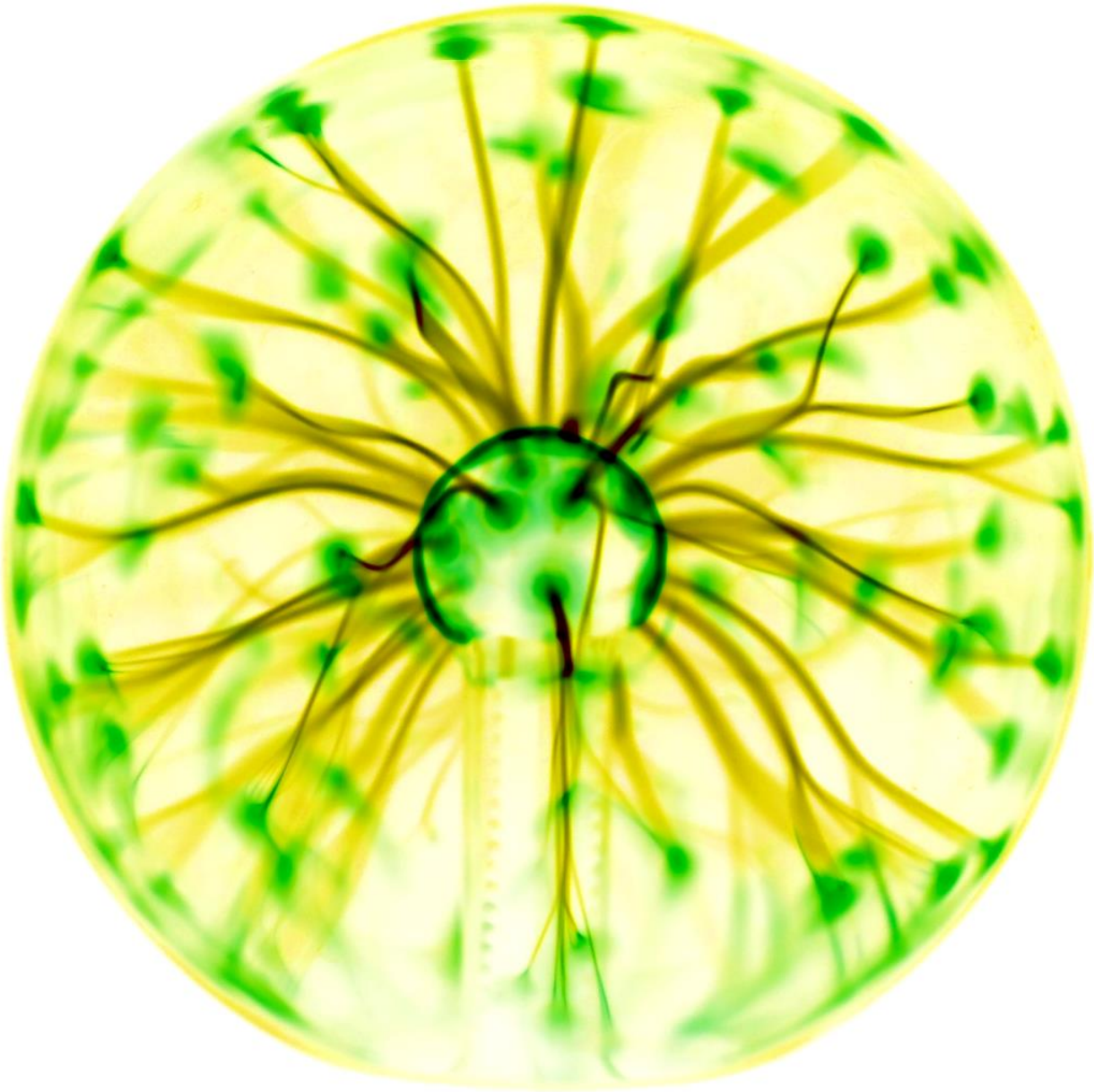
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The socioeconomic impact of inherited retinal dystrophies (IRDs) in the United Kingdom

Retina International

August 2019

Deloitte
Access Economics

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Glossary

Acronym	Full name
AMD	age-related macular degeneration
AWE	average weekly earnings
CBA	cost benefit analysis
CEA	cost effectiveness analysis
CMA	cost minimisation analysis
CPI	consumer price index
CSO	Central Statistics Office
CUA	cost utility analysis
DALY	disability adjusted life year
DLA	disability living allowance
DLTV	Daily Living Tasks Dependent on Vision Questionnaire
DVA	distance visual acuity
DWL	deadweight loss
ENCTS	English National Concessionary Travel Scheme
EOSRD	early onset severe retinal dystrophy
ERG	electroretinography
ESA	employment and support allowance
GDPR	European Union General Data Protection Regulations
GP	general practitioner
GPS	Global Positioning System
HID	handicaps, incapacités, dépendance
HM	Her Majesty's
HRB	Health Review Board
ICER	Institute for Clinical and Economic Review

Acronym	Full name
IRD	inherited retinal dystrophy
LA	local authorities
LCA	Leber congenital amaurosis
LFS	labour force survey
MCPF	marginal cost of public funds
NHS	National Health Service
NICE	National Institute for Health and Care Excellence
OECD	Organization for Economic Cooperation and Development
ONS	Office for National Statistics
PIP	personal independence payment
PPP	purchasing power parity
PSS	personal social services
PSSRU	Personal Social Services Research Unit
RoI	Republic of Ireland
RP	retinitis pigmentosa
RPE	retinal pigment epithelium
SEN	special education needs
SSI	severe sight impairment
UK	United Kingdom
US	United States of America
USH1	Type 1 Usher syndrome
VI	visual impairment
VSLY	value of a statistical life year
wAMD	wet age-related macular degeneration
WHO	World Health Organization
XLRS	X-linked retinoschisis
YLD	years of healthy life lost due to disability

Acronym	Full name
YLL	years of life lost due to premature death

Foreword

This work was funded and supported by IRD COUNTS who recognised the need to better understand the real impact of inherited retinal dystrophies (IRDs) in the United Kingdom (UK) of Great Britain and Northern Ireland, and the Republic of Ireland (RoI).

IRD COUNTS is a consortium of patient-led organisations, industry and clinician partners who assisted in the design of the project and recruitment of survey participants.

IRD COUNTS partners: Retina UK, Fighting Blindness Ireland, Thomas Pocklington Trust, F.Hoffmann-La Roche, MeiraGTx, Novartis Limited UK, ProQR.

IRD COUNTS is managed by Retina International

The socioeconomic impact of inherited retinal dystrophies (IRDs) in the United Kingdom (UK)

Executive summary

Methodology

The socioeconomic burden of inherited retinal dystrophies (IRDs) in the United Kingdom (UK) was estimated using cost-of-illness methodology applying a prevalence approach (Larg & Moss, 2011). This approach involves estimating the number of people with IRDs in a base period (2019) and the costs attributable to IRDs in that period. The analysis was based on a targeted literature review and primary data (survey) collection.

Prevalence

Overall, the prevalence of IRDs in the UK was estimated at 0.03% or 20,814 prevalent cases in 2019. The highest proportion of the overall prevalence was attributed to RP (49.6%, 10,324 people), followed by Usher syndrome (12.4%, 2,590 people) and Stargardt disease (10.1%, 2,109 people).

Table 1: Prevalence of IRDs in the UK (n, 2019)

IRD	Cases
RP	10,324
Usher syndrome	2,590
Stargardt	2,109
LCA/EOSRD	1,587
Best disease	1,003
Cone dystrophy	876
Cone-rod dystrophy	812
Achromatopsia	733
XLRS	483
Choroideremia	298
Total	**20,814**

Total costs

Total costs attributable to IRDs in the UK were estimated to be £523.3 million in 2019, comprising both economic (£327.2 million) and wellbeing costs (£196.1 million).

Of the ten IRDs within scope, RP incurred the greatest proportion of total costs at £262.3 million (50.1%), followed by Usher syndrome (£64.9 million, 12.4%), Stargardt disease (£52.0 million, 9.9%), and LCA/EOSRD (£38.5 million, 7.4%).

Wellbeing costs comprised the largest share of total costs at £196.1 million (37.5%), followed by productivity costs (£114.1 million, 21.8%), carer costs (£84.5 million, 16.1%), other costs (£74.9 million, 14.3%), deadweight losses (£28.7 million, 5.5%) and health system costs (£25.0 million, 4.8%).

Chart 1: Total costs of IRDs (£ million, 2019) in the UK by condition

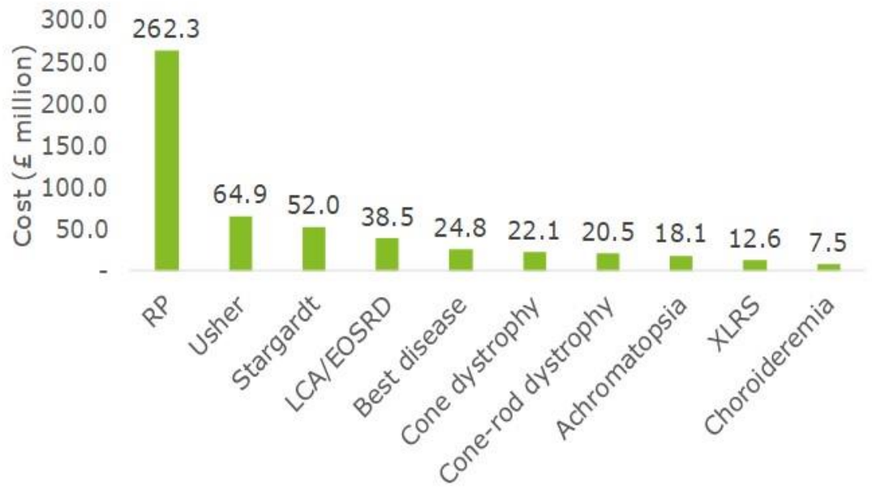


Table 2: Total costs of IRDs (£ million, 2019) in the UK by cost type

Cost type	Percentage (%) of total costs	Cost (£ million)
Wellbeing costs	37.5	196.1
Productivity costs	21.8	114.1
Informal carer costs	16.1	84.5
Other costs	14.3	74.9
Deadweight losses	5.5	28.7
Health system costs	4.8	25.0
Total costs	**100**	**523.3**

Conclusion

IRDs imposed significant economic and wellbeing costs on the UK population in 2019. Persons living with an IRD incur significant economic costs and reductions in their quality of life. Significant economic costs are also borne by families, friends, government, employers and society.

1 Introduction

The socioeconomic burden of inherited retinal dystrophies (IRDs) in the United Kingdom (UK) was estimated using best practice cost-of-illness methodology applying a prevalence approach. The analysis was based on data collected in a targeted literature review and primary (survey) data collection.

1.1 Purpose

Retina International on behalf of the IRD COUNTS consortium engaged Deloitte Access Economics to estimate the disease burden and economic impact of IRDs in the UK and RoI from a societal perspective – a cost-of-illness study. This approach involves estimating the number of people with IRDs in a base period (2019) and the costs attributable to IRDs in that period.

Cost-of-illness studies are conducted with the intent of describing the economic burden imposed by a disease, on a specific population (Larg & Moss, 2011). The proprietary Deloitte Access Economics cost-of-illness methodology estimates both economic and wellbeing costs.

The results from a cost-of-illness study can be used to define the economic burden of a disease, justify investment in preventive or treatment interventions, inform funding allocation and prioritisation, provide a basis for policy and planning, and provide inputs for economic analyses such as cost benefit analyses (CBA); cost effectiveness analyses (CEA), cost utility analyses (CUA) and cost minimisation analyses (CMA) (Rice, 2000; The Joanna Briggs Institute, 2014).

1.2 This report

This report outlines the disease burden and economic impact of IRDs in the UK in 2019.

To facilitate accessibility, non-standard formatting has been used throughout this report. This includes size 14 font and darker headings. In addition, spacing within the tables has been increased and pages containing detailed charts and tables have been converted to landscape.

The remaining sections of this report are set out as follows:

- Chapter 2: Background
- Chapter 3: Methodology
- Chapter 4: Survey results
- Chapter 5: Prevalence
- Chapter 6: Health system costs
- Chapter 7: Individual productivity costs
- Chapter 8: Other costs
- Chapter 9: Wellbeing costs
- Chapter 10: Sensitivity analysis
- Chapter 11: Conclusion
- References
- Appendices.

2 Background

IRDs represent a diverse group of progressive, visually debilitating diseases for which there have traditionally been no effective treatments to restore vision.

IRDs are genetically diverse with over 260 genes currently linked to this group of diseases (Duncan et al, 2018).

This report focuses on the following ten IRDs, a list determined by Retina International and the IRD COUNTS consortium:

- Retinitis pigmentosa (RP)
- Leber congenital amaurosis (LCA)/early onset severe retinal dystrophy (EOSRD)
- Cone dystrophy
- Choroideremia
- Usher syndrome
- Best (vitelliform) macular dystrophy (Best disease)
- Code-rod dystrophy
- Stargardt disease
- X-linked retinoschisis (XLRS)
- Achromatopsia.

2.1 Retinitis pigmentosa (RP)

Retinitis pigmentosa (RP) is a group of chronic, progressive diseases that lead to degeneration of the rod and cone photoreceptors, and the pigmented layer of the retina (retinal pigment epithelium; RPE) (Campochiaro and Mir, 2018). While most cases are non-syndromic, people can experience RP in association with other sensory deficits (syndromic) (Campochiaro and Mir, 2018). Non-syndromic describes conditions that are present not as a result of another syndrome, whereas syndromic describes conditions which occur as a result of another syndrome. Symptoms of RP include impaired night vision (nyctalopia), a loss of peripheral vision and deteriorated central vision (Verbakel et al, 2018).

RP is inherited through autosomal dominant, autosomal recessive or X-linked patterns (Campochiaro and Mir, 2018). There are currently over 80 genes associated with RP, with

most expressed in either the photoreceptors or the RPE cells (Verbakel et al, 2018). Subtypes of RP that commence earlier in life are usually associated with faster progression, while persons with X-linked subtypes tend to experience greater disease severity (Verbakel et al, 2018).

People can undergo genetic testing to determine the presence of genetic mutations associated with RP; however, this is challenging due to significant genetic diversity (Birtel et al, 2019). Other primary diagnostic imaging techniques can be used to diagnose RP and are essential in assessing disease severity and progression (Verbakel et al, 2018).

While there is currently no cure for RP, both surgical and non-surgical (visual aids) approaches can be used to manage symptoms (Fahim et al, 2018). Gene therapy has been authorised by the European Medicines Agency for the treatment of adult and paediatric patients with vision loss due to inherited retinal dystrophy caused by confirmed biallelic RPE65 mutations and who have sufficient viable retinal cells. Gene and stem cell therapies are in the process of being explored as potential forms of treatment for RP (Fahim et al, 2018). Phase I/II trials of gene therapy in affected humans are ongoing (Kumaran et al, 2018a).

2.2 Leber congenital amaurosis (LCA)/early onset severe retinal dystrophy (EOSRD)

Leber congenital amaurosis (LCA) and early onset severe retinal dystrophy (EOSRD) comprise IRDs that are characterised by severe and early visual deterioration (Kumaran et al, 2017). EOSRD usually presents between infancy and five years of age whereas LCA presents between birth and the first few months of life (Kumaran et al, 2018b).

LCA/EOSRD is typically inherited via an autosomal recessive pattern, though an autosomal dominant inheritance pattern can occur in rare cases (Kumaran et al, 2018b). Presently, 25 genes responsible for LCA/EOSRD have been identified; accounting for 70-80% of cases (Kumaran et al, 2017).

Symptoms commonly associated with LCA/EOSRD include early-onset visual impairment, oscillations of the eye (nystagmus), slow/absent pupillary reactions, heightened light

sensitivity (photophobia), farsightedness (hyperopia) and cornea bulging (Kumaran et al, 2017). There are also a number of non-ocular symptoms associated with LCA/EOSRD such as neurodevelopmental delays, intellectual disability, autism and olfactory dysfunction (Chacon-Camacho and Zenteno, 2015).

LCA/EOSRD can be screened for through genetic testing which aims to detect pathogenic variations in the genes most commonly associated with the conditions (Kumaran et al, 2018b). Other diagnostic tools can be used to assess symptoms of LCA/EOSRD (Kumaran et al, 2018b).

Treatment of LCA/EOSRD currently focuses on symptom management such as the correction of refractive error and provision of low vision supports (Kumaran et al, 2018b). Gene therapy has been authorised by the European Medicines Agency for the treatment of adult and paediatric patients with vision loss due to inherited retinal dystrophy caused by confirmed biallelic RPE65 mutations and who have sufficient viable retinal cells.

Ongoing clinical trials aim to determine whether cone cell function can be protected using gene delivery at an early disease stage (Kumaran et al, 2018c). Stem cell therapy is also being investigated as a possible therapeutic option (Kumaran et al, 2018c).

2.3 Cone dystrophy

Cone dystrophy is a progressive IRD characterised by gradual degeneration of the cone photoreceptors (Sisk et al, 2018). Six genes are responsible for the vast majority of cone dystrophy cases; inherited in an autosomal dominant or autosomal recessive pattern (Gill et al, 2019).

Symptoms that most commonly present due to cone dystrophy include difficulties in adjusting to bright light (hemeralopia), colour vision disturbances, reduced visual acuity and nystagmus (Sisk et al, 2018). Persons with cone dystrophy present as either stable or progressively declining. The former typically present with cone dystrophy symptoms during early childhood and remain stable. Progressive cone dystrophy usually develops in late childhood or during early adulthood and worsens over time (Gill et al, 2019).

People can be screened for cone dystrophy disease-causing genes using molecular genetic testing (Gill et al, 2019). There are several diagnostic imaging techniques that can be used to diagnose cone dystrophy (Sisk et al, 2018).

There are currently no treatments available for slowing progression or restoring visual impairment. Management relies on the alleviation of visual symptoms (Gill et al, 2019).

2.4 Choroideremia

Choroideremia is an IRD that causes progressive loss of vision due to degeneration of the choroid, retinal pigment epithelium and photoreceptors (Tsang and Sharma, 2018). Choroideremia is inherited in an X-linked recessive pattern (MacDonald et al, 2015). Males are affected by X-linked recessive disorders much more frequently than females. Early disease manifests as night-blindness and peripheral visual field defects with onset typically in the first two decades of life. As the disease progresses, the visual field constricts relatively symmetrically. Visual acuity is preserved until advanced disease. Symptoms of choroideremia include tunnel vision, night blindness (nyctalopia) and can progress to central blindness (MacDonald et al, 2015).

Genetic testing can be used to screen for the presence of the choroideremia (CHM) gene mutation (MacDonald et al, 2015). Diagnosis of choroideremia is undertaken through diagnostic imaging technologies (Coussa and Traboulsi, 2012).

Current approaches to choroideremia management focus on symptom alleviation through surgical and non-surgical methods (for example, dietary modification and visual aids) (MacDonald et al, 2015). Initial clinical trial results of gene therapy have shown that rod and cone function may be restored, this potential treatment is still under investigation and, as such, is not available to the general population (Kumaran et al, 2018a; Mitsios et al, 2018).

2.5 Usher syndrome

Usher syndrome is an IRD characterised by hearing loss and deteriorating visual function. It is the leading genetic cause of deaf-blindness, representing half of all cases (Mathur and Yang, 2019). Usher syndrome is inherited in an autosomal

recessive pattern with 11 genes currently linked to the condition (Mathur and Yang, 2019).

The visual symptoms of Usher syndrome are identical to those associated with RP. These symptoms include nyctalopia, dark adaptation difficulties, abnormal electroretinography (ERG) responses, peripheral vision loss, abnormal retinal pigmentation, myopia and blindness (Mathur and Yang, 2015). People will also experience a loss of auditory function and may encounter difficulties with balance (Mathur and Yang, 2015).

Usher syndrome can be screened for the presence of chromosomal mutations most commonly associated with the disease (Mathur and Yang, 2015). Diagnosis of the condition can occur through diagnostic imaging modalities (Yang et al, 2012).

There are three major clinical subtypes of Usher syndrome. Persons with Type I (USH1) are characterised as having congenital severe-to-profound deafness, vestibular dysfunction and onset of RP within the first decade of life. While Type II (USH2) persons have congenital moderate-to-severe hearing loss, normal vestibular function and onset of RP within the second decade of life. For USH3 persons, hearing loss, vestibular dysfunction and onset of RP are progressive, sporadic and variable, respectively (Mathur and Yang, 2015).

While the auditory symptoms of the condition can be managed through the use of aid devices, there remains no treatment for the visual component of the condition (Mathur and Yang, 2019).

Viral-mediated gene replacement therapy is emerging as a potential approach to prevent a person from progressing to total blindness resulting from Usher syndrome, though the enormity of the causative genes makes their delivery difficult (Mathur and Yang, 2015; Takahashi et al, 2018).

2.6 Best disease

Best disease is an IRD characterised by a bilateral yellow lesion in the macula which changes over time (Parodi et al, 2014). Hereditary factors present as the primary risk for the condition

with disease-causing gene inherited in an autosomal dominant pattern (Budiene et al, 2014).

The symptoms associated with Best disease include slow decline in visual acuity, worsening central vision and occasionally, distorted vision of straight lines (metamorphopsia) (Zerbib et al, 2016).

Genetic testing can be used to screen for the presence of the BEST1 gene mutation (Parodi et al, 2014). Best disease can be diagnosed using imaging techniques (Budiene et al, 2014).

There are no treatments available to prevent the onset or progression of Best disease, though symptoms can be managed through the use of visual aids (Budiene et al, 2014). Clinical trials, using gene and cell-based therapies, are ongoing (Yang et al, 2015).

2.7 Cone-rod dystrophy

Cone-rod dystrophy is an IRD that is part of the group of pigmentary retinopathies characterised by progressive degeneration of the cone and rod cells (Durlu et al, 2014). The symptoms of cone-rod dystrophy include poor visual acuity, light sensitivity (photophobia), nystagmus, colour blindness (dyschromatopsia) and impaired night vision (nyctalopia) (Gill et al, 2019). Cone-rod dystrophy differs from cone dystrophy in that it is characterised by earlier degeneration of rod photoreceptors (Thiadens, 2012).

People can undergo genetic testing to determine the presence of genetic mutations associated with cone-rod dystrophy. Further, the disease can be diagnosed using various imaging techniques (Gill et al, 2019).

The prognosis of cone-rod dystrophy is typically more severe and rapid relative to other IRDs, leading to earlier total blindness. However, the age of onset varies ranging from the second to the fifth decade of life (Thiadens et al, 2012).

There are no existing treatments to prevent the progression of cone-rod dystrophy (Gill et al, 2019). Care strategies focus on alleviating the symptoms, including refractive correction, tinted lenses for photophobia and low vision aids (Gill et al, 2019).

2.8 Stargardt disease

Stargardt disease is the most prevalent inherited macular dystrophy. It is characterised by rapid deterioration of central vision and a progressive bilateral atrophy of the RPE and neuroepithelium (Tanna et al, 2017). People with the disease experience deterioration of retinal rod and cone photoreceptors, atrophy of the macular RPE, and macular degeneration (Han et al, 2014). Stargardt disease leads to a decline in central visual including an inability to perceive colours (dyschromatopsia) and the emergence of blind spots (scotoma) (Tanna et al, 2017).

ABCA4 is the primary gene associated with Stargardt disease, though there are over 900 disease-causing variants identified in the gene (Tanna et al, 2017). As the gene is inherited in an autosomal recessive pattern, hereditary factors are central to assessing the risk of disease presentation (Tanna et al, 2017).

Genetic testing for Stargardt disease is particularly difficult due to the genetic diversity of the disease (Tanna et al, 2017). Retinal imaging technologies can be used as a diagnostic tool (Ergun et al, 2015).

There is currently no treatment for Stargardt disease, though gene therapy is emerging as a potential option (Han et al, 2014; Kumaran et al, 2018a). Limiting exposure to UV light by wearing UV screening glasses and limiting intake of vitamin A is advised as part of disease management strategies. Stem cell therapies are also being investigated (Tanna et al, 2017). In addition, two late stage clinical trials investigating an oral therapy are underway (Fighting Blindness Foundation, 2019).

2.9 X-linked retinoschisis (XLRS)

X-linked retinoschisis (XLRS) is a rare congenital malformation of the retina characterised by splitting within inner retinal layers, including the nerve fibre layer (Rao et al, 2018). The primary risk factor for XLRS is the RS1 gene, which is inherited in an X-linked manner (Sieving et al, 2014).

XLRS is characterised by central vision loss, nystagmus and/or strabismus (crossed eyes). More severe cases will exhibit symptoms of vitreous haemorrhage, retinal detachment and/or

glaucoma, which can result in extensive vision loss (Vincent et al, 2013).

Diagnosis of XLRS can be made using retinal imaging techniques (Vincent et al, 2013; Sieving et al, 2014). Presently, no cure exists for XLRS but gene replacement therapy is being explored (Kumaran et al, 2018a).

Management of XLRS generally relies on pharmacological or vision-aid interventions (Molday et al, 2012). Surgical options may also be utilised to address more severe complications associated with the condition, such as retinal detachment (Rao et al, 2018).

2.10 Achromatopsia

Achromatopsia is an inherited macular dystrophy characterised by dysfunction or the absence of cone photoreceptors (Yu et al, 2014). While most people experience complete achromatopsia, with no function present in all cones, some people have incomplete achromatopsia in which one or more cone types remains functional (Kohl et al, 2018).

Achromatopsia is diverse with people displaying a range of symptoms; however, the primary symptom is a partial or total absence of colour vision (Hirji et al, 2018). Other symptoms may include photophobia, nystagmus, and poor visual acuity (Hirji et al, 2018).

The genes associated with achromatopsia are inherited in an autosomal recessive pattern indicating that hereditary factors are most significant risk factor for the condition (Kohl and Hammer, 2013). Achromatopsia can be screened for using molecular genetic testing approaches (Kohl et al, 2018). Diagnosis can occur using a variety of imaging techniques (Kohl et al, 2018).

While there is currently no cure for achromatopsia, early phase clinical trials are currently being undertaken to determine the extent to which gene therapy might benefit visual acuity, colour discrimination, photophobia and nystagmus (Kumaran et al, 2018a). Strategies available to manage the symptoms associated with the condition include the use of special-filter glasses and low vision aids (Kohl et al, 2018).

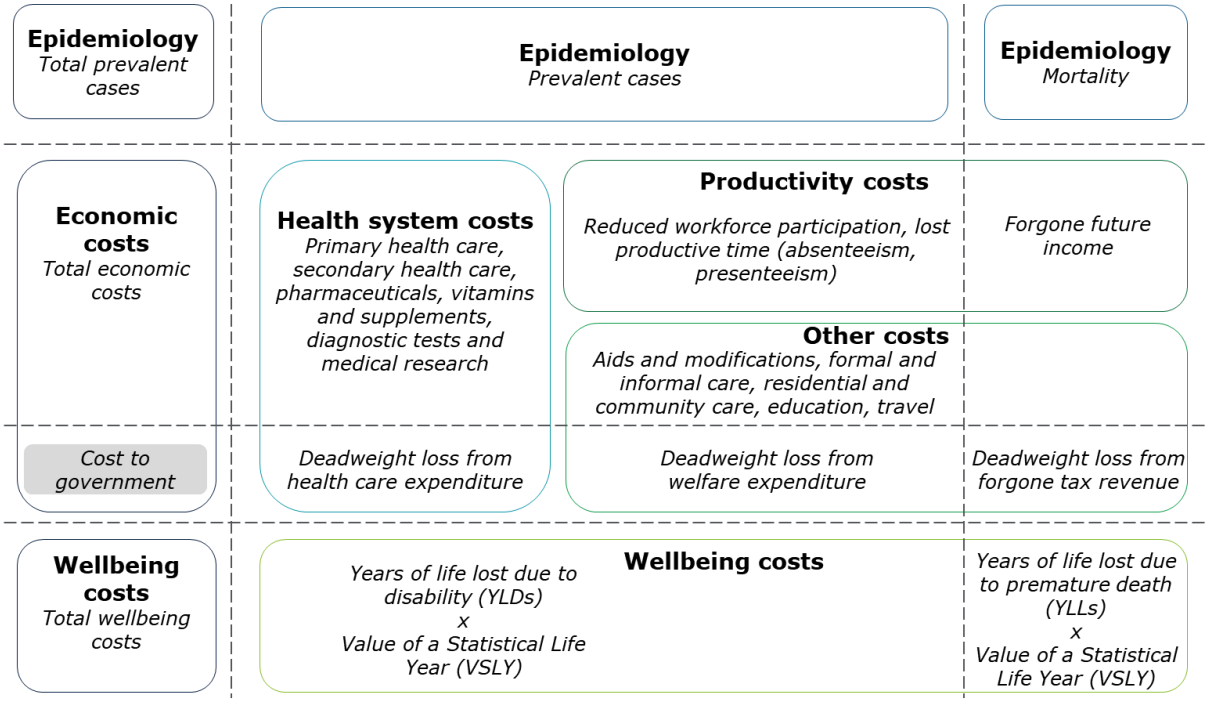
3 Methodology

This section describes the cost-of-illness methodology used to estimate the socioeconomic burden of IRDs in the UK in 2019.

