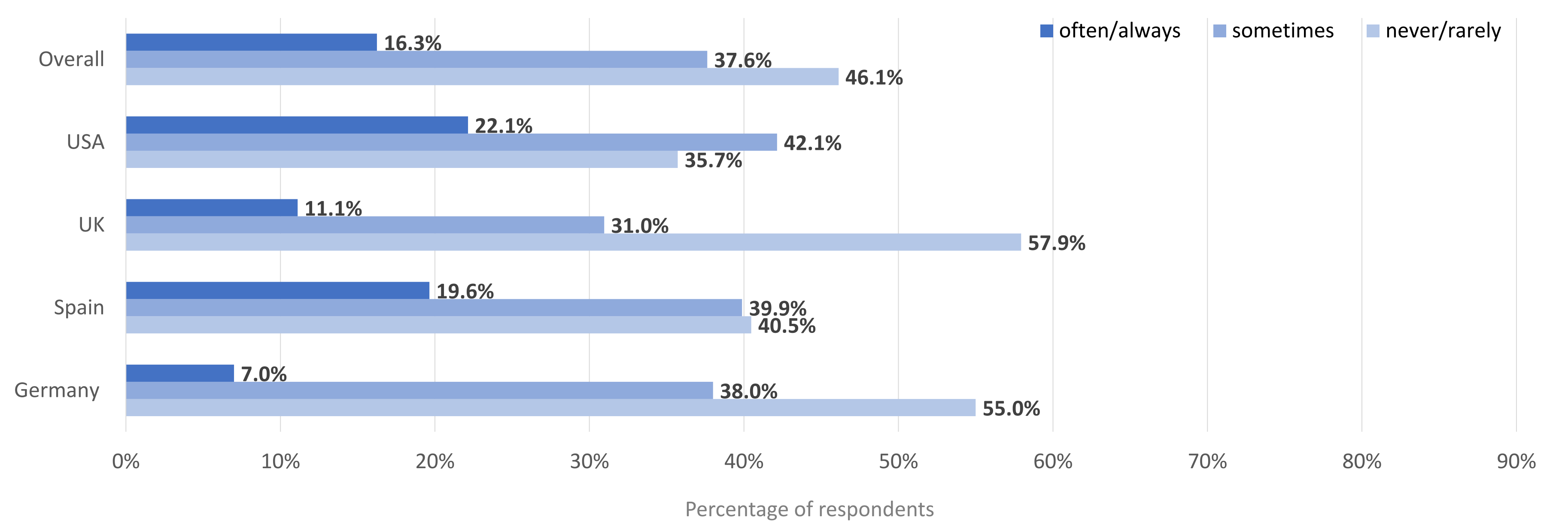


Figure 2. How often do you need to educate your HCPs about RD condition?

