HSD14

X-Linked Retinitis Pigmentosa Impacts Patients' Independence, Work Status, and Quality of Life: Insights From the Cross-Sectional EXPLORE XLRP-1.2 Physician Survey

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INTRODUCTION

X-linked retinitis pigmentosa (XLRP) is a rare, inherited eye disease causing progressive loss of photoreceptors.[1,2] XLRP is among the most aggressive forms of retinitis pigmentosa and patients develop legal blindness at a median age of 45 years.[3]

There is currently no effective treatment for XLRP. Compared with unaffected individuals, patients with XLRP may experience significant challenges in their daily lives, including psychosocial and emotional burdens, barriers to work and career progression, as well as lost productivity and increased healthcare costs.[4]

As potential targeted therapies for XLRP emerge, a more thorough understanding of the disease's impact on patients' quality of life (QoL), work status, and level of independence is needed.

OBJECTIVE

The EXPLORE XLRP MSM survey was conducted to obtain real-world insights into the current standards of clinical practice for XLRP in eight European countries. The objective of this analysis was to understand unmet needs in managing XLRP and to evaluate its impact on patients' independence/autonomy, work status, and QoL.

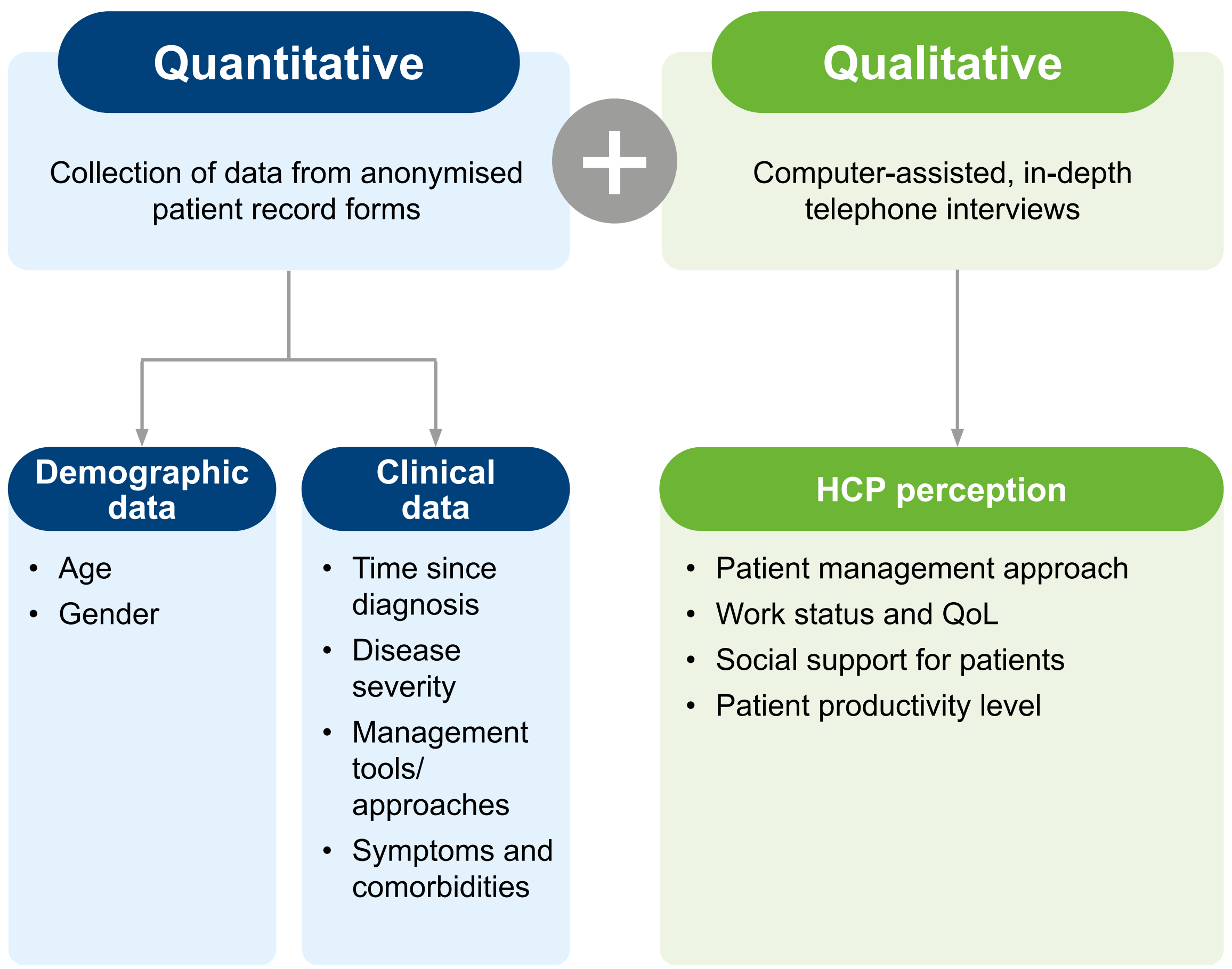
METHODS

EXPLORE XLRP MSM was an exploratory, cross-sectional, physician survey conducted in eight European countries.