The socioeconomic burden of IRDs in the UK was estimated using cost-of-illness methodology applying a prevalence approach (Larg & Moss, 2011).

This approach involves estimating the number of persons living with an IRD in a base period (2019) and the costs attributable to IRDs in that period. Figure 3.1 below provides an overview of the cost-of-illness model. The analysis was based on a targeted literature review and primary data (survey) collection, described in the following sub-sections.

Figure 3.1: Cost-of-illness model overview



Source: Deloitte Access Economics.
Note: There is a lack of available data on the risk of premature mortality attributable to IRDs. As such, no costs associated with premature mortality were estimated.

3.1 Costing methodology

Costs were categorised by type, as:

- Costs to the health system including primary and secondary health care, pharmaceuticals, vitamins and supplements, diagnostic tests and medical research.
- Individual productivity losses due to reduced workforce participation, absenteeism and presenteeism (reduced productivity while at work).
- Other costs such as aids and modifications, formal and informal care, residential and community care, education, travel and deadweight losses (society-wide efficiency losses arising from loss of taxation revenue due to reduced workforce participation and higher government expenditures).
- Wellbeing cost measured by years of healthy life lost to IRDs.

Different costs of disease are borne by different individuals or sectors of society. Understanding how costs are shared helps to make informed decisions regarding interventions. While persons living with an IRD are most severely affected by the condition, family members and other parts of society also face costs attributable to IRDs.

This analysis estimated first round societal impacts. No second round or longer term dynamic impacts were modelled (i.e. changes in wages or labour market outcomes associated with the economic burden of IRDs).

Component figures or calculations may not sum to totals due to rounding.

3.2 Targeted literature review

A targeted review of the scientific literature and publicly available databases was conducted to identify the most relevant inputs for this report. The review involved a targeted (non-systematic) search of the PubMed and Cochrane Library databases to:

- Identify the target indication (IRDs)
- Identify epidemiological parameters of the target indication
- Identify economic parameters of the target indication.

Additional ad-hoc searches were performed, as required. Where possible, inputs specific to IRDs and the UK were used. Where this was not possible, inputs from similar populations in other countries, and diseases of comparable severity and symptoms, but different aetiology, were used.

3.3 Primary data (survey) collection and analysis

A survey was distributed to persons living with an IRD and/or the parents of children (under 18) living with an IRD to:

- Collect more specific and detailed primary data regarding the health care resource utilisation and productivity impacts of persons living with an IRD and the parents of children (under 18) living with an IRD, attributable to IRDs.
- Address any data gaps identified in the targeted literature review.

Retina International and the IRD COUNTS consortium identified relevant stakeholders for recruitment. Participation was restricted to persons living with an IRD and the parents of children (under 18) living with an IRD. Ethical and patient consent procedures complied with all requirements set out in the European Union General Data Protection Regulations (GDPR), and ethical and compliance obligations. Please refer to Section 4 for a breakdown of persons living with an IRD and/or the parents of children (under 18) living with an IRD invited to complete the survey, and responses received.

The survey questionnaire was co-designed with Retina International and the IRD COUNTS consortium, and with reference to the metrics identified in the targeted literature review. Skip logic was incorporated into the programming of the survey to ensure only relevant questions would be asked of participants, depending on the answers provided to previous questions.

Survey responses were cleaned for inappropriate responses including attributing other related costs in questions about specific cost items. For example, referring to travel costs when attending the doctor, travel costs are separately captured in specific questions and, hence, these responses were removed to avoid double counting. Each free text question was further analysed for response variance, outliers were removed and

response from the UK and RoI were pooled to reduce variance were appropriate.

3.4 Sensitivity analysis

In many cases, the inputs underlying the cost-of-illness analysis are uncertain and changes in these inputs may have a significant impact upon the total estimate of the costs of IRDs in the UK in 2019. Published population prevalence estimates of IRDs (overall or condition-specific) are limited. Recent and nationwide prevalence estimates for IRDs in the UK were not identified.

Given the limited published data on the prevalence of IRDs in the UK, sensitivity analysis using an upper bound sensitivity estimate based on the highest published prevalence estimate available was undertaken.

4 Survey results

This section describes the results from the survey of persons living with an IRD and/or the parents of children (under 18) living with an IRD in the UK.

Key findings

- Male UK respondents comprised a slight majority (male: 52.8%, female: 47.2%).
- Respondents from the UK were largely adults over the age of 18 (83%), 17% of responses were received from parents or carers of a child under 18.
- The survey sample was skewed towards older age groups with people aged under 35 making up less than 20% of the sample.
- Over half (56.6%) of persons living with an IRD in 2019 had received a genetic test for their IRD during their lifetime.
- Most persons living with an IRD (62.3%) reported at least one member of their family had seen a specialist to check for IRD symptoms.

4.1 Summary of approach

A survey was distributed to persons living with an IRD and/or the parents of children (under 18) living with an IRD to:

- Collect more specific and detailed primary data regarding the health care resource utilisation and productivity impacts of persons living with an IRD, and the parents of children (under 18) living with an IRD attributable to IRDs.
- Address any data gaps identified in the targeted literature review.

129 survey responses were received from persons living with an IRD and parents of children (under 18) living with an IRD across the UK and RoI.

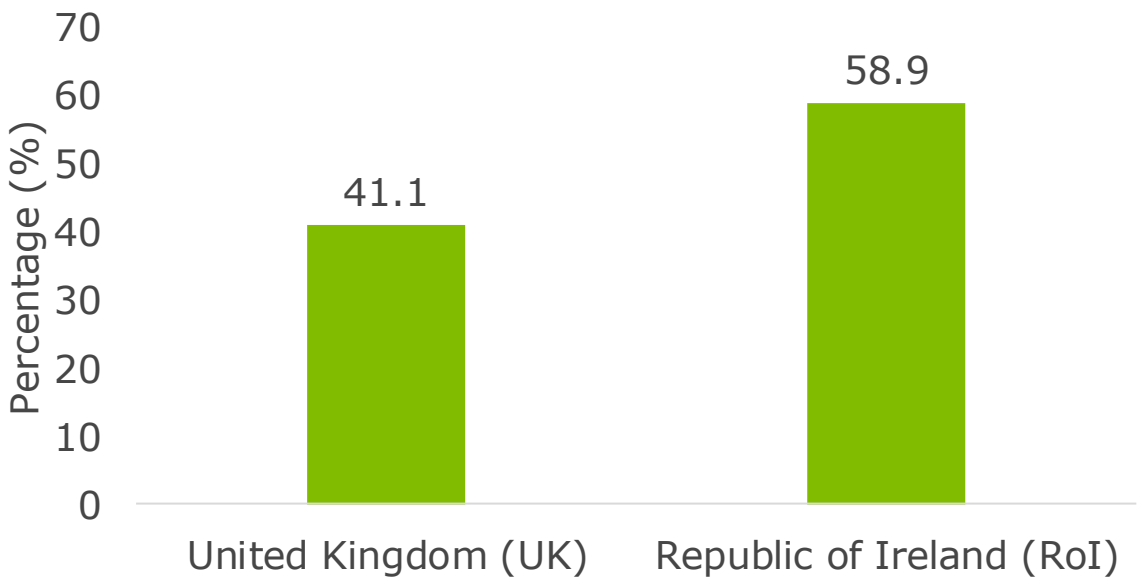
Please refer to Section 3.3 for more detail regarding the survey methodology.

4.2 Participant characteristics

Of the 129 people who responded to the survey, 41.1% were from the UK and 58.9% were from the RoI (Chart 4.1 and Table 4.1).

The remainder of this subsection reports the characteristics of survey participants from the UK.

Chart 4.1: Distribution of survey participants by country (n=129)



Source: Deloitte Access Economics analysis.

Table 4.1: Distribution of survey participants by country (n=129)

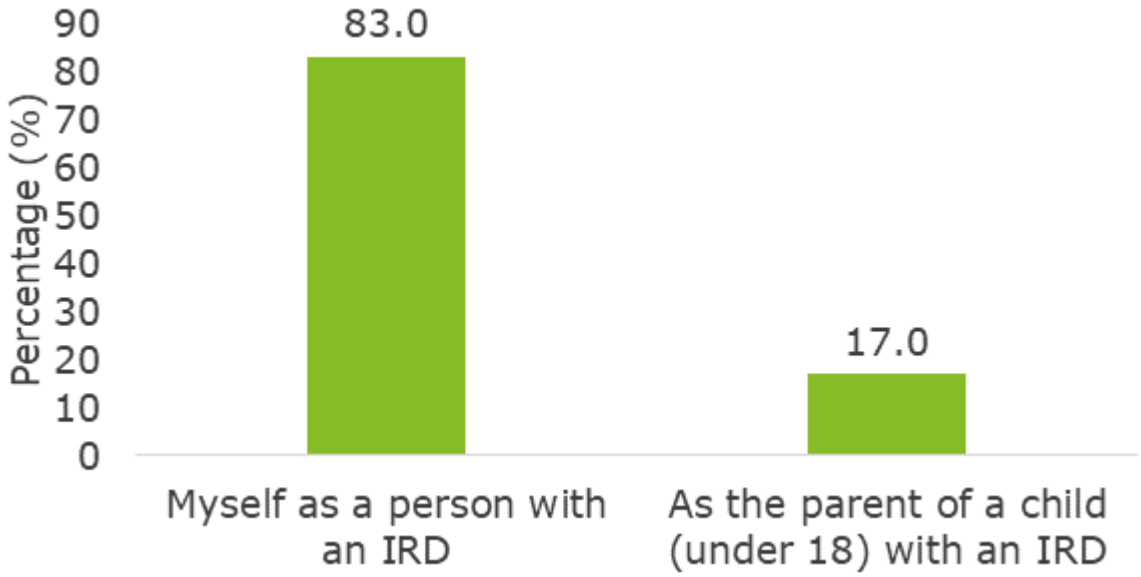
Country	Count (n)	Percentage (%)
Republic of Ireland (RoI)	76	58.9
United Kingdom (UK)	53	41.1
Total	**129**	**100.0**

Source: Deloitte Access Economics analysis.

4.2.2 Participant type (person living with an IRD or parent of a child with an IRD)

Of the 53 UK respondents, 83% were persons living with an IRD and 17% were the parents of children (under 18) living with an IRD (Chart 4.2 and Table 4.2).

Chart 4.2: Distribution of UK survey participants by type (person living with an IRD or parent of a child with an IRD) (n=53)



Source: Deloitte Access Economics analysis.

Table 4.2: Distribution of UK survey participants by type (person living with an IRD or parent of a child with an IRD) (n=53)

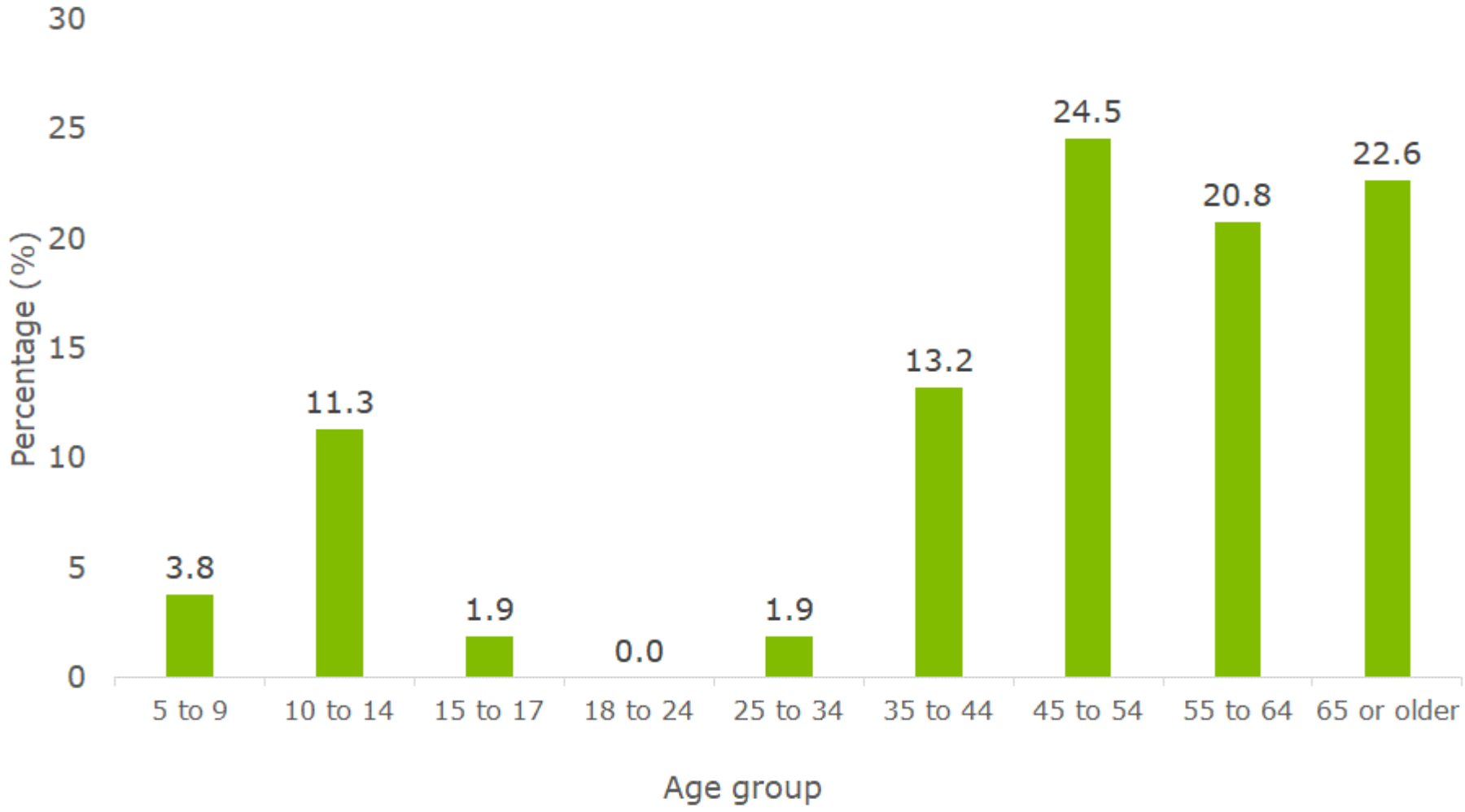
Participant type (Adult or Parent)	Count (n)	Percentage (%)
Myself as a person with an IRD	44	83.0
As the parent of a child (under 18) living with an IRD	9	17.0
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

4.2.3 Age

The survey sample is skewed towards older persons living with an IRD with the majority of UK respondents aged 45 years and older. Multiple five year age groups have been aggregated to preserve participant confidentiality. The 45 to 54 year old age group received the greatest number of responses (24.5%) (Chart 4.3 and Table 4.3).

Chart 4.3: Distribution of UK survey participants by age (n=53)



Source: Deloitte Access Economics analysis.

Table 4.3: Distribution of UK survey participants by age (n=53)

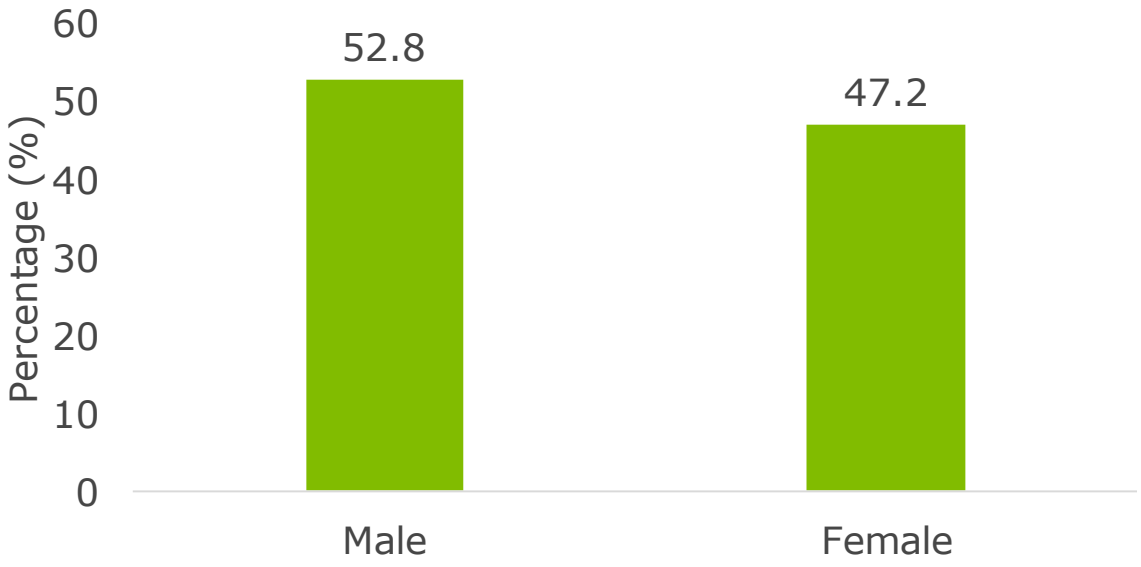
Age group	Count (n)	Percentage (%)
34 or younger	10	18.9
35 to 44	7	13.2
45 to 54	13	24.5
55 to 64	11	20.8
65 or older	12	22.6
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

4.2.4 Sex

Male UK respondents comprised a slight majority (male: 52.8%, female: 47.2%) (Chart 4.4 and Table 4.4).

Chart 4.4: Distribution of UK survey participants by sex (n=53)



Source: Deloitte Access Economics analysis.

Table 4.4: Distribution of UK survey participants by sex (n=53)

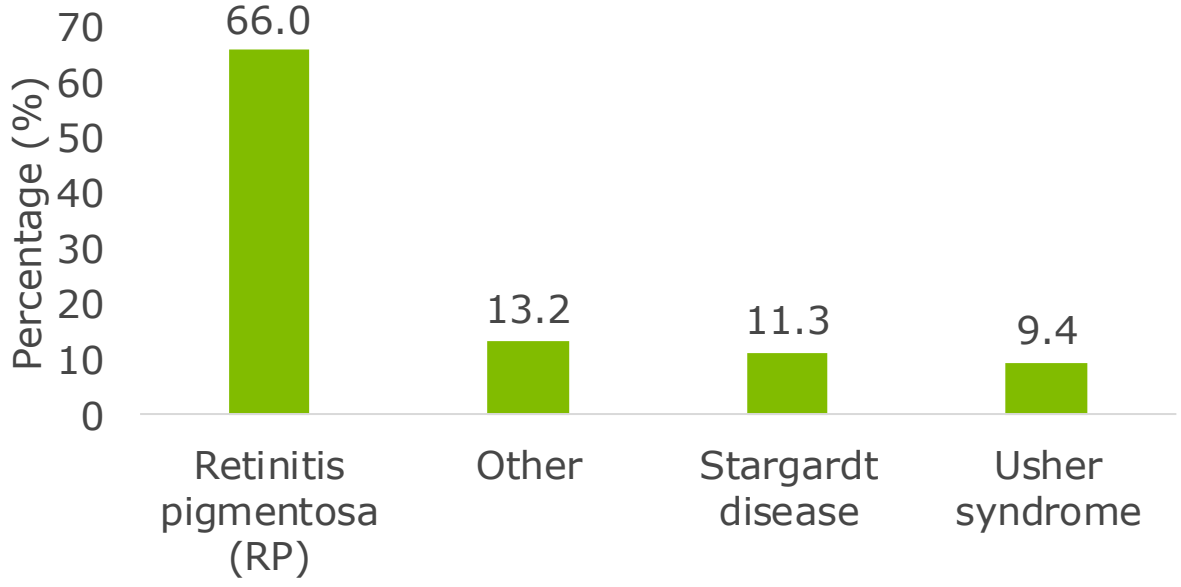
Sex	Count (n)	Percentage (%)
Male	28	52.8
Female	25	47.2
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

4.2.5 Condition

Survey respondents with RP were in the majority (66.0%), followed by Stargardt disease (11.3%) and Usher syndrome (9.4%). There was a small response rate for achromatopsia, cone dystrophy, choroideremia and LCA. Together they accounted for 13.2% of the IRDs reported (Chart 4.5 and Table 4.5). There were no responses across either of the two countries for Best disease or XLRS.

Chart 4.5: Distribution of survey participants by condition in the UK (n=53)



Source: Deloitte Access Economics analysis.

Table 4.5: Distribution of survey participants by condition in the UK (n=53)

IRD	Count (n)	Percentage (%)
RP	35	66.0
Other	7	13.2
Stargardt disease	6	11.3
Usher syndrome	5	9.4
Total	**53**	**100.0**

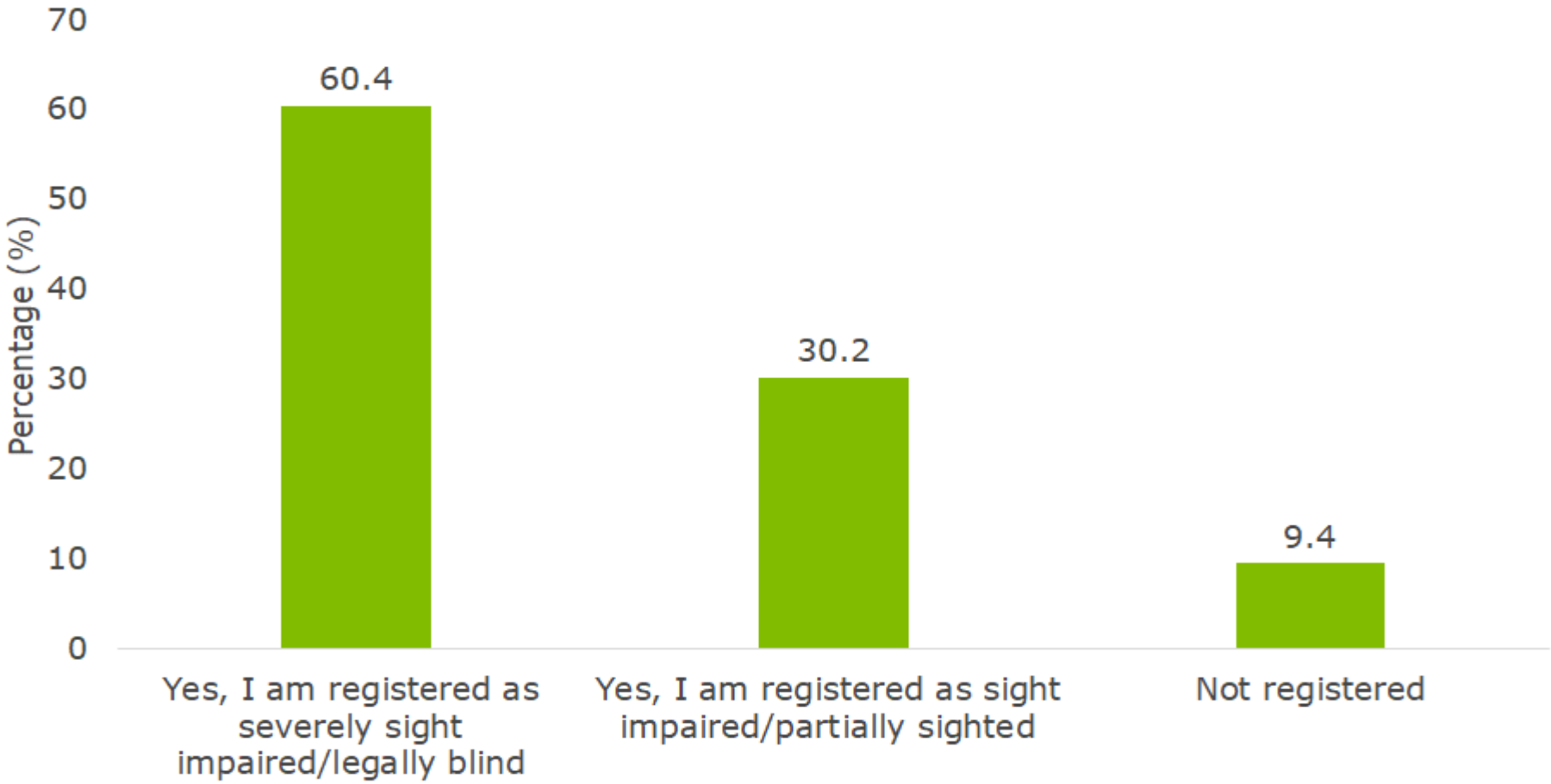
Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

4.2.6 Visual impairment registration

The majority of UK respondents (90.6%) reported they were legally registered with 60.4% registered as severely sight impaired/legally blind and 30.2% sight impaired/partially sighted.[1] Those who were not registered (11.6%) included participants who chose not to be registered and participants whose sight loss did not meet the criteria (Chart 4.6 and Table 4.6). Most of the participants who were not registered reported they had good central vision.

[1] Legally blind was defined as 'best corrected visual acuity of less than 3/60, or less than 6/60 with the presence of contracted field of vision'. Partially sighted was defined as 'best corrected visual acuity of less than 6/60 and greater than 3/60 with full visual field, or less than 6/24 with moderate contraction of visual field, or less than 6/18 with marked contraction of visual field'.

Chart 4.6: Distribution of survey participants by visual impairment registration status in the UK (n=53)



Source: Deloitte Access Economics analysis.

Table 4.6: Distribution survey participants by visual impairment registration status in the UK (n=53)

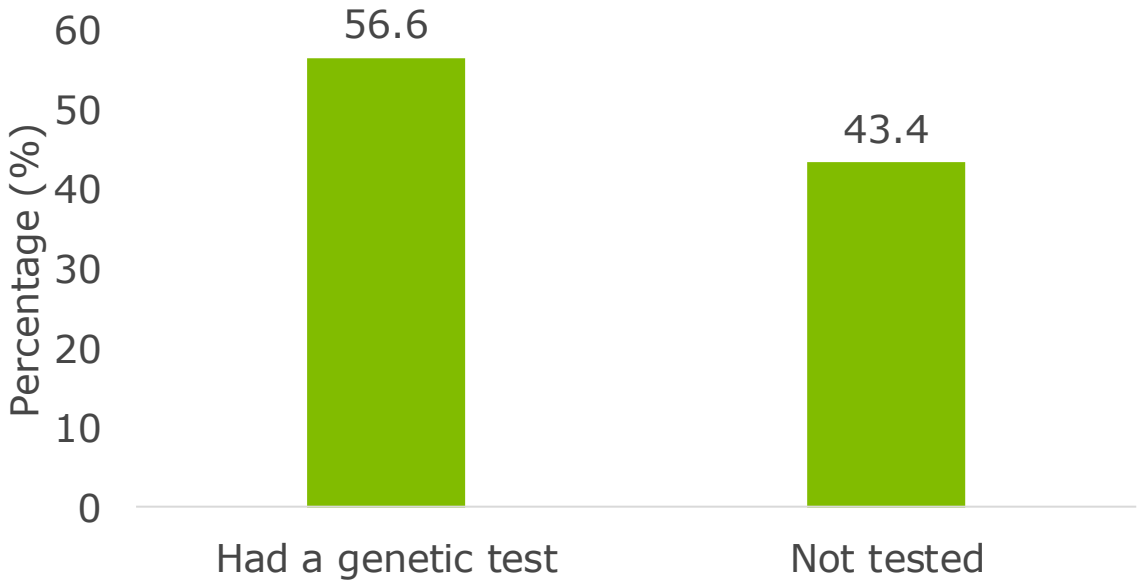
Visual impairment	Count (n)	Percentage (%)
Yes, I am registered as severely sight impaired/legally blind	32	60.4
Yes, I am registered as sight impaired/partially sighted	16	30.2
Not registered	5	9.4
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

4.2.7 Genetic testing

In some instances, persons living with an IRD can receive a genetic test to investigate the underlying genetic cause(s) of their specific IRD. Just over half (56.6%) of survey participants reported they had received a genetic test for the purpose of diagnosing their IRD. (Chart 4.7 and Table 4.7).

Chart 4.7: Genetic test utilisation of survey participants in the UK (n=53)



Source: Deloitte Access Economics analysis.

