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# A microsimulation model to estimate the quality of life and cost of inherited retinal diseases (IRDMOD)

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# IRDMOD

1. What are Inherited Retinal Diseases (IRDs)
2. Primary data collection to estimate costs and quality of life (QoL)
3. IRDMOD
4. Next steps

# Inherited retinal diseases (IRDs)

- IRDs are a group of rare genetic diseases, estimated to affect between 1 in 1,000 and 1 in 2,000 people (Liew et al 2014, Hannay et al 2020).
- IRDs comprise a group of vision disorders which cause retinal dysfunction and degeneration leading to progressive vision loss and blindness (Duncan et e 2018)
- They are now the leading cause of legal blindness in working age adults (Viriato et al 2020).
- They often have early onset of disease and symptoms, leading to a substantial disease burden on both patients and their carers/families (Grigg et al 2020).
- Until recently, there were no treatment options for IRDs
- From around 2019, regulatory bodies, governments and other funders began to approve and fund use of Luxturna, a gene therapy for people with *RPE65 retinopathy*, a rare form of IRD
- So far, results of this gene therapy have been very promising
- However, there is very limited data on the impact of IRDs

# IRDs continued

- In Australia, when the Medical Services Advisory Committee (MSAC) recommended funding it, they acknowledged a lack of utility data and any non-health care related costs
- In the UK, the list price in 2019 was over £600,000 per patient, with the negotiated price for the NHS kept confidential
- There are many other gene therapies for IRDs in trials
- As a result, data is needed to evaluate the cost effectiveness of any gene therapies, as well as access to genetic testing and genome sequencing
- We worked with a team of health economists, geneticists, genetic counsellors and ophthalmologists to collect primary data on IRDs and develop a model to allow decision makers to understand the impact of the disease and the potential benefits of any interventions

# Primary data collection

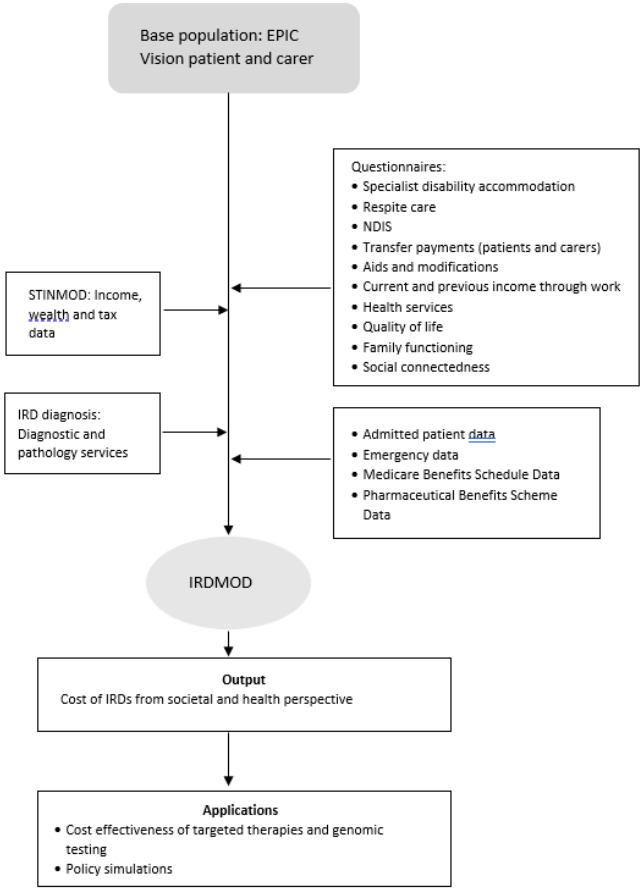
- Tailored questionnaire – the Economic and Psychosocial Impact of Caring for Families Affected by Visual Impairment
- All patients with a clinical diagnoses who attended a genetic of ophthalmic consultation at Children's hospital in Westmead or Save Sight Institute Sydney were invited
- Interviews are performed by research assistants and genetic counsellors, often up to 4 hours in length
- Collects data across every domain, health, social, quality of life, genetics, service use
- Patients under 18 are interviewed by proxy
- Carers and patients are interviewed

**Administrative data**

- For health data, data linkage is used to obtain health service utilisation and costs

