The Five Dimensions of the ISM Patient Experience – Uncovering the "Real-world" **Experience of Patients with Indolent Systemic Mastocytosis**

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Background

- Indolent systemic mastocytosis (ISM) is a clonal mast cell disease driven by the *KIT* D816V mutation in ~95% of adult cases^{1–3}
- Patients with ISM can have lifelong symptoms, which can be debilitating, across multiple organ systems^{4–8}
- Most patients rely on polypharmacy for the management of symptoms with best supportive care (BSC) medications, such as H1/H2 antihistamines and mast cell stabilizers, although in many patients, symptoms are not adequately controlled with BSC medications^{8–9}
- Living with ISM often substantially affects various facets of a patient's life, including medical, emotional, social, and practical domains, thereby negatively impacting their quality of life (QoL). Challenges such as misinformation, gaps in knowledge, and the inherent complexity of this condition can contribute to delays in diagnosis¹⁰
- Collaboration and clear communication between healthcare professionals and patients is key in addressing these issues and improving patient outcomes¹⁰
- To ensure optimal care, it is crucial to have a better understanding of the unmet needs and challenges faced by patients with ISM
- An in-depth qualitative approach to survey-based research has been used to further understand the lived experience of patients with ISM

Figure 1. Patient and caregiver demographics

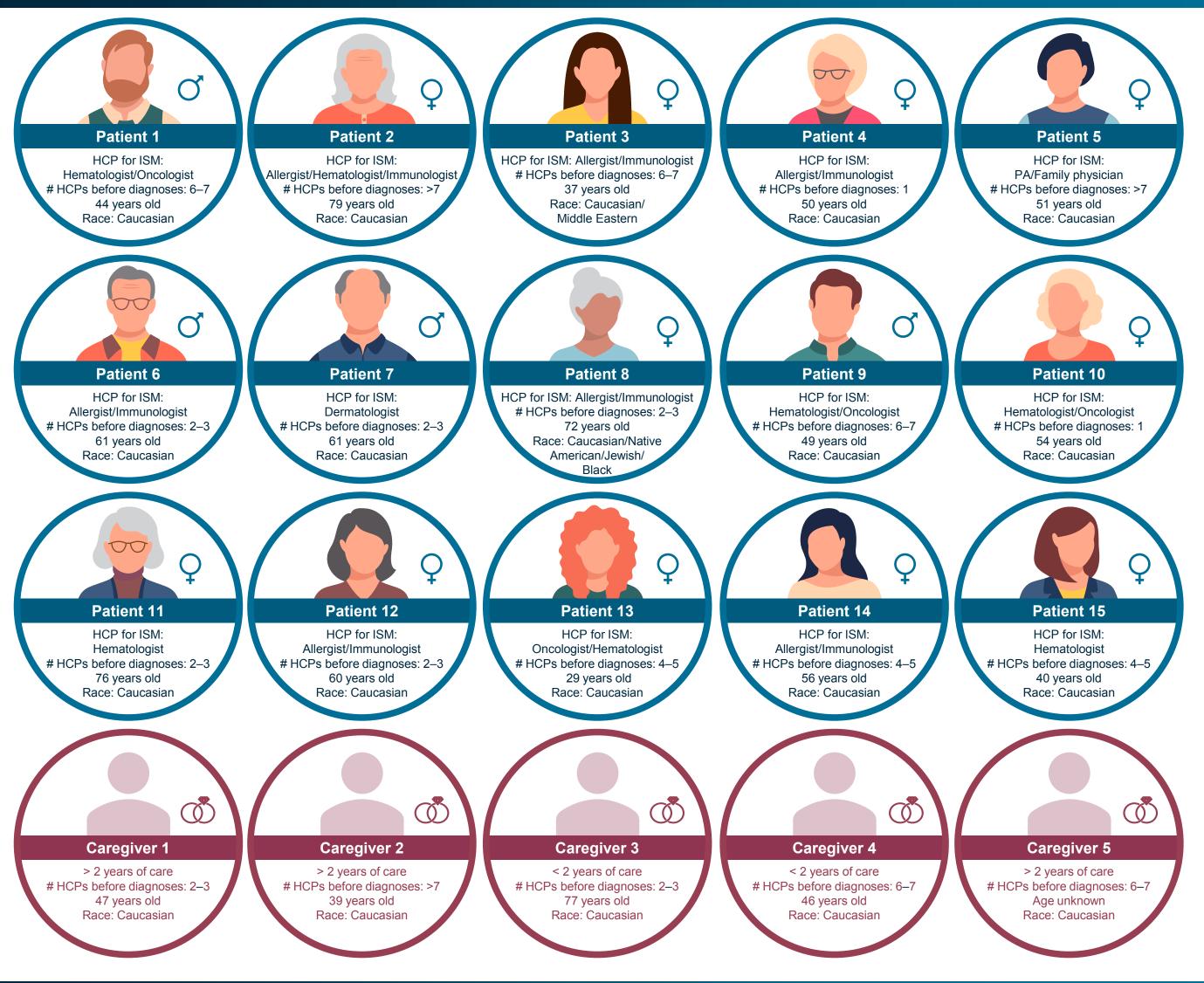
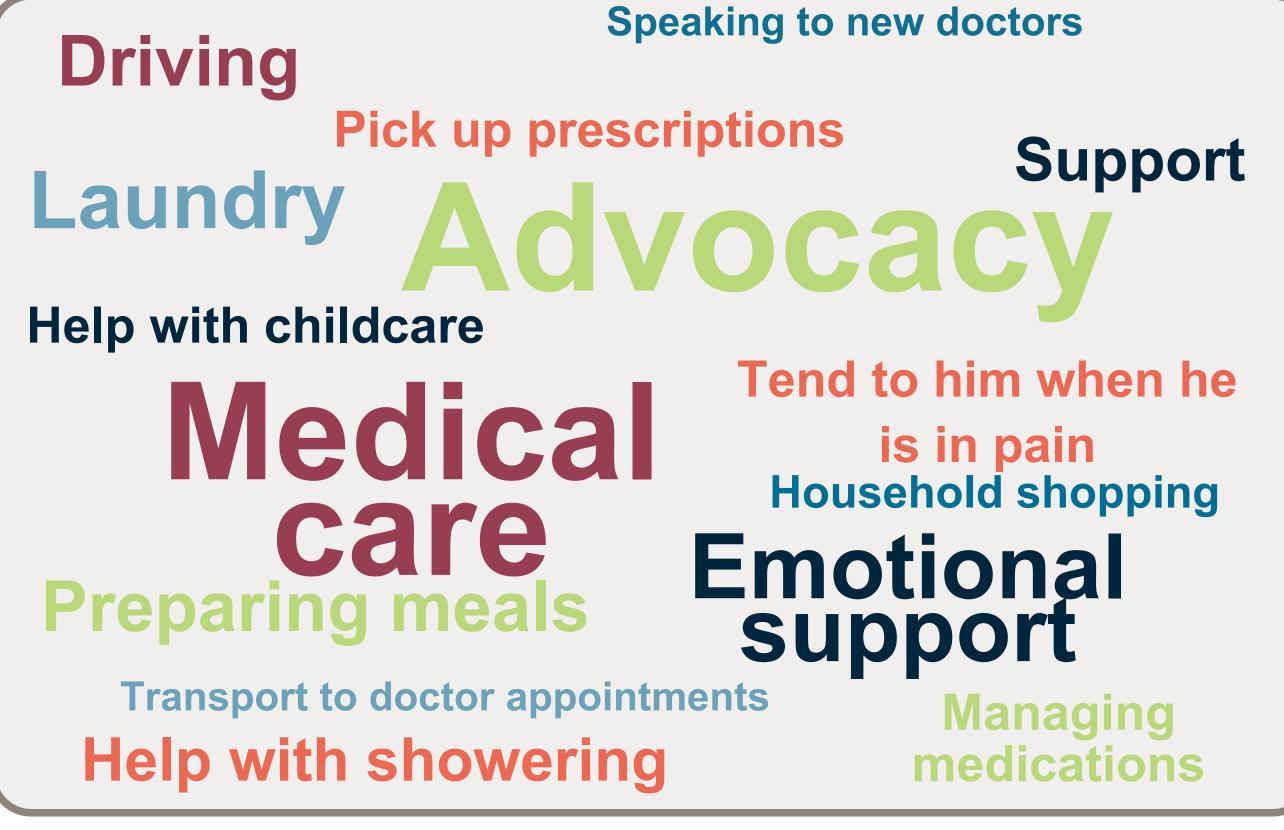
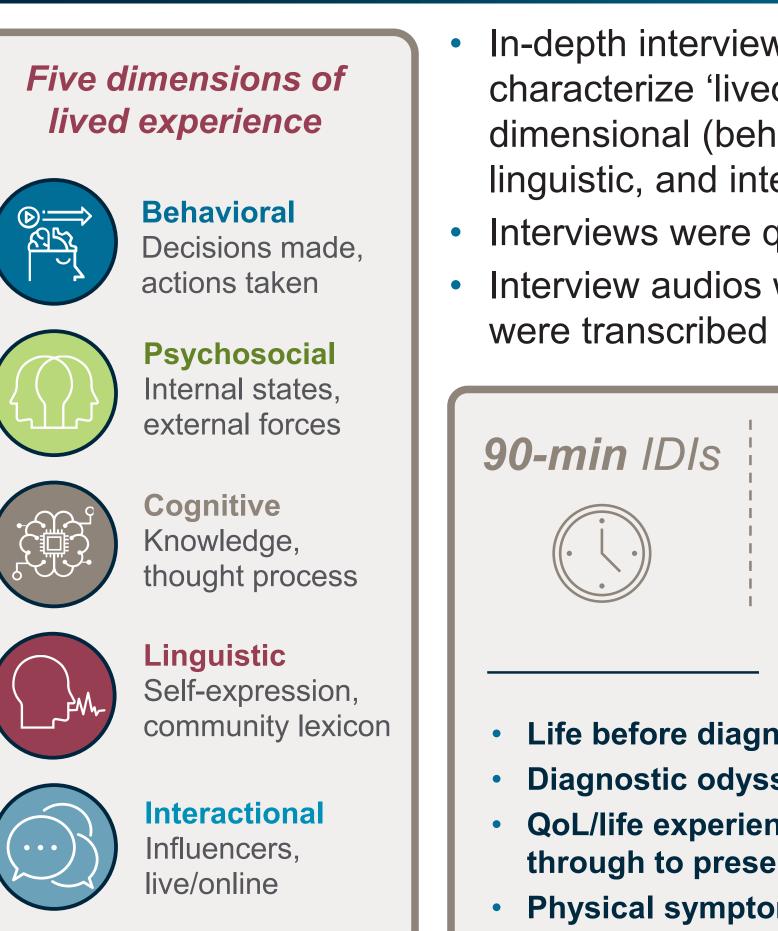