Retina specialists/ophthalmologists (n=15) with experience managing XLRP and geneticists (n=3) were interviewed to gain real-world insights on their patients with XLRP (n=47).

Eligible healthcare providers (HCPs) had a minimum of 5 years' experience managing or seeing patients with XLRP and 50% of their professional time was devoted to direct patient care.

The study was conducted in two phases:



Quantitative

Collection of data from anonymised patient record forms

Qualitative

Computer-assisted, in-depth telephone interviews

Demographic data

- Age
- Gender

Clinical data

- Time since diagnosis
- Disease severity
- Management tools/ approaches
- Symptoms and comorbidities

HCP perception

- Patient management approach
- Work status and QoL
- Social support for patients
- Patient productivity level

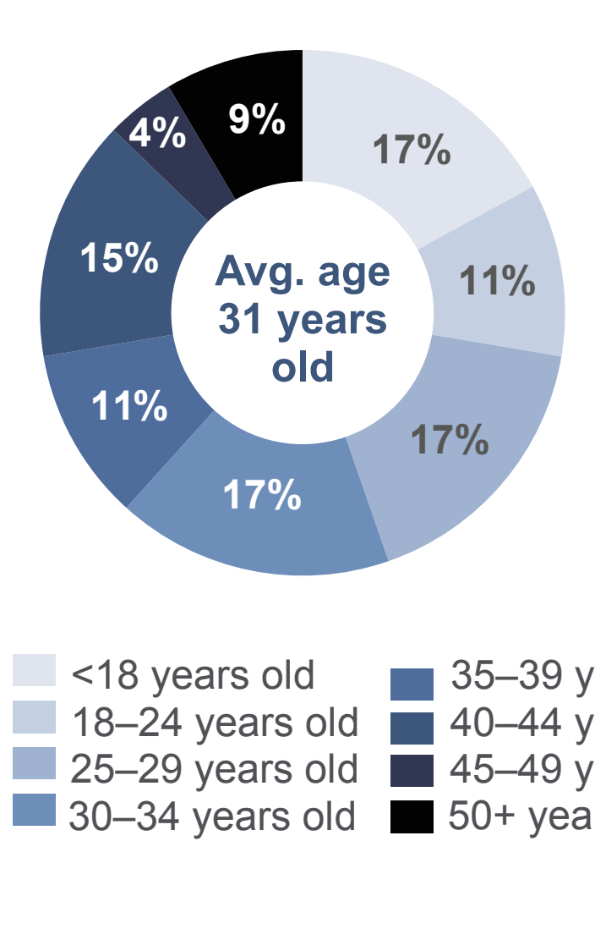
Ethics approval for this study was requested; the Ethics Committee confirmed that this research was out of scope.

RESULTS AND DISCUSSION

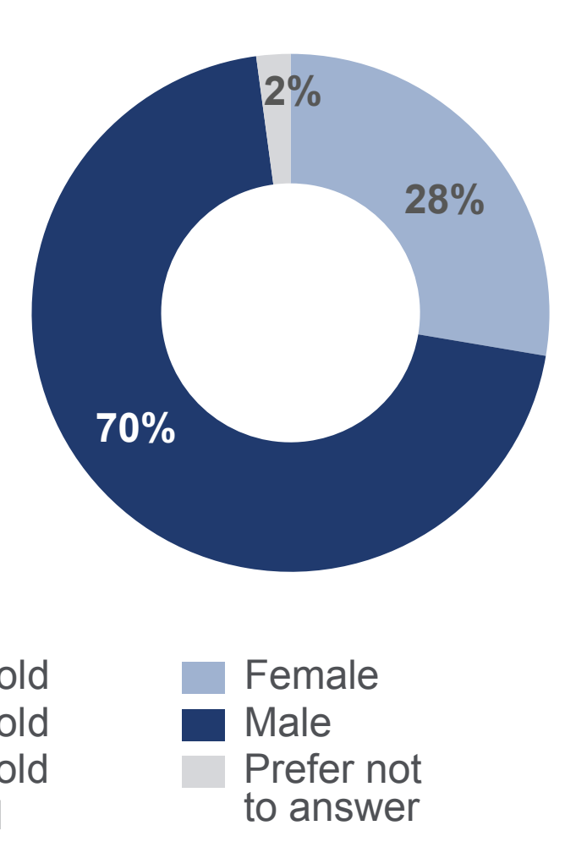
Patient demographics

Most patients with XLRP were male (70%) and aged 18–49 years old (75%); 17% lived alone (53% lived with family).

