

# Impact of Inherited Retinal Disease in the Republic of Ireland and the United Kingdom



## Inherited Retinal Dystrophies Consortium Operating for the Under-Represented for Novel Therapies and Services

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### Purpose:

Accurate data regarding prevalence of Inherited Retinal Diseases (IRDs), impact on individuals and families affected, and cost burden to UK and Irish economies is lacking.

This hinders development and commissioning of clinical services, treatments, and the planning and implementation of clinical treatment trials.

Thus there is a need for a stronger evidence base to support value for money to regulatory bodies for treatments recently approved, and treatments progressing through clinical trials towards market.

### Methods:

This cost of illness study relates to the following 10 IRDs in the Republic of Ireland (ROI) and United Kingdom (UK) in 2019:

- |                       |                            |
|-----------------------|----------------------------|
| 1. Achromatopsia      | 6. LCA/EOSRD               |
| 2. Best Disease       | 7. Retinitis Pigmentosa    |
| 3. Choroideremia      | 8. Stargardt Disease       |
| 4. Cone Dystrophy     | 9. Usher Syndrome          |
| 5. Cone-Rod Dystrophy | 10. X-Linked Retinoschisis |

The socioeconomic burden of these IRDs in the ROI and UK was estimated using a cost of illness methodology<sup>(1,2)</sup>.

The analysis was based on a targeted literature review and primary data collection via survey.

**Patient involvement was incorporated through each stage of project design and implementation, including the research brief, survey design and review, and accessibility testing.**

### Results:

**Table (1) Estimated Prevalent cases (number of people) with IRDs in the Republic of Ireland and the United Kingdom**

Region	Republic of Ireland		United Kingdom	
	Base Cases	Upper Band	Base Cases	Upper Band
Retinitis Pigmentosa	755	1274	10,324	17,422
Usher Syndrome	189	320	2,590	4,371
Stargardt Disease	154	260	2,109	3,559
LCA/EOSRD	116	196	1,587	2,679
Best Disease	73	124	1,003	1,693
Cone Dystrophy	64	108	876	1,478
Cone Rod Dystrophy	59	100	812	1,370
Achromatopsia	54	90	733	1,237
X-linked Retinoschisis	35	60	483	814
Choroideremia	22	37	298	504
<b>Total</b>	<b>1,521</b>	<b>2,569</b>	<b>20,815</b>	<b>35,126</b>

**Table (2) Survey responses in relation to Genetic Testing and Counseling**

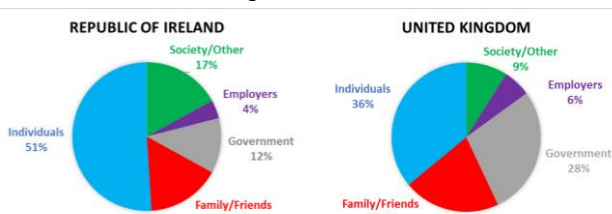
Region	Republic of Ireland	United Kingdom
Never received a genetic test	40.8%	43.4%
Never received genetic counselling	69.7%	50.9%

**Table (3) Breakdown of IRD cost from highest to lowest cost type**

Republic of Ireland			United Kingdom		
Cost Type	% Total Costs	Cost (€ Million)	Cost Type	% Total Costs	Cost (£ Million)
Wellbeing Costs	33.8	16.0	Wellbeing Costs	38.4	196.1
Productivity Costs	19.8	9.4	Productivity Costs	22.3	114.1
Deadweight losses	16.7	7.9	Carer Costs	16.5	84.5
Carer Costs	14.2	6.7	Other	13.5	68.7
Other Costs	11.9	5.6	Deadweight losses	5.4	27.4
Health System Costs	3.6	1.7	Health System Costs	3.9	19.9
<b>Total</b>	<b>100</b>	<b>47.3</b>	<b>Total</b>	<b>100</b>	<b>510.7</b>

- Approximately **1/3 of costs** were attributed to **wellbeing costs**.
- Compared to the general population persons with an IRD in the Republic of Ireland and the United Kingdom were **55.7%**, and **40.2%** less likely to be in paid employment.

**Figure (1) Percentage of IRD costs by payer in the Republic of Ireland and the United Kingdom**



### Conclusions:

IRD patients and their families have common issues including, the need for timely equitable access to genetic testing and counselling, equality in accessing employment, and a revision of the assessment process for reimbursement of therapies currently focused on the cost-of-illness to the healthcare system.

IRD patients do not frequently engage the healthcare system and as such suggests a cost-of-illness model from a societal perspective may be a better format.

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### References:

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