Figure 2. Types of support provided by caregivers



ISM, indolent systemic mastocytosis; HCP, health care provider; PA, physician associates.

Figure 3. Methods



^aParticipants were unaware of the involvement of Blueprint Medicines in the survey, and Blueprint Medicines did not know the identity of the participants. IDI, in-depth interview; QoL quality of life.

Figure 4. Prevalence of patient-reported symptoms (%)

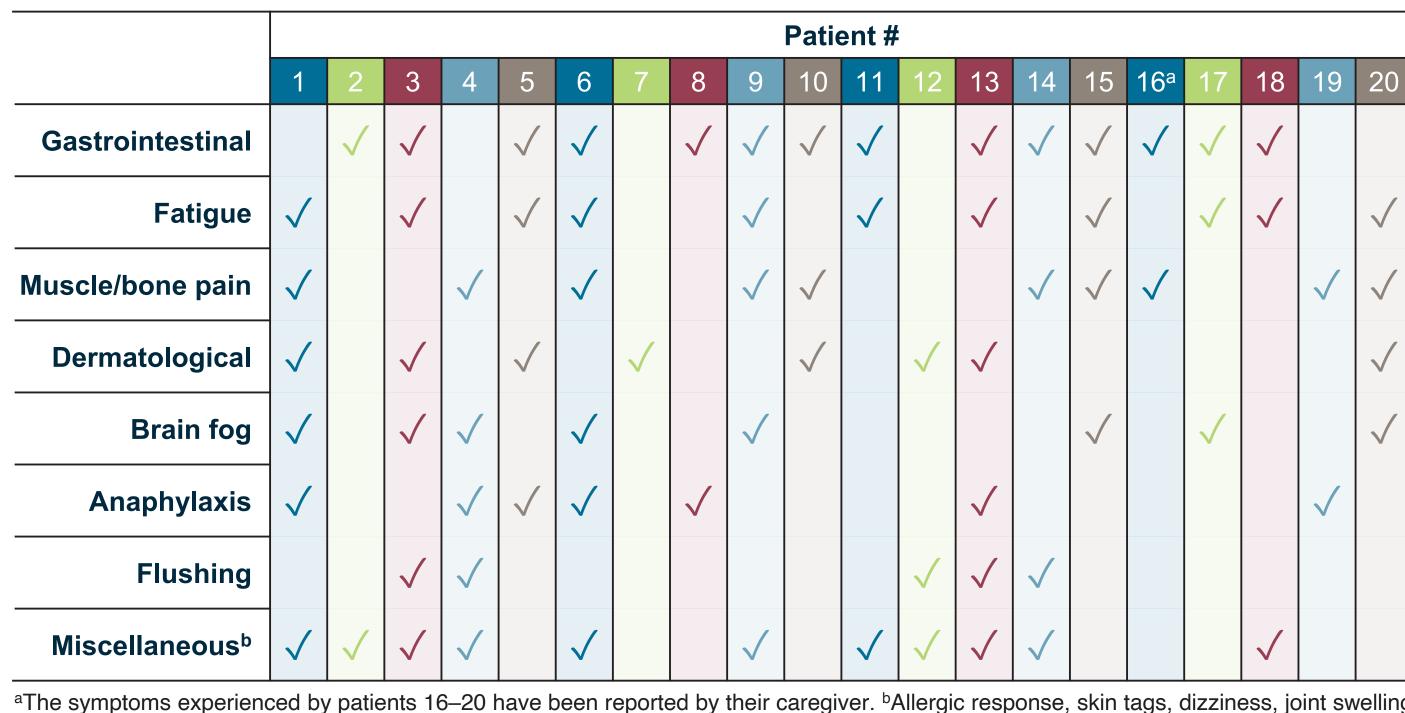
- Gastrointestinal (GI) symptoms were the most common, with 70% of patients reporting symptoms
- GI symptoms, fatigue, muscle and bone pain, and dermatological symptoms were experienced by over half of the patients

Gastrointestinal						70%
Fatigue						60%
Muscle/bone pain						60%
Dermatological						55%
Brain fog						45%
Anaphylaxis						35%
Flushing						25%
Miscellaneous ^a						45%

^aAllergic response, skin tags, dizziness, joint swelling, inflammation, rapid heartbeat, burning tongue, difficulty breathing, sweating.

Figure 5. Combination of symptoms reported

experiencing as many as six different symptoms



In-depth interviews (IDIs) were conducted to characterize 'lived' experience through a fivedimensional (behavioral, psychosocial, cognitive, linguistic, and interactional) analysis tool Interviews were qualitive, double blind,^a and 1:1 Interview audios were recorded, and summaries



IDI topics

- Life before diagnosis
- Diagnostic odyssey
- QoL/life experience from inception of symptoms through to present
- Physical symptoms psychological consequences

Many patients reported experiencing a combination of symptoms, with some

inflammation, rapid heartbeat, burning tongue, difficulty breathing, sweating.

Figure 6: Eleven core insights

- IDIs with patients and caregivers resulted in eleven core insights relating to one or more of the dimensions of lived experience (Figure 3)
- Patients experienced a spectrum of time to diagnosis ranging from one to two years up to more than ten years
- Patients reported a range in the number of HCPs seen before receiving a diagnosis between one and more than seven HCPs

The search for answers is ongoing after diagnosis: Fear of disease worsening





"My allergist didn't know a lot about it but he just knew that these were some of the symptoms that accompanied systemic mastocytosis...so yeah, a lot of encouragement to research it myself."

Supporting self-advocacy and empowerment for patients

When considering support for patients with ISM and their unmet needs, the efforts for further education must not end at the moment of diagnosis

The quality of physician relationship was not related to the degree of patient health literacy



"I went on the internet and the National Institute **6 6)** of Health website and just a lot of places like that...I gained a lot of knowledge on this myself."

