

# Diagnosis Patterns in Patients With Systemic Mastocytosis in Europe and United Kingdom

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## Introduction

- Systemic mastocytosis (SM) is a rare, clonal mast cell disease primarily driven by the *KIT* D816V mutation and characterized by unpredictable and debilitating skin, gastrointestinal, and systemic symptoms<sup>1,2</sup>
- Previously, a United States (US)-based survey study (Touchstone) reported insights from health care professionals (HCPs) treating SM patients. In that survey, responses from 119 HCPs revealed that SM diagnosis in the US is challenging and often delayed, involving a variety of specialists, notably allergists/immunologist and hematologists/oncologists<sup>2</sup>
- There is limited insight regarding HCP profiles and their perceptions on the diagnosis and management of SM patients in Europe and the United Kingdom (UK)
- The Perceptions Realities and Insights on Systemic Mastocytosis (PRISM) survey study seeks to examine the experiences of SM (advanced and non-advanced [eg, indolent]) patients, as well as gain perspectives from HCPs treating SM in 7 countries in Europe
- Here, we report interim data from PRISM from HCPs in Germany, UK, and Austria and their perspectives on the impact of SM on patients

## Methods

- PRISM was designed by an international steering committee of SM clinical experts and patient advocates
- The study was composed of 2 independent surveys:
  - A Patient Survey (119 questions) on patient diagnostic experiences, symptom burden (via a validated ISM symptom assessment form [ISM-SAF] generating a Total Symptom Score [TSS]), quality of life (QoL) via the 12-item short form survey (SF-12), Euro-QoL five-dimension (EQ-5D), Euro-QoL visual analogue scale (EQ-VAS), and work productivity and activity impairment questionnaire (WPAI)
  - An HCP Survey (103 questions) on approaches to SM diagnosis, care, and management
- Study materials were vetted by independent scientific review and approved by designated ethics committees for the 7 participating countries: France, Switzerland, Spain, Italy, Austria, Germany, and the UK
- From November 2022 through August 2023, patients ≥18 years of age with an SM diagnosis and HCPs who treated patients with SM were recruited via social media, advocacy groups, and word of mouth in the study countries
- Recruited participants provided consent, were enrolled in the study, and completed the relevant survey
- This analysis presents interim HCP results available as of March 2023 for 3 countries (Germany, the UK, and Austria)

## Results

- A total of 296 HCP responses were collected as part of the PRISM survey, from Germany, the UK, and Austria
- HCP respondents were primarily hematologists/oncologists (H/Os, n=87) and general practitioners (GPs, n=87) (Table 1) with the largest number of HCP responders from Germany (n=123) (Table 1)
- The number of SM patients seen by provider type varied across countries (Figure 1); a variety of healthcare specialties diagnose and manage SM patients, likely due to the heterogeneous nature of the disease
- HCP perceptions of time to diagnosis of non-advanced SM averaged around 17.2 months (Figure 2)
- Most HCPs perceived significant SM impact on patient QoL and employment opportunities, with 68.2% (n=203) reporting SM affected patients' lives "quite a bit" or "a great deal"; 59% (n=175) reported patients lost employment opportunities due to SM (Figure 3)
- Across all countries, HCPs reported their top primary goals for treatment were focused on better QoL and improved survival (Figure 4)

## PRISM HCP Responses

Table 1. HCP Demographics and Office Setting

|                         | All (n=296) | Germany (n=123) | UK (n=110) | Austria (n=63) |
|-------------------------|-------------|-----------------|------------|----------------|
| <b>Specialty</b>        |             |                 |            |                |
| General Practitioner    | 87 (29.4)   | 33 (26.8)       | 41 (37.3)  | 13 (20.6)      |
| Hematologist/Oncologist | 87 (29.4)   | 31 (25.2)       | 42 (38.2)  | 14 (22.2)      |
| Dermatologist           | 35 (11.8)   | 17 (13.8)       | 9 (8.2)    | 9 (14.3)       |
| Allergist/Immunologist  | 28 (9.5)    | 14 (11.4)       | 11 (10.0)  | 3 (4.8)        |
| Gastroenterologist      | 23 (7.8)    | 15 (12.2)       | 3 (2.7)    | 5 (7.9)        |
| Internist               | 18 (6.1)    | 7 (5.7)         | 0 (0)      | 11 (17.5)      |
| Other                   | 18 (6.1)    | 6 (4.9)         | 4 (3.6)    | 8 (12.7)       |
| <b>Office Setting</b>   |             |                 |            |                |
| Academic Hospital       | 126 (42.6)  | 39 (31.7)       | 56 (50.9)  | 31 (49.2)      |
| Community Hospital      | 48 (16.2)   | 10 (8.1)        | 27 (24.5)  | 11 (17.5)      |
| Solo Private Practice   | 103 (34.8)  | 65 (52.8)       | 18 (16.4)  | 20 (31.7)      |
| Other                   | 19 (6.4)    | 9 (7.3)         | 9 (8.2)    | 1 (1.6)        |

