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RESEARCH ARTICLE

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A cost of illness model to estimate the annual societal costs of X-linked retinitis pigmentosa (XLRP) in the United Kingdom (UK)

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ABSTRACT

Objective: To estimate the annual per-patient costs in the United Kingdom (UK) for each stage (disease severity) of X-linked retinitis pigmentosa (XLRP), a rare retinal disease that progressively leads to visual impairment/blindness.

Methods: The EXPLORE XLRP-2 study enrolled patients with XLRP ($n = 168$) and their informal caregivers ($n = 68$) in 10 countries. EXPLORE XLRP-2 outcomes including health care costs/resource use (e.g. health care visits, concomitant medications), comorbidities, productivity loss, out-of-pocket costs, welfare payments, and XLRP management costs were used to estimate the annual costs for mild, moderate, or severe XLRP. A human capital approach was used to calculate patients' productivity losses; an opportunity cost approach was used to calculate the value of time spent on informal caregiving.

Results: Total annual costs of XLRP in the UK were estimated to be £57,386 per patient for mild XLRP, £72,372 for moderate XLRP, and £103,127 for severe XLRP. The cost of managing XLRP is greatest for those with worse disease. For all XLRP severity levels, the largest drivers of total costs were caregivers' opportunity costs and patients' productivity losses.

Conclusion: Our UK-focused cost model demonstrates that the societal costs associated with XLRP tend to be higher in patients with more severe disease and are driven primarily by considerable opportunity costs for caregivers and productivity losses for patients. Health care resource use has little impact on the total cost. With no treatment currently available for XLRP, the high economic burden of the disease rests not with health care payers but mainly with patients and their caregivers.

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XLRP; X-linked retinitis pigmentosa; cost model; economic model; economic burden

Introduction

X-linked retinitis pigmentosa (XLRP) is a rare, progressive, hereditary retinal disease that primarily affects males[1–4]. XLRP leads to visual impairment and often progresses to legal blindness based on visual acuity criteria by age 40–45 or even earlier; some XLRP studies using visual field-based definitions reported blindness by a mean age of 25 years[4–10]. More than 70% of XLRP cases are caused by mutations in the retinitis pigmentosa GTPase regulator (RPGR) gene, leading to death of photoreceptors, primarily affecting the rods[10,11].

Patients living with XLRP often experience nyctalopia (difficulty seeing in low light conditions) as an early symptom, followed by progressive loss of peripheral vision and impairment of visual acuity as the disease worsens; these visual impairments are associated with decreased patient autonomy and mobility. As XLRP progresses in severity, worsening visual impairment can be attributed to thinning and loss of central foveal cones, which subsequently affects central vision. In XLRP, annual rates of visual acuity decline range from −8.2% (0.079 logMAR/year) to −3.5% (0.015

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logMAR/year); greater rates of annual visual acuity decline are typically observed among patients population in the second decade of life, a finding that highlights the rapid progression of XLRP[4–6]. No treatment is currently available for XLRP; its management includes the use of low-vision aids and ultra-low vision rehabilitative strategies, as well as treatment of disease complications and comorbidities (e.g. cataract, cystoid macular edema)[5,12–17].

Database studies have demonstrated that patients living with inherited retinal diseases like XLRP experience significantly worse overall survival, higher likelihood of requiring medical visits, and decreased quality of life compared to patients without inherited retinal disease[18]. However, data describing details of the specific impact of XLRP on patients and their caregivers are limited; there is a need for a better real-world understanding of patient/caregiver experiences and the costs of XLRP to patients, health systems, and society[4,19]. Because XLRP is a very rare disease, the potential number of patients and (especially) their informal caregivers from whom to collect outcome and cost data is small. To address these data gaps, the exploratory, multicenter, non-interventional EXPLORE XLRP-2 study combined patient chart review with cross-sectional surveys administered to patients living with XLRP caused by *RPGR* mutations and their informal caregivers in 10 countries across Europe and Israel[20–22]. EXPLORE XLRP-2 demonstrated that the real-world impacts (including those related to mental health, employment, mobility, and daily activities) of XLRP on patients are more pronounced in patients with severe XLRP. The study showed that informal caregivers spend an average of over 28 hours per week caring for patients with XLRP; for patients with severe XLRP, even more time is spent on caregiving[20–22].