Supporting self-advocacy and empowerment for patients

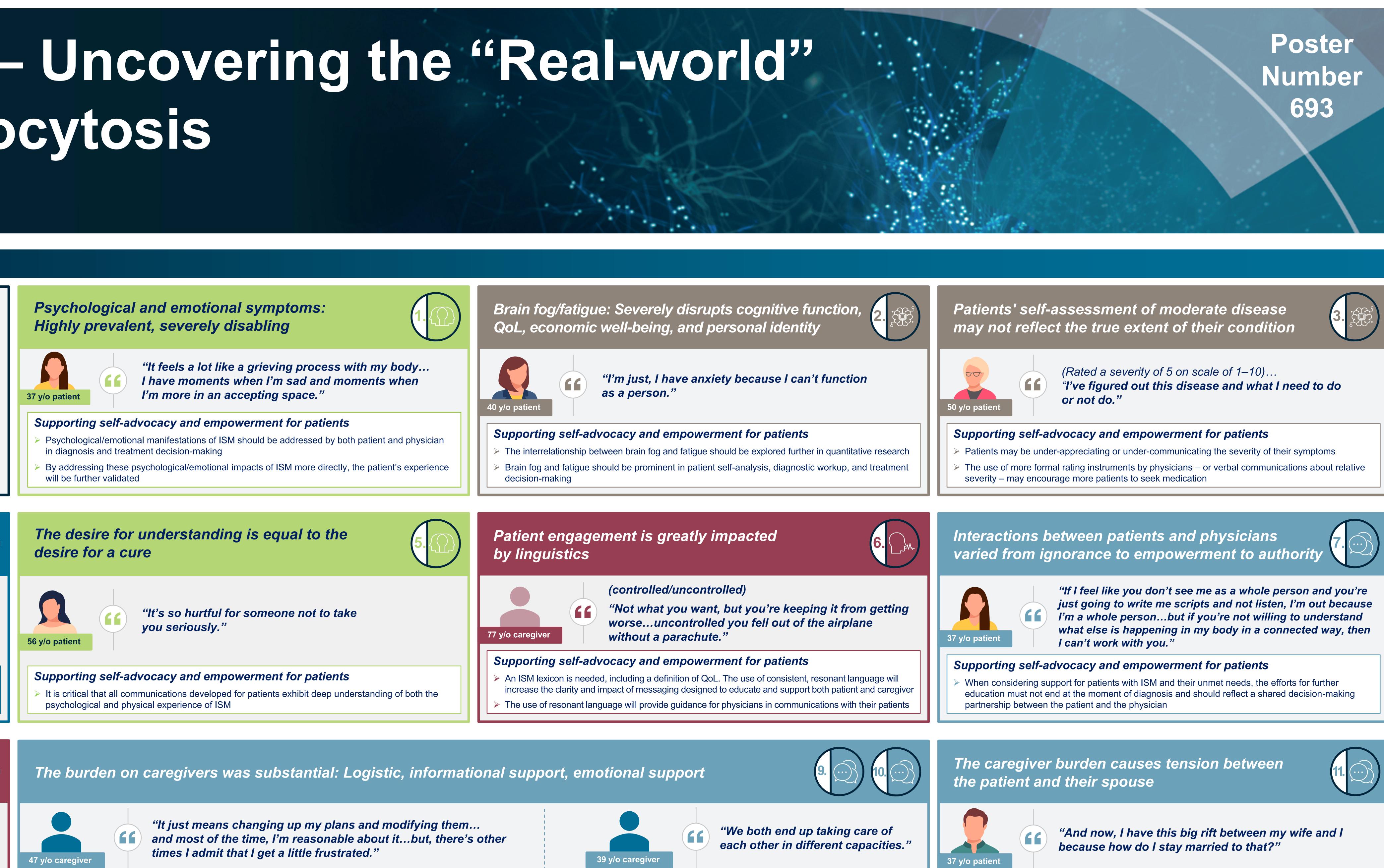
- The lack of association between degree of literacy and patient adherence and outcomes means the a different set of criteria for "literacy" must be created
- A better understanding of which information surrounding ISM should be prioritized is needed