Figure 1. Mean Number of Patients With SM Seen by HCP Type Across Countries

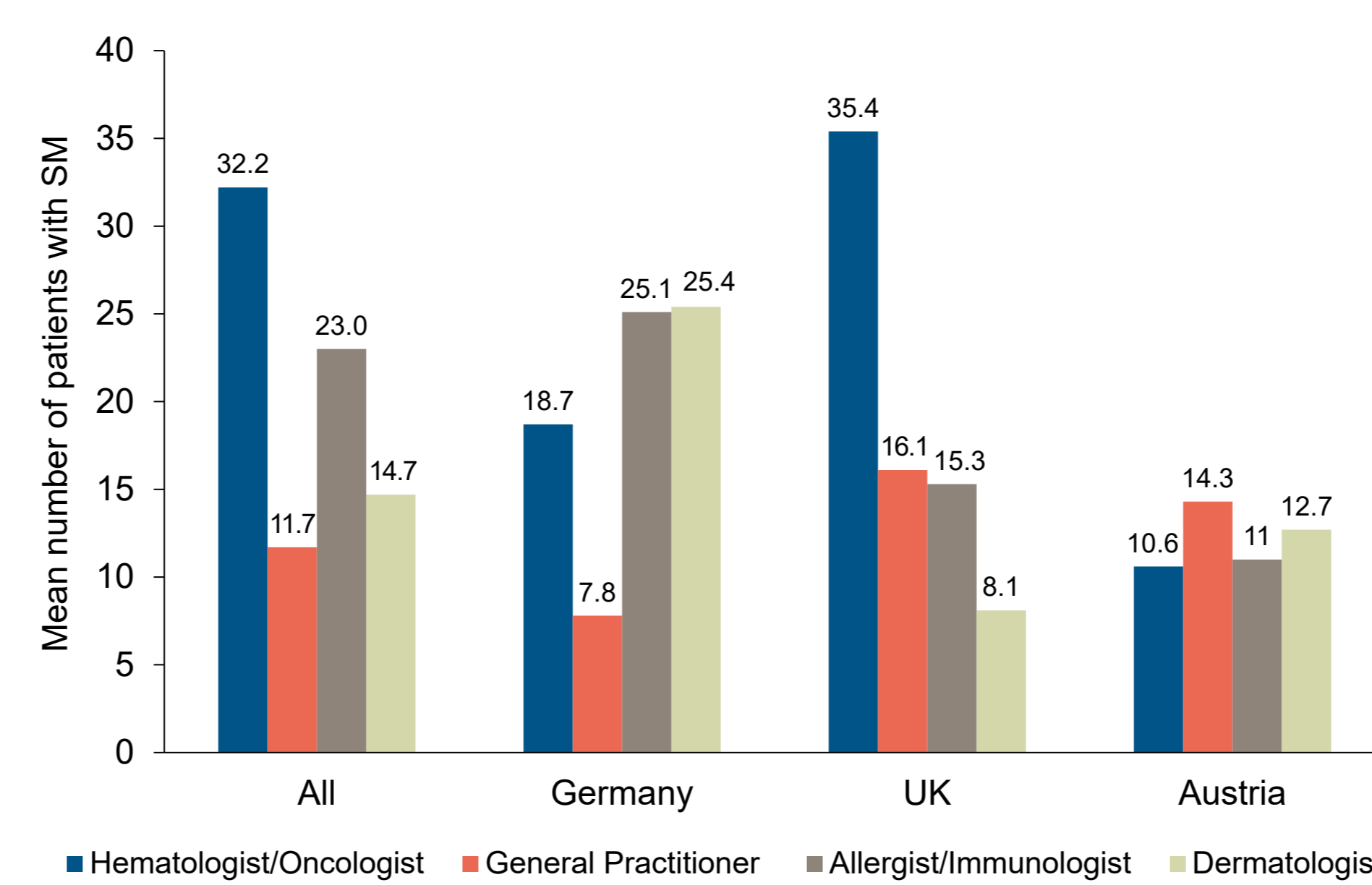


Figure 2. HCP-Perceived Time From Patient Non-Advanced SM Symptom Onset to Diagnosis by HCP Type Across Countries

