



Rare Disease Patient and Caregiver Perspectives On Rare Disease Diagnostic Journey: Results From An International Survey

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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 available diagnostic treatments and tools.
- Diagnostic tools are limited due to the lack of research fundings in rare diseases, therefore many patients live undiagnosed for many years.
- Access to appropriate care, treatment, and diagnosis is difficult due to a lack of specialists or/and financial limitations.
- On average it takes about 4.8 years and 7 specialists for an accurate diagnosis. The meantime is accompanied by misdiagnosis.

Objective

- To understand the diagnostic journey of RD patients by asking RD patients and their caregivers about insights and experiences in the healthcare system.

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 diagnostic journey, access to healthcare, and perspectives on living with a RD 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

Overall, the diagnostic journey for RD stakeholders has been prolonged and arduous, often fraught with difficulty finding the right RD specialist, difficulty receiving diagnostic treatments, or their RD being misdiagnosed. The burden associated with RD diagnostic journey varied dramatically across the countries, with patients often experiencing higher burden in one of the studied European countries, warranting closer scrutiny in improving health care access and delivery for the RD stakeholders in relevant geographies.

References

Lancet Global Health. (2024) The landscape for rare diseases in 2024. Available at: [https://www.thelancet.com/journals/langlo/article/PIIS2214-109X\(24\)00056-1/fulltext](https://www.thelancet.com/journals/langlo/article/PIIS2214-109X(24)00056-1/fulltext)

Adachi, T. et al. (2023) Enhancing equitable access to rare disease diagnosis and treatment around the world: A review of evidence, policies, and challenges, International journal of environmental research and public health. Available at: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10049067>

Baynam, G. et al. (2024) Global Health for Rare Diseases Through Primary Care, The Lancet Global Health. Available at: <https://www.sciencedirect.com/science/article/pii/S2214109X24001347>

Faye, F. et al. (2024) Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: Results of a rare Barometer Retrospective Patient survey, Nature News. Available at: <https://www.nature.com/articles/s41431-024-01604-z>

Results

- Survey participants included 100 from Germany, 168 from Spain, 252 from the UK and 280 from the U.S.
- Median age of participants was 44 years; 85% were female; 78% were RD patients, 15% were caregivers of a RD patient, and 7% represented both patient and caregiver. Over 300 unique RDs were represented within the study cohort.
- Over 72% reported difficulty in finding a doctor specialized in the RD of interest, and it ranged between 67% (U.S) and 80% (Germany).
- Overall, 66% took up to 5yrs to receive a correct RD diagnosis (Fig 3); 21% took >10yrs to receive a correct RD diagnosis and it ranged between 15% (U.S) and 28% (UK).
- Over 54% reported to have been misdiagnosed at least once in their diagnostic journey. (Fig 1)
- Majority had to see >=3 specialists, to receive their RD diagnosis. (Fig 2)
- 34% reported being denied diagnostic treatment during their diagnostic journey. (Fig 4)