Economic data describing details of the total cost to society of XLRP are sparse, and no data have been identified that provide estimated societal costs per disease stage (i.e. stage of XLRP severity). To address this gap, we have developed a cost of illness model that uses findings from EXPLORE XLRP-2 to estimate the annual per-patient societal costs (including direct and indirect costs) for patients with mild, moderate, or severe XLRP using the United Kingdom (UK) as a base case scenario.

Methods

Data sources

Outcome data from EXPLORE XLRP-2 were used as inputs for the cost of illness model, supplemented by targeted literature searches for additional data inputs[23–30]. Full details of the EXPLORE XLRP-2 study design have been reported previously[20–22]. The study received local ethical committee approval at each study site and adhered to the principles of the Declaration of Helsinki. All study participants provided informed consent. Questionnaires administered in EXPLORE XLRP-2 and whose outcomes contributed inputs to our cost model included the Work Productivity and Activity Impairment General Health v2.0 [WPAI-GH2], the Hospital Anxiety and Depression Scale [HADS], and long-term impact patient questionnaires (LIPQ) and long-term impact caregiver questionnaires (LICQ). The HADs and the WPAI-GH2 are validated questionnaires. The LIPQ and LICQ questionnaires were developed by the study sponsor with input from specialists and patient experts.

EXPLORE XLRP-2 outcomes used for the cost model included comorbidities (from health care resource use data and HADS outcomes), productivity losses (from WPAI, LIPQ, and LICQ outcomes), out-of-pocket costs (from LIPQ outcomes), welfare payments (from LIPQ outcomes), and XLRP management costs (from health care resource use data and LIPQ outcomes).

EXPLORE XLRP-2 study data describing XLRP management costs (i.e. health care utilization costs specific to XLRP) were limited; it is probable that not all health care visits were collected in the study. The relevant EXPLORE XLRP-2 study data were complemented by data from targeted literature review, including additional literature data relevant to comorbidities, productivity losses, and costs[23–30].

Cost model details

A cost of illness model was prepared to estimate annual per-patient costs of each stage of XLRP (i.e. mild, moderate, or severe) from a societal perspective, including direct costs (XLRP management and health care provider costs) and indirect costs (patients' and caregivers' productivity losses and out-of-pocket costs) (Table 1).

Results

Health care resource use

Relevant study outcomes from EXPLORE XLRP-2 describing health care resource use data (i.e. XLRP management including GP and specialist visits and frequencies of ophthalmic comorbidities and XLRP complications [e.g. cataract, cystoid macular edema]) for study participants are summarized in Table 2.

The cost model's inputs and assumptions (derived from EXPLORE XLRP-2 study data[20]) used to calculate

Table 1. Summary of cost model inputs.

Direct costs (health care provider costs related to XLRP management)	Indirect costs (privately incurred costs)
Comorbidity costs: mental health	Patients' productivity losses
Comorbidity costs: concomitant medications	Caregivers' opportunity costs
Comorbidity costs: injuries	Out-of-pocket costs

Table 2. Key health care resource use findings from EXPLORE XLRP-2[20] (all study participants).

	$N = 168$[a]
Average number of GP visits in past year, mean (SD)	4.0 (8.7)
Average number of other HCP visits in past year, mean (SD)	7.4 (12.2)
Number of patients with injuries due to XLRP in past year, n (% of patients)	55 (35%)
Average number of hospitalizations for injury in past year, mean (SD)	0.2 (0.6)
Average number of days hospitalized for injury in past year, mean (SD)	0.3 (1.0)
Patients with other ophthalmic comorbidities, n (% of patients)	83 (49%)

[a]Not all outcomes listed were available for/reported by all study participants.