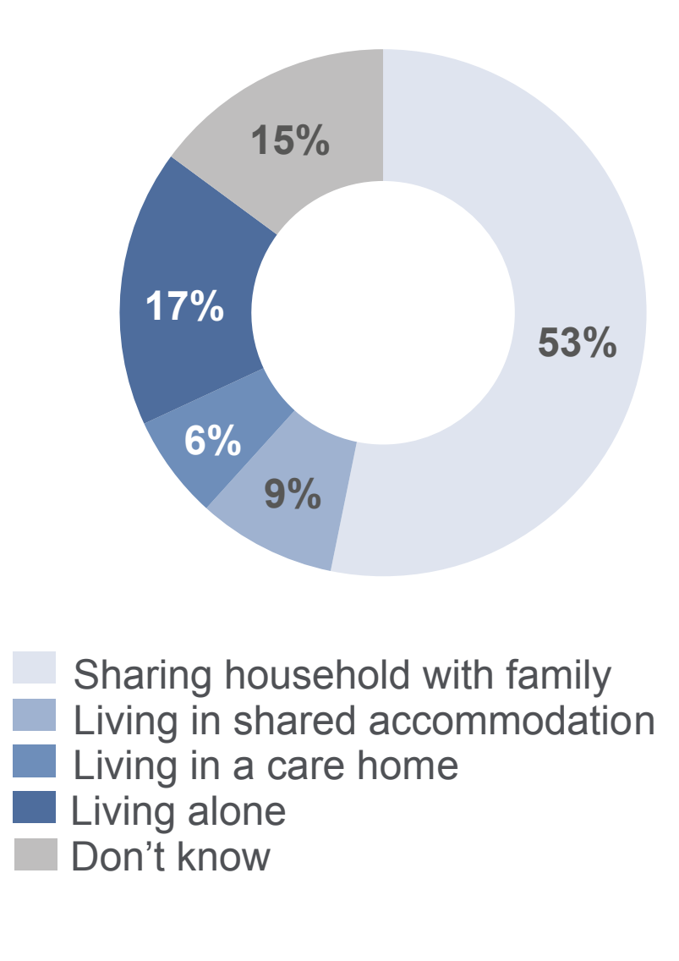
Age



Gender



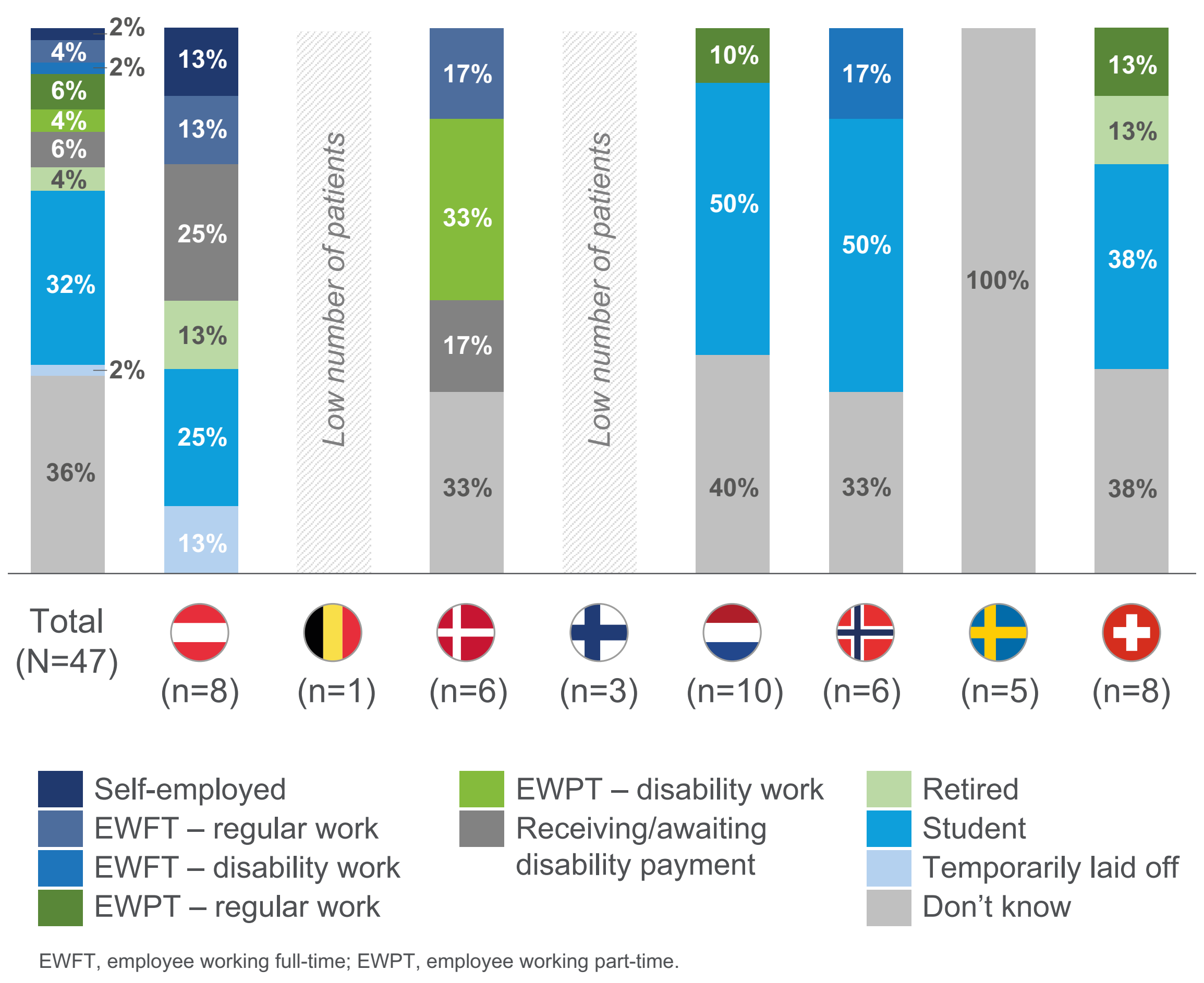
Family status



Patient independence and workforce participation

At the time of the survey, 18% of patients were employed (32% were students; 10% were retired/on disability payment).