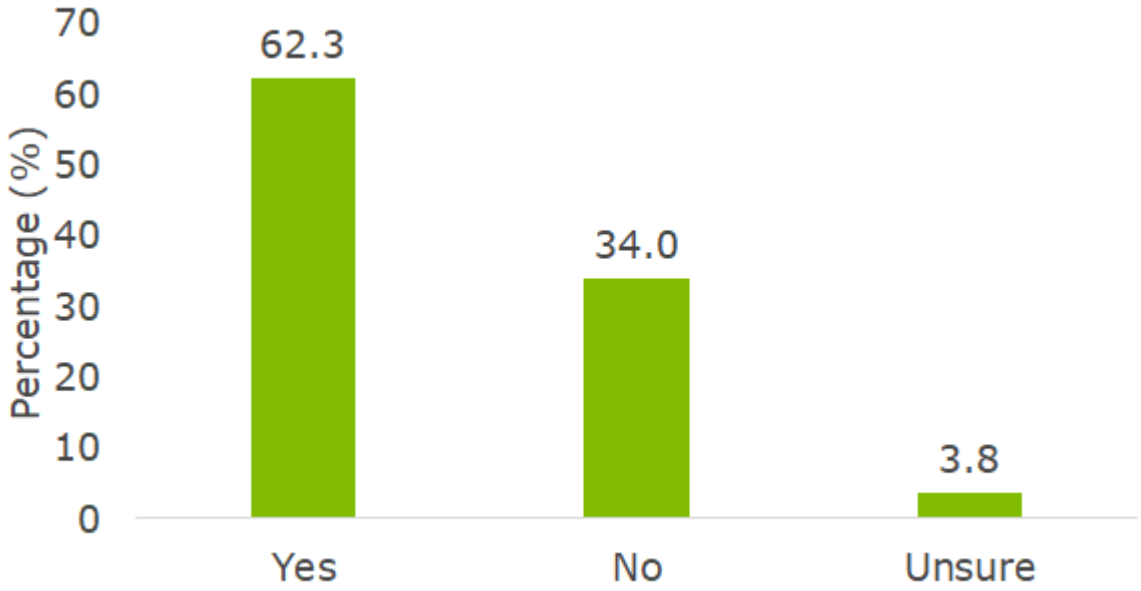
Table 4.7: Genetic test utilisation of survey participants in the UK (n=53)

Genetic test status	Count (n)	Percentage (%)
Had a genetic test	30	56.6
Not tested	23	43.4
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Most persons living with an IRD (62.3%) reported at least one member of their family had seen a specialist to check for IRD symptoms (Chart 4.8 and Table 4.8).

Chart 4.8: Distribution of familial access to specialist care for IRDs in the UK (n=53)



Source: Deloitte Access Economics analysis. Note: Components may not sum to totals due to rounding.

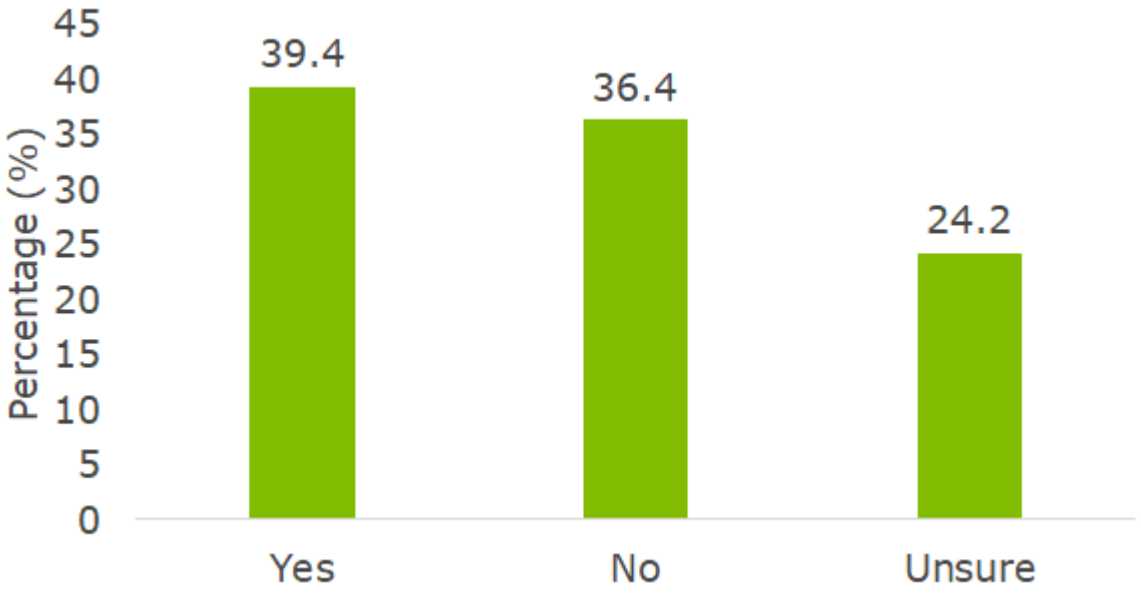
Table 4.8: Distribution of familial access to specialist care for IRDs in the UK (n=53)

Family has sought specialist advice	Count (n)	Percentage (%)
Yes	33	62.3
No	18	34.0
Unsure	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

39.4 per cent of respondents who had family members that sought advice also received genetic testing for an IRD (Chart 4.9 and Table 4.9).

Chart 4.9: Distribution of familial access to genetic testing for IRDs in the UK (n=33)



Source: Deloitte Access Economics analysis. Note: Components may not sum to totals due to rounding.

Table 4.9: Distribution of familial access to genetic testing for IRDs in the UK (n=33)

Family genetic test	Count (n)	Percentage (%)
Yes	13	39.4
No	12	36.4
Unsure	8	24.2
Total	**33**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

5 Prevalence

The prevalence of IRDs refers to the proportion of a population who have IRDs in a given time period. This section describes the approach used to estimate the prevalence of IRDs by age and sex in the UK in 2019.

Key findings

- The overall prevalence of IRDs was estimated to be 0.03% which represented 20,814 cases in the UK in 2019.
- Across the 10 IRDs included in this analysis, RP contributed the highest proportion of cases at 49.6%, or 10,324 cases in the UK.
- The lowest proportion of cases was for choroideremia (1.4%) with 298 cases in the UK.
- Prevalence of IRDs was estimated to be slightly higher in males (52.4%) and tended to increase with age up to the 60-69 year age group.

5.1 Summary of approach

Published population prevalence estimates of IRDs (overall or condition-specific) are limited. Recent and nationwide prevalence estimates for IRDs in the UK were not identified. National registry data on the prevalence of five out of the 10 IRDs was identified from a population-based study conducted in Denmark (Bertelsen et al, 2014). This included a prevalence estimate of RP, LCA/EOSRD, cone-rod dystrophy, choroideremia, and Usher syndrome. In addition, an observational study of Best disease conducted in Denmark informed the estimated population prevalence of Best disease in the UK (Bitner et al, 2012).

To estimate the prevalence of the remaining IRDs (cone dystrophy, Stargardt disease, XLRS, and achromatopsia) included in this analysis, the relative prevalence of the identified IRDs was derived from published results of the Target 5000 study conducted in Ireland and applied to the

relative prevalence of the remaining IRDs (Dockery et al, 2017; Bocquet et al, 2013).

Age and sex specific prevalence rates for the IRDs were utilised where they were available from the literature. Data from the eyeGENE registry, which reports on a sample of 4,635 participants with various IRDs in the United States, was used to inform the sex distribution in the absence of condition-specific literature (National Eye Institute, 2019).

The number of IRD cases in the UK in 2019 was estimated by applying prevalence rates to population data sourced from the UK Office for National Statistics (ONS).

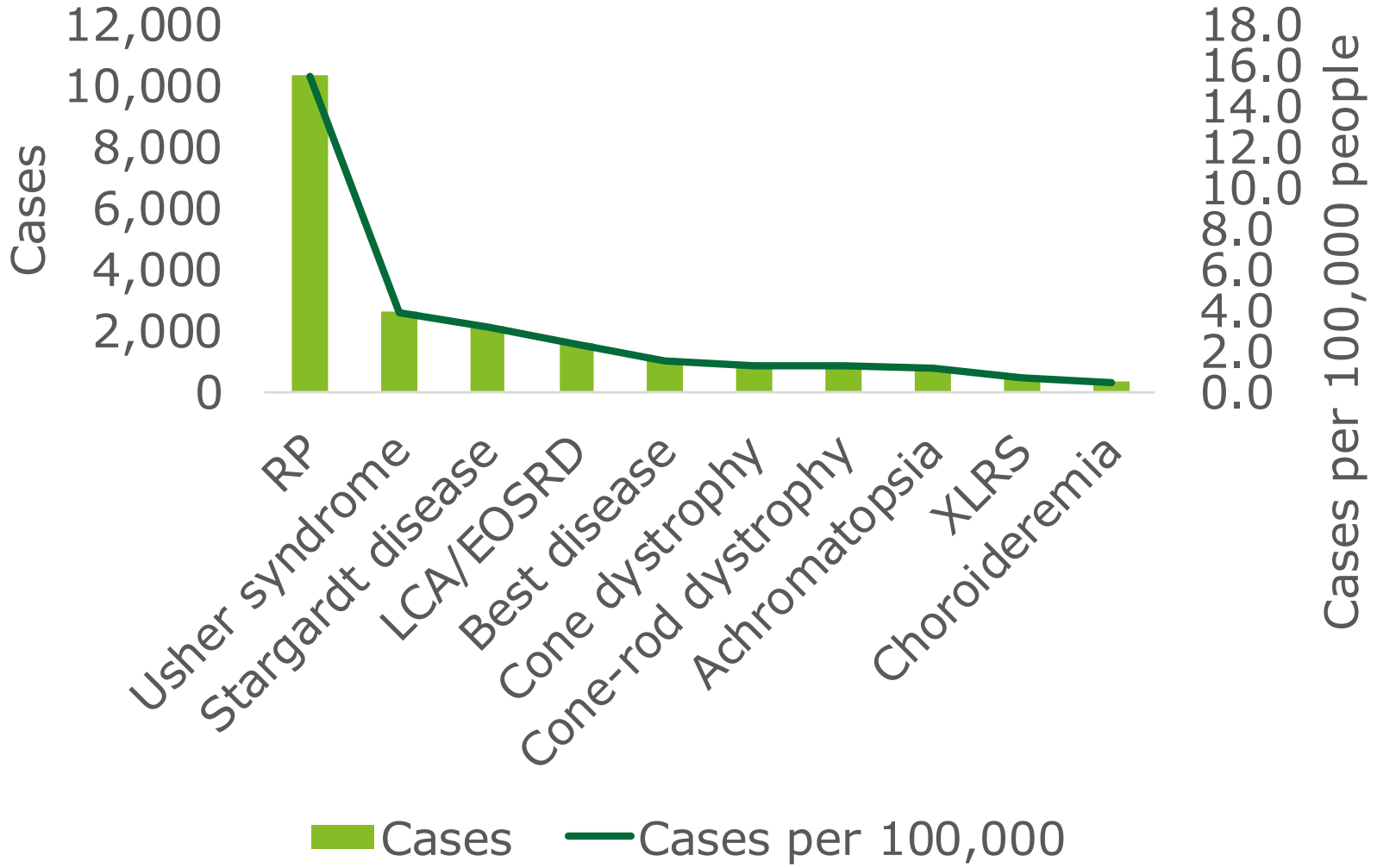
Please refer to Appendix B for further detail regarding the sources informing overall prevalence, and age and sex distribution estimates by condition.

5.2 Results

Overall, the prevalence of IRDs in the UK was estimated at 0.03% or 20,814 prevalent cases in 2019. The highest proportion of the overall prevalence was attributed to RP (49.6%, 10,324 people), followed by Usher syndrome (12.4%, 2,590 people) and Stargardt disease (10.1%, 2,109 people). Chart 5.1 and Table 5.1 provide detailed prevalence estimates by condition, including the prevalence rate, number of cases, and proportion represented by each condition.

Overall prevalence of IRDs in the UK by age and sex is shown in Chart 5.2. The prevalence rate of IRDs increases progressively with age until the 60-69 year age group, after which prevalence gradually declines. Overall prevalence is slightly higher in males (52.4%) compared to females (47.6%).

Chart 5.1: Prevalence of IRDs in the UK (2019) by condition



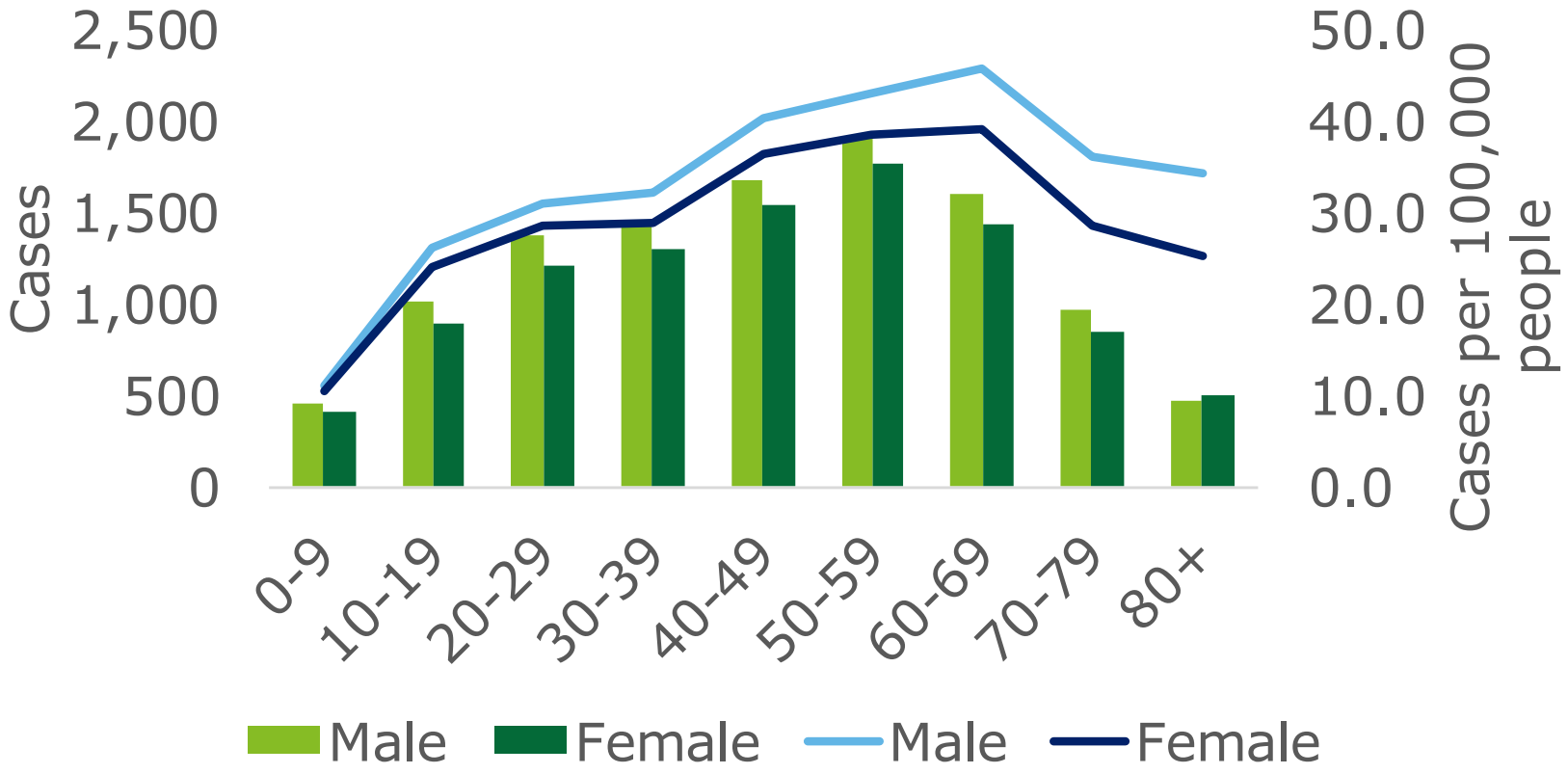
Source: Deloitte Access Economics analysis.

Table 5.1: Prevalence of IRDs in the UK (2019) by condition

IRD	Rate (%)	Cases	Proportion (%)
RP	0.0154	10,324	49.6
Usher syndrome	0.0039	2,590	12.4
Stargardt	0.0032	2,109	10.1
LCA/EOSRD	0.0024	1,587	7.6
Best disease	0.0015	1,003	4.8
Cone dystrophy	0.0013	876	4.2
Cone-rod dystrophy	0.0012	812	3.9
Achromatopsia	0.0011	733	3.5
XLRS	0.0007	483	2.3
Choroideremia	0.0004	298	1.4
Total IRDs	**0.0311**	**20,814**	**100.0**

Source: Deloitte Access Economics analysis based on published literature.
Note: Components may not sum to totals due to rounding.

Chart 5.2: Prevalence of IRDs in the UK (2019) by age and sex



Source: Deloitte Access Economics analysis.

6 Health system costs

This section describes the approach used to estimate the health system costs – including primary and secondary health care, diagnostic tests, pharmaceuticals, vitamins and supplements, and medical research – associated with IRDs in the UK in 2019.

Key findings

- Total health system costs associated with IRDs in the UK in 2019 were estimated to be £25.0 million.
- The largest component of health system costs was secondary health care (£9.0 million), followed by pharmaceuticals (£4.6 million). The least proportion of cost was accounted for by vitamins and supplements at £0.7 million.

6.1 Summary of approach

Health system costs include primary and secondary care, diagnostic tests, pharmaceuticals, vitamins and supplements, and medical research. Health system costs in the UK are primarily paid for by government; however, other sources include out-of-pocket patient payments and funding from other parties such as private health insurers.

The targeted literature review identified no sources estimating the health system cost of IRDs in the UK. International literature was also scarce. The approaches and evidence for estimating categories of health system costs are described in each subsection below. Where available, utilisation of health services was derived from country specific survey results and unit costs were sourced from national reference prices.

While survey data collected provides an estimate of out-of-pocket expenditures, it does not provide a method for attributing costs between society, government, family members and the individual. For this reason, the breakdown of cost by payer was based on the OECD (2017) who publish an estimated share of health system costs borne by the

government, compulsory health insurance, out-of-pocket, voluntary health insurance and other.

6.2 Primary health care

Primary health care includes general practitioner (GP) visits, allied health services, and other community-based services.

Primary health care costs were estimated by applying the average utilisation of primary health care services to the incremental unit cost of each service. Average utilisation was estimated using country specific survey results of the proportion of the IRD population accessing each service and the annual per-person frequency of service use. Out-of-pocket travel costs for each service were obtained from the survey results. Where available, incremental unit costs were sourced from country-specific sources, including the National Schedule of Reference Costs (National Health Service (NHS), 2018a), National Tariff Payment System (NHS, 2019; NHS, 2019b), Personal Social Services Research Unit (PSSRU) (Curtis and Burns, 2018) and relevant other sources as referenced.

6.2.1 Utilisation of primary health care services

In order to inform the annual frequency of health service use among persons living with an IRD, data was taken from the combined survey results. It was found that among people accessing services, the most commonly seen included a psychologist or counsellor (12.00 times per year), an eye clinic liaison officer (1.67 times per year), and a sight support volunteer (2.17 times per year) (Table 6.1).

Survey data was also used to inform the annual frequency of health service use among persons living with an IRD. Health services accessed by respondents in the past 12 months included an optometrist (32%), habilitation specialist (13%)[2], sight support volunteer (6%), GP (4%), genetic counsellor

[2] Habilitation involves one-to-one training for children and young people with a vision impairment, it aims to develop their personal mobility, navigation and independent living skills (Wall, 2017).

(6%), psychologist or counsellor (2%), eye clinic liaison officer (6%), and occupational therapist (2%) (Table 6.1).

Table 6.1: Primary health care service utilisation of survey participants in the UK (n=53)

Health service	Percentage (%) accessing service	Number of visits per year
Optometrist/optician	32	1.2
Habilitation, rehabilitation specialist or service, or low vision specialist	13	10.3
Eye clinic liaison officer	6	1.7
Sight support volunteer	6	2.2
Genetic counsellor	6	1.0
GP	4	1.0
Psychologist, psychiatrist or counsellor	2	12.0
Occupational therapist	2	1.0

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages did not sum to 100.

6.2.2 Incremental unit costs of primary health care services

The incremental unit cost for each service was assumed to comprise a reimbursed amount, out-of-pocket expenditure, and travel costs. Where possible, the reimbursed amount for each service was estimated based on the national reference price (Curtis and Burns, 2018; NHS, 2019; NHS, 2018; PSSRU, 2018).

For services where a national reference price could not be located, a cost estimate was obtained from literature or based on an average across a sample of provider-reported prices. For sight support volunteers, no official estimate could be obtained. Therefore, the average hourly cost of sight support volunteers was estimated based on annual salary data adjusted for non-wage costs and annual leave entitlements (Rabiee et al, 2015; Sight Support Derbyshire, 2018).

Out-of-pocket expenditures and out-of-pocket travel costs for each service were taken from survey responses. Where applicable, a measure of health inflation was utilised to convert unit values to 2019 terms.

Incremental health resource unit costs by service type are provided in Table 6.2.

Table 6.2: Primary health care service incremental unit costs (£ 2019)

Health service	Average unit cost
Genetic counsellor	172
Psychologist, psychiatrist or counsellor	160
Optometrist/optician	105
Occupational therapist	94
GP	62
Habilitation, rehabilitation specialist or service, or low vision specialist	45
Eye clinic liaison officer	43
Sight support volunteer	38

Source: Deloitte Access Economics analysis.

The average primary health care cost for persons living with an IRD is summarised in Table 6.3.

Table 6.3: Average cost of primary health care per person with an IRD (£ 2019)

Service	Cost
Average primary health care costs per person with an IRD	162

Source: Utilisation from Deloitte Access Economics IRD survey. Unit costs from Baxter and Rabiee (2015), Curtis and Burns (2018), Gene Health UK (2019), NHS (2018a), NHS (2019), PSSRU (2018), Sight Support UK (2018), Subramanian et al (2011), Deloitte Access Economics analysis.

6.3 Secondary health care

Secondary health care includes hospital inpatient and outpatient services. Inpatient services include inpatient hospitalisations related to a primary or secondary diagnosis of an IRD, and falls attributable to IRDs. Outpatient services include ophthalmologist attendances and associated procedures performed in an outpatient clinic.

The total cost associated with secondary health care services was estimated by applying the average cost of services to the number of services delivered.

6.3.1 Inpatient services

The number of inpatient services related to a primary or secondary diagnosis of an IRD was estimated using data from the NHS (2019c), adjusted to reflect the proportion of IRDs in-scope and scaled to the UK population (Bocquet et al, 2013). A weighted average cost associated with these services was taken from NHS (2019b) reference prices.

The cost of inpatient services (excluding emergency department visits) was estimated based on national reference prices and admissions data published by the NHS (2019). It was assumed that admissions were equal to the number of consultant episodes where H35.5 (hereditary retinal dystrophy) was listed as a primary diagnosis. This measure of admissions

was adjusted using the proportion reported by Bocquet et al (2013) of IRDs in scope, and scaled using the ratio of the English population to total UK population. For secondary admissions, a similar method was used; however, further adjustment was made to account for the estimated number of comorbidities (2.26) based on Frick et al (2012).

The estimated number totalled 112 admissions relating to IRDs as a primary cause, with a further 1,393 admissions relating to IRDs as a secondary cause. This yielded a total number of 1,505 admissions attributed to IRDs.

The average cost of an admission was calculated as an average of retinal procedure costs weighted by the relative frequency of services for each code (NHS, 2019). For secondary cause admissions, a weighted average was calculated based on eye procedure codes. The estimated cost averaged £2,070.17 for primary cause admissions, and £2,358.08 for secondary cause admissions.

To estimate the total cost of inpatient services, the estimated number of admissions was applied to the average cost of an admission. The estimated cost totalled £3.5 million.

6.3.2 Outpatient services

The number of outpatient admissions was based on utilisation data from the country-specific survey results, applied to the number of prevalent cases. An average cost for a specialist ophthalmologist consult in an outpatient clinic (code 130) was taken from the NHS (2018a) National Schedule of Reference Costs, inflated to 2019 terms.

Out-of-pocket expenditures and travel costs were taken from the survey responses and added to the average service cost to estimate the total incremental unit cost.

Table 6.4: Average cost of outpatient health care per person with an IRD (2019)

Parameter	Source	Estimate
Percentage (%) accessing service	Deloitte Access Economics survey	71
Number of times per year	Deloitte Access Economics survey	1.2
Incremental unit cost of outpatient consultation (£)	NHS (2018a), Deloitte Access Economics survey	134
Total average annual cost of outpatient services (£) per person with an IRD		29

Source: Deloitte Access Economics, NHS (2018a).

6.3.3 Cost of falls

Falls may result from the visual impairment associated with IRDs. In order to estimate the cost of these falls, a population attributable fraction approach was used to estimate the proportion of attributable falls (NHS Digital, 2018). An average cost was then estimated (NHS Improvement, 2018; 2017; 2016; 2015; 2014) and inflated to 2019 terms (ONS, 2019).

6.3.4 Average cost of secondary health care

The average secondary health care cost for persons living with an IRD is summarised in Table 6.5.

Table 6.5: Average cost of secondary health care per person with an IRD (£, 2019)

Service	Cost
Average secondary health care costs per person with an IRD	326

Source: Utilisation from Deloitte Access Economics IRD survey. Unit costs from ONS (2019), NHS (2019b), Deloitte Access Economics modelling.

6.3.4.2 Average cost of secondary care for family members

The cost of secondary care for family members of persons living with an IRD was assumed to include the cost of specialist outpatient consultation and genetic counselling. Survey data was used to inform the proportion of persons with an IRD whose family members had accessed these services, as shown in Table 6.6.

The number of family members accessing specialist consultation was estimated by calculating the number of persons with an IRD who reported having one or more family members who had accessed specialist consultation. It was then assumed that one of these family members had accessed this service within the past year.

To derive the number of family members accessing genetic counselling, it was assumed that family members accessed this service in-line with the ratio of genetic counselling utilisation to genetic testing utilisation among persons with an IRD.

Family members were assumed to access one service per annum. Incremental unit costs for genetic counselling and specialist outpatient consultation were assumed to equal the respective unit costs for these services among people with an IRD.

Table 6.6: Average cost of secondary health care for family members per person with an IRD (£, 2019)

Service	Estimate
Proportion of people with an IRD whose family member received genetic counselling	14.8%
Proportion of people with an IRD whose family member received a specialist consultation	62.3%
Average secondary health care costs for family members per person with an IRD	109

Source: Utilisation from Deloitte Access Economics IRD survey. Unit costs from ONS (2019), NHS (2019b), Deloitte Access Economics modelling.

6.4 Diagnostic tests

The cost of genetic testing for persons with an IRD was estimated at £850.00 based on the average cost of retinal dystrophy 235 gene exome panel sequencing of the entire coding region of genes (NHS, 2019d).

The number of family members accessing genetic testing was estimated by calculating the number of persons with an IRD who reported having one or more family members who had accessed genetic testing. It was then assumed that one of these family members had accessed this test within the past year.

The unit cost of genetic testing for family members was estimated at £157.50 based on retinal dystrophy 235 gene exome panel testing for known mutations in family members (NHS, 2019d). Utilisation and frequency of service use for family members was based on survey results.

The cost of genetic testing for persons living with an IRD (£850.00) and for family members (£157.50) was applied to the utilisation and frequency of service use, based on country specific survey results, to derive an average cost per person.

The average cost of diagnostic tests for persons living with an IRD and their family members is summarised in Table 6.7.

Table 6.7: Average cost of diagnostic tests (£ 2019)

Service	Cost
Average cost of diagnostic tests per person with an IRD	71
Average cost of diagnostic tests for family members per person with an IRD	132

Source: NHS (2019), Deloitte Access Economics analysis.

6.5 Pharmaceuticals

The cost of pharmaceuticals attributable to IRDs was derived from available literature. Frick et al (2012) estimated the additional pharmaceutical costs associated with RP in a US population to be $336 per person. This value was converted to UK terms using health purchasing power parity (OECD, 2011) and inflated to 2019 terms using a measure of health inflation in the UK.

Table 6.8: Average cost of pharmaceuticals per person with an IRD (£ 2019)

Service	Cost
Average pharmaceutical costs per person with an IRD	223

Source: Frick et al (2012), OECD (2011), Deloitte Access Economics analysis.