Figure 1. Number of times patients were misdiagnosed

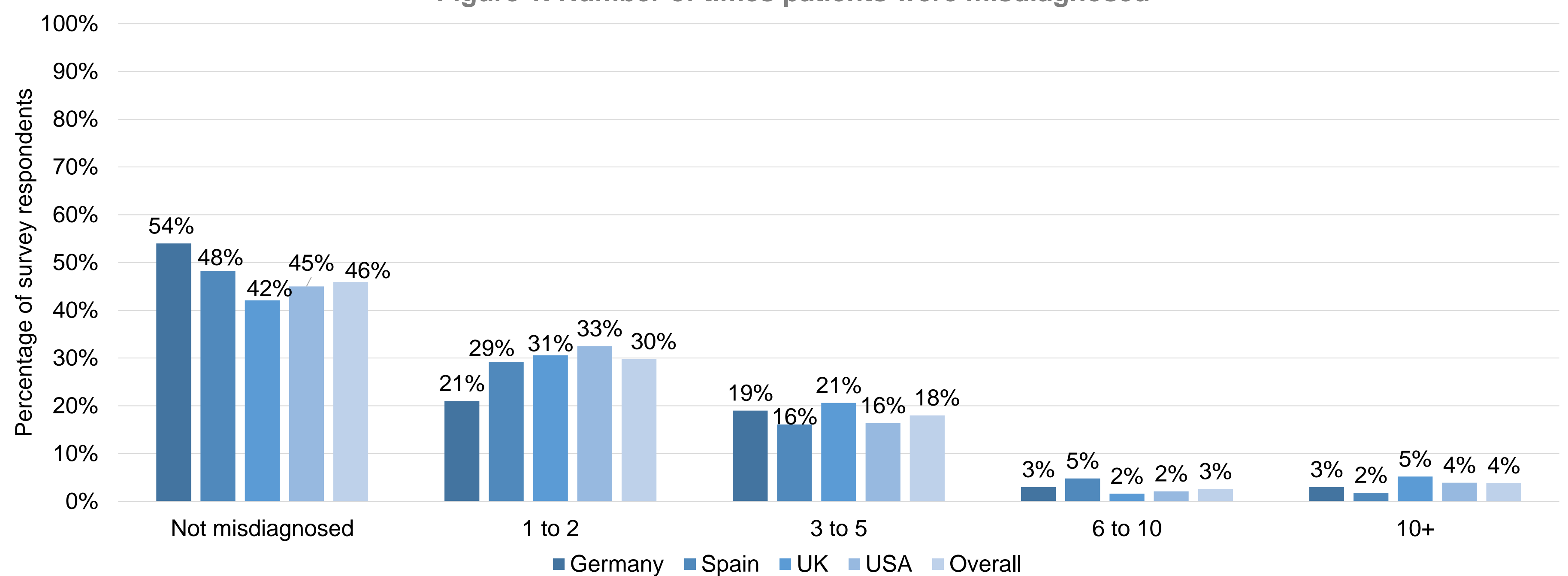


Fig1. reflects the number of patients that were misdiagnosed

Figure 2. Number of specialists patients consulted before their confirmed diagnosis

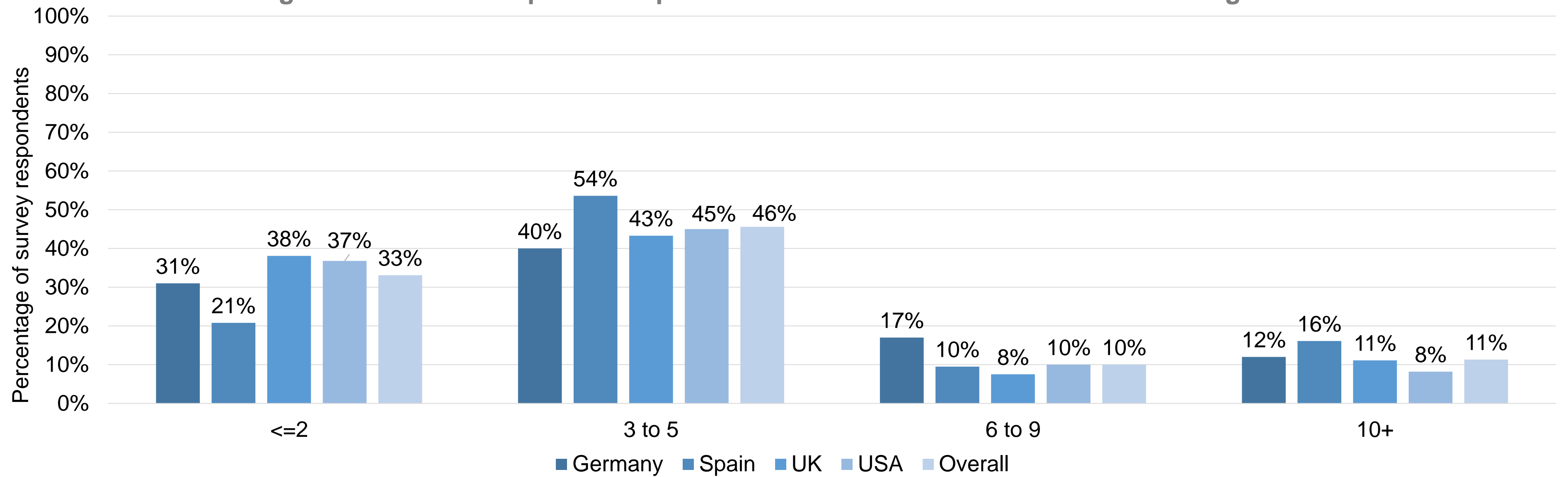


Fig2. reflects the number of specialists patients consulted before receiving a diagnosis