Work status per country

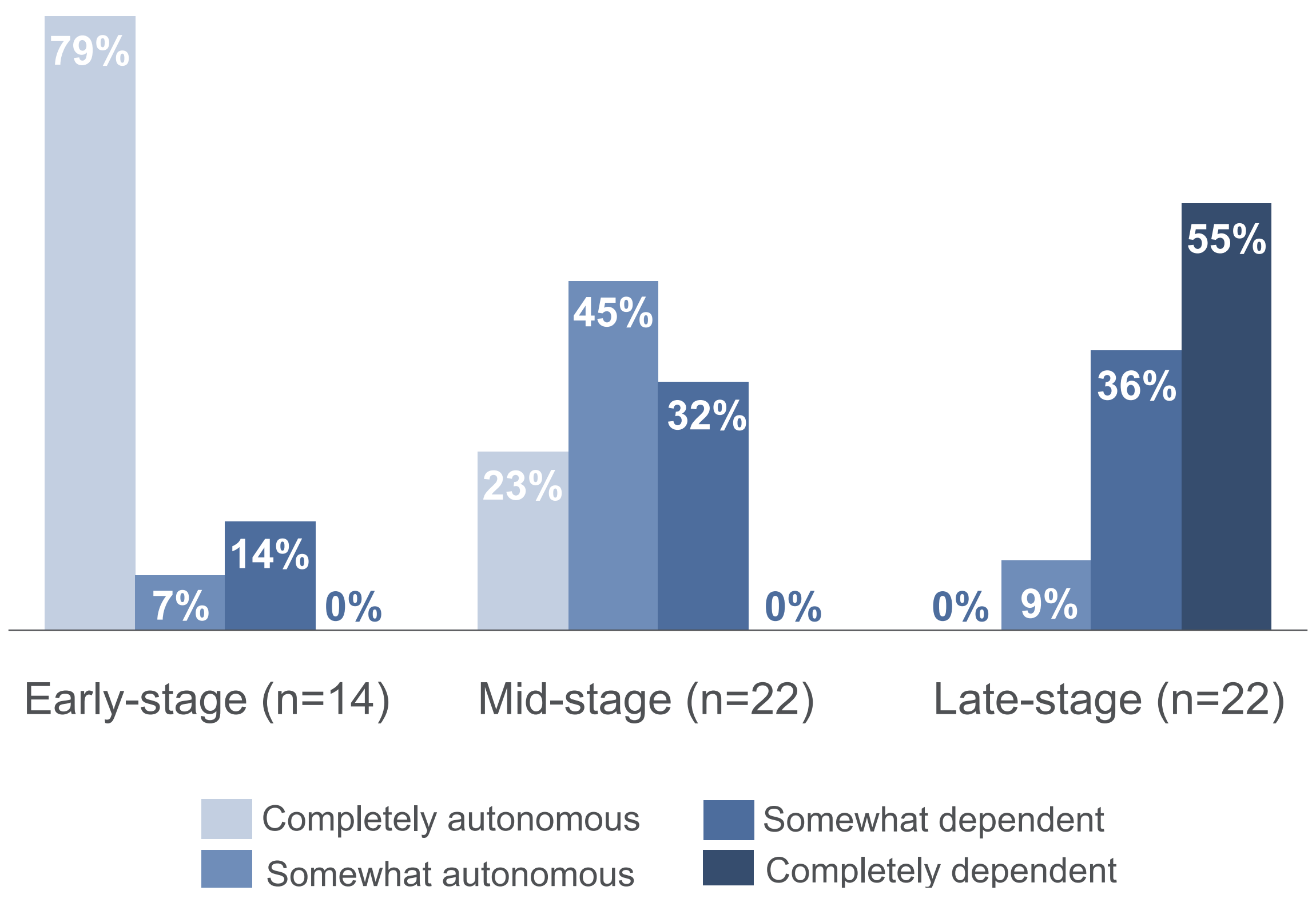


EWFT, employee working full-time; EWPT, employee working part-time.

> It might not be fair, but I always prioritise younger patients who are actively working. They have the biggest need for treatment.
>
> *- Retina specialist, Sweden*

