"*If I have Sjögren's syndrome, I want to know it as early as possible*": The perspective of first-degree relatives of patients with Sjögren's syndrome from an international survey

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First-degree relatives of patients with Sjögren's syndrome (SS) have a higher risk of developing SS and other autoimmune diseases (AD) [1–3]. The Pre-Sjögren's Syndrome Targeted Immunology Evaluation (PreSStige) study is the first preclinical study in SS and it has been designed to perform a clinical and immunological characterization of subjects at risk for developing SS, including FDRs [4]. An online survey was co-designed in English and translated into 14 languages by rheumatologists and patient research partners to explore the perceived risk to develop SS or another AD and the attitude on health-related decisions of SS FDRs. The survey was distributed in 2023 via patient associations, forums and support groups (Appendix A). FDRs without a diagnosis of SS/other ADs were confronted with the hypothetical scenario of being offered a rheumatology consultation free of charge and if during the consultation SS "red flags" were identified, they would be advised to undergo additional exams (within a variable range of costs). Data were analysed using IBM-SPSS 28.0 software. The PreSStige study has been approved by the Ethics Committee of the Medical University of Graz (Austria).

We gathered 1219 valid responses from 41 countries (Fig. S1, Tables S1 and S2). The majority of respondents ($n = 674$, 55%) were parents, 329 (27%) were siblings and 216 (18%) were offspring of SS patients. In total, 254 (21%) and 152 (12%) of respondents already received a diagnosis of SS or another AD respectively (Table S3).

Of the 965 respondents not diagnosed with SS/AD, 79–81% disagreed or felt neutral about the statement "*Since my parent/child/sibling has SS, I will certainly develop it or another AD*" (Fig. 1). In line with this, 62% of respondents would like to know their risk of developing SS/other ADs whereas 36% of respondents would rather avoid thinking about SS and their possible risk to develop it and mentioned being worried (39%), sad (12%) or scared (9%) about the possibility of developing SS in the future.

When confronted with the hypothetical scenario, 88% ($n = 715$) respondents would accept a rheumatology consultation free of charge with the main reasons being: "*If I have SS/other AD, I want to know it as early as possible*" (84%) (Fig. 2). Of these, 91% would perform additional exams if advised by the rheumatologist based on the detection of SS red flags. Those who accepted the free rheumatology consultation but would not proceed further mentioned the economic aspect (namely pay for the additional exams) as the main barrier.

Lack of symptoms (74%) and lack of time (16%) were the two main reasons to decline a free rheumatology consultation.