Table 3. Summary of the cost model's inputs/assumptions: direct costs (health care resource use).

Cost model inputs (XLRP management)	Mild	Moderate	Severe
Average number of health care provider visits in the past year			
XLRP specialist visits	1.00	1.00	1.00
Other specialist visits	1.00	1.00	1.00
General practitioner visits	6.25	1.67	4.15
Eye doctor/ophthalmologist visits	0	1.00	1.00
Physiotherapy	0	0	1.00
Average number of Injuries in the past year			
All injuries	14.00	13.00	28.00
Injuries requiring hospitalization	6.00	3.00	5.00
Injuries not requiring hospitalization	8.00	10.00	23.00
Mental health care inputs			
Depression prevalence, %			
Mild depression	17.2%	12.5%	15.3%
Moderate depression	3.4%	17.5%	11.9%
Severe depression	0%	5.0%	3.4%
Anxiety prevalence, %			
Mild anxiety	19.0%	20.0%	28.8%
Moderate anxiety	13.8%	15.0%	20.3%
Severe anxiety	5.2%	7.5%	3.4%
Annual mental health resource utilization rate	0.624	1.5	0.816
Annual mental health cost, £	£27.56	£66.25	£36.04

annual per-patient treatment costs for each health state (XLRP severity) are provided in Table 3. The cost model's inputs and assumptions (derived from EXPLORE XLRP-2 study data[20]) used to calculate annual patient productivity loss costs and caregiver opportunity costs are provided in Table 4.

Annual societal costs of XLRP

Using these inputs and assumptions (derived from EXPLORE XLRP-2 study data), the cost model's estimated societal costs of XLRP in the UK are shown in Table 5, broken down by cost category and XLRP severity. For all XLRP severity levels, the largest drivers of costs were caregiver opportunity costs (the loss of potential income because of time spent caregiving) and patient productivity losses (the decline in employment productivity and income due to the disease) (Figure 1).

Table 4. Summary of the cost model's inputs/assumptions: indirect costs (patient productivity loss costs/caregiver opportunity costs).

Cost model inputs (productivity losses)	Mild	Moderate	Severe
Patient productivity loss inputs[a]			
Currently unemployed, %	37.0%	42.9%	53.7%
Absenteeism	0	4.3	8.9
Presenteeism	15.2	33.5	34.8
Productivity loss	16.3	35.4	35.3
Caregiver opportunity cost inputs[b]			
Time spent caregiving (hours/week)	22.06	22.41	38.13
Time spent caregiving (hours/year)	1147.12	1165.32	1982.76

[a]Derived from EXPLORE XRLP-2 study data (WPAI outcomes).
[b]Derived from EXPLORE XRLP-2 study data (LICQ outcomes).

Discussion

This study estimates the annual societal costs of XLRP using data from the EXPLORE XLRP-2 study and demonstrates that a substantial financial burden is borne by patients and caregivers. Although cost estimates were calculated using UK-specific costing data, the EXPLORE XLRP-2 resource use data were from patients recruited from 9 European countries and Israel with heterogeneous healthcare systems. Using the resource data from the overall EXPLORE XLRP-2 study cohort was deemed appropriate due to the sample size difference between the overall cohort (patient $n = 168$ and their informal caregivers $n = 68$) vs UK only ($n = 25$ and $n = 8$ respectively), with sample size of participants from the UK for some questionnaires per severity level being ≤ 2. Care pathways vary across European countries and Israel, including differences in the first point of contact for ophthalmic care and for comorbid conditions such as depression or anxiety. Therefore, healthcare resource use and costs are likely to depend on health system organization, which should be considered when interpreting the UK data and also when extrapolating these findings beyond the UK.

Our cost model using the UK as a base case scenario is the first comprehensive effort to develop an

Table 5. Cost model outputs: summary of total annual costs per patient by XLRP severity.