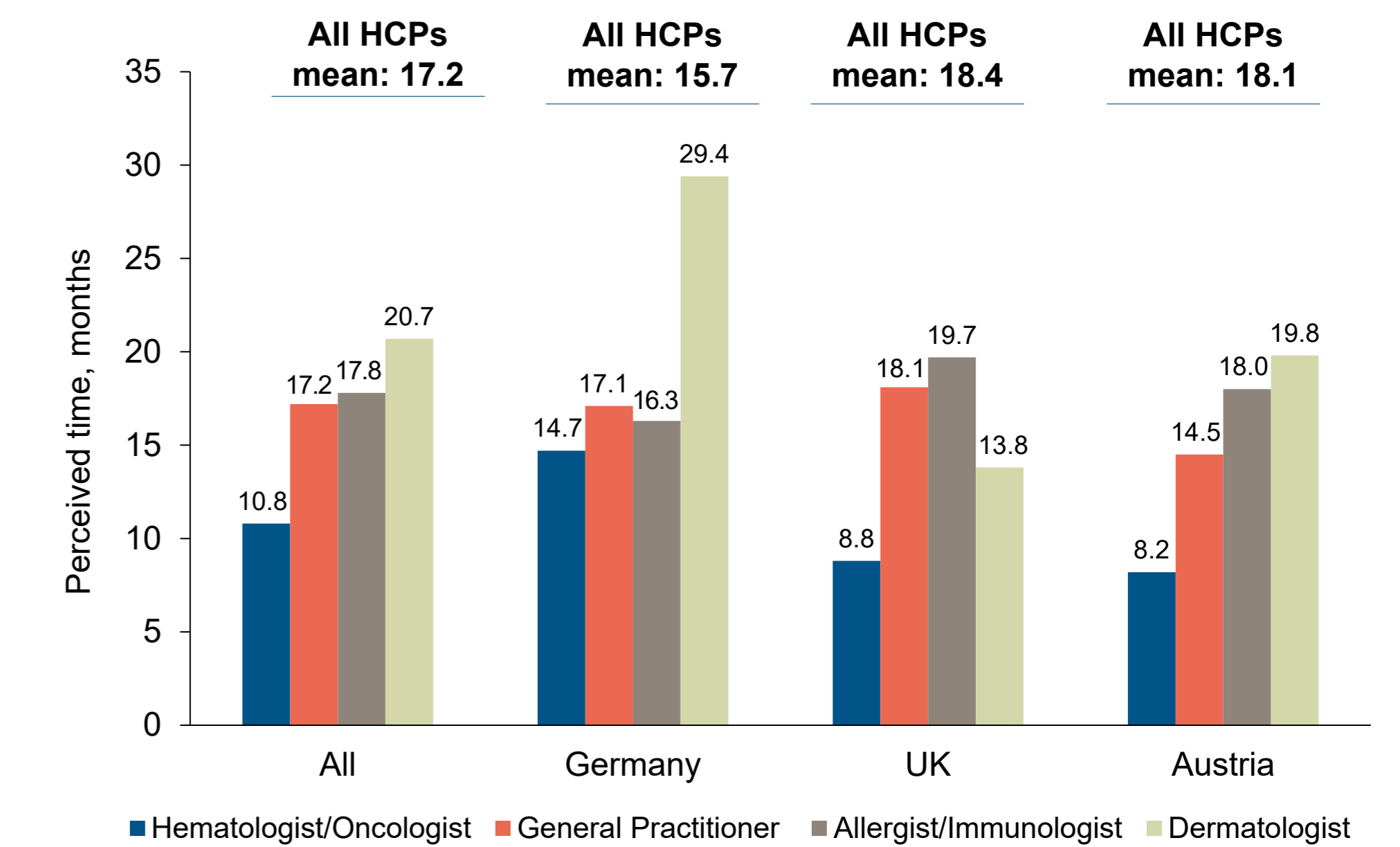


Figure 3. Most HCPs Perceive SM to Have a Large Impact on Patient's Lives and Employment Opportunities