6.5.1 Gene therapy

One gene therapy – Luxturna – has been authorised by the European Medicines Agency (EMA) for the treatment of adult and paediatric patients with vision loss due to inherited retinal dystrophy caused by confirmed biallelic RPE65 mutations and who have sufficient viable retinal cells. At present this product

is not reimbursed in the UK or the RoI and as such is not included in this analysis.

6.6 Vitamins and supplements

Country specific survey results were used to estimate the utilisation and out-of-pocket expenditures incurred by persons living with an IRD on vitamins and supplements.

Table 6.9: Average cost of vitamins and supplements per person with an IRD (£ 2019)

Service	Cost
Average cost of vitamins and supplements per person with an IRD	31

Source: Deloitte Access Economics.

6.7 Medical research

Medical research costs represent the average annual value of all grants relating to IRDs that are active in 2019 (Medical Research Council, 2019). This cost does not vary with prevalence.

The value of medical research costs associated with IRDs was estimated by filtering the grants database published by the Medical Research Council (2019) for key terms. An average of the past five years of grants related to these conditions was calculated to estimate average annual medical research costs in 2019 (Table 6.10).

This estimate is likely to be conservative since it does not include broader research related to blindness or visual acuity which may partly be attributable to IRDs. For example, UK blindness certifications suggest that IRDs account for 5.7% of blindness and visual impairment in the UK (Quartilho et al, 2016; Bunce et al, 2010; Bunce & Wormald, 2006). If the value of grants related to blindness and visual impairment was allocated on this basis, the total value of grant funding for IRDs may be estimated at £5,087,055.

Table 6.10: Total medical research costs associated with IRDs (£ million 2019)

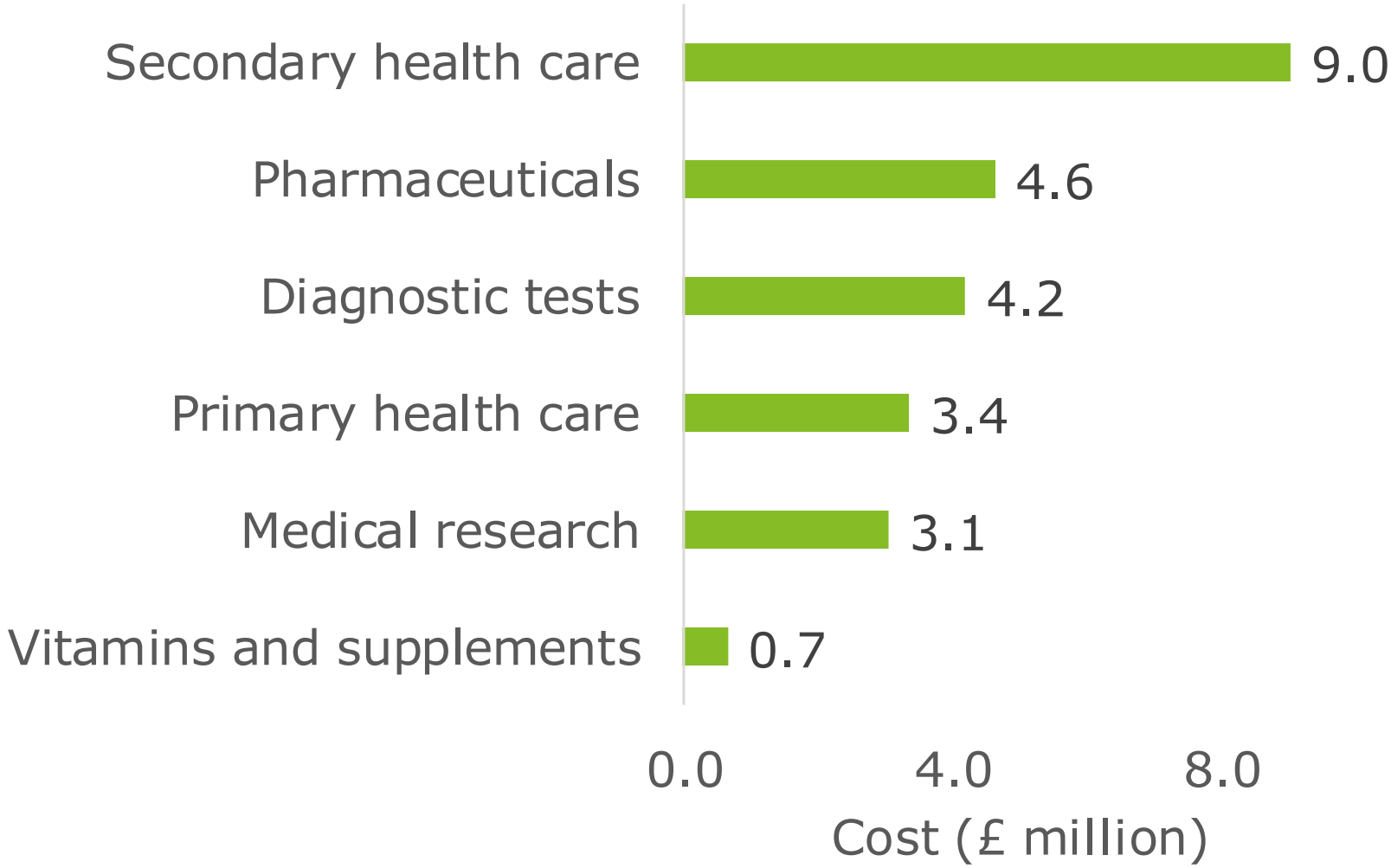
Input	Cost
Total medical research costs	3.1

Source: Medical Research Council (2019), Deloitte Access Economics analysis.

6.8 Results

Total health system costs associated with IRDs in the UK in 2019 were estimated to be £25.0 million. The largest component of health system costs were for secondary health care (£9.0 million), followed by pharmaceuticals (£4.6 million). The lowest proportion of the costs was accounted for by vitamins and supplements at £0.7 million (Chart 6.1, Table 6.11).

Chart 6.1: Total health system costs of IRDs (£ million, 2019) in the UK by cost type



Source: Deloitte Access Economics.

Table 6.11: Total health system costs of IRDs (£ million, 2019) in the UK by cost type

Cost type	Cost
Secondary health care	9.0
Pharmaceuticals	4.6
Diagnostic tests	4.2
Primary health care	3.4
Medical research	3.1
Vitamins and supplements	0.7
Total costs	**25.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

7 Individual productivity costs

This section describes the approach used to estimate individual productivity costs – including reduced workforce participation and lost productive time – associated with IRDs in the UK in 2019.

Key findings

- In 2019, the individual productivity cost of IRDs in the UK were estimated to be £114.1 million.
- 78% of the individual productivity cost were due to forgone income as a result of reduced workforce participation.
- In the UK, persons with an IRD were 40.2% less likely to be in paid employment than the general population.
- IRDs resulted in a 9.6% reduction in productivity while at work.

7.1 Summary of approach

Persons living with an IRD can experience reduced capacity to effectively participate in the workforce. Consequently, there are significant productivity costs in terms of reduced workforce participation and lost productive time associated with IRDs.

A human capital approach was adopted to estimate the productivity costs attributable to IRDs. This involved calculating the difference in employment rate between persons living with an IRD and the general population, multiplied by average weekly earnings (AWE). Similarly, costs incurred through lost productive time are estimated by multiplying the average number of weeks of productive time lost by AWE.

Survey results informing this section were validated through triangulation with literature estimating the productivity costs of IRDs, or conditions with comparable aetiology, in the UK and internationally.

7.2 Reduced workforce participation

Persons living with an IRD may experience reduced workforce participation relative to the general population due to disadvantages in job-seeking or self-selection out of the labour force. This can lead to significant productivity losses through lost wages.

Most survey participants were not in the workforce[3] (54.5%), followed by those in full-time employment (29.5%) and those employed part-time, casual, or under employed (15.9%) (Table 7.1). As such, survey participants were employed at a rate of 45.5% compared to the general population employment rate of 76.1% (ONS, 2019). This translated to a reduced workforce participation of 40.2% for persons living with an IRD in the UK.

Costs incurred through reduced workforce participation are estimated by applying the time lost to reduced workforce participation to the UK general population employment rates and AWE by age and sex.

Table 7.1: Distribution of survey participants by employment status in the UK (n=44)

Employment status	Count (n)	Percentage (%)
Not in the labour force	24	54.5
Employed full-time	13	29.5
Employed part-time, casual, or under employed	7	15.9
Total	**44**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

[3] 'Not in the labour force' included participants who indicated they were retired, studying or not employed and not looking for work.

The following studies estimating the employment rates of persons living with an IRD or condition of a comparable aetiology in the UK were found:

- Slade and Edwards (2015) noted that adults with a visual impairment were employed 46% less than the general population, who were employed at a rate of 27%. This study was informed by interviews with 1,223 registered blind and partially sighted people in the UK between November 2014 and April 2015.
- Using data from the UK Labour Force Survey (LFS) between October 2012 and July 2015, Hewett and Keil (2016) calculated that the employment rate for the general population in the UK without a disability was 73.8%, approximately 30% higher than those with a self-reported seeing difficulty (45.1%) – see Table 7.2 for the employment rates distributed by age.

Table 7.2: Employment rate (%) UK (2012-2015) by disability status and age

	Employment rate	
Age	**Seeing disability (%)**	**General population (%)**
16-25	25.6	54.0
26-44	47.6	81.2
45-54	56.4	85.9
55-64	46.3	70.1

Source: Hewett and Keil (2016).

7.2.1 Evidence of reduced workforce participation – international

The following studies estimating the employment rates of persons living with an IRD or condition of a comparable aetiology in countries other than the UK were found:

- Chaumet-Riffaud et al (2017) surveyed 148 patients, aged 25-50, in France diagnosed with RP between 2013 and 2015. The average employment rate across three levels of visual acuity was 75.3% (see Table 7.3). This was marginally lower than the employment rate (80.2%) of the general French population aged 25-49 in 2014.
- Based on the 2016 RoI census data, the Central Statistics Office (CSO) (2017) estimated that 36.4% of people with VI and/or blindness were employed. By contrast, the employment rate of the general population was estimated to be 87%.
- Sherrod et al (2014) used data from 19,849 participants in the 1999-2008 United States (US) National Health and Nutrition Examination Survey to examine the association of vision with work status. The employment rate was estimated to be 42% in participants with a severe visual impairment compared to 69.5% in participants with a normal level of visual function.

Sander et al (2005) utilised the French national *Handicaps, incapacités, dépendance* (HID) survey to estimate people with severe VI and blindness were employed at a rate of 29%, while those with a mild impairment were employed at a rate of 65%.

The results reported by each identified study are outlined in Table 7.3 and 0. These studies were excluded from this analysis in favour of the IRD specific survey results.

Table 7.3: Employment rate for people with RP (%) France (2017) by level of vision

Vision	Employment rate (%)
Mild deficiency	78.8
Low vision	79.5
Blind	67.6
Average	**75.3**

Source: Chaumet-Riffaud et al (2017).

Table 7.4: Summary of evidence on reduced workforce participation due to IRDs or VI/blindness

			Employment		
Study	**Condition**	**Country**	**No VI (%)**	**VI (%)**	**Reduction (% points)**
Chaumet-Riffaud et al (2017)	RP	France	80.2	70.1	10.1
CSO (2017)	VI – General	Ireland	87.1	36.4	50.7
Hewett and Keil (2016)	VI – General	UK	73.8	45.1	28.7
Slade and Edwards (2015)	VI – General	UK	73.0	27.0	46.0
Sherrod et al (2014)	VI – General	US	69.5	42.0	27.5
Sander et al (2005)	VI – General	France	65.0	29.0	36.0

Source: Chaumet-Riffaud et al (2017), CSO (2017), Hewett and Keil (2016), Slade and Edwards (2015), Sherrod et al (2014) and Sander et al (2005).

7.3 Lost productive time

Lost productive time is comprised of presenteeism and absenteeism.

7.3.1 Presenteeism

Presenteeism is the average number of hours per day that an employee loses to reduced performance or impaired function as a result of their condition. This is measured as a reduction in the quality and efficiency of work produced. Relative to absenteeism, presenteeism may occur more frequently and have a larger effect (Van den Heuvel et al, 2010). Persons living with an IRD may be more likely to experience reduced productivity at work as a result of their condition.

Costs incurred through presenteeism are estimated by applying the time lost to presenteeism to the UK general population employment rates and AWE by age and sex.

Schakel et al (2018) collected cross-sectional data through interviews and surveys of visually impaired and normally sighted adults in the Netherlands between 2015 and 2016. Using a scale between one and ten, visually impaired and normally sighted respondents placed a value on their level of efficiency while at work. Deloitte Access Economics used the relative decline in self-reported work efficiency scores – measured on a scale of 1-10 – between the visually impaired (6.6) and normally sighted (7.3) respondents to estimate presenteeism of 9.6%.

The estimate, based on the study by Schakel et al (2018), was considered a more robust estimate of IRD-related presenteeism than the survey due to its inclusion of a control group (normally sighted people) with which to compare the self-reported decline in work efficiency of people with a visual impairment against. As such, the estimate based on Schakel et al (2018) was selected as the input for the calculation.

7.3.2 Absenteeism

Absenteeism is the average number of days per year that an employee is away from work due to their condition.

Absenteeism incurs a productivity cost to:

- **Employers:** Lost output from work absences whilst incurring leave entitlements, management time in processing employee absences, line manager time in rearranging work, time of back office personnel, and overhead costs of employees such as office space and insurance costs.
- **Government:** Sickness benefit entitlement paid through social insurance.
- **Individuals:** Lost wages due to the gap between paid wages and the sick leave entitlement received from employers and/or government.

The survey results indicate that persons living with an IRD did not take any additional days of leave due to their condition, indicating that IRD-related absenteeism was zero in the UK.

By comparison, one study estimated that people with sight loss in the US are likely to take an average of 4.1 additional days off work annually by Pezzullo et al (2018). This study was excluded from this analysis in favour of the survey results that are specific to the IRD population.

7.4 Other productivity costs not estimated

Premature mortality results in productivity losses due to forgone potential earnings. This can also result in increased costs associated with employee turnover such as search, hiring and training costs. There is a lack of available data on the risk of premature mortality associated with IRDs. As such, no cost has be allocated to forgone income or search, hiring and training costs due to IRDs in the RoI.

By comparison, some literature estimating the cost of premature mortality associated with VI and/or blindness is available. Wang et al (2001) reported a 70% increased chance of mortality with the presence of any sight loss, accounting for comorbidities. Deloitte Access Economics (2014) previously estimated that the total present value loss from premature mortality and retirement associated with vision impairment in the UK was £2.14 million in 2013.

7.5 Results

Total productivity costs due IRDs in the UK in 2019 were estimated to be £114.1 million. Lost income as a result of reduced workforce participation resulted in the greatest

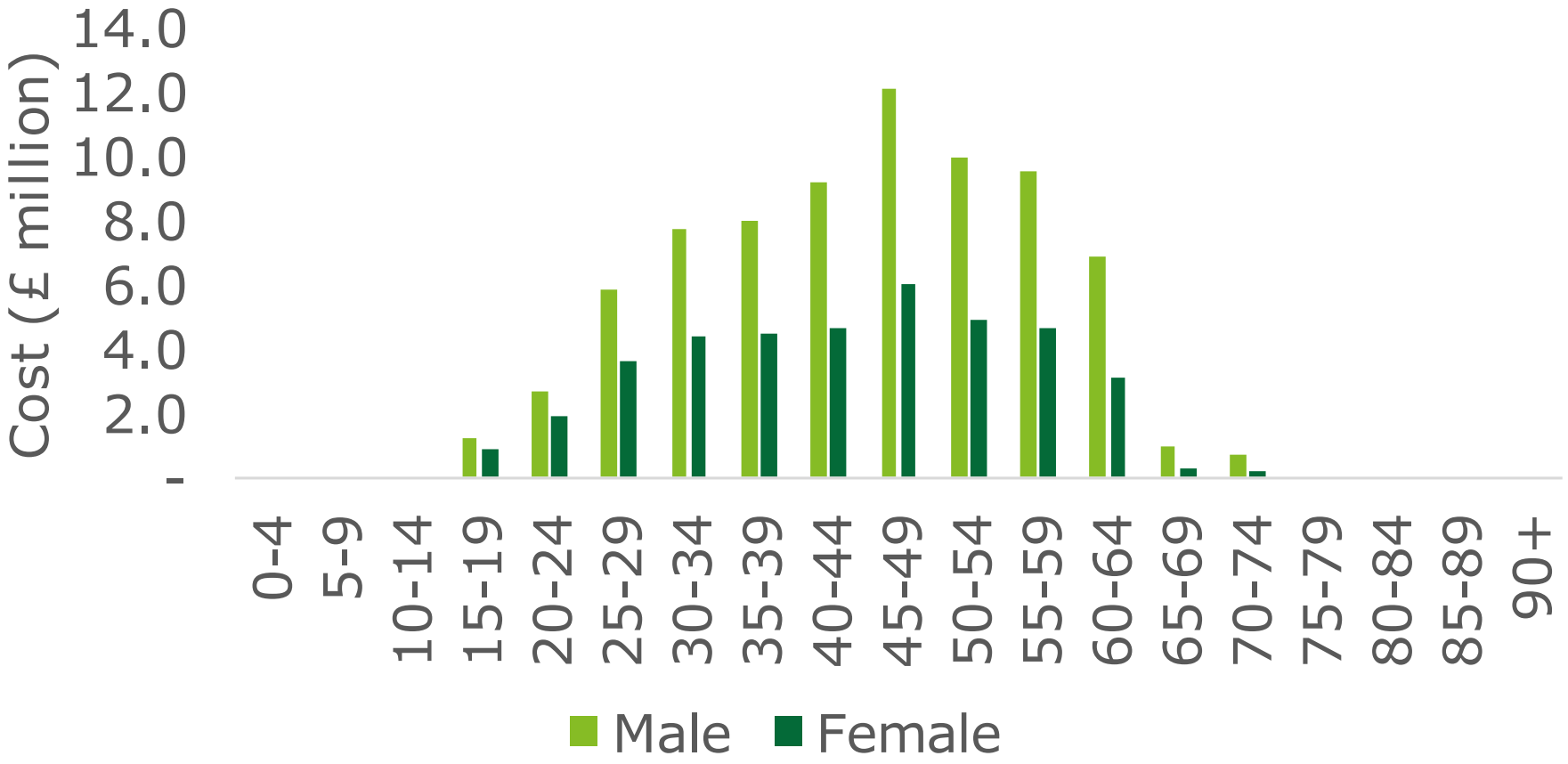
proportion of productivity costs at £89.0 million (78%) (Table 7.5). Productivity costs were highest for people aged 45-49 years, reflecting the high earnings and employment rates in that age group (Chart 7.1).

Table 7.5: Total productivity costs of IRDs (£ million, 2019) in the UK by cost type

Cost type	Cost
Reduced workforce participation	89.0
Presenteeism	25.1
Absenteeism	-
Total	**114.1**

Source: Deloitte Access Economics analysis.

Chart 7.1: Total productivity costs of IRDs (£ million, 2019) in the UK by age and sex



Source: Deloitte Access Economics analysis.

8 Other costs

This section describes the approach used to estimate the other costs – including aids and modifications, education, travel, formal and informal care, and deadweight losses of taxation payments – associated with IRDs in the UK in 2019.

Key findings

- In 2019, other costs attributable to IRDs in the UK were estimated to be £188.1 million, including formal and informal carer costs of £97.6 million, aid and modifications costs of £44.7 million, deadweight losses of £28.7 million, travel costs of £8.6 million, residential care costs of £4.5 million and education costs of £4.0 million.
- Carer costs associated with carers of persons with an IRD include £13.1 million of formal care and £84.5 million of informal care.

8.1 Summary of approach

Other costs attributable to IRDs include aids and modifications, education, travel, formal and informal care, and deadweight losses of taxation payments.

A range of informal care activities may be provided to people with an IRD. Impairment and disability not only affects the individual, but can also affect their family and friends. This can limit the ability of the individual to engage in day-to-day and self-care activities, requiring others to do these activities in their place. People with an IRD and/or their carers may receive welfare payments.

The act of taxation and redistribution creates distortions and inefficiencies in the economy, so transfers also involve real net costs to the economy, known as deadweight loss. This section calculates the deadweight loss resulting from lost taxation (which must be raised elsewhere) and government expenditure on welfare payments and the health system.

Survey results informing this section were supplemented with data obtained from grey literature (i.e. government publications and research reports). Where possible, these estimates were validated through triangulation with published literature estimating the other costs of IRDs, or conditions with comparable aetiology, in the UK and internationally.

8.2 Aid and modifications

Aids and modifications include items such as a guide dog, home modifications, magnifying glasses, Global Positioning System (GPS), electronic mobility devices and other similar products used to assist persons living with an IRD with their daily living.

8.2.1 Aids and modifications

The greatest number of survey participants reported using a white/long cane (58.5%), followed by handheld magnifiers (56.6%), high intensity lamps (45.3%), book alternatives (45.3%), customised clocks, watches or timers (43.4%), screen reading software (37.7%), and screen magnification technology (35.8%). In the absence of sufficient survey response data, it was conservatively assumed that all persons living with an IRD used a magnifying glass, contrast enhancing filters, green or blue blocking sunglasses, and high vision lamps. Other aids are outlined in Table 8.1.

The number of aids used per annum was also informed based on country-specific survey response data. Of people using a specific aid, the largest quantity of aids used per annum was recorded for book alternatives (10.0), followed by tactile or large print labels (4.4), handheld magnifiers (2.3), high intensity lamps (2.3), portable note takers (2.0), and screen magnification technology (1.8). Other aids are outlined in Table 8.1.

Table 8.1: Aids and modification used by survey participants in the UK (n=53)

Aids and modifications	Percentage (%)*	Number used per annum
Contrast enhancing filters	100.0	1.0
Green or blue blocking sunglasses	100.0	1.0
High vision lamps	100.0	1.0
Magnifying glasses	100.0	1.0
White cane	58.5	1.0
Handheld magnifiers	56.6	2.3
Book alternatives	45.3	10.0
High intensity lamps	45.3	2.3
Customised clocks, watches or timers	43.4	1.7
Screen reading software	37.7	1.7
Screen magnification technology	35.8	1.8
Magnifying mirrors	28.3	1.8
Other	18.9	2.1
Large keyboards	15.1	1.4
Portable note takers	13.2	2.0
Guide dog	11.3	1.0
Mini guide or other electronic mobility device	9.4	1.0

Aids and modifications	Percentage (%)*	Number used per annum
Tactile or large print labels	9.4	4.4

Source: Deloitte Access Economics analysis.
* This question allowed for the selection of multiple answers, hence, the percentages did not sum to 100.

8.2.2 Costs of aids and modifications

The cost of aids was estimated by applying the average utilisation of aids to their incremental unit cost. Average utilisation was estimated using survey data of the proportion of the IRD population using each aid and the average number of aids used per annum. The cost of each aid was obtained from the out-of-pocket expenditures recorded by the survey, or online shops.

For higher cost items, an annual depreciable amount was used as the basis for estimating an annual cost. This method is used to allocate the cost of assets over their useful life, for example, guide dogs were assumed to have a working life of 8.5 years – meaning that in a given year, it was assumed that, on average, 11.8% of lifetime guide dog costs would be incurred.

The cost of modifications were derived from a study estimating the non-medical costs associated with visual impairment in the UK Lafuma et al (2006). No studies estimating the cost of modifications of persons living with an IRD in the UK were identified. Unit costs were applied to the utilisation obtained from the survey. This yielded an estimated cost of modifications of £356.24 per person in 2004 terms. This estimate was adjusted to 2019 terms using a price index published by the ONS (2019) for furniture, household equipment and routine repair of house.

The average cost for each aid was collected based on prices reported by online shops. For higher cost items, the total purchase cost was attributed to an annual value using a measure of straight line depreciation. A useful life of five years was assumed for electronic products including large keyboards,

screen magnification technology, screen reading software, portable note takers and electronic mobility devices. This assumption was informed by a paper published by Rincon-Aznar et al (2017) commissioned by the Office of National Statistics. An annual depreciable value of guide dogs was also assumed, based on the working life of 8.5 years reported by a UK Guide Dogs (2019) publication. The average annual unit cost of each item is summarised in Table 8.2.

Table 8.2: Aids and modification unit costs for persons living with an IRD in the UK (£ 2019)

Aids and modifications	Annual unit cost
Guide dog	6,588
White cane	788
Mini guide or other electronic mobility device	650
Screen magnification technology	407
Screen reading software	168
Portable note takers	108
Contrast enhancing filters	91
Green or blue blocking sunglasses	62
Handheld magnifiers	54
High intensity lamps	51
High vision lamps	48
Customised clocks, watches or timers	34
Magnifying glasses	28

Aids and modifications	Annual unit cost
Magnifying mirrors	20
Large keyboards	20
Tactile or large print labels	15
Book alternatives	10

Source: Deloitte Access Economics analysis.

These unit costs were multiplied by the survey utilisation data to estimate the cost of aids and modifications in the UK in 2019.

The average cost of aids and modifications for persons living with an IRD is shown in Table 8.3. Overall, expenditure on aids and modifications was estimated to be £44.7 million.

Table 8.3: Average cost of aids and modifications per person (£ 2019)

Product	Cost
Average cost of aids per person	1,702
Average cost of modifications per person	445

Source: Deloitte Access Economics analysis.

8.2.3 Other aids and modifications costs not estimated

Persons living with an IRD may also require aids and modifications for education and employment settings. These products were conservatively excluded from this study to avoid double counting with aids and modifications used in general settings. For example, someone who requires a guide dog for their home use is likely also able to use the same guide dog if required at their place of work.

The survey results indicate that persons living with an IRD pay an average of £123.6 for aids and modifications to assist with

work, and £30.8 on aids and modifications to assist with study. When applied to prevalence estimates, these expenditures total £1.1 million in employment-related aids and modifications, and £0.1 million in education-related aids and modifications.

8.3 Education

Persons living with an IRD can experience significant barriers to effective learning, thus necessitating specialist support and additional resourcing.

In England, funding for students aged up to 25 years of age with special education needs (SEN) is provided by the national government to Local Authorities (LAs) who distribute funds to individual schools in their jurisdiction based on a needs assessment.

As at January 2019, approximately 1.3 million students in England were classified as having SEN with £6.1 billion in funding allocated to schools to support students with high needs in 2018-19 (Department for Education, 2019; Long and Roberts, 2019). This equated to an average estimated cost of £4,638 per student with SEN. Of those students, approximately 12,700 students required SEN due to a visual impairment. The prevalence of IRDs in the UK as a proportion of people with VI was applied to this number to estimate that approximately 721 students may have received SEN support for an IRD in 2019. This translated to a total estimated cost of £3.3 million for SEN attributable to IRDs in England.

A UK-England scale factor was applied to arrive at an estimated cost of £4.0 million for SEN attributable to IRDs in the UK in 2019.

8.4 Travel

Persons living with an IRD can experience significant barriers to driving a motor vehicle, meaning they must rely on public transport as their primary mode of travel. In the UK, persons with an IRD may receive access to free public transport.

The survey asked respondents to estimate how much money they had spent on public transport in the previous month for

reasons other than health care visits.[4] Respondents were asked to estimate these costs before any refunds, reimbursements or financial support that they may have received.

On average, respondents indicated that they spent £58.47 on public transport in a given month, which was multiplied by 12 to derive the average annual expenditure on public transport (£701.60). Respondents who indicated that they did not pay anything for public transport, for example due to having a free travel concession card were excluded from the calculation of the average cost. This aims to encapsulate the full cost of public transport to society and not just the out of pocket costs incurred by individuals.

Of all respondents, 67.9% indicated that they used public transport, which was multiplied by the prevalence of IRDs in the UK, and the average expenditure to estimate the total cost of travel in 2019. The total travel costs for purposes other than health visits were estimated to be £2.4 million.

These travel costs compare well with a top down approach to estimating travel costs, although they are higher. In 2017-18, 9.4 million persons received free transport through the English National Concessionary Travel Scheme (ENCTS) in England at total cost of £1.12 billion. This equated to an average annual per person cost of £119 to the government (Department for Transport, 2019).

Of those who used the ENCTS, approximately 900,000 were classified as having a disability (Department for Transport, 2019). The prevalence of IRDs in the UK as a proportion of people with a disability was applied to this number to estimate that approximately 6,100 persons living with an IRD used the ENCTS in 2017-18.