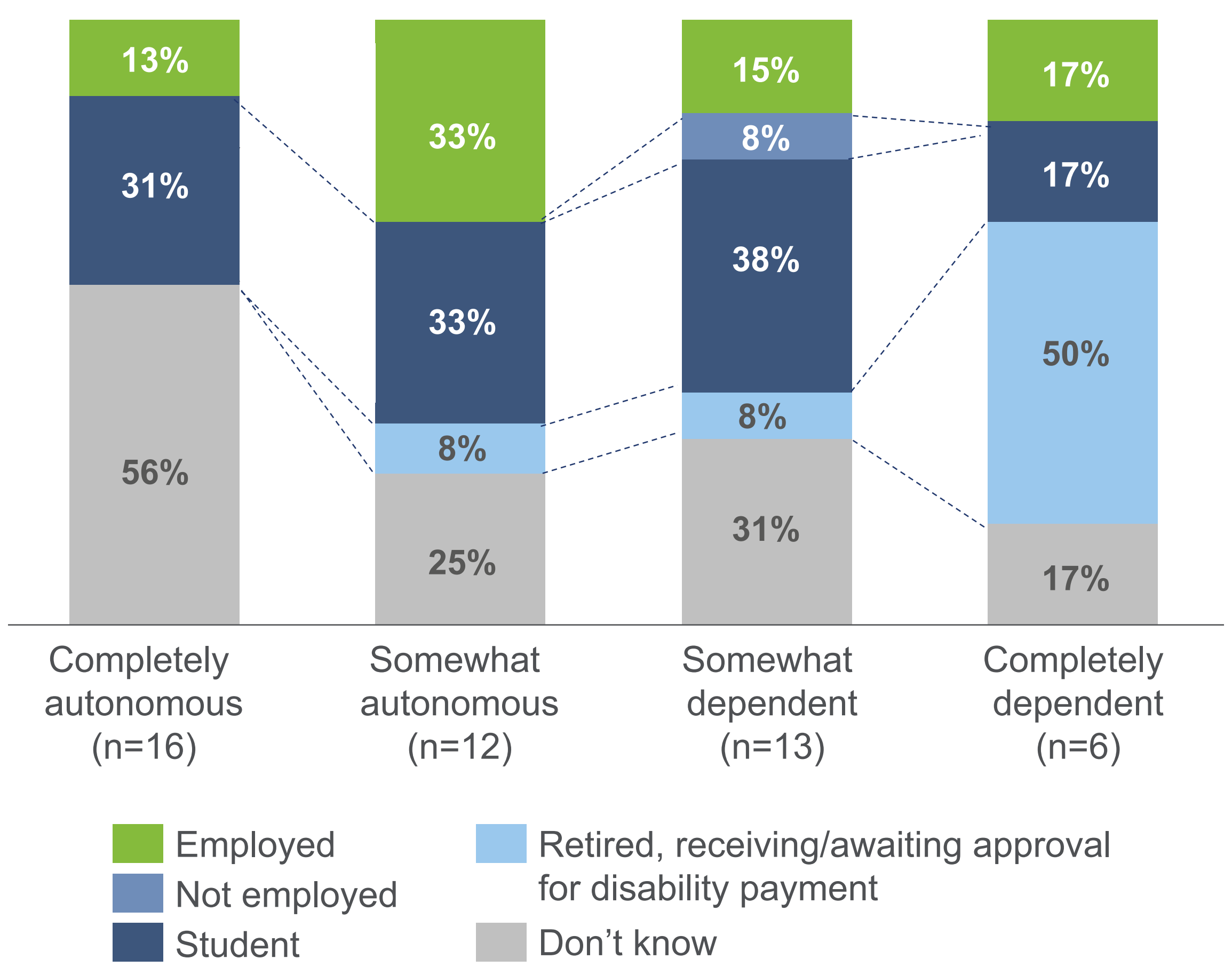
Independence decreased with XLRP progression: 86% of early-stage patients (defined by nyctalopia/night blindness) were "completely or somewhat autonomous", and only 9% of late-stage patients (with central visual impairment/blindness) remained "completely or somewhat autonomous".

