INTRODUCTION

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

There is currently no effective treatment for XLRP. The recommended management includes use of low-vision aids, treatment of complications, and blindness rehabilitation strategies.\(^\text{3,4}\) 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.

RESULTS

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

Most patients presenting with a suspicion of XLRP were recommended by the retina specialists to undergo genetic testing. However, only in 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:

1. Financial reasons
2. Lack of knowledge or awareness
3. Feeling sceptical about the outcome of the test and worrying about impact on insurance
4. Fear of the test result and its impact on their life

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. Support for medical writing and editing of this poster was provided by Louise Müller and Davis Renshaw from IQVIA, Inc.

REFERENCES


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