A UK-England scale factor was applied to arrive at an estimated cost of £0.87 million in travel support due IRDs in the UK in 2019. It is possible that the survey identifies higher costs as persons living with an IRD may use transport services more frequently than other ENCTS recipients.

[4] Travel costs for health care visits were included in section 6.

8.5 Formal and informal care

IRDs can result in impairment and disability that not only affects those persons living with the condition, but can also affect their family and friends. Individuals with severe impairment resulting from an IRD can have limited ability to engage in day-to-day and self-care activities, requiring others to do these activities in their place via formal or informal care.

8.5.1 Formal care

Formal care can include help from a private nurse or assistance with activities such as childcare, housekeeping and shopping – these costs are not subsidised by private health insurance or the government but instead are out-of-pocket expenses borne by the individual.

The cost of formal care for persons living with an IRD can be estimated by establishing the:

- Proportion of persons living with an IRD that receive formal care
- Number of hours of formal care each person with an IRD receives
- Cost per hour of formal care for a person with an IRD.

The utilisation and average weekly hours of formal care were estimated using the survey data (see Table 8.4, and Table 8.5). The hourly cost of formal care was calculated based the UK's PSSRU (Curtis and Burns, 2018) – an annual report that identifies the cost of health services and professionals in the UK (see Table 8.6).

Table 8.4: Percentage of survey participants who receive formal care in the UK (n=53)

Formal care received	Count (n)	Percentage (%)
Does not receive formal care	51	96.2
Receives formal care	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Table 8.5: Average weekly hours of formal care received by survey participants receiving formal care in the UK and RoI (n=8)

Service	Hours
Average weekly hours of formal care	14.1

Source: Deloitte Access Economics analysis.

Table 8.6: Hourly cost of formal care (£ 2019) in the UK

Service	Source	Cost
Hourly cost of formal care	Curtis and Burns (2018)	22.0

Source: Deloitte Access Economics analysis.

These inputs were multiplied together to estimate the cost of community based formal care in the UK in 2019. Overall, formal care costs were estimated to be £13.1 million.

8.5.1.1 Evidence of the proportion of people receiving formal care – UK

The following studies estimating the utilisation of formal care by persons living with an IRD or condition with a comparable aetiology in the UK were found:

- Slade and Edwards (2015) found that 39% of adults with a visual impairment and/or blindness in the UK used a paid support worker. This study was informed by interviews with 1,223 registered blind and partially sighted people between November 2014 and April 2015.
- Ke et al (2007) studied the care use amongst persons aged 50 years and over with age-related macular degeneration (AMD) in Northern Ireland. The average utilisation of formal care was 44.6% across different levels of distance visual acuity (DVA). The utilisation rates by participants' level of DVA in the better eye are presented in Table 8.7.

Table 8.7: Formal care utilisation (%) Northern Ireland (2007) by DVA

DVA (logMAR)	Percentage of people with the level of DVA utilising formal care (%)
-0.2 – 0.3	34.9
0.4 – 0.9	51.6
1.0 or worse	55.6
Average	**44.6**

Source: Ke et al (2007).

8.5.1.2 Evidence of the proportion of people receiving formal care – International

The following studies estimating the utilisation of formal care by persons living with an IRD or condition with a comparable aetiology in countries other than the UK were found:

- Using data from the Canadian Longitudinal Study on Aging Comprehensive Cohort on the population aged 45-85, Aljied et al (2019) found that 13.3% of people with a visual impairment (binocular acuity worse than 20/60) utilised formal care at home.
- Schmier et al (2006) conducted a survey of 803 patients with AMD in Canada using the Daily Living Tasks Dependent on Vision Questionnaire (DLTV). The average level of formal care usage by all patients was 16.6%. Table 8.8 shows the utilisation of formal care by participants' levels of visual acuity.

Table 8.8: Formal care utilisation (%) Canada (2006) by visual acuity

Visual acuity	Percentage of people with the level of visual acuity utilising formal care (%)
>3.5	1.5
>3.0 – 3.5	3.9
>2.55 – 3.0	9.6
>2.0 – 2.55	19.0
>1.55 – 2.0	32.4
≤1.5	33.3
Average	**16.6**

Source: Schmier et al (2006).

8.5.1.3 Evidence of the hours of formal care received – international

The following study estimating the hours of formal care received by persons living with an IRD or condition with a comparable aetiology in a country other than the UK were found:

- Using data from the Canadian Longitudinal Study on Aging Comprehensive Cohort for the population aged 45-85, Aljied et al (2019) estimated that those who used formal care for a visual impairment did so for an average of 5 hours per week. However, this figure did account for potential comorbidities.

8.5.2 Informal care

A range of informal care activities may be provided to persons living with an IRD. Informal care activities depend on the level of impairment, and can include:

- Collecting relevant prescriptions and organising and timing the administration of medication

- Assistance in daily domestic activities such as cooking and laundry
- Ad-hoc tasks, such as shopping, transport and cleaning activities
- Monitoring of the patient's physical and mental wellbeing.

The opportunity cost method was used to estimate the cost of informal care. This method assumes that time spent providing informal care could be used to engage in paid employment or in leisure activities.

The cost of informal care for persons living with an IRD was estimated by using the:

- Proportion of persons living with an IRD that receive informal care
- Number of hours of informal care each person with an IRD receives
- Hourly cost of providing informal care to a person with an IRD.

The utilisation and average weekly hours of informal care were estimated using the survey data (Table 8.9 and Table 8.10). The hourly cost of informal care was calculated based on the AWE in the UK (see Table 8.11).

Table 8.9: Percentage of survey participants who receive informal care in the UK (n=53)

Informal care received	Count (n)	Percentage (%)
Does not receive informal care	42	79.3
Receives informal care	11	20.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Table 8.10: Average weekly hours of informal care received by survey participants receiving informal care in the and UK (n=42)

Service	Hours
Average weekly informal care	12.9

Source: Deloitte Access Economics analysis.

Table 8.11: Hourly cost of informal care (£ 2019) in the UK

Service	Cost
Hourly cost of informal care	13.1

Source: Deloitte Access Economics analysis.

These inputs were multiplied together to estimate the cost of informal care in the UK in 2019. Overall, informal care costs were estimated to be £84.5 million.

8.5.2.1 Evidence of the proportion of people receiving informal care – UK

The following studies estimating the utilisation of informal care by persons living with an IRD or condition with a comparable aetiology in the UK were found:

- Slade and Edwards (2015) found that 48% of adults with a visual impairment and/or blindness in the UK required support from a family member, friend or volunteer. This study was informed by interviews with 1,223 registered blind and partially sighted people between November 2014 and April 2015.
- Ke et al (2007) studied the care use amongst persons aged 50 years and over with age-related macular degeneration (AMD) in Northern Ireland. The average utilisation of informal care was 60.5% across different levels of DVA. The utilisation rates by participants' level of DVA in the better eye are presented in Table 8.12.

Table 8.12: Informal care utilisation (%) Northern Ireland (2007) by DVA

DVA (logMAR)	Percentage of people with the level of DVA utilising informal care (%)
>0.3	37.3
0.4 – 0.9	69.9
1.0≤	88.9
Average	**60.5**

Source: Ke et al (2007).

8.5.2.2 Evidence of the proportion of people receiving informal care – international

The following studies estimating the utilisation of informal care by persons living with an IRD or condition with a comparable aetiology in countries other than the UK were found:

- Using data from the Canadian Longitudinal Study on Aging Comprehensive Cohort for the population aged 45-85, Aljied et al (2019) noted that 15.1% of those with a visual impairment used informal care at home.
- From a study of 546 visually impaired individuals from Portuguese hospitals between 2014 and 2016, it was noted that 39.6% reported using informal care (Marques et al, 2018). Only outpatients with a latest recorded visual acuity of 6/12 or worse in their better seeing eye took part in the study.
- The Irish Health Review Board's (HRB) Annual Report of the National Physical and Sensory Disability Database Committee 2017 showed that 39.0% of the population with a visual deficit utilised informal care (Doyle and Carew, 2017).
- Based on the 2006 National Disability Survey, the CSO (2010) noted that 61.3% of respondents with a visual impairment self-reported requiring some form of informal support from a family member or friend in order to perform daily functions.

- A study of people with visual impairment in Australia showed 61.4% (62) self-reported relying on informal 'home care' to achieve daily functions (Keeffe et al, 2009).
- Schmier et al (2006) conducted a survey of 803 patients with AMD in Canada using the DLTV. The average level of informal care usage by all patients was 41.2%. Table 8.13 shows the utilisation of formal care by participants' levels of visual acuity.
- Based on biennially collected panel data from 1993 to 1994 in the USA on people aged 70 and over, Shih and Lustig (2002) found that 64.2% of those with a visual impairment relied on the use of an informal caregiver.

Table 8.13: Informal care utilisation (%) Canada (2006) by visual acuity

Visual acuity	Percentage of people with the level of visual acuity utilising care (%)
>3.5	3.3
>3.0 – 3.5	21.0
>2.55 – 3.0	39.5
>2.0 – 2.55	68.1
>1.55 – 2.0	62.2
≤1.5	53.3
Average	**41.2**

Source: Schmier et al (2006).

8.5.2.3 Evidence of the number of hours of informal care received – international

The following studies estimating the utilisation of informal care by persons living with an IRD or condition with a comparable aetiology in countries other than the UK were found:

- Using data from the Canadian Longitudinal Study on Aging Comprehensive Cohort (for people aged 45-85), Aljied et al (2019) found that people with a visual impairment used informal care at home for an average of 8.3 hours per week.
- Khan et al (2016) undertook a study of the burden among caregivers of people with a visual impairment in Canada. Of the 236 participants (32% legally blind; 68% low vision), carers reported providing a daily average of 2.2 hours of close supervision, or 15.4 hours per week.
- Weyer-Wendl and Walter (2016) surveyed 150 informal carers of patient with wet age-related macular degeneration (wAMD) in Germany. The average amount of care provided was 6.4 hours per week – see Table 8.14 for the average weekly hours by care recipients' visual acuity.
- Green et al's (2016) study on the cost of blindness in RoI used data from the government data to estimate that blind people utilise an average of 14.7 hours of informal care per week as shown in Table 8.14.
- A study of people with VI in Australia revealed that the 101 participants required an estimated combined total of 15,369 hours of informal care during one year. This translated to 2.93 hours of informal care per person, per week (Keeffe et al, 2009).

Table 8.14: Informal care utilisation by people with wAMD (hours) by visual acuity, Germany (2016)

Visual acuity	Hours
>0.3	4.3
≤0.3 - >0.1	5.6
≤0.1	11.9
All	**6.4**

Source: Weyer-Wendl and Walter (2016).

Table 8.15: Informal care utilisation by blind people (hours) Ireland (2016)

Hours per week	Hours	Percentage utilising informal care (%)
1-14 hours	1	58.0
15-28 hours	15	11.0
29-42 hours	29	6.0
43+ hours	43	25.0
Average	**14.7**	**100.0**

Source: Green et al (2016).

8.6 Residential care

Persons living with an IRD can experience severe (visual) impairment or disability requiring continuous care and supervision. Such persons may require long term care in the form of a residential care service (Wang et al, 2003). Residential care includes the independent sector residential care, local authority residential care and nursing care.

Publicly funded residential care services in the UK are provided at a local level by Councils with Social Services Responsibilities. While some services are provided by the NHS, the majority are provided by councils. The costs are often shared by the local council and person receiving care. Contributions are paid to councils and recorded as revenue with the size of the contribution determined by financial means test which is based on income, capital and assets.

A targeted literature review was conducted to estimate the cost of residential care due to IRDs in the UK. Where IRD-specific literature was unavailable, the scan was expanded to include conditions with comparable aetiology and international sources.

Residential care in England is provided through Personal Social Services (PSS). PSS expenditure in England totalled approximately £13.06 billion in 2015-16 (NHS Digital, 2016),

which includes community care. Of this amount, £74.7 million was spent on long term residential care for people requiring sensory support which is defined as visual or hearing impairments. This expenditure is allocated across nursing, residential, and supported accommodation. See Table 8.16 for the expenditure by category.

Table 8.16: Total expenditure on long term residential care for sensory conditions (£ million 2016) by age, England

Age	Nursing	Residential	Supported	Total
18-64	1.3	13.1	1.3	15.8
65+	16.5	41.3	1.1	58.9
All	**17.9**	**54.5**	**2.4**	**74.7**

Source: NHS Digital (2016).

The ratio of registered blind or visually impaired persons to those with a hearing difficulty in England is an estimated 5.16 to 1.[5] Applying this ratio to the total cost of long term care for a sensory condition, an estimated £62.6 million was spent on long term residential care for blindness or visual impairment. Then applying the prevalence of IRDs in the UK to this figure, an estimated £3.6 million was spent on long term care for persons living with an IRD in England in 2016 pounds (£3.8 million in 2019 pounds). This figure was then scaled using the population of England relative to the UK to arrive at an estimated residential and community care cost of IRDs in the UK of £4.5 million. See Table 8.17 and Table 8.18 for the cost distribution by age and payer, respectively.

[5] This calculated based on the number of registered blind and partially sighted people (HSCIC, 2014) and people registered as deaf or hard of hearing (NHS Digital, 2010).

Table 8.17: Total expenditure on long term residential care for IRDs (£ million, 2019) UK

Age	Total cost
18-64	1.0
65+	3.6
All	**4.5**

Source: NHS Digital (2016) and Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Table 8.18: Total expenditure on long term residential care for IRDs (£ million 2019)

Payer	Percentage (%)	Total cost
Individuals	56.4	2.6
Government	43.6	2.0
Total	**-**	**4.5**

Source: NHS Digital (2016) and Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

8.7 Deadweight losses (DWL) of taxation payments

This section calculates the deadweight losses arising from raising and administering taxation (i.e. transfer) payments, including government and social insurance expenditure on health and welfare.

While transfer payments alone do not impose a net cost to society, the act of raising and redistributing tax distorts the natural pricing mechanisms in the economy leading to inefficient resource allocation. Taxes alter the price and quantity of goods sold compared to what they would be if the market were not distorted. In a practical sense, this distortion

reveals itself as a loss of efficiency in the economy, which means that raising £100 of taxation revenue requires consumers and producers to give up more than £100 of value. This, in turn, creates a reduction in consumer and producer surplus by diminishing the value of trade between these parties that would otherwise be achieved – a deadweight loss. As such, deadweight losses represent a real net cost to the economy.

8.7.1 Welfare payments

The cost of welfare payments themselves are not included in the cost of IRDs. However, the deadweight losses arising from raising and administering welfare payments are included in the total cost of IRDs.

Persons living with an IRD may be eligible for several forms of government support:

- Attendance Allowance
- Constant Attendance Allowance
- Disability Living Allowance (DLA)/Personal Independence Payment (PIP)
- Disability Premium
- Disabled Student's Allowance
- Employment and Support Allowance (ESA)
- Enhanced Disability Premium
- Incapacity Benefits
- NHS Low Income Scheme
- Severe Disability Premium.

Additionally, informal carers of a person or persons living with an IRD may be eligible to receive the 'Carer's Allowance'.

A description of each of these welfare payments is provided in Appendix C.

Welfare payments were estimated by applying the average utilisation of welfare payments to the weekly entitlement associated with each payment. Average utilisation was estimated using country specific survey data of the proportion of respondents who received each payment. The weekly entitlement amount for each welfare payment was calculated

based on the weighted average amount received by all recipients of that welfare type[6].

The majority of participants from the UK (83.0%) reported receiving support from their government (Table 8.19).

Table 8.19: Welfare payment utilisation of survey participants in the UK (n= 53)

Government support utilisation	Count	Percentage (%)
Supported	44	83.0
Unsupported	9	17.0
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

[6] Individuals receive different amounts for the same welfare type depending on their level of need.

Table 8.20: Welfare payments received by persons living with an IRD (£ 2019) by welfare type

Type	Welfare	Weekly entitlement
Individual	ESA	107.43
	DLA/PIP	102.50
	Attendance Allowance	71.78
	Severe Disability Premium	65.85
	Enhanced Disability Premium	16.80
Carer	Carer's Allowance	66.15

Source: UK Department of Work and Pensions (2019).

These average welfare payments were applied to data on individual payment types to estimate the total cost of welfare payments to persons living with an IRD, which was £115.5 million in 2019. In addition, the total cost of welfare payments to carers of persons living with an IRD was estimated to be £2.7 million in 2019. Therefore, the total welfare payments due to IRDs were estimated to be £118.2 million in 2019.

8.7.2 Reduced taxation revenue

Lower employment participation and lower output (e.g. due to absenteeism or presenteeism) reduce the possible taxation revenue government can collect. The reduction in taxation revenue was estimated by applying an average personal income or company tax rate – which were assumed to be 23.4% and 19.0%, respectively – to lost individual or company earnings.

The total reduction in taxation revenue was estimated to be £45.4 million in 2019.

8.7.3 Deadweight loss of taxation payments and administration

The deadweight loss due to lost taxation revenue (given an assumption of no change in spending) or additional expenditure on government programs (e.g. health or welfare) can be estimated by applying the marginal burden of taxation to the total of lost taxation and government expenditures.

Kleven and Kreiner (2006) provide estimates of the marginal welfare cost of raising additional tax revenue (marginal cost of public funds (MCPF) for five European countries using micro data on taxes, benefits paid, and labour supply elasticities across different income levels. Within this study, nine estimates of the MCPF were provided for the UK, ranging from 0.93 to 1.36 (where 1 represents zero MCPF) and based on alternative scenarios regarding alternative income elasticity scenarios.

For the purposes of this study we have used the simple average of the MCF across the nine scenarios, calculated as 1.12. Consequently for every additional £1 raised by the UK government to fund costs associated with sight loss and blindness it has been assumed there is £0.12 in lost welfare due to deadweight loss.

The total deadweight loss due to IRDs were estimated to be £27.4 million in 2019, as shown in Table 8.21.

Table 8.21: Deadweight losses due to IRDs (£ million 2019)

Cost type	Total cost	Resulting deadweight loss
Welfare payments	118.2	14.7
Government programs (e.g. health)	56.9	7.1
Lost individual taxes	20.8	2.6
Lost carer taxes	19.8	2.5
Lost company taxes	4.8	0.6
Total	**220.5**	**27.4**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

8.8 Results

Total other costs due IRDs in the UK in 2019 were estimated to be £188.1 million. Informal care resulted in the greatest proportion of other costs at £84.5 million (45%), followed by aids and modifications (£44.7 million, 24%) and deadweight losses (£28.7 million, 15%) (Table 8.21). Other costs by type are summarised in Table 8.22.

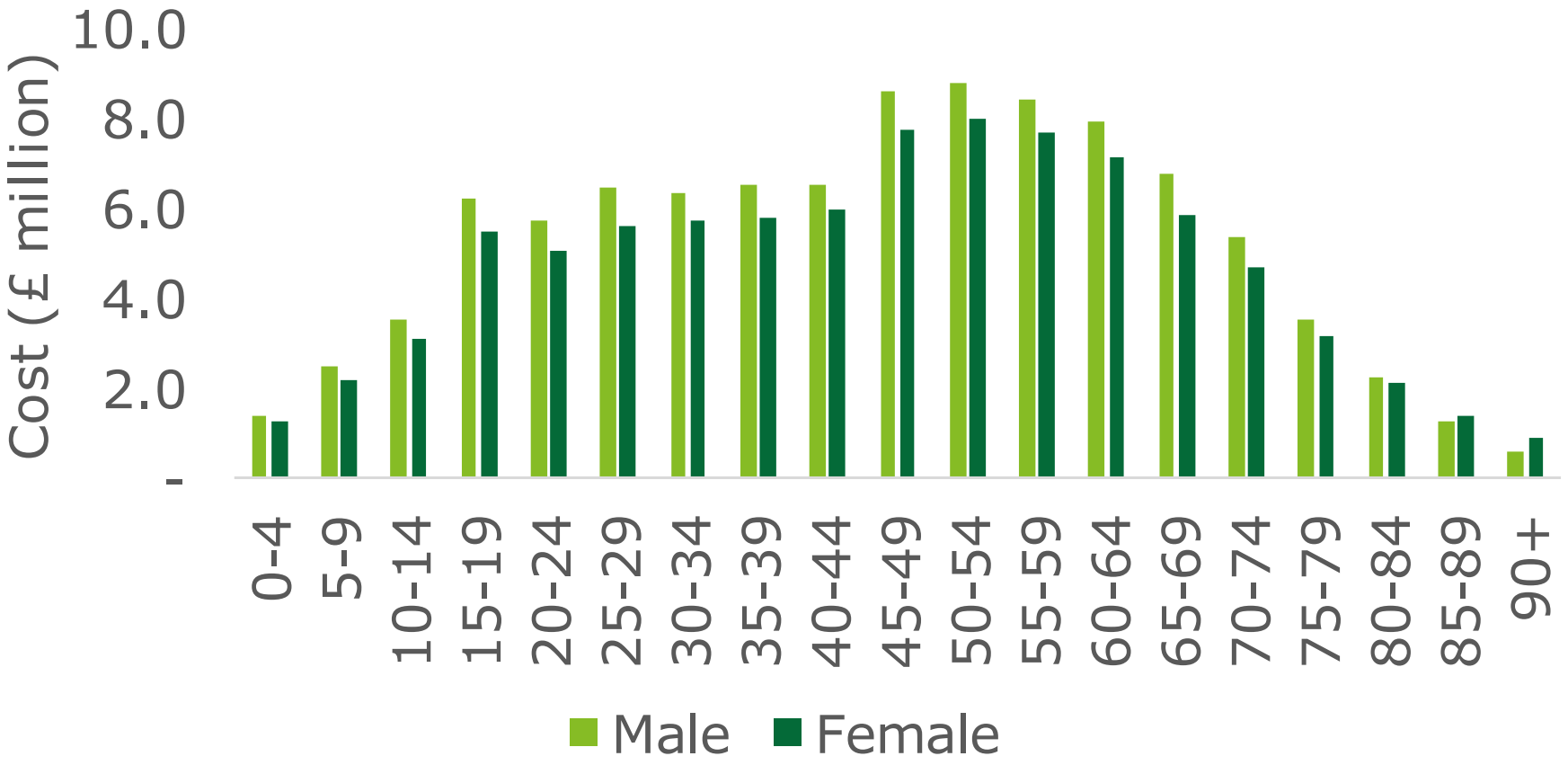
The largest component of this cost is borne by families (36%), with government, individuals and society incurring 25%, 24% and 15% of the costs, respectively. Other costs by payer are summarised in Chart 8.2.

Table 8.22: Total other costs of IRDs (£, 2019) in the UK by cost type

Cost type	Cost per person (£)	Total cost (£ million)
Informal care	4,059	84.5
Aids and modifications	2,148	44.7
Deadweight losses	1,379	28.7
Formal care (community)	628	13.1
Travel	413	8.6
Residential care	218	4.5
Education	191	4.0
Total		**188.1**

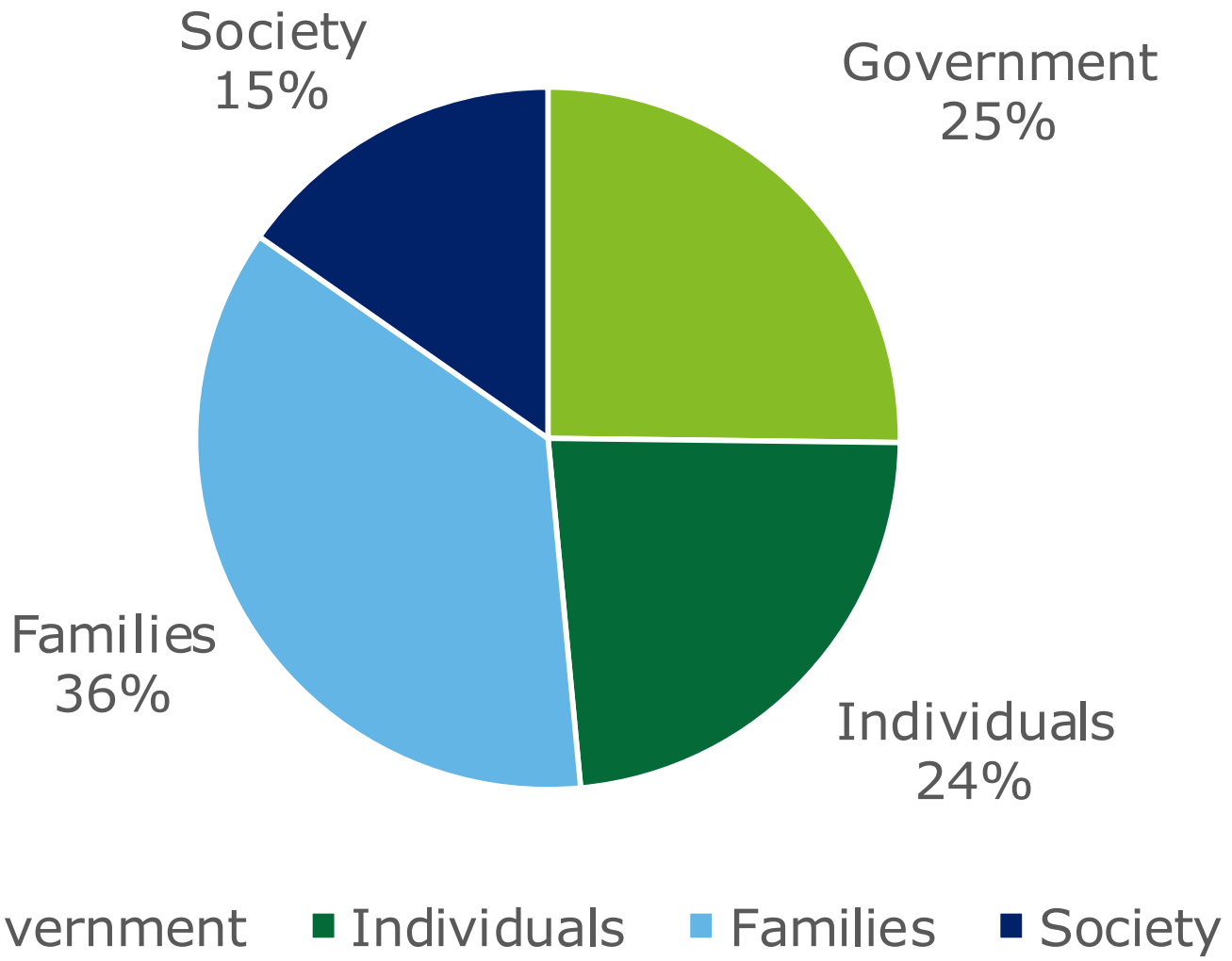
Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Chart 8.1: Total other costs of IRDs (£ million, 2019) in the UK by age and sex



Source: Deloitte Access Economics analysis.

Chart 8.2: Proportion (%) of other costs of IRDs in the UK by payer



Source: Deloitte Access Economics analysis.

9 Wellbeing costs

This section describes the approach used to estimate wellbeing costs associated with IRDs in the UK in 2019.

Key findings

- In 2019, persons living with an IRD in the UK were estimated to experience a total of 3,193 disability adjusted life years (DALYs).
- Total wellbeing costs associated with IRD in 2019 were estimated to be £9,421 per person and £196.1 million in total.

9.1 Summary of approach

Wellbeing costs were estimated using the World Health Organization (WHO) burden of disease methodology. This is a non-financial approach, where pain, suffering and premature mortality are measured in terms of DALYs.

DALYs are composed of premature mortality (years of life lost due to premature death – YLL) and morbidity (years of healthy life lost due to disability – YLD) components. DALYs are calculated by assigning disability weights to various health states, where zero represents a year of perfect health and one represents death. Other health states are given a weight between zero and one to reflect the quality of life that is lost due to a particular condition. For example, a disability weight of 0.2 is interpreted as a 20% loss in the quality of life relative to perfect health for the duration of the condition.

Disability weights were obtained from the IHME's *Global Burden of Disease Study 2017* and are classed into three levels of severity (see 0). A disability weight were applied to the prevalence of IRDs in the UK and discounted at a rate of 3% consistent with WHO methodology (WHO, n.d.).

Table 9.1: Disability weights per person due to 'other vision loss' (2017)

Sequela	Health state (severity of VI)	Health state description	Disability weight
Moderate vision impairment due to other vision loss	Distance vision, moderate impairment	Vision problems that make it difficult to recognize faces or objects across a room	0.031
Severe vision impairment due to other vision loss	Distance vision, severe impairment	Severe vision loss, which causes difficulty in daily activities, some emotional impact, and some difficulty going outside the home without assistance	0.184
Blindness due to other vision loss	Distance vision blindness	Completely blind, which causes great difficulty in some daily activities, worry and anxiety, and great difficulty going outside the home without assistance	0.187

Source: IHME (2018).