Current disease stage and level of independence



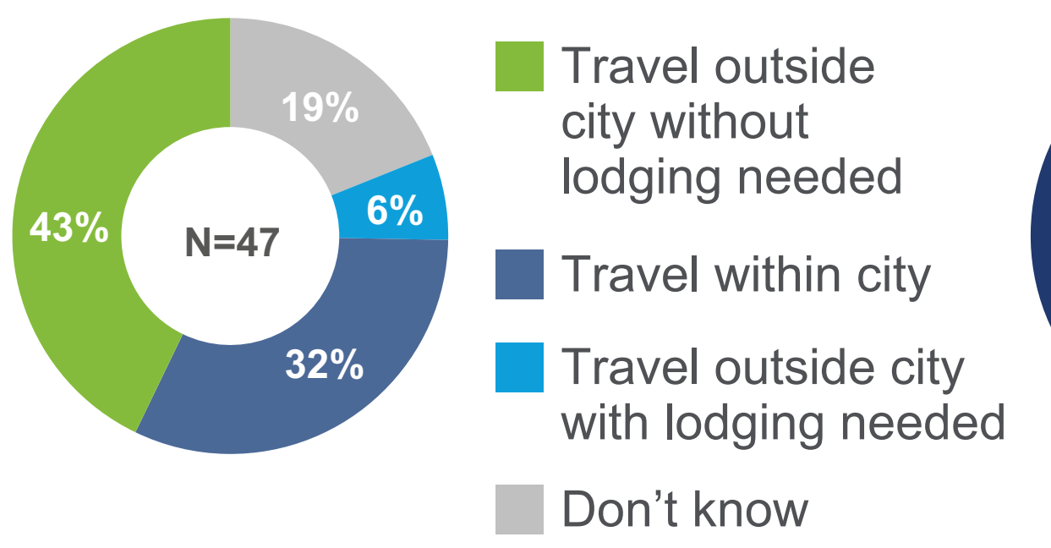
With declining independence, patients were less active in the workplace, especially those "completely dependent" on friends/family. However, the results indicate that HCPs were unaware of independence and employment status for many patients.

Level of independence and employment status

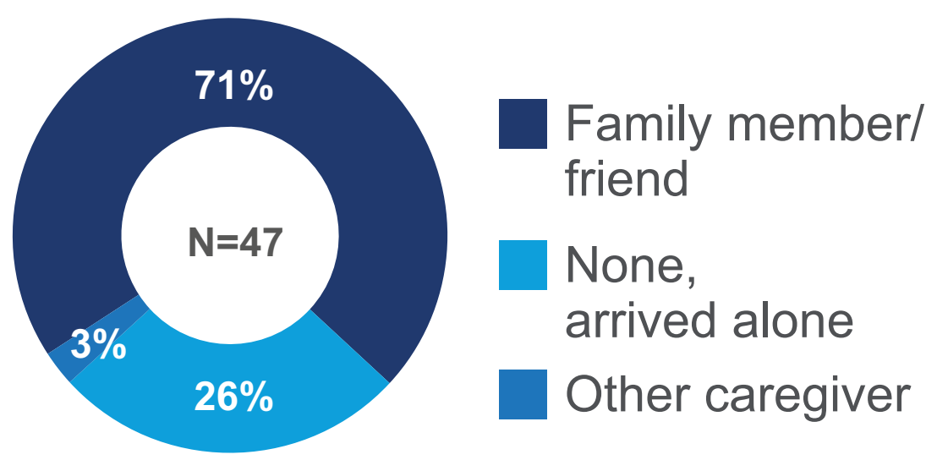


Specialist visits involved travel to a different city for 49% of patients, usually requiring support from family members.