Figure 3. Number of years to receive a confirmed diagnosis

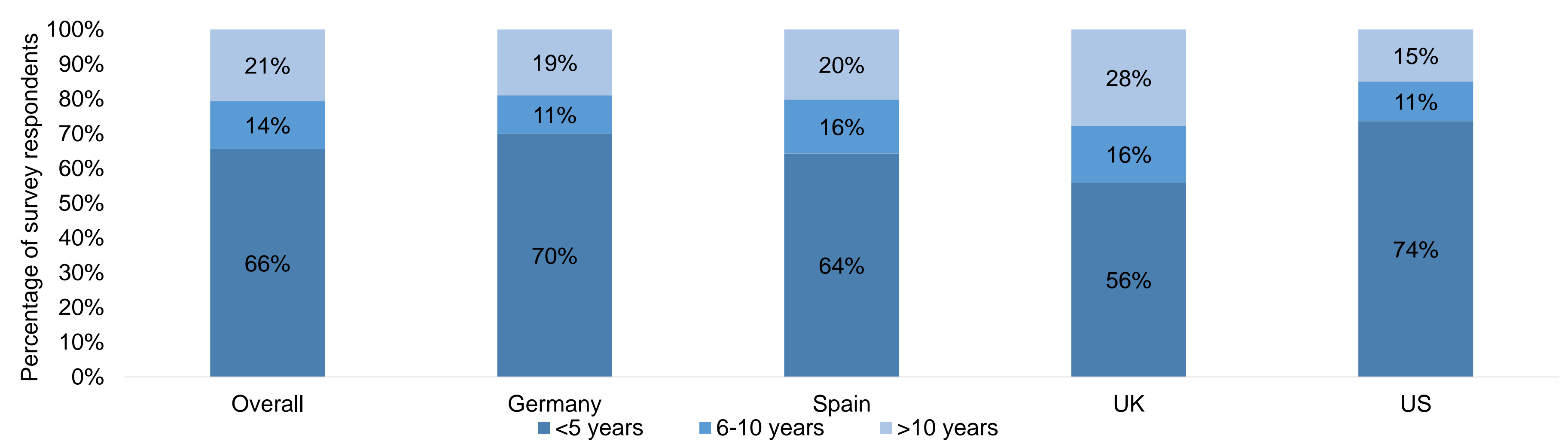


Fig3. reflects the number of years it took for patients to receive a diagnosis

Figure 4. Proportion of Patients denied a diagnostic test

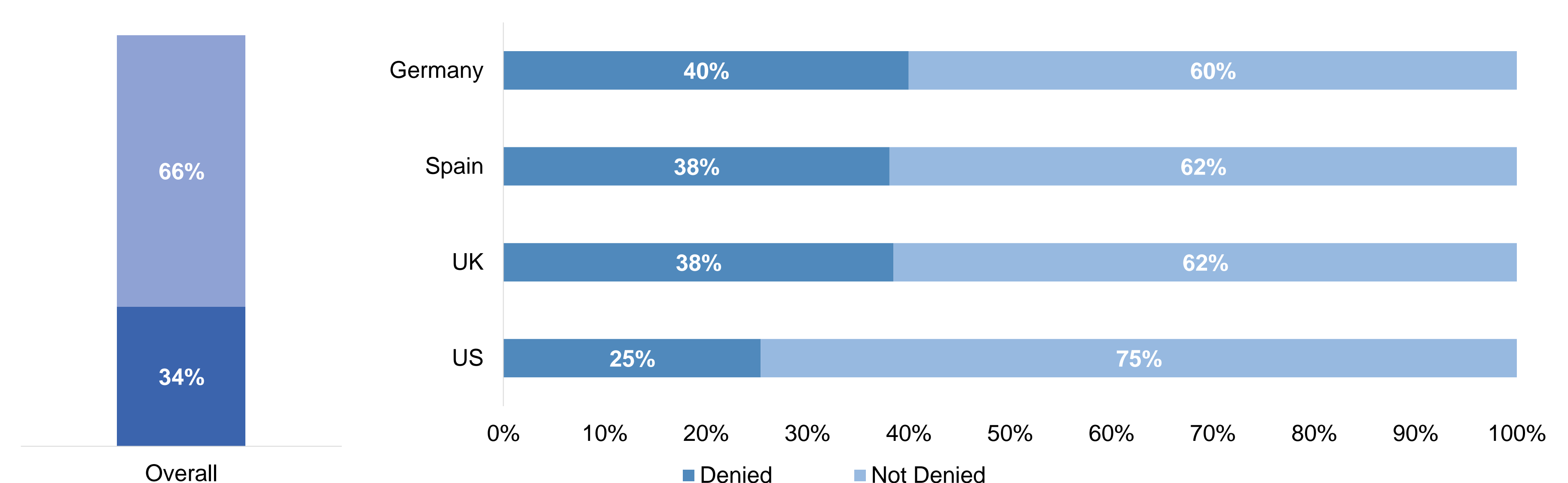


Fig4. reflects the proportions of patients that have been denied diagnostic treatment

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