Cost category	Mild	Moderate	Severe	Data sources from EXPLORE XLRP-2
XLRP management costs				EXPLORE XLRP-2 chart review
	£649	£505	£629	EXPLORE XLRP-2 LIPQ
Mental health costs	£28	£66	£36	EXPLORE XLRP-2 HADS
Concomitant medication costs				EXPLORE XLRP-2 chart review
	£46	£33	£44	EXPLORE XLRP-2 LIPQ
Cost of injuries	£38	£73	£76	EXPLORE XLRP-2 LIPQ
Indirect costs: patients' productivity loss	£16,345	£25,859	£32,592	EXPLORE XLRP-2 WPAI
Indirect costs: caregivers' opportunity costs	£30,972	£31,464	£ 53,535	EXPLORE XLRP-2 LICQ
Out-of-pocket costs	£7,862	£11,579	£4,835	EXPLORE XLRP-2 LIPQ
Total annual costs (excluding welfare costs)	**£55,940**	**£69,579**	**£91,746**	
Welfare costs[a]	£1,446	£2,793	£11,381	EXPLORE XLRP-2 LIPQ
Total annual costs (including welfare costs)	**£57,386**	**£72,372**	**£103,127**	

[a]Welfare costs included monthly social subsidies, generally paid by the government/health system.

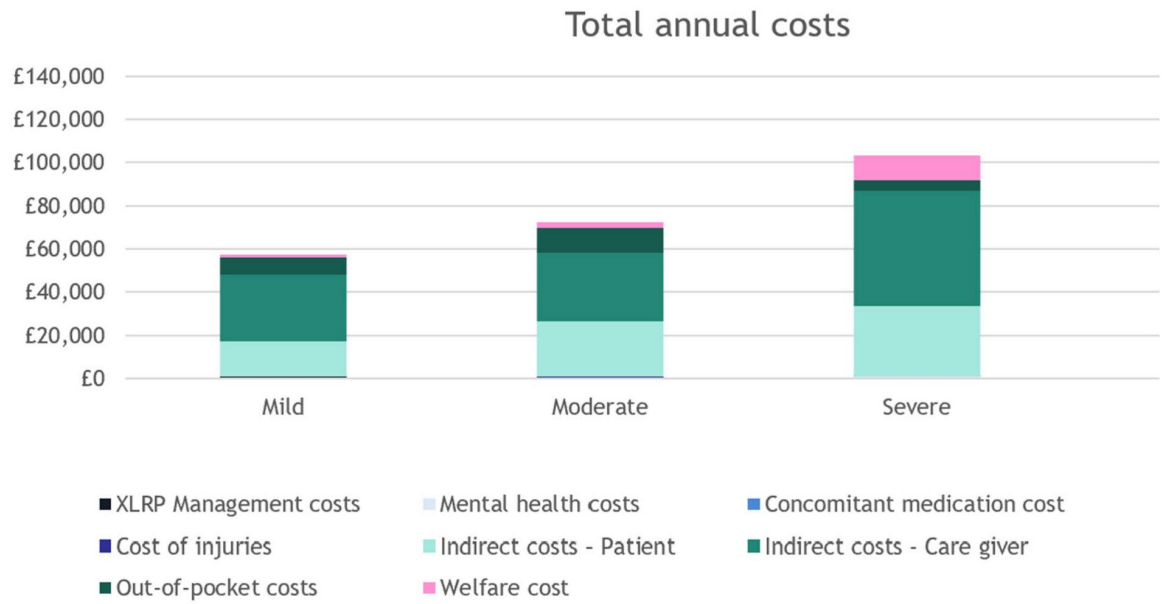

Figure 1. Proportions of total costs by cost category and XLRP severity.

economic model for XLRP. This cost model demonstrates that the total annual costs associated with XLRP increase with severity of vision loss, driven primarily by opportunity costs for informal caregivers and by productivity loss costs and welfare payments for patients. Health care resource use has little impact on the total cost. Such a model (which includes indirect costs and out-of-pocket costs incurred by patients living with XLRP) is appropriate for understanding these costs, because with no treatment currently available for XLRP[5,12–17], the economic burden of the disease mainly sits with patients, their caregivers, and wider society; and not with health care payers.