Travel distance to consultation



Travel assistance

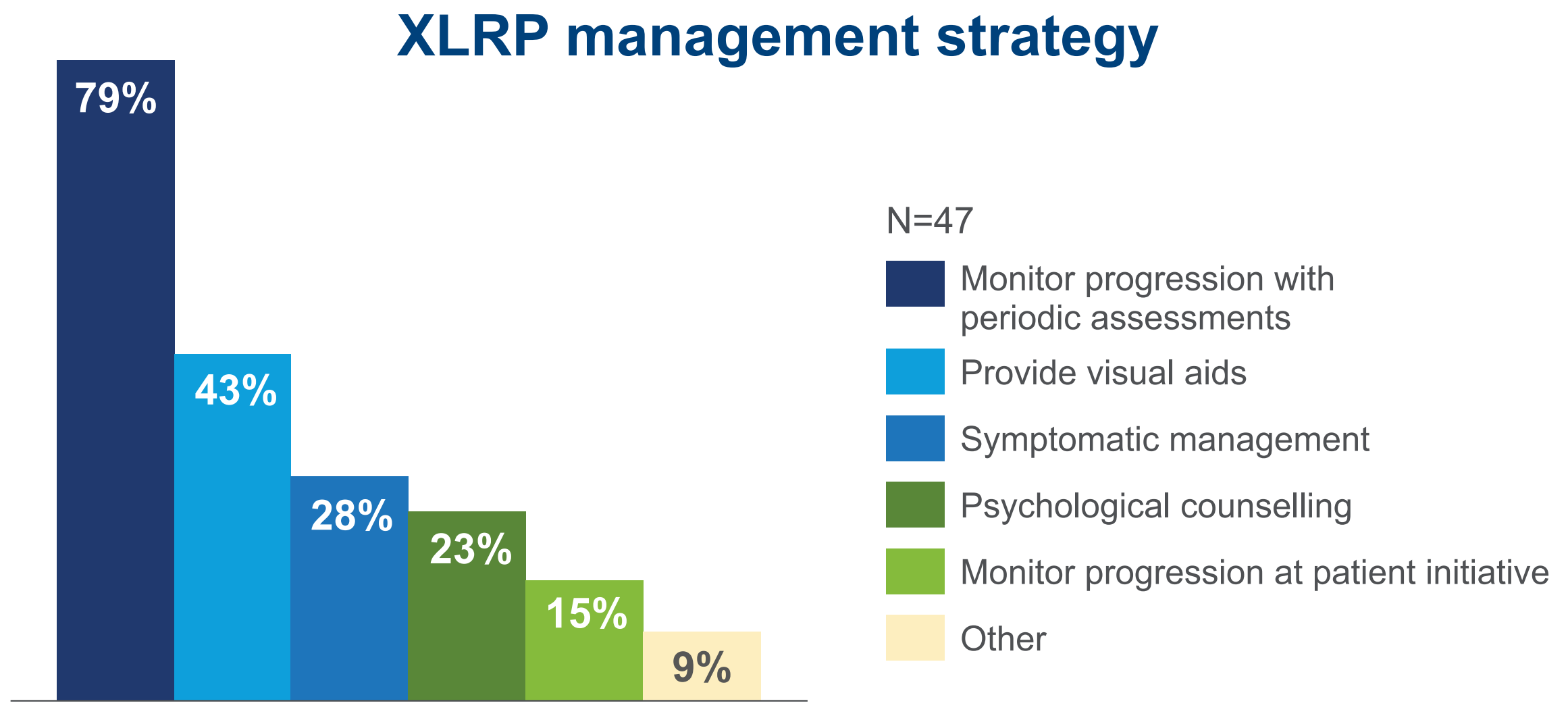


> It's not many patients who are lost to follow-up, but for the ones who are, it is very often because of travel.
>
> *- Retina specialist, Norway*

QoL and unmet needs in XLRP management

Regular assessment, symptomatic management, and providing visual aids was at the core of XLRP patient management.

