HSD119

Patient Pathways, Genetic Testing, and Diagnosis of X-Linked Retinitis Pigmentosa in Europe: 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. The recommended management includes use of low-vision aids, treatment of complications, and blindness rehabilitation strategies.[4,5]

As potential targeted therapies for XLRP emerge, early diagnosis and access to genetic testing for both patients and family members will likely be topics of key importance, as will efforts to streamline the patient journey.

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 the pathways by which European patients with XLRP reach retina specialists, ophthalmologists, and geneticists for genetic testing and diagnosis.

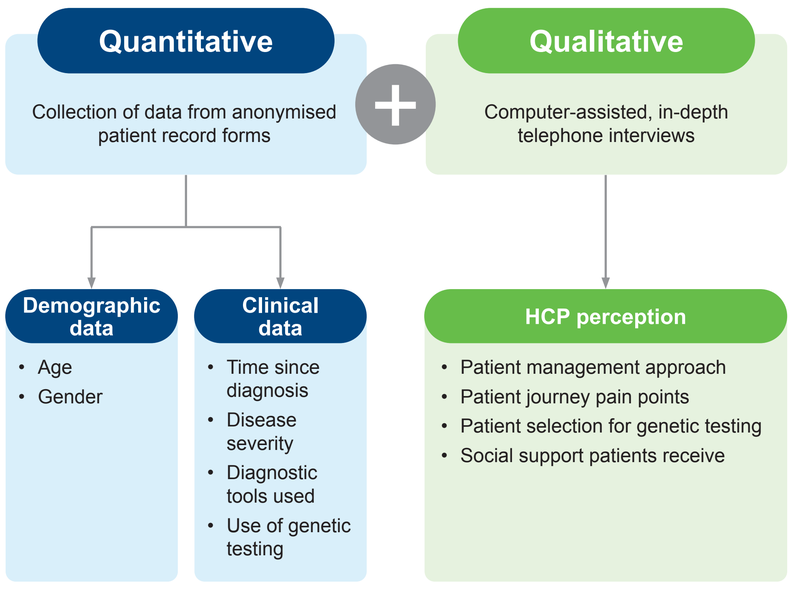
METHODS

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:

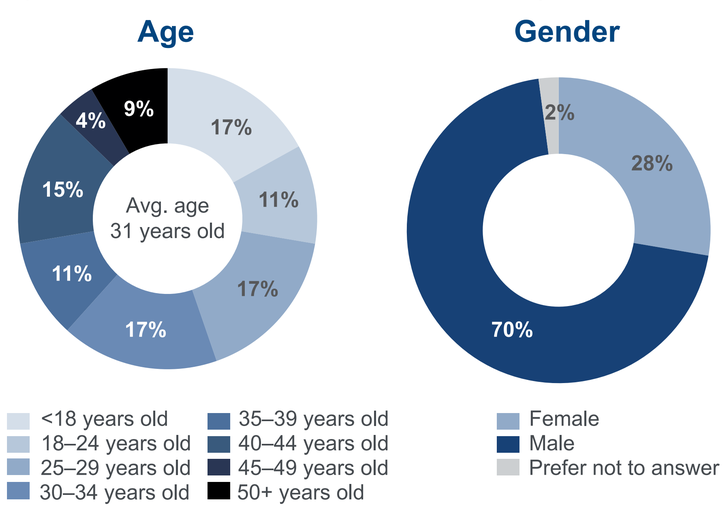


Ethics approval for this study was requested; the Ethics Committee confirmed that this research was out of scope.