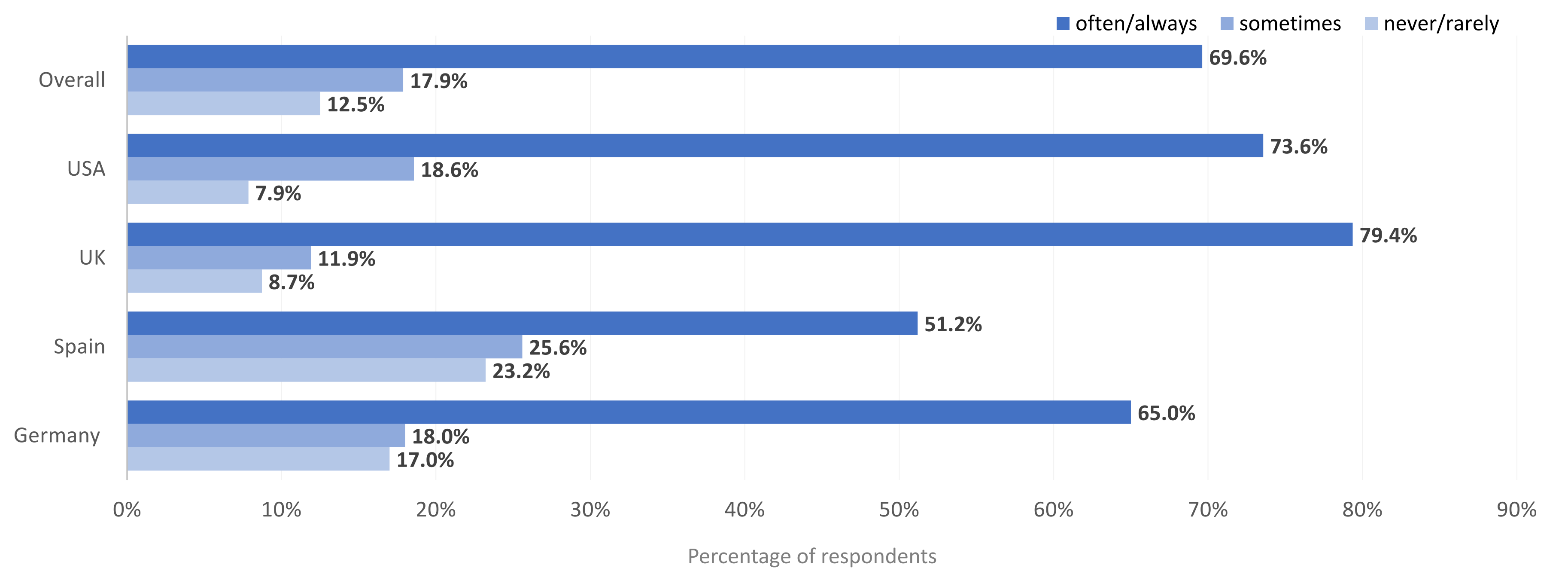


Figure 3. Difficulty in finding information about patient's RD condition

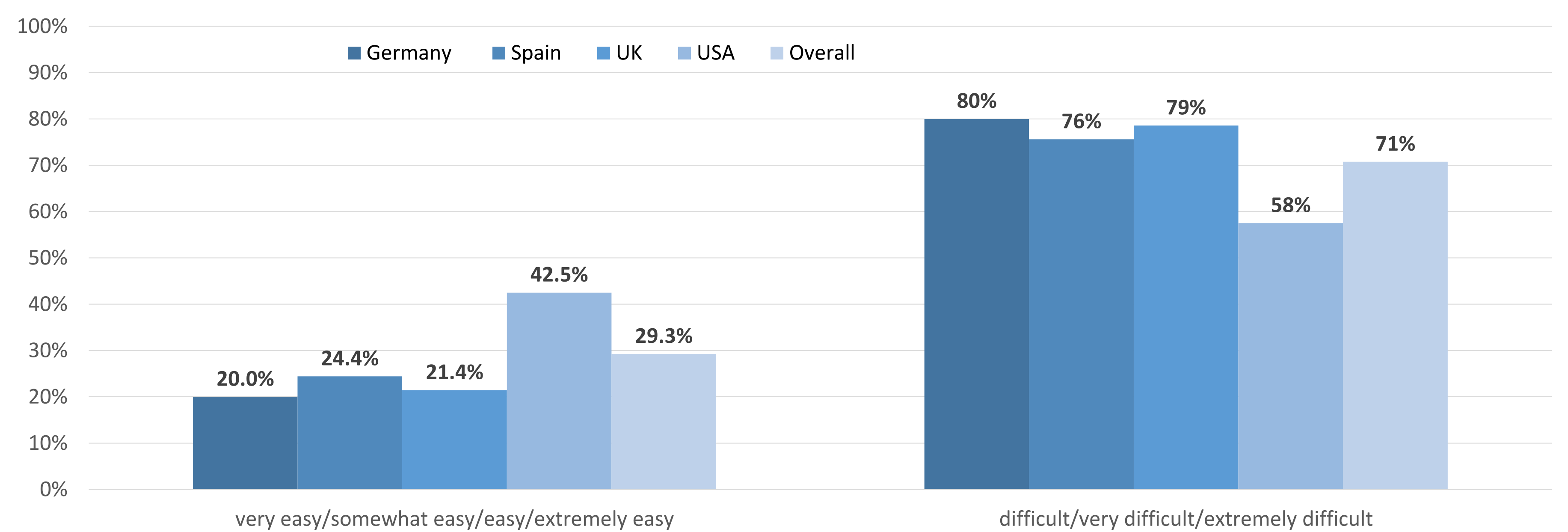
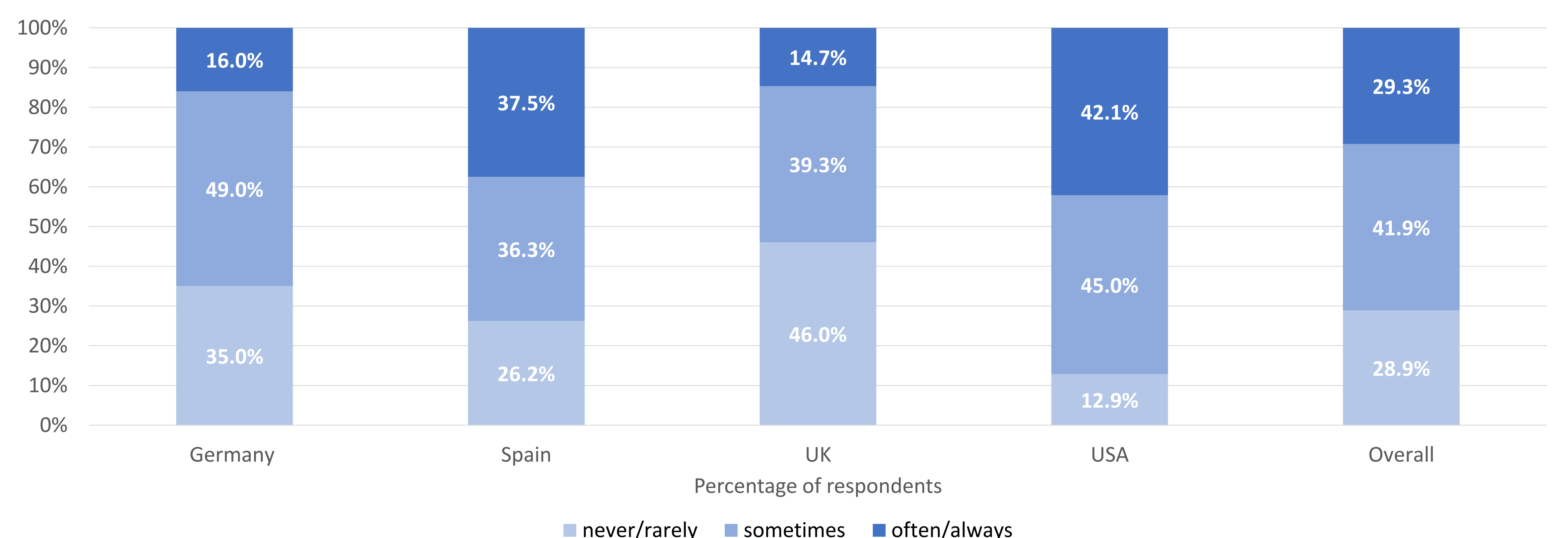


Figure 4. Difficulty in finding information about patient's RD condition



For more information, contact: [snarayanan@avant-health.com](mailto:snarayanan@avant-health.com)