Impact of symptoms and communication gaps

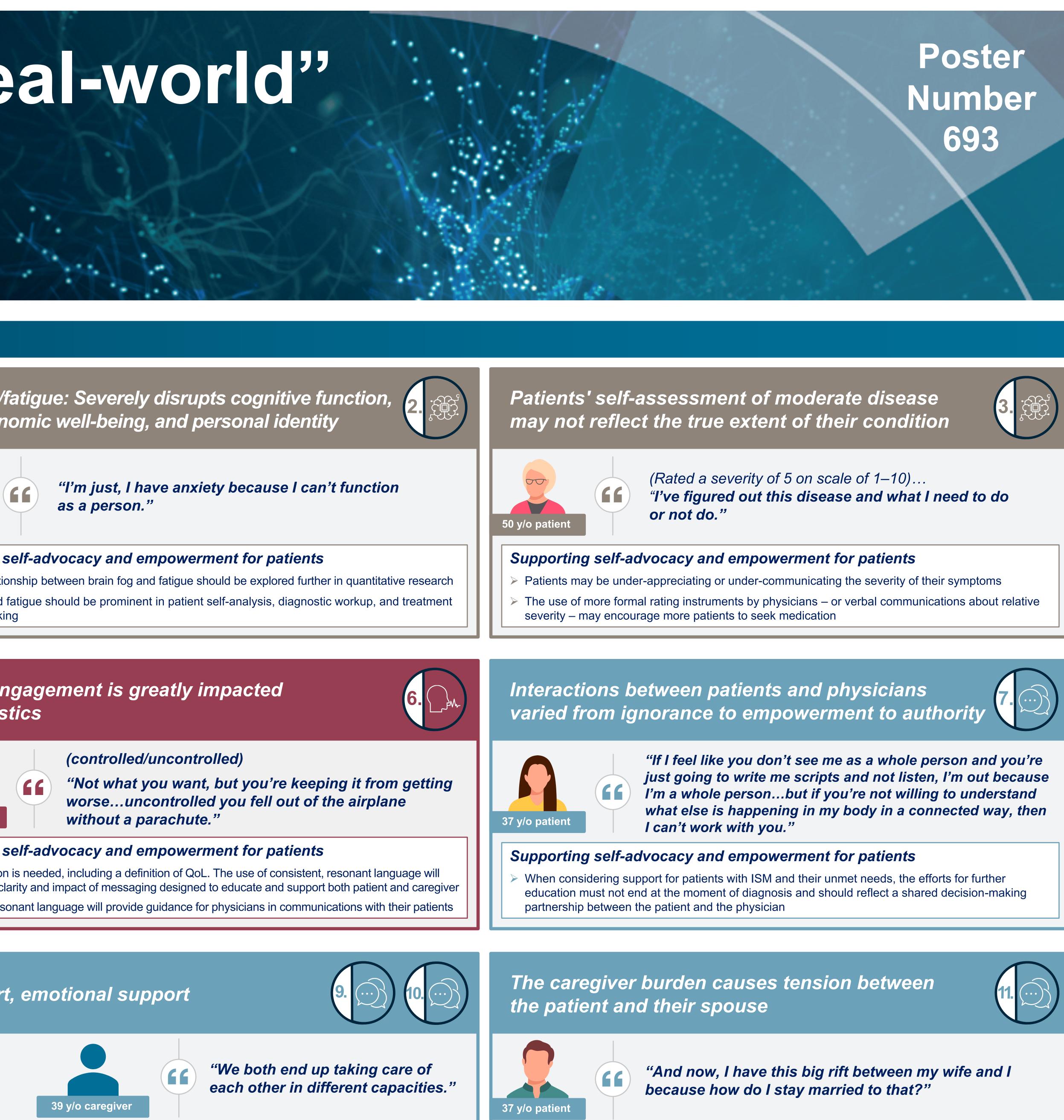
- Psychological and emotional symptoms affect patients' cognitive function, QoL, and economic stability, yet are often underestimated
- There is a notable disconnect in patient–physician communication, leading to challenges in achieving a shared understanding of the full impact of the disease
- Some patients saw up to seven HCPs before receiving a diagnosis, which contributes to poor QoL and causes frustration and confusion

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Supporting self-advocacy and empowerment for patients

- Logistical challenges can impact the caregiver, as their QoL is limited along with the patient's
- > As the patient's symptoms improve (sensitivity to triggers, brain fog, fatigue, and emotional consequences of the condition), the caregiver's QoL will improve proportionately Supporting the caregiver will require substantial and sustained investment of resources beyond merely educational efforts

Conclusions

continue to face uncertainties and fears about their condition

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invisible nature of their illness

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Psychological burdens are caused by both the caregiver's stress of changing lifestyle and the burdens of supporting a patient whose negative emotions can create tension and stress in the relationship

Supporting self-advocacy and empowerment for patients

Support for caregivers must include emotional and practical strategies

Patient empowerment and the need for understanding

Caregiver burden

- Patients actively seek information and take charge of their health journey, but Caregivers of patients face substantial logistical and emotional challenges, impacting their relationship with the patient and adding to the overall burden of the illness • They desire greater empathy and understanding, particularly due to the
 - ISM as a rare chronic, and debilitating disease has an impact beyond the patient

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