

X-Linked Retinitis Pigmentosa Negatively Affects Patients' Work Status, Independence, and Quality of Life: Results From the Cross-Sectional EXPLORE XLRP 1 Physician Survey

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INTRODUCTION

- X-linked retinitis pigmentosa (XLRP), a form of inherited retinal dystrophy, is a rare eye disease associated with the progressive loss of photoreceptors. XLRP is among the most aggressive forms of retinitis pigmentosa, accounting for approximately 15% of all cases, leading to legal blindness in the third or fourth decade of life.^{1,4}
 - As an X-linked recessive disorder, XLRP primarily affects men.^{1,3,6}
 - Women who are heterozygous for an XLRP mutation are often considered unaffected carriers and may be asymptomatic, but some female carriers can experience significant visual symptoms, a phenomenon thought to be related to variable X-inactivation patterns.^{1,3,9}
- Approximately 70-80% of XLRP cases are caused by mutations in the *retinitis pigmentosa GTPase regulator* (RPGR) gene.^{1,5,8}
- For inherited retinal diseases such as XLRP, genetic testing is strongly recommended to provide a definitive diagnosis.^{10, 11}
- No treatment is currently available for XLRP. The recommended management includes use of low-vision aids, treatment of the complications (e.g., cataract, cystoid macular edema), and blindness rehabilitation strategies.¹¹⁻¹⁶
- Limited published data are available that describe details of the XLRP patient journey (including assessment, genetic testing, diagnosis, and management); there is a need for a better real-world understanding of the current standards of clinical practice and the potential obstacles to diagnosis and genetic testing.
- 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. The EXPLORE XLRP 1 survey interviewed retina specialists (n=20) and geneticists (n=5) in Germany, France, Italy, Spain, and the UK to provide insights on patients with XLRP (n=80), to better understand the pathways by which patients in Europe with XLRP are referred to these specialists for diagnosis, and to examine the impact of COVID-19 on patient management.

METHODS

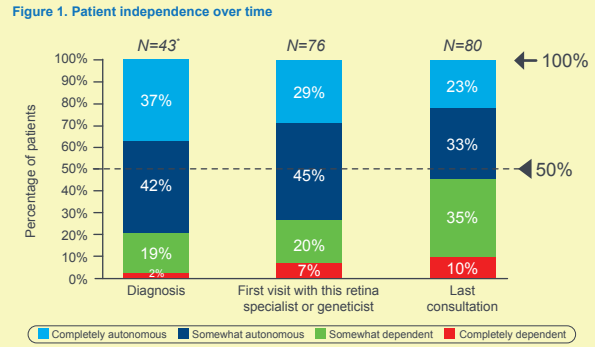
- EXPLORE XLRP 1 was an exploratory, cross-sectional, physician survey conducted in Germany, France, Italy, Spain, and the UK.
 - Retina specialists and geneticists in these countries were identified who had at least 5 years' experience managing or seeing patients with XLRP, and who were responsible for the recent management of patients with XLRP (retina specialists) or who were responsible for the genetic testing of patients with XLRP (geneticists).
- Retina specialists provided anonymized information for four recent patients diagnosed with XLRP that each had personally managed, including details of the diagnostic and referral process and decisions to pursue genetic testing.
 - The anonymized patient information was collected through an online survey completed by the retina specialists and stored in a secure database.
 - Individual 60-minute telephone interviews were then conducted with the retina specialists and geneticists to capture their perspectives on management approaches for patients with XLRP.

RESULTS

- A total of 20 retina specialists and five geneticists from France, Germany, Italy, Spain, and the UK participated in the survey (four retina specialists and one geneticist from each country). Patient information was collected for 80 patients (16 from each country).
- The retina specialists surveyed reported that their patients with XLRP were mostly male (91%) and that 57% were aged 18-40 years.

Patient independence and workforce participation

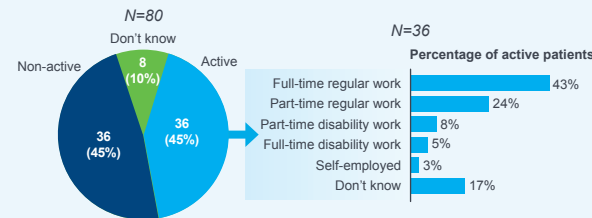
- Most of the patients (79%) described by the retina specialists lived with their families; for 65% of patients, visits to retina specialists required traveling to a different city.
- Retina specialists noted that patient independence decreased over time; most patients (79%) were "completely or somewhat autonomous" at diagnosis, but this had declined to only 56% at the time of the most recent consultation (Figure 1).
- A total of 37% of patients were "completely autonomous" at diagnosis (with 2% being "completely dependent" on family/friends), but only 23% were "completely autonomous" at their most recent consultation (with 10% being "completely dependent"). At the time of the survey, the average length of time the patients were managed by these retina specialists was 4.9 years.



*Number of patients does not add up to 80 due to some respondents answering "don't know." Number of this response at diagnosis and first visit was 37 and 4, respectively.