RESULTS AND DISCUSSION

Patient demographics

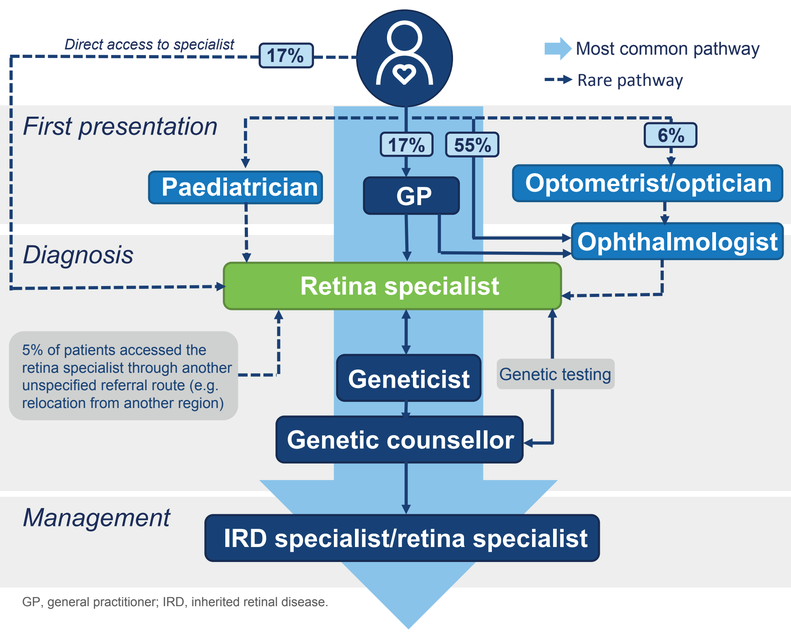
Most patients with XLRP were male and under 34 years old.



Patient pathway

- In the surveyed countries, XLRP was usually diagnosed by retina specialists (47%) or ophthalmologists (26%).
- Patients often consulted several HCPs before a retina specialist confirmed XLRP diagnosis.

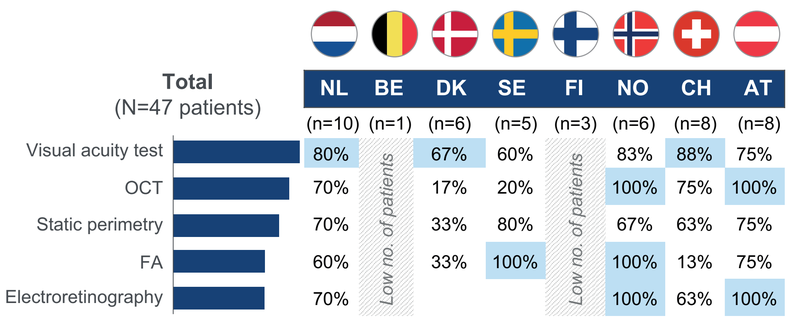
Referral pathways to retina specialist



GP, general practitioner; IRD, inherited retinal disease.

> "In a small country there are not many patients, so there is no common treatment protocol or treatment pathway."
> *- Ophthalmologist, Finland*

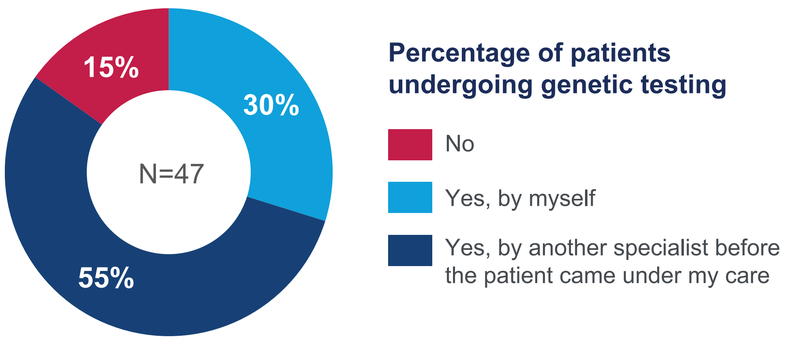
Some of the most common diagnostic tests performed on suspicion of XLRP included visual acuity, optical coherence tomography, and static perimetry tests.



OCT, optical coherence tomography; FA, fundus autofluorescence.
Only diagnostic tests administered to >50% of patients are shown.