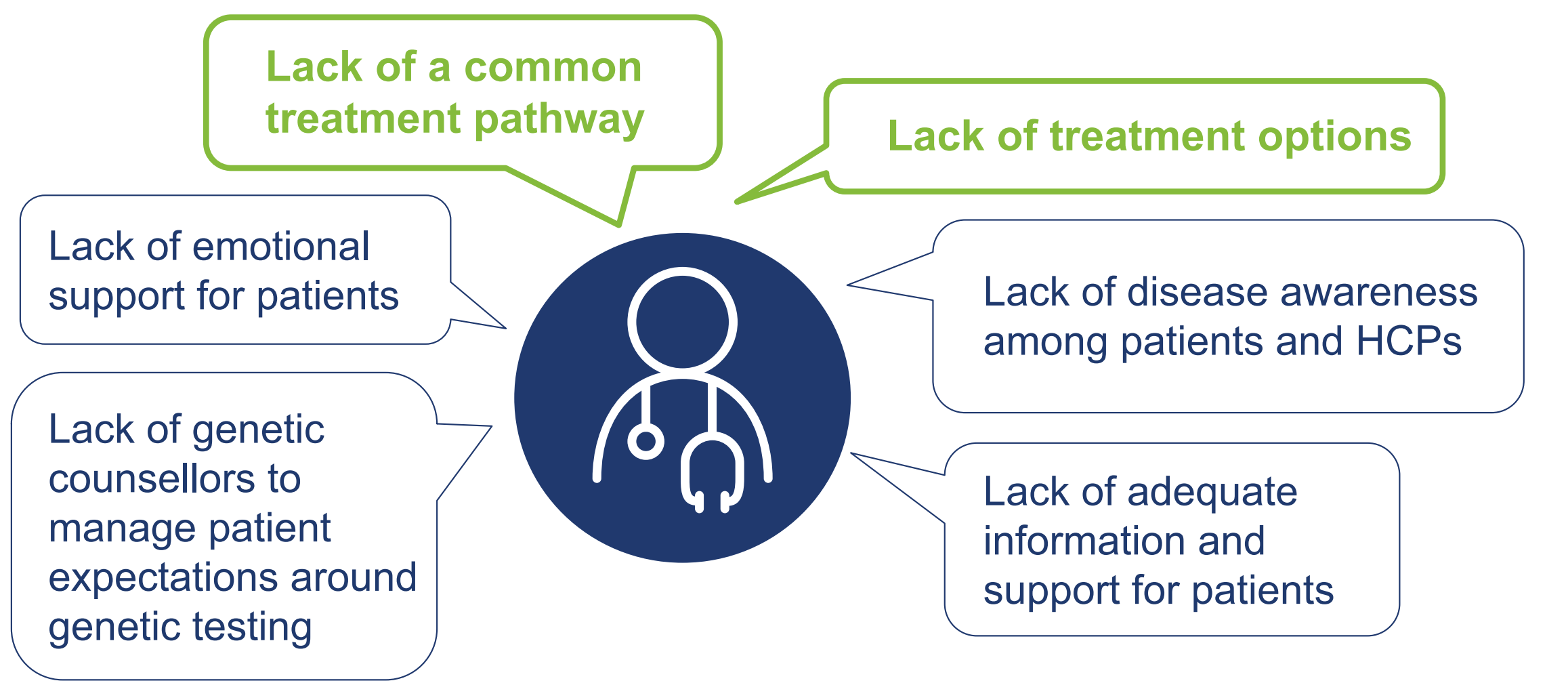
XLRP management strategy



However, despite the impact of XLRP on patients' lives:

- **only 28%** of HCPs reported that they monitor QoL, with monitoring being largely informal
- **only 47%** of patients were offered social, emotional, and/or financial support.

When asked about **challenges** and **unmet needs** in the management of patients with XLRP, the HCPs listed some of the following concerns[a]:



- **Lack of a common treatment pathway**
- **Lack of treatment options**
- Lack of emotional support for patients
- Lack of disease awareness among patients and HCPs
- Lack of genetic counsellors to manage patient expectations around genetic testing
- Lack of adequate information and support for patients

[a]The most frequently reported concerns are in green type.

CONCLUSIONS

- For patients with XLRP, work status and QoL are impacted by disease progression and reduced patient autonomy, which are likely to worsen over time.
- Some patients with XLRP are active in the workforce and policies to enable workplace participation are called for; visual impairment should not be a reason for high unemployment in the 21st century.
- Despite being exploratory, this cross-sectional survey demonstrated that XLRP has a major impact on patients' lives and provides valuable real-world insights that may not be generated by clinical studies or health economic research.

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DISCLOSURES

TD, JL, and KP are employees of Janssen Pharmaceutica N.V., a pharmaceutical company of Johnson & Johnson. AF and KA are employees of IQVIA, Inc.

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