For the purposes of this report, a single disability weight of 0.153 was applied to DALYs. This reflects an average of the three disability weights by severity, weighted by the estimated distribution of prevalent IRD cases in the UK across the three levels of severity.[7]

DALYs were converted into pounds using an estimate of the value of a statistical life year (VSLY). The VSLY is an estimate of the value society places on an anonymous life. The VSLY in the UK was sourced from 'The Green Book', published by HM Treasury, which is the 'central government guidance on appraisal and evaluation' in the UK (HM Treasury, 2018). The value was £60,000 per person as at April 2018 and was inflated using the UK Health Consumer Price Index (CPI) to reflect the VSLY as at April 2019 (see Table 9.2).

Table 9.2: UK VSLY (£)

Date	VSLY
April 2018	60,000
April 2019	61,411

Source: HM Treasury (2018), ONS (2019), Deloitte Access Economics analysis.

These estimates are supported by the survey results wherein the vast majority of participants reported experiencing negative impacts on their wellbeing as a result of their IRD, with 86.6% reporting experiencing anxiety, 67.1% depression, 50.0% social isolation, 45.1% financial stress and 14.6% 'other' wellbeing effects (Table 9.3). The 'other' category included negative impacts on persons living with an IRD wellbeing due

[7] The distribution of cases of VI/blindness across the severities was estimated based on the average of studies by Bunce (2006), Bunce (2010) and Quartilho (2016), using vision loss certifications in the UK (NHS, 2017) and the estimated prevalence of IRDs in the UK.

to fear, stress, fatigue, hopelessness, loneliness, panic attacks, suicidal thoughts and the development of tics.

Table 9.3: Wellbeing status of survey participants in the UK (n=53)

Wellbeing (persons living with an IRD)	Percentage (%)*
Anxiety	86.8
Depression	67.9
Social isolation	56.6
Financial stress	45.3
Other	13.2

Source: Deloitte Access Economics analysis
*This question allowed for the selection of multiple answers, hence, the percentages did not sum to 100.

Furthermore, the wellbeing effects of IRDs are not limited to persons living with an IRD. 51.2% of survey participants reported that close family members had experienced feelings of depression, anxiety, or another mental health condition as a result of their IRD. The parents of children (under 18) living with an IRD reported feelings of anxiety (17.1%), depression (13.4%), and 'other' mental health impacts (3.7%) (Table 9.4). The 'other' category included negative impacts on the parents of children (under 18) living with an IRD wellbeing due to stress, loneliness and concerns for the safety of their child.

In addition, the majority of persons living with an IRD (73.2%) and the parents of children (under 18) living with an IRD (75%) were frustrated by the lack of awareness and support for IRDs (Table 9.5).

Table 9.4: Wellbeing status of survey participants (the parents of children (under 18)) in the UK (n=9)

Wellbeing (the parents of children (under 18) living with an IRD)	Percentage (%)*
Anxiety	77.8
Depression	77.8
Other mental health impacts	22.2

Source: Deloitte Access Economics analysis.
*This question allowed for the selection of multiple answers, hence, the percentages did not sum to 100.

Table 9.5: Degree of survey participant frustration with the lack of awareness and support for IRDs in the UK (n=53)

	Percentage (%)				
Participant type	**Strongly disagree**	**Disagree**	**Neutral**	**Agree**	**Strongly agree**
Persons living with an IRD	7.5	1.9	17.0	30.2	26.4
The parents of children (under 18) living with an IRD	1.9	0.0	1.9	7.5	5.7

Source: Deloitte Access Economics analysis.

9.2 Results

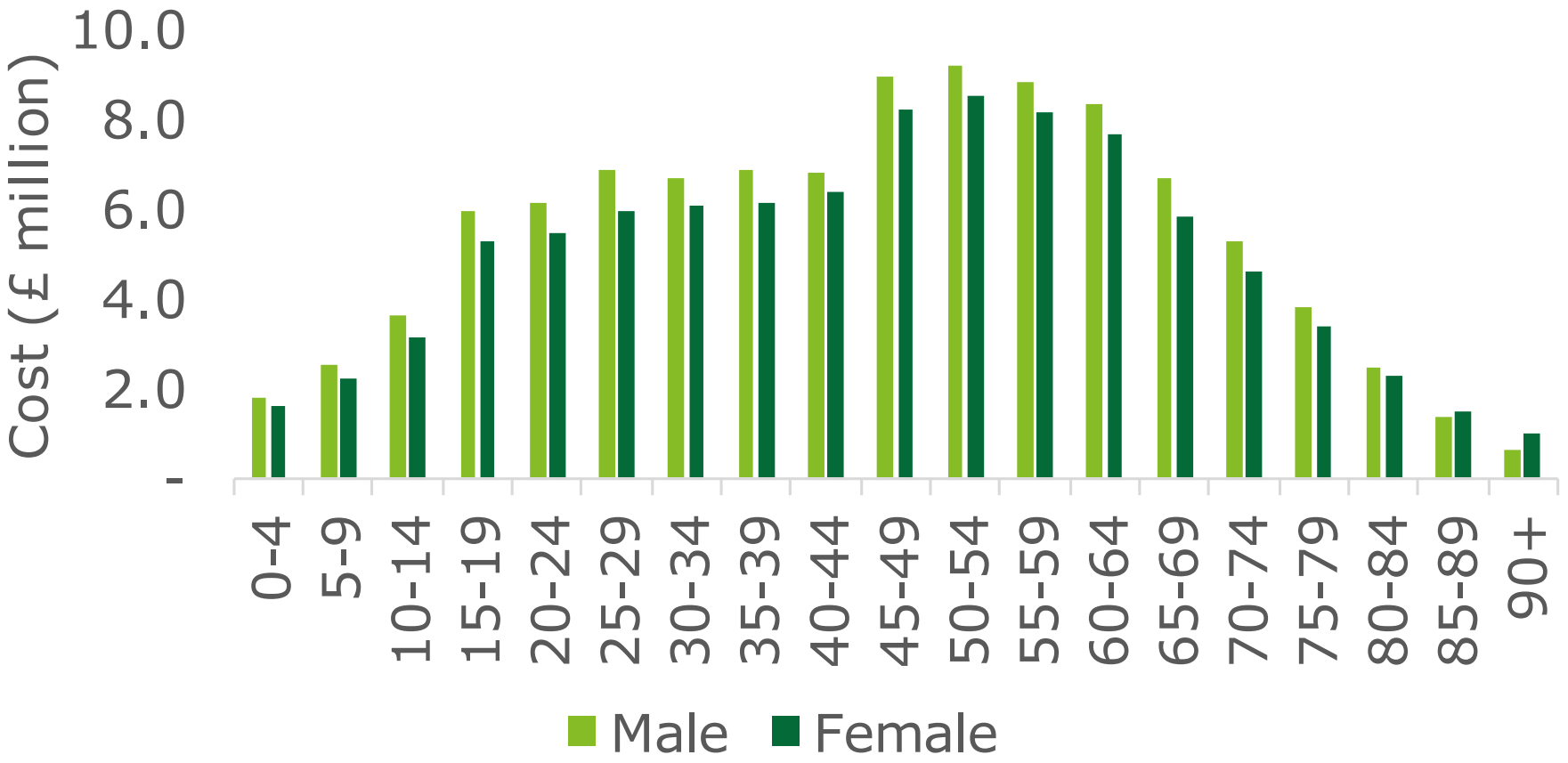
In 2019, persons living with an IRD in the UK were estimated to experience 3,193 DALYs overall. This equated to total wellbeing costs of £196.1 million or £9,421 per person (see Table 9.6). Wellbeing costs by age and sex are displayed in Chart 9.1.

Table 9.6: Total DALYs and wellbeing costs of IRDs (2019) in the UK

Parameter	Per person (£)	Total (£ million)
DALYs	0.15	3,193
Wellbeing costs	**9,421**	**196.1**

Source: Deloitte Access Economics analysis based on IHME (2016), HM Treasury (2018).

Chart 9.1: Total wellbeing costs of IRDs (£ million, 2019) in the UK by age and sex



Source: Deloitte Access Economics analysis based on IHME (2016), HM Treasury (2018).

10 Sensitivity analysis

In many cases, the inputs underlying the cost-of-illness analysis are uncertain and changes in these inputs may have a significant impact upon the total estimate of the costs of IRDs in the UK in 2019. Given the limited published data on the prevalence of IRDs in the UK, sensitivity analysis using an upper bound sensitivity estimate based on the highest published prevalence estimate available was undertaken.

The overall upper bound prevalence rate was based on Orphanet (2019), which reports estimated prevalence for a range of rare diseases, including IRDs. While this source reports that its underlying research is based on a systematic review of PubMed, grey literature, and expert consultation (Orphanet, 2019), primary data informing these estimates is difficult to ascertain and verify, and therefore does not inform the base estimates.

Using this approach, an upper bound prevalence rate of 0.0525% was estimated for the UK – 68.8% higher than the base prevalence estimate of 0.0311% reported in this analysis.

The upper bound prevalence rate of RP is in line with that reported by O'Neill et al (2007), who estimated the prevalence of RP in Northern Ireland. However, this study was excluded from further analysis since the full text could not be located. Similarly, Stone (2007) conducted a study which examined the prevalence of LCA in the United States. This study was also excluded from analysis in favour of European-specific prevalence results provided by Bertelsen et al (2014).

0 and Table 10.2, below, outline the base case and upper bound prevalence estimates for each IRD included in this study.

Table 10.1: Prevalence of IRDs in the UK (2019) by condition (base case and upper bound)

	Base case			Upper bound		
IRD	**Rate (%)**	**Cases**	**Proportion (%)**	**Rate (%)**	**Cases**	**Proportion (%)**
RP	0.0154	10,324	49.6	0.0261	17,422	49.60
Usher syndrome	0.0039	2,590	12.4	0.0065	4,371	12.44
Stargardt	0.0032	2,109	10.1	0.0053	3,559	10.13
LCA/EOSRD	0.0024	1,587	7.6	0.0040	2,679	7.63
Best disease	0.0015	1,003	4.8	0.0025	1,693	4.82
Cone dystrophy	0.0013	876	4.2	0.0022	1,478	4.21
Cone-rod dystrophy	0.0012	812	3.9	0.0020	1,370	3.90
Achromatopsia	0.0011	733	3.5	0.0018	1,237	3.52
XLRS	0.0007	483	2.3	0.0012	814	2.32
Choroideremia	0.0004	298	1.4	0.0008	504	1.43
Total IRDs	**0.0311**	**20,814**	**100.0**	**0.0525**	**35,126**	**100.00**

Source: Deloitte Access Economics analysis.

Table 10.2: Prevalence of IRDs in the UK (2019) by age and sex (upper bound)

	Male		Female		Total	
Age group	**Rate (%)**	**Cases**	**Rate (%)**	**Cases**	**Rate (%)**	**Cases**
0-9	0.0188	776	0.0174	686	0.0181	1,462
10-19	0.0439	1,709	0.0407	1,505	0.0424	3,214
20-29	0.0524	2,322	0.0483	2,046	0.0504	4,368
30-39	0.0545	2,422	0.0489	2,186	0.0517	4,608
40-49	0.0679	2,829	0.0614	2,609	0.0646	5,438
50-59	0.0725	3,233	0.0649	2,988	0.0686	6,221
60-69	0.0776	2,696	0.0660	2,416	0.0717	5,111
70-79	0.0610	1,627	0.0481	1,434	0.0542	3,060
80+	0.0582	792	0.0426	851	0.0489	1,644
Total	**0.0557**	**18,406**	**0.0494**	**16,720**	**0.0525**	**35,126**

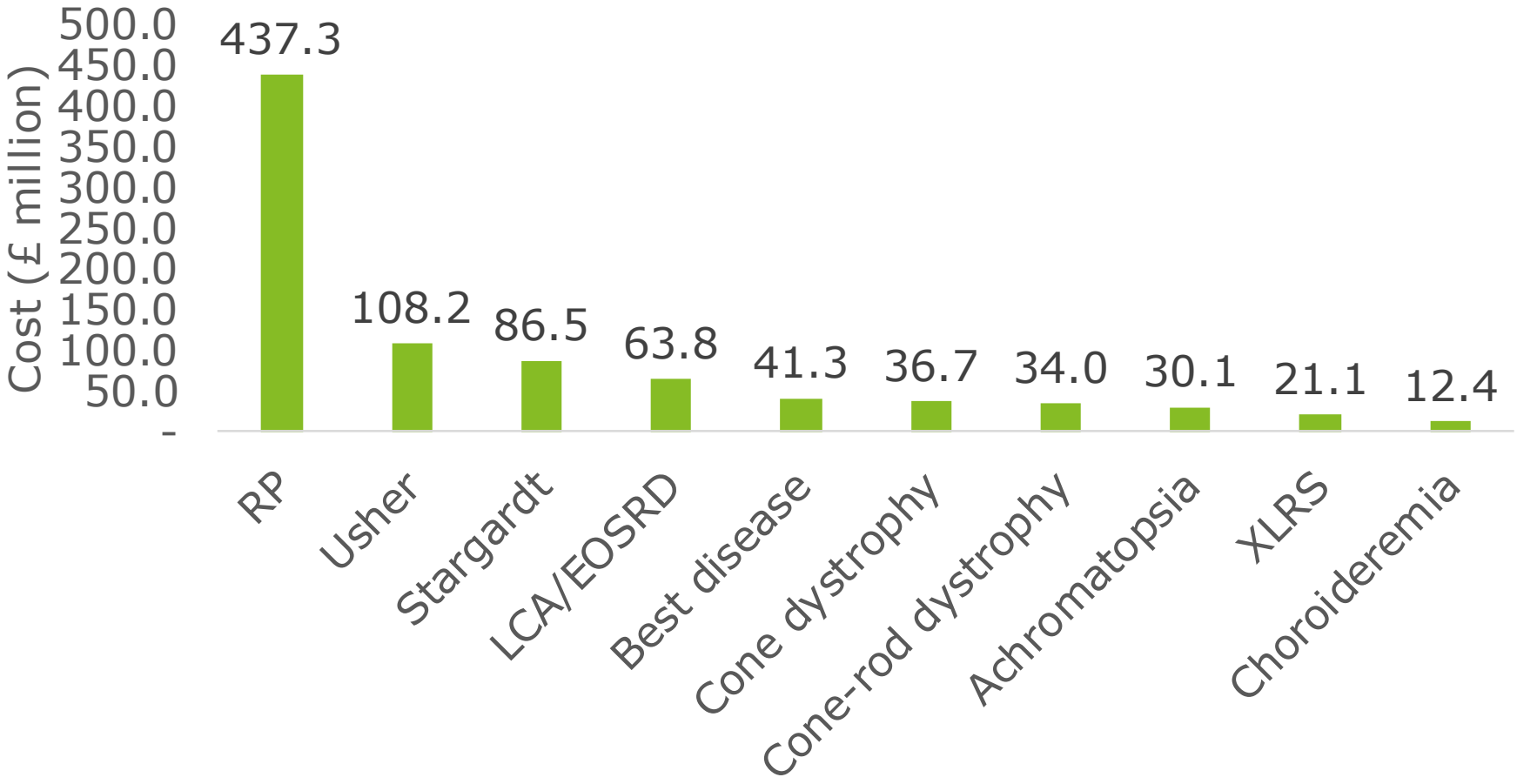
Source: Deloitte Access Economics analysis.

10.1 Results

The upper bound sensitivity scenario yielded an overall estimate of the cost of IRDs in the UK of £871.6 million. The variance between upper and lower bound costs by condition are shown in Chart 10.1 while Table 10.3 shows the upper bound and base estimates by condition.

The most sensitive components of cost included the wellbeing cost (£330.9 million), followed by productivity costs (£192.6 million) (Chart 10.2). Table 10.4 shows the upper bound and base estimate costs, by component.

Chart 10.1: Sensitivity analysis results (£ million, 2019) in the UK by condition



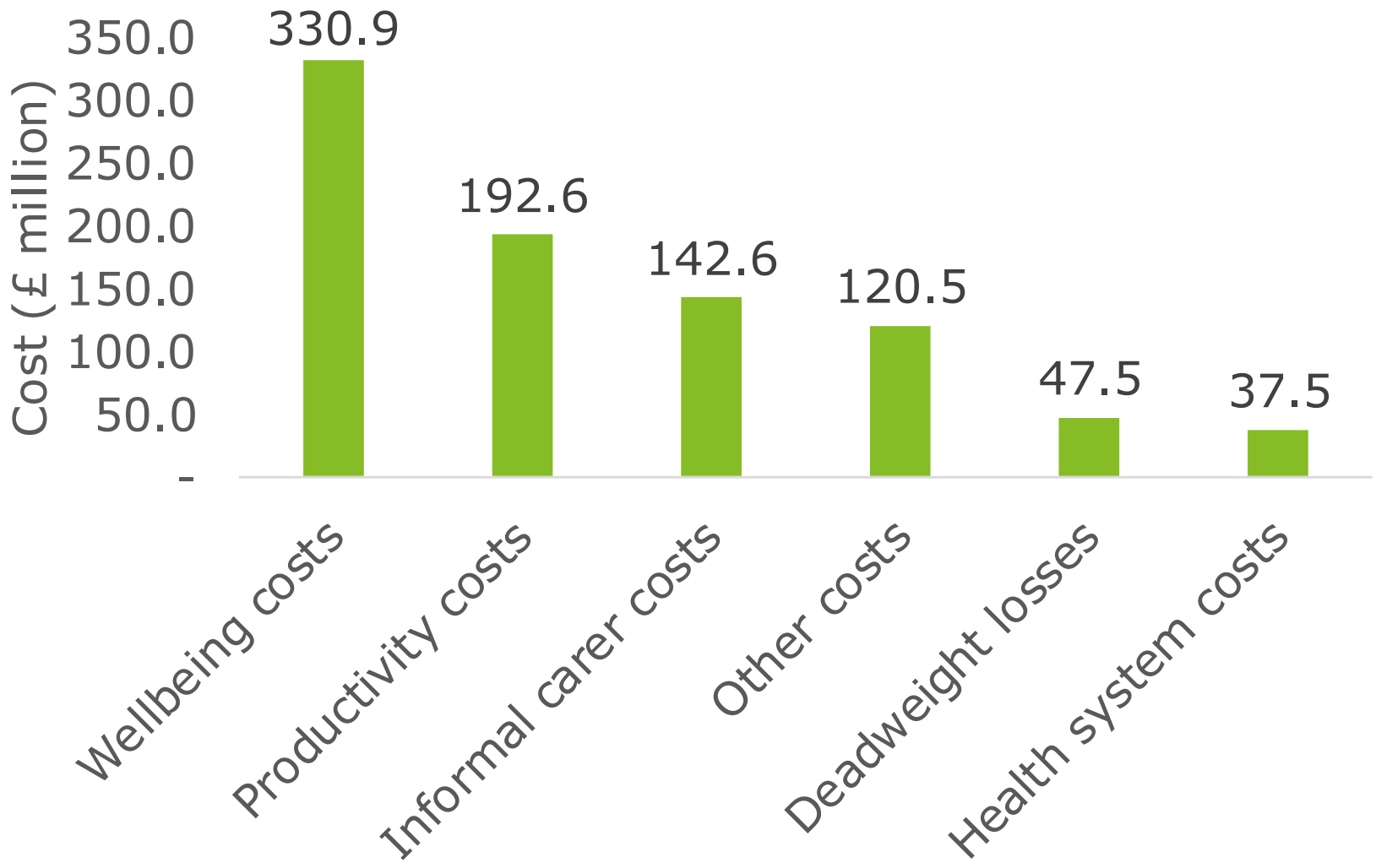
Source: Deloitte Access Economics analysis.

Table 10.3 Sensitivity analysis results (£ million, 2019) in the UK by condition

IRD	Base case	Upper bound
RP	262.3	437.3
Usher	64.9	108.2
Stargardt	52.0	86.5
LCA/EOSRD	38.5	63.8
Best disease	24.8	41.3
Cone dystrophy	22.1	36.7
Cone-rod dystrophy	20.5	34.0
Achromatopsia	18.1	30.1
XLRS	12.6	21.1
Choroideremia	7.5	12.4
Total IRDs	**523.3**	**871.6**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Chart 10.2: Sensitivity analysis results (£ million, 2019) in the UK by cost type



Source: Deloitte Access Economics analysis.

Table 10.4: Sensitivity analysis results (£ million, 2019) in the UK by cost type

Cost type	Base case	Upper bound
Wellbeing costs	196.1	330.9
Productivity costs	114.1	192.6
Informal carer costs	84.5	142.6
Other costs	74.9	120.5
Deadweight losses	28.7	47.5
Health system costs	25.0	37.5
Total costs	**523.3**	**871.6**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

11 Conclusion

11.1 Summary of total costs

This analysis found that IRDs imposed significant economic and wellbeing costs on the UK population in 2019. As it currently stands, persons living with an IRD incur significant economic costs and reductions in their quality of life. In addition to the costs incurred by these persons, their families, friends, government, employers and society all incur significant economic costs due to IRDs.

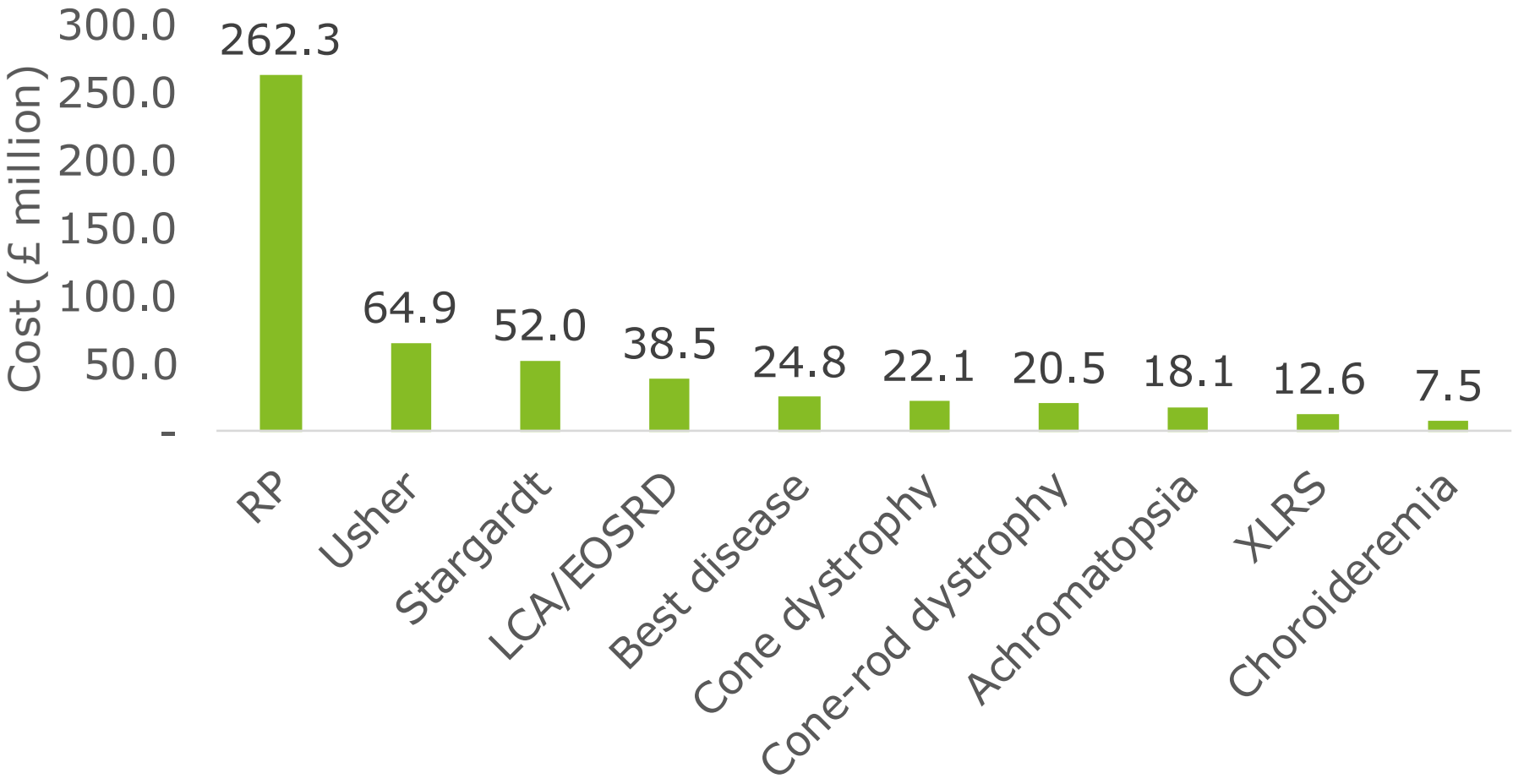
Total costs attributable to IRDs in the UK were estimated to be £523.3 million in 2019, comprising both economic costs (£327.2 million) and wellbeing costs (£196.1 million).

Of the ten IRDs within scope, RP incurred the greatest proportion of total costs at £262.3 million (50.1%), followed by Usher syndrome (£64.9 million, 12.4%), Stargardt disease (£52.0 million, 9.9%), and LCA/EOSRD (£38.5 million, 7.4%) (Chart 11.1 and Table 11.1).

Wellbeing costs comprised the largest share of total costs at £196.1 million (37.5%), followed by productivity costs (£114.1 million, 21.8%), informal carer costs (£84.5 million, 16.1%), other costs (£74.9 million, 14.3%), deadweight losses (£28.7 million, 5.5%) and health system costs (£25.0 million, 4.8%) (Chart 11.2 and Table 11.2). This means that in addition to imposing significant economic costs, IRDs result in pain and suffering that leads to a significant wellbeing cost for persons living with an IRD.

Individuals bear the largest share of total costs at 36%, followed by government (28%), family and friends (21%), society/others (9%), and employers (6%) (Chart 11.3).

Chart 11.1: Total costs of IRDs (£ million, 2019) in the UK by condition



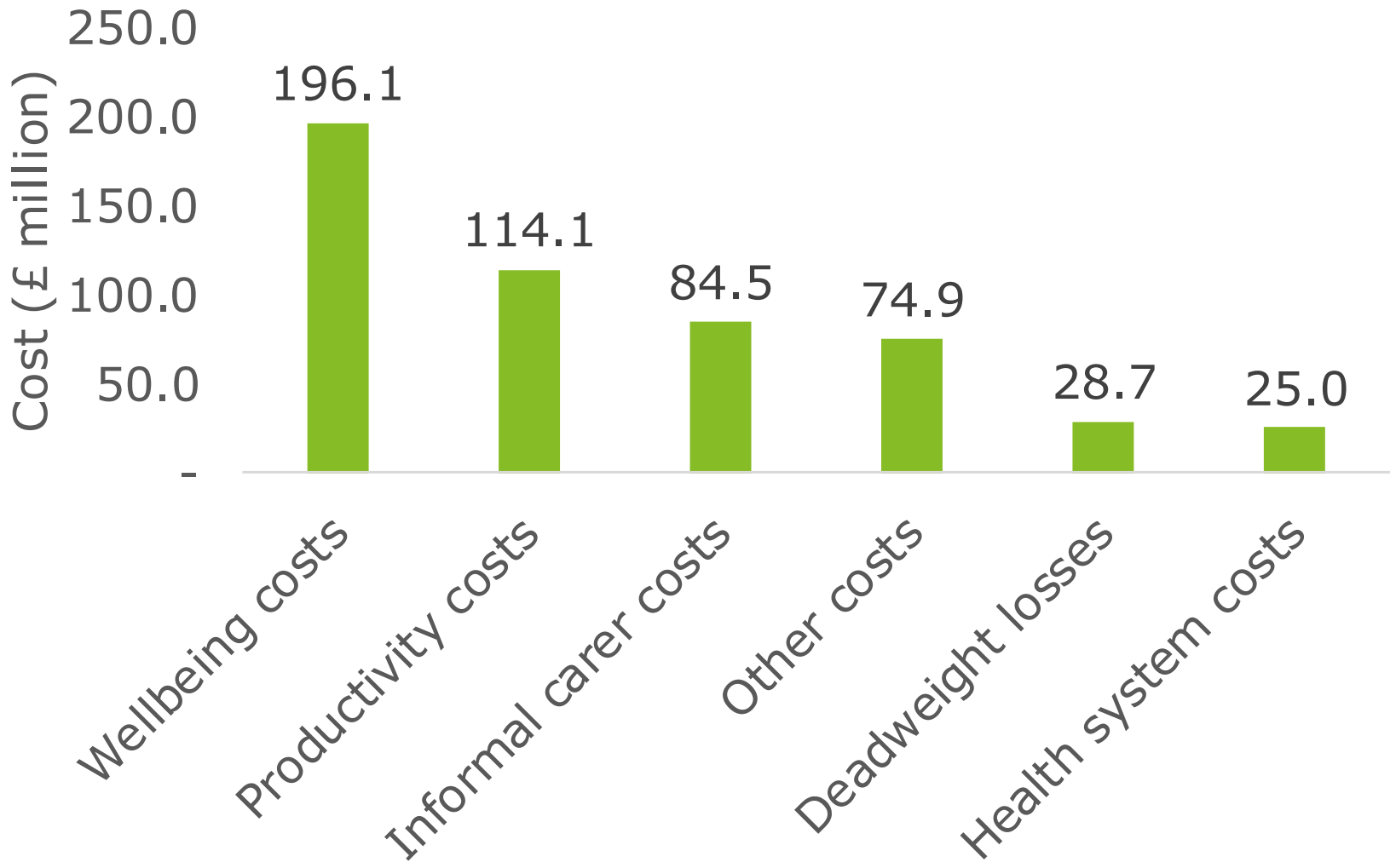
Source: Deloitte Access Economics analysis.