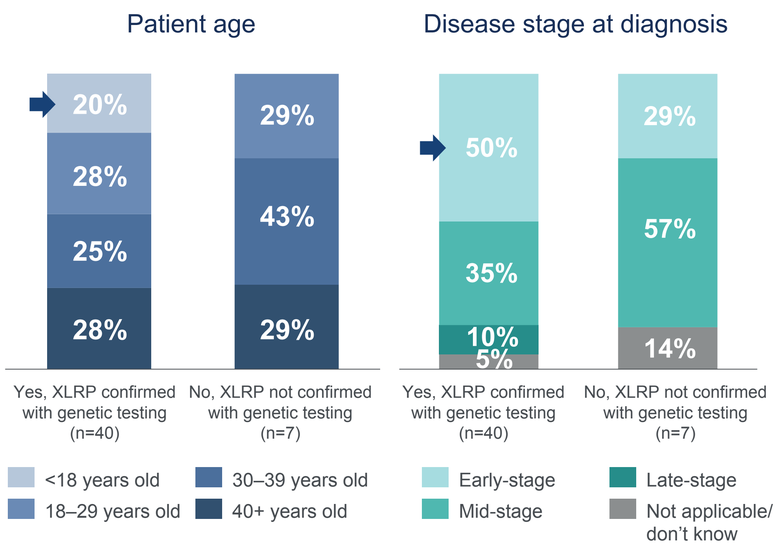
Genetic testing

Overall, 85% of patients with XLRP represented in this survey received a confirmatory genetic diagnosis.



Overall, younger patients (particularly those <18 years old) and those with early-stage XLRP were more likely to undergo genetic testing.

Ratio of genetic testing by age and disease stage



Most patients presenting with a suspicion of XLRP were recommended by the retina specialists to undergo genetic testing. However, only **1 in 3 families** opted to be tested.

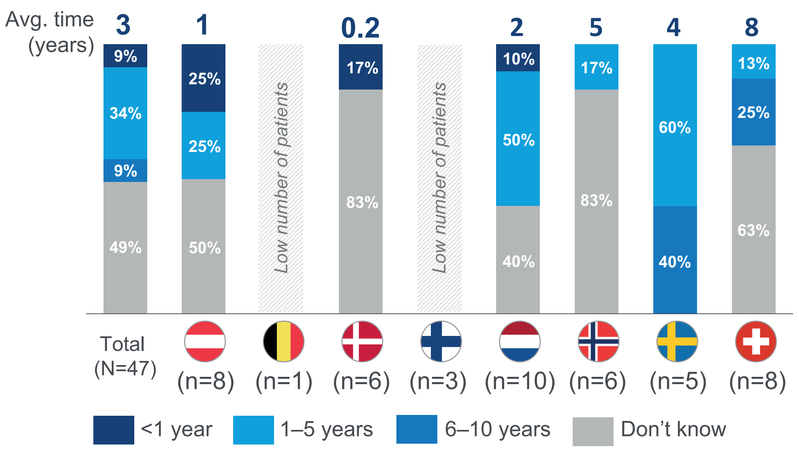
Retina specialists/ophthalmologists stated some of the following reasons why patients may be reluctant to undergo testing[a]:

[a]The graphic illustrates a selection of representative responses.

Diagnosis

There was an average delay of 3 years between symptom onset and genetic diagnosis reported in this survey.[b]

Time from symptom onset to genetic diagnosis



[b]We note that in some regions, this information was not available.

> "In my experience, I can say one of the diagnoses was quick, it was after 8 months, which was rapid"
> *- Ophthalmologist, The Netherlands*

Retina specialists/ophthalmologists estimated that it took 1–8 months (2.8 months average) to receive test results, whereas geneticists estimated it took 1 month. Reasons for testing delays included **lack of patient awareness** and **lack of reimbursement** for testing costs by insurance providers.

CONCLUSIONS

- The pathways by which patients with XLRP in the surveyed countries are referred to retina specialists and geneticists are complex, lengthy, and vary considerably by country.
- XLRP diagnosis was confirmed by genetic testing for most patients treated by retina specialists; however, delays in receiving results accounted for incomplete uptake, especially among older patients.
- Early diagnosis is important for patients to enable them to understand how the diagnosis will impact their life and family, and to facilitate participation in clinical trials.
- 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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