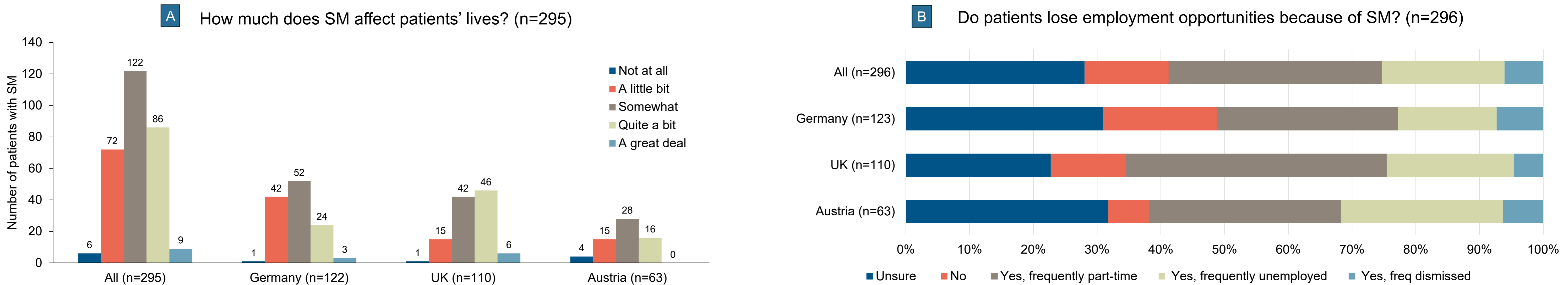
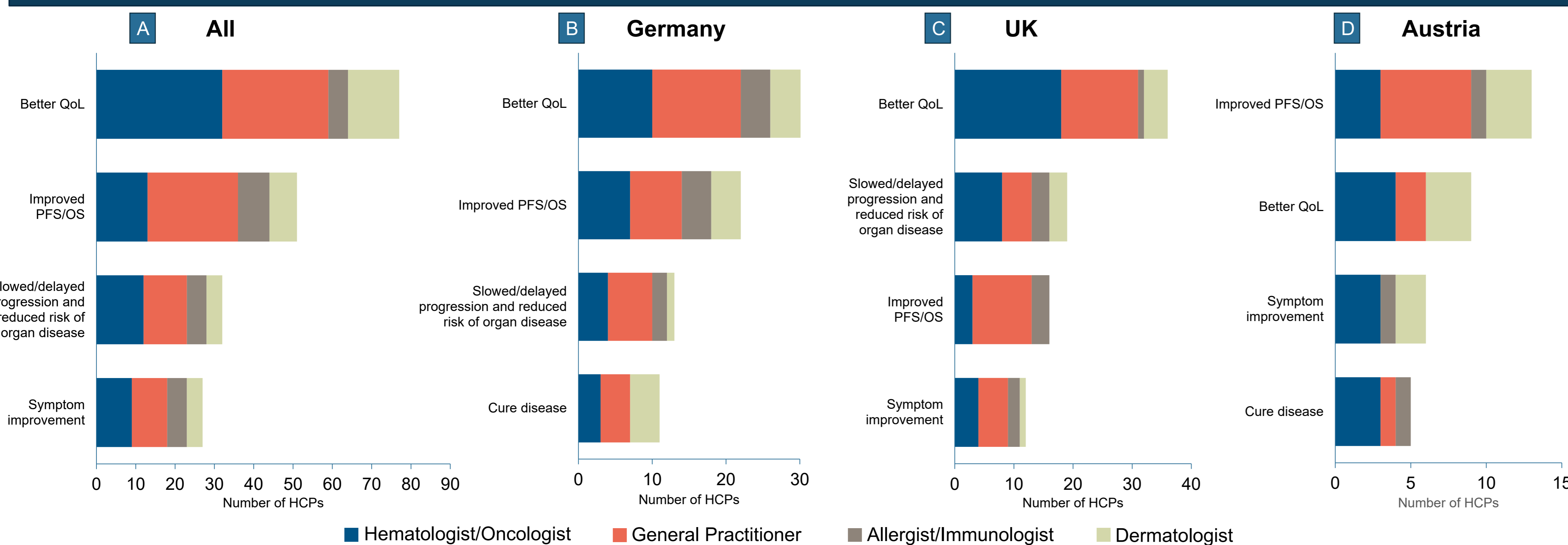


Figure 4. HCPs Report Their Primary Goals for Treatment Are to Improve Quality of Life (QoL) and Survival



Abbreviations: OS, overall survival; PFS, progression-free survival; QoL, quality of life.

- References  
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## Disclosures

Study sponsored by Blueprint Medicines. TG, DP, and CGS are employees of Blueprint Medicines and own stock in Blueprint Medicines.

## Conclusions

This interim data set from PRISM HCP responders in Germany, the UK, and Austria showed that

- A variety of healthcare specialties diagnose and manage SM patients due to the heterogeneous nature of the disease
- HCP perception of time to diagnosis varied by specialty but overall was considerably shorter than that reported by SM patients in a previous US-based survey (Touchstone)<sup>3</sup>
- HCPs perceive SM to have a considerable negative impact on patients' QoL and ability to work
- Improving QoL and survival were the primary treatment goals of HCPs for their patients