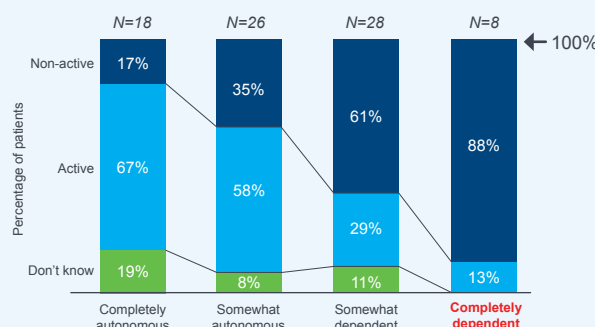
- Retina specialists reported that at the time of the last visit, 45% of patients were active in the workforce, of whom 43% worked full time, 24% worked part time, and 13% participated in disability work (Figure 2).

Figure 2. Work status and form of work



- Workforce participation was related to the extent of independence/autonomy; 67% of "completely autonomous" patients had active working status, compared with 13% of "completely dependent" patients (Figure 3).

Figure 3. Patient dependence and employment status

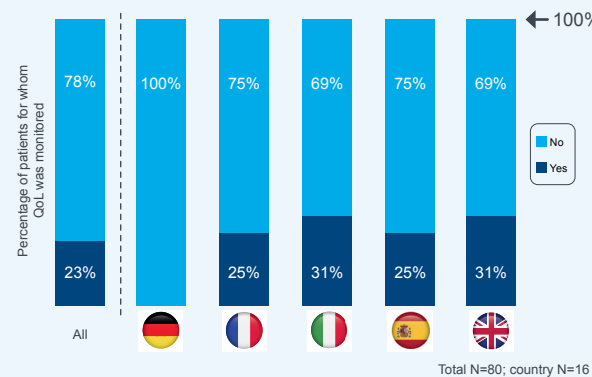


- The retina specialists and geneticists indicated that their female patients with XLRP experience similar challenges with independence/autonomy and their ability to work. Some retina specialists and geneticists responded that their female patients often progressed to late-stage disease at an earlier age than did their male patients.

QoL and unmet needs in XLRP management

- Retina specialists indicated the importance of the impact of XLRP on patients' QoL, but many responded that they evaluate QoL only informally; QoL was monitored with validated instruments in only 23% of patients (Figure 4).
- QoL instruments utilized by some retina specialists include the visual function (VF) QoL, the VF index (VF-14), and the 25-item visual function (VFQ-25) questionnaires.

Figure 4. QoL assessment



- Retina specialists in the UK estimated that social/emotional/psychological support was offered to the majority (>90%) of patients with XLRP, while retina specialists in France, Spain, Germany, and Italy indicated that it was offered to patients only rarely (<20%) (Figure 5).

Figure 5. Social/emotional support

Country	% of mentions on offered support	Type of support
Germany	N=3/16 (19%)	<ul style="list-style-type: none"> Psychological care and social support available within the clinic Support groups Local self-care organizations
France	N=3/16 (19%)	<ul style="list-style-type: none"> GP arranges psychological support Patients' associations
Italy	N=0/16 (0%)	<ul style="list-style-type: none"> The Blind People's Institute Appears to be limited support for XLRP patients
Spain	N=2/16 (13%)	<ul style="list-style-type: none"> There is no strong psychological support or emotional support center
UK	N=15/16 (94%)	<ul style="list-style-type: none"> The Low Vision Liaison Officer provides information on support and referral to "low-vision aids clinic"

- The retina specialists and geneticists generally agreed that the patients most in need of new XLRP treatment options are younger patients in the early stages of the disease, because delay of disease progression in these patients would have the greatest effect on preserving vision and QoL.

- Rehabilitation efforts and management of XLRP complications are key treatment goals for older patients who have already experienced significant disease progression and vision loss.

CONCLUSIONS

- For patients with XLRP, work status and QoL are impacted by disease progression and reduced patient autonomy, challenges that are likely to worsen as these patients age.**
- Fewer than 50% of patients with XLRP are active in the workforce, and policies to enable workplace participation (which may include part-time and disability work) for patients with XLRP and other rare eye diseases are called for; visual impairment should not be a reason for high unemployment in the 21st century.
- Unmet needs in XLRP management include more standardized QoL assessment, improved and earlier access to patient support programs, and better treatment options.
- Although this cross-sectional survey was exploratory in nature, it demonstrated that XLRP has a major impact on patients' lives and provides valuable real-world insights from retina specialists and geneticists that may not otherwise be available through clinical studies or health economic research.

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Conflicts of Interest

KP, JL, TD, YK, and RN are employees of Janssen Pharmaceuticals, Inc. KA is an employee of IQVIA and has a consulting agreement with Janssen Pharmaceuticals, Inc. FP has received personal fees for advisory boards from Janssen. Copies of this poster are for personal use only and may not be reproduced without permission from the Professional Society for Health Economics and Outcomes Research and the authors of this poster.

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