# IRDMOD structure

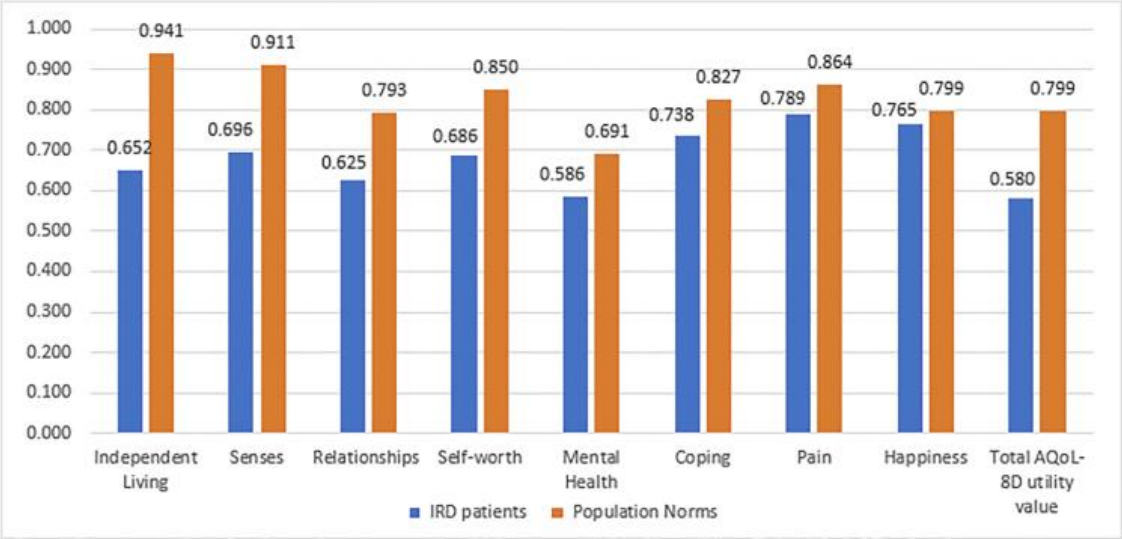
**Figure 1:** Structure of IRDMOD



# IRDMOD

## QUALITY OF LIFE ANALYSIS

- In health economic analysis, health utility values are required for measuring quality of life
- This requires a validated instrument
- In EPIC-Vision we collected the Assessment of Quality of Life – 8 Dimension (AQoL-8D)




Population norm values for dimensions are scores which are unweighted by utilities. Total AQoL-8D refers to AQoL-8D utility value. One sample t-tests were significant at the 0.05 level for all dimensions and total utility values.

AQoL-8D = Assessment of Quality-of-Life 8D

# IRDMOD

## COSTS

- We model the lifetime costs of IRDs using a combination of data collected from EPIC-Vision, linked health data and counterfactual income and tax modelling
- This is done by estimating average annual costs in each age group, multiplied by time in age group, summed over average life expectancy in Australia
- Costs are estimated for each individual with IRD and their carer and spouse
- Microsimulation modelling allows us to be able to change inputs for each individual (if examining an intervention) and their carer/spouse
- Each cost is assigned to a payer:
  - Government – state/federal
  - Patient
  - Carer
  - Other

# IRDMOD

## HEALTH AND SOCIETAL COSTS

- Societal costs are estimated from data collected in EPIC-Vision
- This includes social service use, income support payments, use of the National Disability Insurance Scheme and income losses
- Where possible, costs are taken from published sources, such as current income support levels
- Health costs are a combination of self-reported data and linked administrative health data in Australia

*Counterfactual modelling – patients, carers and spouses*

- We collect data on patient, carer and spouse incomes
- Using the static income model (STINMOD), we match individuals to STINMOD based on age, gender, state of residence and education (IRDs in our cohort did not seem to impact education)
- Income loss is taken as the difference between their current income and the average of the simulations (1,000) from STINMOD
- Similar is performed for tax, with an added step of tax imputation of EPIC-Vision cohort as they did not report tax paid