Table 11.1: Total costs of IRDs (£ million, 2019) in the UK by condition

IRD	Cost
RP	262.3
Usher	64.9
Stargardt	52.0
LCA/EOSRD	38.5
Best disease	24.8
Cone dystrophy	22.1
Cone-rod dystrophy	20.5
Achromatopsia	18.1
XLRS	12.6
Choroideremia	7.5
Total	**523.3**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Chart 11.2: Total costs of IRDs (£ million, 2019) in the UK by cost type



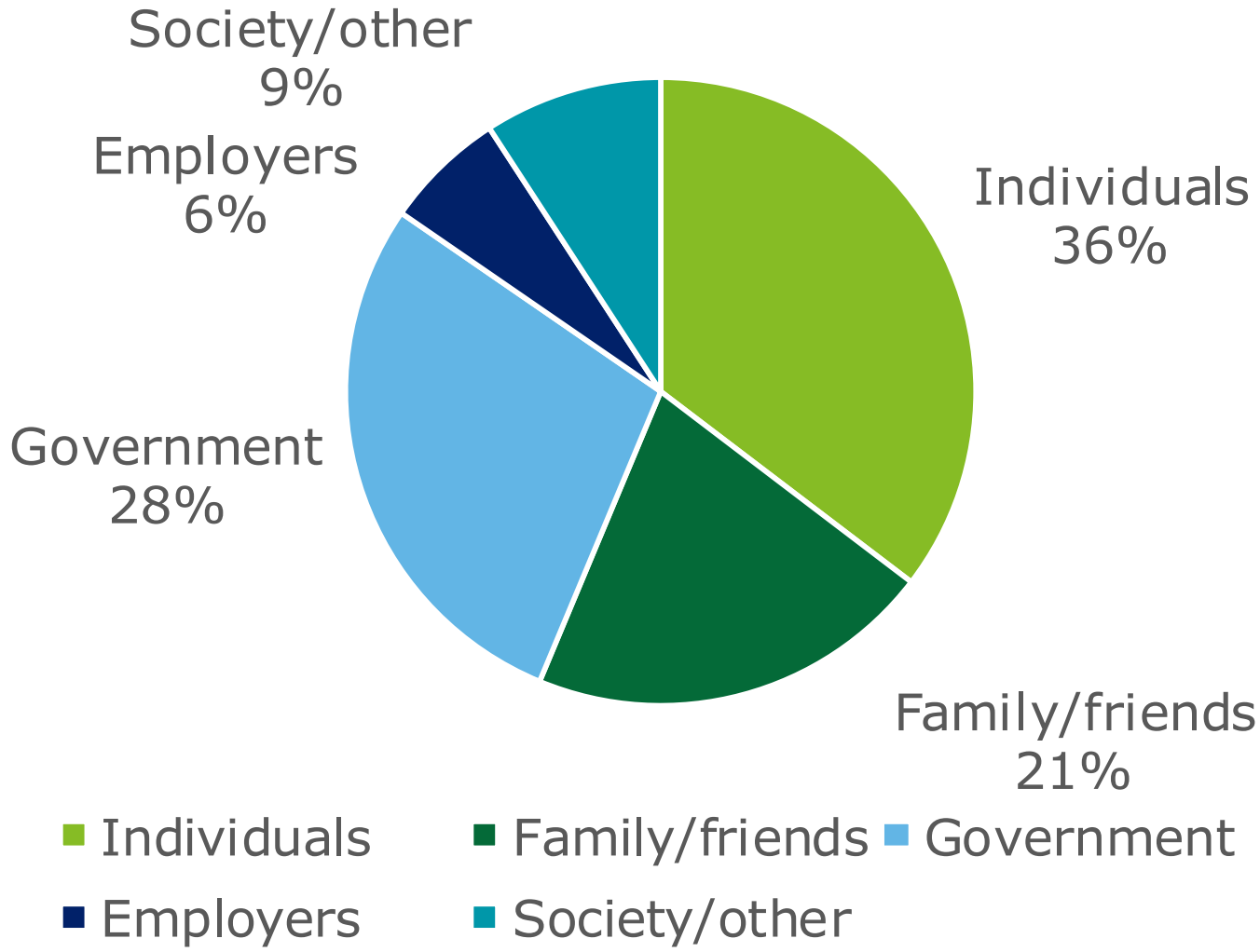
Source: Deloitte Access Economics analysis.

Table 11.2: Total costs of IRDs (£ million, 2019) in the UK by cost type

Cost type	Percentage of total cost (%)	Total cost
Loss of wellbeing	37.5	196.1
Productivity costs	21.8	114.1
Informal carer costs	16.1	84.5
Other costs	14.3	74.9
Deadweight losses	5.5	28.7
Health system costs	4.8	25.0
Total costs	**100.0**	**523.3**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

Chart 11.3: Proportion (%) of total costs of IRDs in the UK by payer (2019)



Source: Deloitte Access Economics analysis.

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Appendix A Additional information on survey results

This appendix details the survey questionnaire distributed to persons living with an IRD and/or the parents of children (under 18) living with an IRD.

The questionnaire comprised 12 modules:

1. About you
2. Your current condition
3. Work
4. Education
5. Management of your condition – health care providers
6. Management of your condition – medications and supplements
7. Management of your condition – vision aids and modifications and travel
8. Formal care
9. Informal care
10. Government support
11. Other impacts of your condition
12. Impact of your child's IRD on your mental wellbeing – from your own perspective as a parent/ guardian.

Skip logic was incorporated into the programming of the survey to ensure only relevant questions were asked of participants, depending on the answers provided to previous questions.

The questions asked, answers available and results received for each question are outlined in this appendix. In instances where a low number of results were received, results are aggregated to protect participant privacy and confidentiality.

A.1. Introduction

Deloitte Access Economics (Australia) is assisting Retina International and its partner organisations to better understand the real impact of inherited retinal dystrophies (IRDs). This survey aims to evaluate the financial and social impact of IRDs.

The findings will inform two reports, one each for the UK and the Republic of Ireland that will estimate the socioeconomic impact of IRDs in the UK and Ireland. The reports, including the summary findings from this survey, will be published on the websites of Retina International and its partner organisations.

Information collected through this survey will inform knowledge and understanding of:

- the impact on the wellbeing of individuals affected and their families; and
- the economic cost of IRDs to health and social care, education and employment and carers.

A.1.1. Purpose of the study

You are invited to participate in an online survey, which is being conducted in order to better understand IRD impacts. The survey is open to people who have or are the parent/guardian of someone with any of the following conditions:

- Retinitis pigmentosa (RP)
- Leber congenital amaurosis (LCA)/early onset severe retinal dystrophy to LCA (LCA/EOSRD)
- Usher syndrome
- Stargardt disease
- Choroideremia
- X linked retinoschisis
- Achromatopsia
- Cone dystrophy
- Cone rod dystrophy
- Best disease.

This survey contains up to 12 sections called modules and may take approximately 50 minutes to complete.

This survey will collect personal information about you including demographic information (such as your age and gender) and details about your work and education. It will also collect personal information about you that may be considered special category (or "sensitive") personal information, including your ethnicity, details about your condition, how you manage your condition, your use of healthcare services and your wellbeing.

Your personal information will be used to estimate the economics costs of IRDs and to describe the characteristics of groups of people (not individuals) who have or are the parent/guardian of someone with a diagnosed IRD.

Do you consent to Retina International and associated partners (IRD COUNTS- a consortium of patient organisations and industry partners advocating on behalf of the IRD community) to contact you in the future for further relevant research?

- Yes
- No

A.1.2. Confidentiality

Your personal information and special category information collected as part of this survey will be kept strictly confidential. Your identity will not be revealed and your confidentiality will be protected in any reviews and reports of this study which may be published.

While we use a third party survey platform provider to assist us in administering this survey, this provider is bound by strict confidentiality and privacy obligations. This means that your information will only be accessed by authorised personnel where required for the purpose of administering the survey and conducting the analysis described above.

A.1.3. The Legal basis for processing personal information

Before you complete this survey you will be asked to provide your consent to the collection and processing of your personal information and special category information for the purpose described above. Your consent will then form the legal basis upon which we are able to process your information.

A.1.4. Voluntary participation/right to refuse or withdraw

Your participation in this survey is voluntary; and you can withdraw at any time if you change your mind. If you decide to withdraw your consent by exiting the survey mid-way through, your responses will not be recorded.

You have various rights in relation to your personal information, including the right to seek access to, or to correct,

your information (for more information, please see our Privacy Statement).

If you are located in Europe, you also have the right to:

- ask that we delete personal information that we hold about you, or restrict the way in which we use your personal information;
- withdraw consent to our processing of your personal information; and
- obtain and/or move your personal information to another service provider.

However, we may still be required to retain a copy of certain information where it is necessary to comply with applicable laws or professional standards.

A.1.5. Contact details should you experience any unsettled feelings during this survey

If you experience any unsettled feelings while completing this survey, please contact SAMARITANS who provide confidential support in Ireland or the UK for free from any phone on 116 123. If you have any concerns about your IRD care and management, please contact your general practitioner/preferred health provider.

Fighting Blindness Ireland has a free counselling service for people and families living with sight loss.

The contact details for participants in Ireland are by email, insight@fightingblindness.ie, or by phone 01 674 6496.

For any questions about the objectives of the survey, how your data will be used or to exercise any of your rights, please contact the Deloitte Privacy Officer at privacy@deloitte.com.au

For any other questions please contact: irds@deloitte.com.au

A.2. Eligibility and Consent

Who are you completing this survey for?

- Myself as a person with an IRD
- As the parent/guardian of a child (aged up to 18 years old) with IRD
- On behalf of an adult with IRD as their carer

If you would like to proceed with participating in this survey, please provide consent.

By providing consent you are telling us that you:

- Understand what you have read.
- You are 18 years or over.
- Consent to take part in the research project.
- Consent to the processing of your personal information for the purpose described in the introduction to this survey
- Consent to the processing of your health and other types of special category information for the purpose described in the introduction to this survey.

Please confirm you have read and understand the information provided. By selecting 'yes' you agree to your involvement in the survey to understand the real impact of IRDs.

- Yes, I voluntarily consent
- No, I do not consent

A.3. Preliminary questions

Which of the following best describes how you will respond to this survey?

- As myself a person affected with an IRD
- As the parent/guardian of a child (aged up to 18 years old) with an IRD

Table A.1: Which of the following best describes how you will respond to this survey? (n=53) (UK specific)

Participant type (Adult or Parent)	Count (n)	Percentage (%)
Myself as a person with an IRD	44	83
As the parent/guardian of a child (aged up to 18 years old) with IRD	9	17
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

A.4. Module 1: About you

Which IRD are you diagnosed with?

- Retinitis pigmentosa (RP)
- Leber congenital amaurosis (LCA)
- Usher syndrome
- Stargardt disease
- Choroideremia
- X linked retinoschisis
- Achromatopsia
- Cone dystrophy
- Cone rod dystrophy
- Best disease

Table A.2: Which IRD are you diagnosed with? (n=53) (UK specific)

Participant condition	Count (n)	Percentage (%)
RP	35	66.0
Other	7	13.2
Stargardt disease	6	11.3
Usher syndrome	5	9.4
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

What form of retinitis pigmentosa (RP) are you diagnosed with?

- Autosomal dominant RP
- Autosomal recessive RP
- X linked RP
- Unsure

Table A.3: What form of retinitis pigmentosa (RP) are you diagnosed with? (n=35) (UK specific)

Participant condition	Count (n)	Percentage (%)
Unsure	15	42.9
Autosomal dominant RP	9	25.7
X linked RP	6	17.1
Autosomal recessive RP	5	14.3
Total	**35**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

What was your biological sex at birth?
- Female
- Male
- Prefer not to say

Table A.4: What was your biological sex at birth (n=53) (UK specific)

Biological sex	Count (n)	Percentage (%)
Female	28	52.8
Male	25	47.2
Not stated	0	0.0
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

What is your age (all ages)?
- 5 to 9
- 10 to 14
- 15 to 17
- 18 to 24

- 25 to 34
- 35 to 44
- 45 to 54
- 55 to 64
- 65 to 74
- 75 or older

Table A.5: What is your age (all ages)? (n=53) (UK specific)

Age	Count (n)	Percentage (%)
Under 18	9	17.0
18-24	0	0.0
25-34	1	1.9
35-44	7	13.2
45-54	13	24.5
55-64	11	20.8
65-74	10	18.9
75+	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

What is your ethnicity?

- White including: English/Welsh/Scottish/Northern Irish/British/Irish/Irish Traveller/Other White background
- Asian/Asian British/Asian Irish including: Indian/Pakistani/Bangladeshi/Chinese/Any other Asian background
- Black/African/Caribbean/Black Irish/Black British including: African/Caribbean/Any other Black/African/Caribbean background

- Mixed/Multiple ethnic groups including: White and Black Caribbean/White and Black African/White and Asian/Any other Mixed/Multiple ethnic background
- Other ethnic group

Table A.6: What is your ethnicity? (n=53) (UK specific)

Participant ethnicity	Count (n)	Percentage (%)
White including: English/Welsh/Scottish/Northern Irish/British/Irish/Irish Traveller/Other White background	50	94.3
Non-white	3	5.7
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

Which country do you reside in?
- England
- Wales
- Scotland
- Northern Ireland
- Republic of Ireland

Table A.7: Which country do you reside in? (n=53) (UK specific)

Participant condition	Count (n)	Percentage (%)
England	43	81.1
Northern Ireland	6	11.3
Other	4	7.5
Total	**53**	**100**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

A.5. Module 2: Your current condition

Which of these best describes the vision (if any) you have remaining?

- No vision
- Light perception only (or shadows only)
- Some useful central vision
- Good central vision
- Some useful peripheral vision
- Good peripheral vision
- I still have good overall vision
- Other – please specify

Table A.8: Which of these best describes the vision (if any) you have remaining? (n=53) (UK specific)

Participant condition	Count (n)	Percentage (%)
Some useful central vision	24	45.3
Good central vision	11	20.8
Some useful peripheral vision	11	20.8
Light perception only (or shadows only)	7	13.2
Good peripheral vision	3	5.7
I still have good overall vision	1	1.9
No vision	0	0.0
Total	**53**	**100**

Source: Deloitte Access Economics analysis. Note "Other" responses were reclassified into the relevant categories. This question allowed for the selection of multiple answers, hence, the percentages did not sum to 100.

Are you registered as severely sight impaired/legally blind or sight impaired/partially sighted?

- Yes, I am registered as severely sight impaired/legally blind (defined as best corrected visual acuity of less than 3/60, or less than 6/60 with the presence of contracted field of vision)
- Yes, I am registered as sight impaired/partially sighted (defined as best corrected visual acuity of less than 6/60 and greater than 3/60 with full visual field, or less than 6/24 with moderate contraction of visual field, or less than 6/18 with marked contraction of visual field)
- No, my degree of sight loss does not meet the criteria
- No, I have chosen not to be registered

Table A.9: Are you registered as severely sight impaired/legally blind or sight impaired/partially sighted? (n=53) (UK specific)

Participant condition	Count (n)	Percentage (%)
Yes, I am registered as severely sight impaired/legally blind (defined as best corrected visual acuity of less than 3/60, or less than 6/60 with the presence of contracted field of vision)	32	60.4
Yes, I am registered as sight impaired/partially sighted (defined as best corrected visual acuity of less than 6/60 and greater than 3/60 with full visual field, or less than 6/24 with moderate contraction of visual field, or less than 6/18 with marked contraction of visual field)	16	30.2
No, I have chosen not to be registered	5	9.4
No, my degree of sight loss does not meet the criteria	0	0.0
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

Has a doctor or specialist said that you have hearing loss?
- Yes, mild hearing loss (hearing impairment in the better ear of 20 to 34 dBHL)
- Yes, moderate hearing loss (hearing impairment in the better ear of 35 to 64 dBHL)
- Yes, severe hearing loss (hearing impairment in the better ear of 65 to 94 dBHL)
- Complete hearing loss (hearing impairment in the better ear of greater than 95 dBHL)
- Yes, unsure of level of hearing loss
- No

Table A.10: Has a doctor or specialist said that you have hearing loss (n=5) (UK specific)

Diagnosed hearing loss	Count (n)	Percentage (%)
Yes, any level of hearing loss	5	100.0
No	0	0.0
Total	**5**	**100.0**

Source: Deloitte Access Economics analysis.

Which of these best describes the hearing (if any) you have remaining?
- I do not have any hearing loss
- I do not have problems hearing what is said in a quiet environment, although I have difficulty following a conversation in a noisy environment
- I have difficulty hearing a normal voice in a quiet environment, and I have difficulty taking part in conversation in a noisy environment
- I can hear loud speech directly in my ear in a quiet environment, and I have great difficulty taking part in conversation in a noisy environment
- I am unable to hear in a quiet environment

Table A.11: Which of these best describes the hearing (if any) you have remaining? (n=53) (UK specific)

Self-reported hearing	Count (n)	Percentage (%)
No	48	90.6
Any hearing difficulties	5	9.4
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

A.6. Module 3: Work

Which of the following best describes your current state of work and study?

- Full time work
- Part time work
- Casual work (Not working on a regular basis)
- Underemployed (e.g. working part time but desiring full time work, or working in a role that does not use your skills or qualifications)
- Student only
- Retired (not working)
- Not employed and not studying, and looking for work
- Not employed, and not looking for work

Table A.12: Which of the following best describes your current state of work and study? (n=44) (UK specific)

Employment/study status	Count (n)	Percentage (%)
Retired (not working)	17	38.6
Part time work	13	29.5
Underemployed (e.g. working part time but desiring full time work, or working in a role that does not use your skills or qualifications)	7	15.9
Student only	4	9.1
Full time work	2	4.5
Casual work (Not working on a regular basis)	1	2.3
Not employed and not studying, and looking for work	0	0.0
Not employed, and not looking for work	0	0.0
Total	**44**	**100.0**

Source: Deloitte Access Economics analysis.

What are your average weekly earnings after tax?
- 0 to 250
- 250 to 500
- 500 to 1,000
- 1,000 to 1,500
- 1,500 or more
- Prefer not to say

Table A.13: What are your average weekly earnings after tax? (n=18) (employed persons in the UK)

Average weekly earnings (post tax, £)	Count (n)	Percentage (%)
0 to 500 £	6	33.3
More than 500 £	8	44.4
Prefer not to say	4	22.2
Total	**18**	**100**

Source: Deloitte Access Economics analysis.

How many hours does your employer expect you to work in a typical 7 day week? If your hours vary, estimate the average. If you are self-employed, estimate the number of hours you would consider a full work week. If you have more than one job, combine total number of hours for all jobs.

- Text response

Table A.14: How many hours does your employer expect you to work in a typical 7 day week? (n=18) (UK specific)

	Average	Standard Deviation
Expected work hours	31	13

Source: Deloitte Access Economics analysis.

Now please think of your work experiences over the past 4 weeks. In the past 4 weeks, how many full and partial days of work (example 1.5 days) did you miss because of your IRD? Please include only days missed for your own health, not someone else's health.

- Text response

Table A.15: How many full and partial days of work did you miss because of your IRD? (n=18) (UK specific)

	Average	Standard Deviation
Work days missed due to IRDs	0	0

Source: Deloitte Access Economics analysis.

About how many hours altogether did you work in the past 4 weeks? If you did not work at all in the past 4 weeks, enter "0".

- Text response

Table A.16: How many hours altogether did you work in the past 4 weeks? (n=18) (UK specific)

	Average	Standard Deviation
Actual work hours	92.2	64.9

Source: Deloitte Access Economics analysis.

On a scale from 0 to 10 where 0 is the worst job performance anyone could have at your job and 10 is the performance of a top worker, how would you rate the usual performance of most workers in a job similar to yours?

- 0-10 scale

Table A.17: How would you rate the usual performance of most workers in a job similar to yours? (n=18) (UK specific)

Perceived workplace performance rating	Count (n)	Percentage (%)
6	2	11.1
7	5	27.8
8	5	27.8
9	5	27.8
10	1	5.6
Total	**18**	**100.0**

Source: Deloitte Access Economics analysis.

Thinking about the impact of your IRD on your work, how would you rate your overall job performance on the days you worked during the past 4 weeks, using the same 0 to 10 scale?

- 0-10 scale

Table A.18: How would you rate your overall job performance on the days you worked during the past 4 weeks? (n=18) (UK specific)

Self-assessed performance rating	Count (n)	Percentage (%)
5	2	11.1
6	2	11.1
7	3	16.7
8	8	44.4
9	0	0.0
10	3	16.7
Total	**18**	**100.0**

Source: Deloitte Access Economics analysis.

How would you compare your overall job performance on the days you worked during the past 4 weeks with the performance of most other workers who have a similar type of job?

- You were a lot better than other workers
- You were somewhat better than other workers
- You were a little better than other workers
- You were about average
- You were a little worse than other workers
- You were somewhat worse than other workers
- You were a lot worse than other workers

Table A.19: How would you compare your overall job performance on the days you worked during the past 4 weeks with the performance of most other workers who have a similar type of job? (n=18) (UK specific)

Performance comparison	Count (n)	Percentage (%)
You were a lot worse than other workers	0	0.0
You were somewhat worse than other workers	1	5.6
You were a little worse than other workers	1	5.6
You were about average	4	22.2
You were a little better than other workers	3	16.7
You were somewhat better than other workers	6	33.3
You were a lot better than other workers	3	16.7
Total	**18**	**100.0**

Source: Deloitte Access Economics analysis.

Please select any modifications or aids that you currently use in your working environment specifically or including for your IRD.

- Books, including books with enlarged font, and audio-books
- Laptops (only select if specialised for you)
- Magnifiers
- Spoken word processor, verbal calculator or screen reader
- Braille display
- Ergonomic adaptations e.g. raised screens
- Modifications to mobile phones such as applications (apps)
- Other – please describe

- I do not use any additional items to support my work because I do not need them
- I do not use any additional items to support my work because they are not available

Table A.20: What modifications or aids used in the working environment specifically or including for an IRD? (n=18) (UK specific)

Modification or aid	Count (n)	Percentage (%)
Magnifiers	7	38.9
Laptops (only select if specialised for you)	4	22.2
Ergonomic adaptations e.g. raised screens	4	22.2
Modifications to mobile phones such as applications (apps)	4	22.2
Other – please describe	4	22.2
I do not use any additional items to support my work because I do not need them	4	22.2
Spoken word processor, verbal calculator or screen reader	3	16.7
Books, including books with enlarged font, and audio-books	1	5.6
I do not use any additional items to support my work because they are not available	1	5.6
Braille display	0	0.0
Total	**18**	**100.0**

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

Approximately how much money did you spend in the past year on items related to your condition to assist you with your working?

- Text response

Table A.21: Approximately how much money (£) did you spend in the past year on items related to your condition to assist you with your working? (n=53) (UK specific)

	Average (£)	Standard Deviation
Money spent on aids and modifications for work	48	169

Source: Deloitte Access Economics analysis.

A.7. Module 4: Education

Are you currently studying or learning?

- Yes
- No

Table A.22: Are you currently studying or learning? (n=53) (UK specific)

Study Status	Count (n)	Percentage (%)
Not Studying	44	83.0
Studying	9	17.0
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Where are you currently learning?

- Higher education (university)
- Further education
- Secondary education/post primary education
- Primary education
- Pre school/early childhood education

- Other education provider

Table A.23: Where are you currently learning? (n=9) (UK specific)

Study Status	Count (n)	Percentage (%)
Higher education (university)	0	0.0
Further education	0	0.0
Preschool/early childhood education	0	0.0
Other education provider	0	0.0
Primary education	6	66.7
Secondary education/post primary education	3	33.3
Total	**9**	**100.0**

Source: Deloitte Access Economics analysis.

About how many hours altogether did you spend at your place of learning in the past 4 weeks?

- Text response

Table A.24: How many hours altogether did you spend at your place of learning in the past 4 weeks? (n=9) (UK specific)

	Average	Standard Deviation
Hours	97.56	38.13

Source: Deloitte Access Economics analysis.

How many hours do you spend on education related activities (e.g. studying, completing homework and/or assignments,

research etc.) outside of your place of learning in a typical 7 day week? If it varies, estimate the average.

- Text response

Table A.25: How many hours do you spend on education related activities (e.g. studying, completing homework and/or assignments, research etc.) outside of your place of learning in a typical 7 day week? (n=9) (UK specific)

	Average	Standard Deviation
Hours	5.72	5.75

Source: Deloitte Access Economics analysis.

In the past 4 weeks how many full and partial days of class (example 1.5 days) did you miss because of your IRD? Please include only days missed for your own health, not someone else's health.

- Text response

Table A.26: In the past 4 weeks how many full and partial days of class (example 1.5 days) did you miss because of your IRD? (n=9) (UK specific)

	Average	Standard Deviation
Hours	0.89	1.27

Source: Deloitte Access Economics analysis.

Please select any modifications or aids that you currently use in your learning environment to assist with your learning.

- Books, including books with enlarged font and audio-books
- Laptops (only select if specialised for you)
- Magnifiers
- Spoken word processor, verbal calculator or screen reader
- Braille display
- Ergonomic adaptations e.g. raised screens
- Modifications to mobile phones such as applications (apps)

- Other – please describe
- I do not use any additional items to support my learning because I do not need them
- I do not use any additional items to support my learning because they are not available

Table A.27: What modifications or aids used in the learning environment specifically for an IRD? (n=9) (UK specific)

Modification or aid	Count (n)	Percentage (%)
Laptops (only select if specialised for you)	8	88.9
Books, including books with enlarged font, and audio-books	7	77.8
Modifications to mobile phones such as applications (apps)	5	55.6
Other – please describe	5	55.6
Magnifiers	4	44.4
Ergonomic adaptations e.g. raised screens	3	33.3
Spoken word processor, verbal calculator or screen reader	2	22.2
Braille display	1	11.1
I do not use any additional items to support my work because I do not need them	0	0.0
I do not use any additional items to support my work because they are not available	0	0.0

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

Approximately how much money did you spend in the past year on items related to your condition to assist you with your learning?

- Text response

Table A.28: How much money did you spend in the past year on items related to your condition to assist you with your learning? (n=53) (UK specific)

	Average (£)	Standard Deviation
Money spent on aids and modifications for learning	31	142

Source: Deloitte Access Economics analysis.

Do you receive additional support from a special needs teacher/assistant in your learning environment?
- Yes
- No

Table A.29: Do you receive additional support from a special needs teacher/assistant in your learning environment? (n=53) (UK specific)

Special needs teacher/assistant	Count (n)	Percentage (%)
No	44	83
Yes	9	17
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

On average, approximately how many hours per week do you receive additional support from a special needs assistant?
- Text response

Table A.30: How many hours per week do you receive additional support from a special needs assistant? (n=9) (UK specific)

	Average	Standard Deviation
Hours	21.00	13.21

Source: Deloitte Access Economics analysis.

A.8. Module 5: Management of your condition – health care providers

Have you seen any health providers specifically for or relating to the management of your IRD in the last year?

- Yes
- No
- Unsure

Table A.31: Have you seen any health providers specifically for or relating to the management of your IRD in the last year? (n=53) (UK specific)

Health system utilisation (total this year)	Count (n)	Percentage (%)
Yes	36	67.9
No	17	32.1
Unsure	0	0.0
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

When was the last time you saw a health provider specifically for or relating to the management of your IRD?