Because there is currently no treatment for XLRP, health care resource use is generally low across all stages of the condition. Most health care visits occur in the mild state when patients seek advice from their GP. However, as the vision loss progresses from mild to moderate/severe, the frequency of visits tends to decrease. This may be due to a lack of effective interventions, reducing the need for frequent follow-up care. Fewer health care visits may also be a consequence of the XLRP-related mobility and independence issues that worsen over time[19].

Patients with moderate XLRP incurred the highest out-of-pocket costs, and patients with severe XLRP incurred the lowest out-of-pocket costs. Patients with moderate XLRP had the highest mean monthly expenditure for items related to vision loss. These higher costs incurred by those with moderate XLRP may reflect patients preparing for their vision deteriorating further (into severe XLRP), such as making additional home adjustments or vision aids purchases in early stages of the disease which may not need to be re-purchased. A substantially higher proportion of those with severe XLRP received welfare payments, indicating that

although few treatment costs are incurred, costs to society due to XLRP manifest in other ways.

Indirect costs attributed to informal caregivers' opportunity costs were substantial and were greater than those attributed to patients' productivity losses, a finding consistent with the fact that many informal caregivers report spending many hours per week caring for patients with XLRP and can experience employment and mental health impacts as well[21].

Limitations

Several limitations should be considered when interpreting the findings of our cost model. Some limitations are related to how patient and caregiver data were collected in EXPLORE XLRP-2: most resource use and cost data were based on questionnaire responses subject to participants' recall bias (and not based on actual economic data collected). The frequency and nature of health care visits (and thus, expenditures by health care providers) may have been under-reported in EXPLORE XLRP-2.

In the EXPLORE XLRP-2 study, data on hospitalization (and hospitalization due to injury) were not collected in sufficient detail for those to be used to calculate hospitalization costs, and resource use related to injury may have been under-reported. Additionally, resource use and cost data related to mental health (e.g. costs of medication or psychological treatment for anxiety or depression) were not directly collected in EXPLORE XLRP-2, and out-of-pocket costs were not collected for caregivers.

Conclusion

We have developed a cost model that estimates the total annual, per-patient societal costs associated with XLRP in the UK for each level of disease severity (mild, moderate, or severe). This is the first research to estimate costs across each health state (XLRP severity); these costs are considerable, and indirect costs including patients' productivity loss costs and caregivers' opportunity costs were the largest drivers of total XLRP-related costs. These total costs are estimated to increase substantially as XLRP severity worsens: patients with severe XLRP have nearly double the societal costs compared to those with mild XLRP. Health care resource use was low overall and had little impact on the total societal cost of XLRP. These cost estimates demonstrate the substantial economic burden imposed on XLRP patients, their caregivers, and wider society, underlining the clear need for effective XLRP therapies.

Transparency

Declaration of funding

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Declaration of financial/other relationships

Authors ML, JVD, TD, TV, and KP are employees of Janssen Pharmaceuticals/Johnson and Johnson Innovative Medicine. Author CVR is an employee of Open Health and has a consulting agreement with Janssen Pharmaceuticals/Johnson and Johnson Innovative Medicine. FP has received consulting fees from AbbVie, Bayer, Janssen, Novartis, and Roche.

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Authors contributions

Authors TD, KP, JVL, ML, and CVR contributed to the design of the cost model. FP contributed to the critical review of the data. All authors contributed to interpretation of the study findings and preparation/reviewing/approval of this manuscript.

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Ethics statement

The EXPLORE XLRP-2 study received local ethical committee approval at each study site and adhered to the principles of the Declaration of Helsinki. All study participants provided informed consent to participate.

Data availability statement

The datasets generated and analyzed for this study are available from the corresponding author upon reasonable request.

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