# IRDMOD Lifetime cost

| N | Age group | Patient costs | | | | | | | | | | | Carer and spouse costs | | | Total costs | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | Commonwealth government | | | | State government | | | Private | | | Other | Commonwealth government | | Private | Total cost per patient | Total cost per household |
| | | Health System | Social | NDIS | Tax receipts | Health system | Social | NDIS | Health | OOP expenses other | Lost income | | Social | Tax receipts | Carer and spouse lost income | | |
| 6 | 0 to 5 | 15,859 | 1,335 | 7,948 | - | 14,482 | 4,004 | 5,755 | 3,655 | 3,266 | - | 79 | 9,356 | 4,587 | 16,210 | 385,906 | 416,372 |
| 17 | 6 to 18 | 1,675 | 7,955 | 7,844 | 282 | 272 | 4,314 | 5,680 | 1,198 | 2,646 | 1,263 | 56 | 13,721 | 11,122 | 32,408 | 1,350,313 | 1,456,916 |
| 21 | 19 to 29 | 874 | 11,430 | 4,106 | 599 | 158 | - | 2,973 | 894 | 544 | 4,286 | 45 | 2,348 | 3,889 | 11,796 | 1,733,090 | 1,869,913 |
| 9 | 30 to 39 | 9,767 | 10,889 | 7,895 | 9,015 | 9,020 | - | 5,717 | 825 | 343 | 31,163 | 105 | 751 | 200 | 1,112 | 2,485,243 | 2,681,447 |
| 15 | 40 to 49 | 2,003 | 10,999 | 5,365 | 11,535 | 933 | - | 3,885 | 1,968 | 416 | 29,009 | 221 | - | 577 | 1,185 | 3,058,440 | 3,299,896 |
| 14 | 50 to 59 | 2,693 | 15,653 | 3,910 | 17,198 | 1,000 | - | 2,831 | 1,533 | 793 | 44,494 | 1,181 | - | 5,250 | 14,841 | 4,020,110 | 4,337,487 |
| 6 | 60 to 69 | 3,382 | 16,683 | 7,612 | 8,373 | 1,079 | 333 | 5,512 | 2,108 | 1,210 | 29,028 | 1,339 | 563 | 4,043 | 15,862 | 4,826,609 | 5,207,657 |
| 6 | 70 - 84 | 2,522 | 17,025 | - | 333 | 615 | - | - | 912 | 1,137 | 2,853 | 630 | - | 2,074 | 12,518 | 5,159,644 | 5,566,985 |

# IRDMOD

## WHAT IS THE COST OF A RARE DISEASE?

| Commonwealth government | |
|---|---|
| Health | 324,429 |
| Social spending excl National Disability Insurance Scheme (NDIS) - patient | 1,016,419 |
| NDIS | 434,688 |
| Social spending - carer | 264,113 |
| Tax receipts - patient | 476,140 |
| Tax receipts - carer/spouse | 340,031 |
| Total | 2,855,820 |
| | |
| State Government | |
| Health | 206,614 |
| Social spending - patient | 79,439 |
| NDIS | 314,774 |
| Total | 600,828 |
| | |
| Individual/household | |
| Health | 120,789 |
| Other including aids/modifications | 100,254 |
| Lost income - patient | 1,440,453 |
| Lost income - spouse/carer | 1,137,364 |
| Total | 2,798,860 |
| | |
| Private health insurance spending** | 38,893 |
| | |
| Total cost (after adjustment)* | 5,159,644 |

| Annual costs by visual acuity | $ |
|---|---|
| Better than or equal to 6/60 (N=45) | 41,357 |
| Worse than 6/60 (N=49) | 83,910 |
| | |
| Annual costs by gender | |
| Female (N=55) | 54,527 |
| Male (N=39) | 76,067 |

# IRDMOD

## WHERE TO NEXT?

- IRMOD has been designed to be adaptable
  - The outputs shown in this presentation were for all IRDs
  - The model can be estimated on sub-groups of this analysis
- Estimating interventions?
  - Determined by the impact of the intervention
  - For example, as the model is a lifetime cost model by age group, it can be used to estimate changes in costs and benefits at different ages or disease progression
- *Societal costs*
  - The outputs on whole IRD costs as well as QoL impacts shown the significant disease burden of IRDs
  - There are substantial QoL impacts
  - Most costs are societal, these must be considered in evaluations

# References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