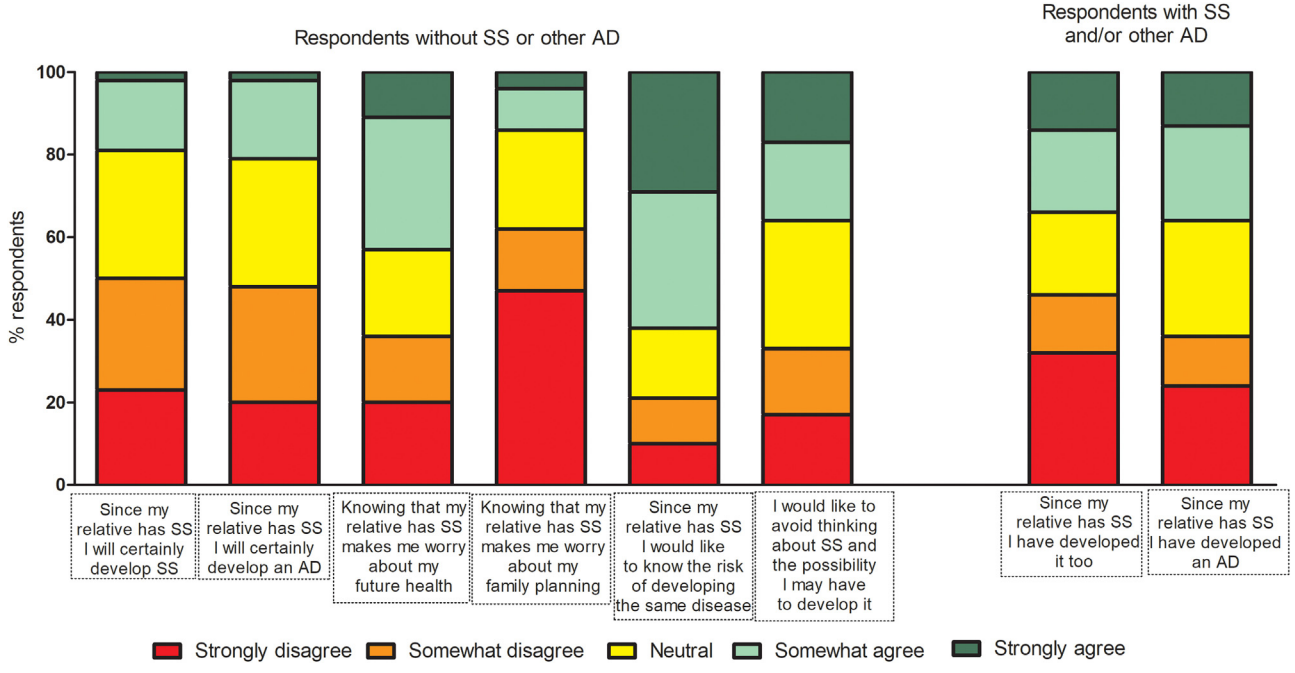

Fig. 1. Agreement with statements indicating the perceived risk and beliefs of respondents with ($n = 406$) or without ($n = 813$) a diagnosis of Sjögren's syndrome (SS) and/or other autoimmune disease (AD).

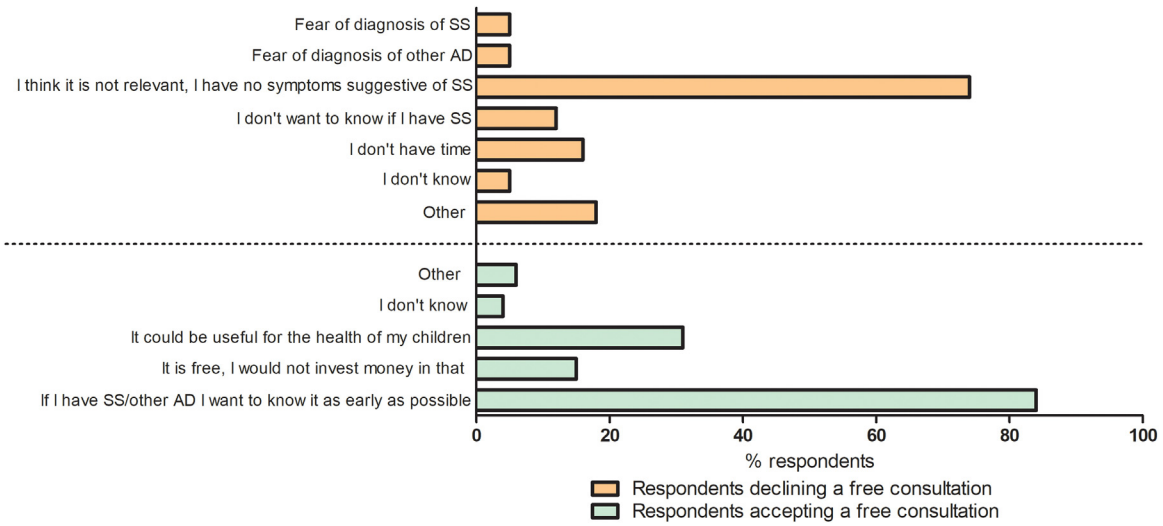

Fig. 2. Reasons for accepting or declining a free rheumatology consultation from respondents without a diagnosis of Sjögren's syndrome (SS) and/or other autoimmune disease (AD) ($n = 813$).

This is the first study focused on SS FDRs from Europe and beyond aimed to set the stage for large scale preclinical research in SS at-risk individuals. Although the majority of respondents would like to know their risk of developing SS and to undergo rheumatology consultations/additional exams to achieve an early diagnosis of SS, our results point out that before proposing to a SS FDR the enrolment into a preclinical study, researchers should carefully explore the emotional attitude towards the disease and explain how epidemiological data on large cohorts can be transferred into rheumatology daily practice. The recurrence of "lack of symptoms" and "lack of time" as arguments to decline a free rheumatology consultation highlights that education on the rationale of preclinical research may be needed also to prompt individuals to prioritize health in today's fast-paced world.

Our study has some limitations such as the survey-based nature and a possible selection bias, however it is the first of its kind and provides the perspective of a large and geographically spread group of SS FDRs. In conclusion, our results support that recruitment of SS FDRs in preclinical studies across Europe and beyond is feasible and identified potential barriers to be overcome to facilitate study enrolment and prevent discomfort or economic discrimination among eligible subjects.

Disclosure of interest

The authors declare that they have no competing interest.

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Data availability statement

All relevant data are included in the main manuscript and supplementary material.

Author contribution

All authors contributed and approved the final version of the manuscript.

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Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.jbspin.2024.105695.

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