- 1 to 2 years
- 2 to 3 years
- 4 or more years
- Unsure

Table A.32: Last health system visit for those who had not seen anyone on the last year (n=17) (UK specific)

Health system utilisation (total)	Count (n)	Percentage (%)
1 to 2 years	6	35.3
4 or more years	6	35.3
2 to 3 years	5	29.4
Unsure	0	0.0
Total	**17**	**100.0**

Source: Deloitte Access Economics analysis.

Which health providers have you seen for management specifically or including for your IRD in the last year?

- General practitioner
- Ophthalmologist
- Optometrist/optician
- Eye Clinic Liaison Officer (ECLO)
- Sight support volunteer
- Psychologist, psychiatrist or counsellor
- Genetic counsellor
- Occupational therapist
- Hospital emergency department
- Admitted to hospital as an inpatient
- Specialist outpatient clinic or unit
- Habilitation, Rehabilitation specialist or service, or low vision specialist
- Other – please describe
- Unsure

Table A.33: Which health providers have you seen for management specifically or including for your IRD in the last year? (n=36) (UK specific)

Health System Utilisation (specific)	Count (n)	Percentage (%)
Ophthalmologist	27	75.0
Optometrist	17	47.2
Specialist outpatient clinic	8	22.2
Habilitation specialist	7	19.4
Eye clinic liaison officer	3	8.3
Sight support volunteer	3	8.3
Genetic counsellor	3	8.3
General practitioner	2	5.6
Psychologist	1	2.8
Occupational therapist	1	2.8
Emergency department	1	2.8
Admitted to hospital (inpatient)	0	0.0

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

Approximately how often do you attend appointments with health providers specifically for or related to your IRD?

- Weekly
- Fortnightly
- Monthly
- Every 3 to 6 months
- Yearly
- Every 2 years
- Never

Table A.34: How often do you attend appointments with health providers specifically for or related to your IRD? (n = 36) (UK specific)

Health System Utilisation (visits)	Visits per year (n)
Psychologist	12.0
Habilitation specialist	10.3
Sight support volunteer	2.2
Eye clinic liaison officer	1.7
Ophthalmologist	1.6
Specialist outpatient clinic	1.4
Optometrist	1.2
General practitioner	1.0
Genetic counsellor	1.0
Occupational therapist	1.0
Emergency department	1.0
Admitted to hospital (inpatient)	0.0

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

Approximately how much money did your last visit to your health provider cost you out of pocket, after refunds, reimbursements and financial support?

- Text response

Table A.35: Approximately how much money did your last visit to your health provider cost you out of pocket, after refunds, reimbursements and financial support? (n = 36) (UK specific)

Health System Utilisation (cost)	Average (£)
Psychologist	50.00
Ophthalmologist	32.96
Optometrist	28.53
Specialist outpatient clinic	16.50
Genetic counsellor	5.00
General practitioner	0.00
Eye clinic liaison officer	0.00
Sight support volunteer	0.00
Occupational therapist	0.00
Emergency department	0.00
Admitted to hospital (inpatient)	0.00
Habilitation specialist	0.00

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

Approximately how long does it take for you to travel one way to your doctor/clinician for management of your IRD? Estimate

this time for the doctor/clinician you visit most often for your condition.

- Less than 15 minutes
- Between 15 minutes and 30 minutes
- Between 30 minutes and an hour
- Between 1 hour and 2 hours
- Greater than 2 hours

Table A.36: Approximately how long does it take for you to travel one way to your doctor/clinician for management of your IRD? (n=36) (UK specific)

Average hours spent travelling to medical appointments	Count (n)	Percentage (%)
Between 30 minutes and an hour	11	30.6
Between 1 hour and 2 hours	10	27.8
Between 15 minutes and 30 minutes	9	25.0
Greater than 2 hours	4	11.1
Less than 15 minutes	2	5.6
Total	**36**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how much money does it usually cost you per appointment to travel (e.g. either using public transport or a car) to your doctor/clinician for management of your IRD? Please estimate any accommodation, public transport or petrol costs before any refunds, reimbursements and financial support were received. If you are unsure please answer not applicable.

- Text response

Table A.37: How much money (£) does it usually cost you per appointment to your doctor? (n=33) (UK specific)

	Average (£)	Standard Deviation
Average cost of the last medical appointment	16.74	16.50

Source: Deloitte Access Economics analysis.

Do you take time off work or education to attend these appointments?

- Yes, for all of my appointments
- Yes, for about three quarters of my appointments
- Yes, for about half of my appointments
- Yes, for about one quarter of my appointments
- No, I do not take time off work or education to attend my appointments

Table A.38: Do you take time off work or education to attend these appointments? (n=36) (UK specific)

Time off for appointments (total)	Count (n)	Percentage (%)
No, I do not take time off work or education to attend my appointments	17	47.2
Yes, for all of my appointments	16	44.4
Yes, for about half of my appointments	2	5.6
Yes, for about three quarters of my appointments	1	2.8
Yes, for about one quarter of my appointments	0	0.0
Total	**36**	**100**

Source: Deloitte Access Economics analysis.

Do you attend consultations alone or with a friend, family member or carer?

- I attend consultations alone
- I attend consultations with a friend or family member
- I attend consultations with a formal carer
- I attend consultations with a friend or family member and a formal carer

Table A.39: Do you attend consultations alone or with a friend, family member or carer? (n=36) (UK specific)

Appointment attendance type	Count (n)	Percentage (%)
I attend consultations with a friend or family member	24	66.7
I attend consultations alone	12	33.3
I attend consultations with a formal carer	0	0.0
I attend consultations with a friend or family member and a formal carer	0	0.0
Total	**36**	**100**

Source: Deloitte Access Economics analysis.

Does your friend, carer or family member take time off work to attend these appointments?

- Yes, for all of my appointments
- Yes, for about three quarters of my appointments
- Yes, for about half of my appointments
- Yes, for about one quarter of my appointments

Table A.40: Does your friend, carer or family member take time off work to attend these appointments? (n=24) (UK specific)

Appointment companion time off	Count (n)	Percentage (%)
Yes, for all of my appointments	15	62.5
Yes, for about one quarter of my appointments	7	29.2
Yes, for about three quarters of my appointments	1	4.2
Yes, for about half of my appointments	1	4.2
Total	**24**	**100.0**

Source: Deloitte Access Economics analysis.

How many appointments have you ever had with a genetic counsellor for or related to your IRD?

- None
- One
- Two
- Three or more
- Unsure

Table A.41: How many appointments have you ever had with a genetic counsellor for or related to your IRD? (n=53) (UK specific)

Genetic counsellor appointments	Count (n)	Percentage (%)
None	27	50.94
One	13	24.53
Three or more	8	15.09
Two	5	9.43
Unsure	0	0.00
Total	**53**	**100**

Source: Deloitte Access Economics analysis.

Have you ever received a diagnostic genetic test specifically for or including for your IRD?

- Yes
- No
- Unsure

Table A.42: Have you ever received a diagnostic genetic test specifically for or including for your IRD? (n=53) (UK specific)

Genetic test (lifetime)	Count (n)	Percentage (%)
Yes	30	56.6
No	21	39.6
Unsure	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Did you receive a diagnostic genetic test specifically for or including for your IRD within the last year?

- Yes
- No
- Unsure

Table A.43: Did you receive a diagnostic genetic test specifically for or including for your IRD within the last year? (n=30) (UK specific)

Genetic test (within the year)	Count (n)	Percentage (%)
No	22	73.3
Yes	8	26.7
Unsure	0	0.0
Total	**30**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how much money did you spend on travel (e.g. airfares or petrol or bus fare or train fare or taxi fare) to receive the results of your genetic test? If you are unsure please answer not applicable.

- Text response

Table A.44: How much money did you spend on travel to receive the results of your genetic test? (n=8) (UK specific)

	Average (£)	Standard Deviation
Average cost of travel for genetic testing	21.88	37.79

Source: Deloitte Access Economics analysis.

Have any other members of your family seen a specialist to check for IRD symptoms?

- Yes
- No
- Unsure

Table A.45: Have any other members of your family seen a specialist to check for IRD symptoms? (n=53) (UK specific)

Family specialist check	Count (n)	Percentage (%)
Yes	30	56.6
No	23	43.4
Unsure	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

How many of your family members have seen a specialist to check for IRD symptoms?

- 1
- 2
- 3
- 4
- 5 or more

Table A.46: How many of your family members have seen a specialist to check for IRD symptoms? (n=33) (UK specific)

Family specialist check (number)	Count (n)	Percentage (%)
One	10	30.3
Three	7	21.2
Five or more	7	21.2
Two	6	18.2
Four	3	9.1
Total	**33**	**100.0**

Source: Deloitte Access Economics analysis.

Have your family members ever received a genetic test specifically or including for IRD?
- Yes
- No
- Unsure

Table A.47: Have your family members ever received a genetic test specifically or including for IRD? (n=33) (UK specific)

Family genetic testing	Count (n)	Percentage (%)
Yes	13	24.5
No	12	22.6
Unsure	8	15.1
Total	**33**	**100.0**

Source: Deloitte Access Economics analysis.

A.9. Module 6: Management of your condition – medications and supplements

Do you use any medications to help manage your IRD?

- Yes
- No

Table A.48: Do you use any medications to help manage your IRD? (n=53) (UK specific)

Medication usage	Count (n)	Percentage (%)
No	51	96.2
Yes	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

What medications do you use to manage your IRD? Please separate any medications using commas.

- Text response

Table A.49: What medications do you use to manage your IRD? (n=2) (UK specific)

Not for publication due to confidentiality implications.

Approximately how much money, on average, have you spent on medications specifically or including for your IRD, after refunds, reimbursements and financial supports in the last month? Please only include out of pocket costs.

- Text response

Table A.50: How much money have you spent on medications specifically or including for your IRD, after refunds, reimbursements and financial supports in the last month? (n=2) (UK specific)

	Average (£)	Standard Deviation
Average cost of medication	12.89	15.09

Source: Deloitte Access Economics analysis.

Do you use any vitamins or nutritional supplements to help manage your IRD?
- Yes
- No

Table A.51: Do you use any vitamins or nutritional supplements to help manage your IRD? (n=53) (UK specific)

Vitamin usage	Count (n)	Percentage (%)
No	47	88.7
Yes	6	11.3
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

What vitamins or nutritional supplements do you use to manage your IRD? Please separate any vitamins or nutritional supplements using commas.
- Text response

Table A.52: What vitamins or nutritional supplements do you use to manage your IRD? (n=53) (UK specific)

Not for publication due to confidentiality implications.

Approximately how much money, on average, have you spent on vitamins or nutritional supplements specifically or including for your IRD, after refunds, reimbursements and financial supports in the last month? Please only include out of pocket costs.

- Text response

Table A.53: How much money have you spent on vitamins or nutritional supplements specifically or including for your IRD in the last month? (n=6) (UK specific)

	Average (£)	Standard Deviation
Average cost of vitamins and supplements	23.17	19.55

Source: Deloitte Access Economics analysis.

A.10. Module 7: Management of your condition – vision aids and modifications and travel

In the last year, approximately how much money have you spent on the following aids and modifications? If you do not use the vision aid please answer not applicable.

- Magnifying glasses
- Contrast enhancing filters
- Green or blue blocking sunglasses
- High vision lamps

Table A.54: In the last year, approximately how much money have you spent on the following aids and modifications? (n=53) (UK specific)

Aid or Modification	Count (n)	Percentage (%)	Average (£)
Green or blue blocking sunglasses	32	60.4	61.78
High vision lamps	28	52.8	47.71
Magnifying glasses	26	49.1	28.42
Contrast enhancing filters	22	41.5	21.91

Source: Deloitte Access Economics analysis.

Are you a guide dog owner?
- Yes
- No

Table A.55: Are you a guide dog owner? (n=53) (UK specific)

Guide dog ownership	Count (n)	Percentage (%)
No	47	88.7
Yes	6	11.3
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how many days in total did you spend learning with a trained professional to work with your guide dog? If you did not receive any training please answer not applicable.
- Text response

Table A.56: Approximately how many days in total did you spend learning with a trained professional to work with your guide dog? (n=9) (UK and RoI combined)

Guide dog owners	Average (days)
Average training time	16.38

Source: Deloitte Access Economics analysis.

Approximately how much money have you spent caring for your guide dog in the last month?
- Text response

Table A.57: Approximately how much money have you spent caring for your guide dog in the last month? (n=9) (UK and RoI combined)

Guide dog owners	Average (£)
Money spent for your guide dog in the last month	89.60

Source: Deloitte Access Economics analysis.

Do you use a 'white' cane to aid your mobility?
- Yes
- No

Table A.58:Do you use a 'white' cane to aid your mobility] (n=53) (UK specific)

White cane	Count (n)	Percentage (%)
Yes	31	58.5
No	22	41.5
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how many days in total did you spend learning with a trained professional to use your cane? If you did not receive any training please answer not applicable

- Text response

Table A.59: How many days in total did you spend learning with a trained professional to use your cane? (n=53) (UK specific)

White cane users	Average (days)
Average training time	16.38

Source: Deloitte Access Economics analysis.

Do you use the aid of a mini guide or other electronic mobility devices (GPS or sensor devices)?

- Yes
- No

Table A.60: Do you use the aid of a mini guide or other electronic mobility devices (GPS or sensor devices)? (n=53) (UK specific)

GPS or Mini guide	Count (n)	Percentage (%)
No	48	90.6
Yes	5	9.4
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

What was the approximate cost of this device? If you are unsure please answer not applicable.

- Text response

Table A.61: What was the approximate cost of this device? (n=129) (UK and RoI combined)

	Average (£)
GPS or Mini guide cost	318.13

Source: Deloitte Access Economics analysis.

Approximately how many hours did you spend training with the instructor for your mobility device? If you did not receive any training please answer not applicable.

- Text response

Table A.62: Approximately how many hours did you spend training with the instructor for your mobility device? (n=53) (UK specific)

	Average (days)
GPS or Mini guide cost training time	0

Source: Deloitte Access Economics analysis.

Do you have customised door frames or light switches in your home specifically or including for your IRD?

- Yes
- No

Table A.63: Do you have customised door frames or light switches in your home specifically or including for your IRD? (n=53) (UK)

Customised door frames or light switches	Count (n)	Percentage (%)
No	50	94.3
Yes	3	5.7
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

If you have customised door frames or light switches in your home specifically or including for your IRD, please estimate the cost of this modification. If you are unsure please answer not applicable.

- Text response

Table A.64: What was the estimated cost of a modification? (n=129) (UK and RoI combined)

	Average (£)
Customised door frames or light switches	131.99

Source: Deloitte Access Economics analysis.

Do you have customised bathtubs, showers or other bathroom modifications in your home specifically or including for your IRD?

- Yes
- No

Table A.65: Do you have customised bathtubs, showers or other bathroom modifications in your home specifically or including for your IRD? (n=53) (UK Specific)

Bathroom modifications	Count (n)	Percentage (%)
No	51	96.2
Yes	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

If you have customised bathtubs, showers or other bathroom modifications in your home specifically or including for your IRD, please estimate the cost of these modifications. If you are unsure please answer not applicable.

- Text response

Table A.66: What was the estimated cost of any modifications? (n=3) (UK and RoI combined)

	Average (£)
Customised bathtubs, showers or other bathroom modifications cost	1844.0

Source: Deloitte Access Economics analysis.

Provide an estimate of the number, if any, of the following items that you use at home specifically for your IRD

- Magnifying mirrors
- High intensity lamps
- Book alternatives (large print or audiobooks)
- Handheld magnifiers
- Tactile or large print labels
- Large keyboards
- Screen magnification technology
- Screen reading software
- Portable note takers

- Customised clocks, watches or timers
- Other

Table A.67: Provide an estimate of the number, if any, of the following items that you use at home specifically for your IRD (n=53) (UK specific)

Aid or modification	Count (n)	Percentage (%)
Handheld magnifiers	30	56.6
High intensity lamps	24	45.3
Book alternatives	24	45.3
Customised clocks or timers	23	43.4
Screen reading software	20	37.7
Screen magnification technology	19	35.9
Magnifying mirrors	15	28.3
Large keyboards	8	15.1
Portable note takers	7	13.2
Tactile or large print labels	5	9.4

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

On average, approximately how often do you use public transport for reasons other than medical appointments because of your IRD?

- More than twice per day
- Twice per day
- Once per day
- A few times per week
- Once per week
- Once every fortnight
- Once per month
- Once every three months or infrequently
- Never, I do not use public transport
- Never, I am still able to drive
- Unsure

Table A.68: How often do you use public transport for reasons other than medical appointments because of your IRD? (n=44, children were not asked this question) (UK specific)

Public transport frequency (non-IRD purpose)	Count (n)	Percentage (%)
A few times per week	15	34.1
Never, I do not use public transport	6	13.6
Once every three months or infrequently	4	9.1
More than twice per day	3	6.8
Once per week	3	6.8
Once every fortnight	2	4.5
Once per day	2	4.5
Once per month	2	4.5
Never, I am still able to drive	1	2.3

Twice per day	5	11.4
Unsure	1	2.3
Total	**44**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how much money have you spent on public transport in the last month for reasons other than medical appointments because of your IRD? Please estimate these costs before refunds, reimbursements and financial support were received. If you are unsure please answer not applicable.

- Text response

Table A.69: Approximately how much money have you spent on public transport in the last month for reasons other than medical appointments because of your IRD? (n=44) (UK specific)

	Average (£)	Standard Deviation
Average spend on public transport in the last month for non-IRD reasons	58.47	46.27

Source: Deloitte Access Economics analysis.

A.11. Module 8: Formal care

Do you have a formal carer (a person who is paid to give care) to assist you with your activities of daily living specifically or including for your IRD?

- Yes
- No

Table A.70: Do you have a formal carer (a person who is paid to give care) to assist you with your activities of daily living specifically or including for your IRD? (n=53) (UK specific)

Formal care	Count (n)	Percentage (%)
No	51	96.2
Yes	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how many hours per week do you receive assistance from a formal carer specifically or including for your IRD?

- Less than 5 hours
- 5 to 9 hours
- 10 to 19 hours
- 20 to 29 hours
- 30 to 39 hours
- 40 hours or more

Table A.71: Approximately how many hours per week do you receive assistance from a formal carer specifically or including for your IRD? (n=8) (UK and RoI combined)

Formal care hours	Count (n)	Percentage (%)
Less than 5 hours	2	25.0
5 to 9 hours	3	37.5
10 to 19 hours	1	12.5
20 to 29 hours	1	12.5
40 hours or more	1	12.5
Total	**8**	**100.0**

Source: Deloitte Access Economics analysis.

A.12. Module 9: Informal care

Do you receive any assistance from an informal carer? An informal carer includes any person, such as a family member, friend or neighbour, who is giving regular, ongoing assistance to another person without payment for the care given. If so, from whom? (Please identify your primary informal carer if you have more than one informal carer).

- Spouse
- Family member or relative
- Friend or other informal carer
- I do not receive any assistance from an informal carer

Table A.72: Do you receive any assistance from an informal carer? (n=53) (UK specific)

Informal carer type	Count (n)	Percentage (%)
Spouse	23	43.4
Family member or relative	17	32.1
I do not receive any assistance from an informal carer	11	20.8
Friend or other informal carer	2	3.8
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Approximately how many hours per week do you receive assistance from an informal carer?

- Less than 5 hours
- 5 to 9 hours
- 10 to 19 hours
- 20 to 29 hours
- 30 to 39 hours
- 40 hours or more

Table A.73: Approximately how many hours per week do you receive assistance from an informal carer? (n=42) (UK specific)

Informal care hours	Count (n)	Percentage (%)
Less than 5	9	21.4
5-9	16	38.1
10-19	8	19.0
20-29	3	7.1
30-39	3	7.1
40+	3	7.1
Total	**42**	**100.0**

Source: Deloitte Access Economics analysis.

What is the sex/gender of your informal carer? (Please choose the option as it applies to your primary informal carer if you have more than one informal carer)

- Male
- Female
- Other/unsure

Table A.74: What is the sex/gender of your informal carer? (n=42) (UK specific)

Informal carer gender	Count (n)	Percentage (%)
Female	23	54.8
Male	19	45.2
Total	**42**	**100.0**

Source: Deloitte Access Economics analysis.

What is the approximate age of your informal carer? (Please choose the option as it applies to your primary informal carer if

you have more than one informal carer and guess if you are unsure exactly)

- 15 to 24
- 25 to 34
- 35 to 44
- 45 to 54
- 55 to 64
- 65 to 74
- 75 or older

Table A.75: What is the approximate age of your informal carer? (n=42) (UK specific)

Informal carer age	Count (n)	Percentage (%)
15-24	2	4.8
25-34	3	7.1
35-44	9	21.4
45-54	8	19.0
55-64	9	21.4
65-74	8	19.0
75+	3	7.1
Total	**42**	**100.0**

Source: Deloitte Access Economics analysis.

Is your informal carer employed and if so, in what capacity? (Please choose the option as it applies to your primary informal carer if you have more than one informal carer).

- Full time work
- Part time work
- Casual work (Not working on a regular basis)
- Underemployed (e.g. working part time but desiring full time work; working in a role that does not use their skills or qualifications)

- Retired
- My informal carer is a student
- My informal carer is not employed, and not looking for work
- My informal carer is not employed, and is looking for work

Table A.76: Is your informal carer employed and if so, in what capacity? (n=42) (UK specific)

Informal carer employment	Count (n)	Percentage (%)
Full time work	17	40.5
Retired	13	31.0
Part time work	9	21.4
My informal carer is a student	2	4.8
My informal carer is not employed, and not looking for work	1	2.4
Casual work (Not working on a regular basis)	0	0.0
Underemployed (e.g. working part time but desiring full time work; working in a role that does not use their skills or qualifications)	0	0.0
Total	**42**	**100.0**

Source: Deloitte Access Economics analysis.

Does your informal carer receive any government support specifically or including for your IRD? If so, which of the following forms of assistance does your informal carer receive?

- Carer's allowance
- Carer's Benefit
- Carer's Support Grant
- Other – please specify
- Unsure

- My informal carer does not receive any government support for my condition

Table A.77: Which of the following forms of assistance does your informal carer receive? (n=42) (UK specific)

Government Payment	Count (n)	Percentage (%)
My informal carer does not receive any government support for my condition	40	95.2
Carer's allowance	2	4.8
Carer's Benefit	0	0.0
Carer's Support Grant	0	0.0
Other – please specify	0	0.0
Unsure	0	0.0

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

A.13. Module 10: Government support

Do you currently receive any government support specifically or including for your IRD? If so, which of the following forms of assistance do you receive?

- Disability Living Allowance (Personal Independence Payment)
- Attendance allowance
- Constant attendance allowance
- Disability premium
- Enhanced disability premium
- Severe disability premium
- NHS Low income scheme
- European health insurance card
- Disabled Students' Allowance
- Other – please specify
- Unsure
- I do not receive any government support for my condition

Table A.78: Do you currently receive any government support specifically or including for your IRD? (n=53) (UK specific)

Government Payment	Count (n)	Percentage (%)
Disability Living Allowance (Personal Independence Payment)	43	81.1
European health insurance card	2	3.8
Enhanced disability premium	1	1.9
Attendance allowance	1	1.9
Severe disability premium	1	1.9

Source: Deloitte Access Economics analysis.
Note: This question allowed for the selection of multiple answers, hence, the percentages do not sum to 100.

A.14. Module 11: Other impacts of your condition

Have you ever experienced feelings of depression because of your IRD?

- Yes

- No

Table A.79: Have you ever experienced feelings of depression because of your IRD? (n=53) (UK specific)

Experience of depression	Count (n)	Percentage (%)
Yes	36	67.9
No	17	32.1
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Have you ever experienced feelings of anxiety because of your IRD?
- Yes
- No

Table A.80: Have you ever experienced feelings of anxiety because of your IRD? (n=53) (UK specific)

Experience of anxiety	Count (n)	Percentage (%)
Yes	46	86.8
No	7	13.2
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Have you ever experienced feelings of another mental health condition because of your IRD?
- Yes, please describe
- No

Table A.81: Have you ever experienced feelings of another mental health condition because of your IRD? (n=53) (UK specific)

Experience of other mental health conditions	Count (n)	Percentage (%)
No	46	86.8
Yes	7	13.2
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Have any of your close family members ever experienced feelings of depression, anxiety, or another mental health condition as a result of your IRD?

- Yes
- No

Table A.82: Have any of your close family members ever experienced feelings of depression, anxiety, or another mental health condition as a result of your IRD? (n=53) (UK specific)

Family experience of other mental health conditions	Count (n)	Percentage (%)
Yes	27	50.9
No	26	49.1
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

On a scale of 1 to 5 with 5 being strongly agree and 1 being strongly disagree, please answer the following statement. I feel frustrated from the lack of awareness and support for IRDs.

- 1 - I strongly disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 2 - I disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 3 - I neither agree nor disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 4 - I agree that I feel frustrated from the lack of awareness and support for IRDs.
- 5 - I strongly agree that I feel frustrated from the lack of awareness and support for IRDs.

Table A.83: Feeling of frustration from the lack of awareness and support for IRDs (n=53) (UK specific)

Frustration	Count (n)	Percentage (%)
1 – I strongly disagree that I feel frustrated	4	7.5
2 - I disagree that I feel frustrated	1	1.9
3 - I neither agree nor disagree	9	17.0
4 - I agree that I feel frustrated	16	30.2
5 - I strongly agree that I feel frustrated	14	26.4
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Do you experience any financial stress because of your IRD?

- Yes
- No
- Unsure

Table A.84: Do you experience any financial stress because of your IRD? (n=53) (UK specific)

Experience of other mental health conditions	Count (n)	Percentage (%)
No	25	47.2
Yes	24	45.3
Unsure	4	7.6
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

Have you experienced social isolation because of your IRD? Social isolation is a state of complete or near complete lack of contact between an individual and society.

- Yes
- No
- Unsure

Table A.85: Have you experienced social isolation because of your IRD? (n=53) (UK specific)

Experience of social isolation	Count (n)	Percentage (%)
No	25	47.2
Yes	24	45.3
Unsure	4	7.6
Total	**53**	**100.0**

Source: Deloitte Access Economics analysis.

A.15. Module 12: Impact of your child's IRD on your mental wellbeing – from your own perspective as a parent/ guardian:

Have you (the parent/guardian) ever experienced feelings of depression due to your caring requirements for your child with an IRD?

- Yes
- No

Table A.86: Have you (the parent/guardian) ever experienced feelings of depression due to your caring requirements for your child with an IRD? (n=9) (UK specific)

Experience of depression	Count (n)	Percentage (%)
Yes	7	77.8
No	2	22.2
Total	**9**	**100.0**

Source: Deloitte Access Economics analysis.

Have you ever experienced feelings of anxiety due to your caring requirements for your child with an IRD?

- Yes
- No

Table A.87: Have you ever experienced feelings of anxiety due to your caring requirements for your child with an IRD? (n=9) (UK specific)

Experience of anxiety	Count (n)	Percentage (%)
Yes	7	77.8
No	2	22.2
Total	**9**	**100.0**

Source: Deloitte Access Economics analysis.

Have you ever experienced feelings of another mental health condition due to your caring requirements for your child with an IRD?

- Yes, please specify
- No

Table A.88: Have you ever experienced feelings of another mental health condition due to your caring requirements for your child with an IRD? (n=9) (UK specific)

Experience of other mental health conditions	Count (n)	Percentage (%)
No	7	77.8
Yes	2	22.2
Total	**9**	**100.0**

Source: Deloitte Access Economics analysis.

[Parent] On a scale of 1 to 5 with 5 being strongly agree and 1 being strongly disagree, please answer the following statement. I feel frustrated from the lack of awareness and support for IRDs.

- 1 - I strongly disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 2 - I disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 3 - I neither agree nor disagree that I feel frustrated from the lack of awareness and support for IRDs.
- 4 - I agree that I feel frustrated from the lack of awareness and support for IRDs.
- 5 - I strongly agree that I feel frustrated from the lack of awareness and support for IRDs.

Table A.89: Parent feeling of frustration from the lack of awareness and support for IRDs (n=53) (UK specific)

Frustration	Count (n)	Percentage (%)
1 – I strongly disagree that I feel frustrated	1	11.1
2 - I disagree that I feel frustrated	0	0
3 - I neither agree nor disagree	1	11.1
4 - I agree that I feel frustrated	4	44.4
5 - I strongly agree that I feel frustrated	3	33.3
Total	**9**	**100.0**

Source: Deloitte Access Economics analysis.

Appendix B Additional information on prevalence

B.1. Summary of approach

A targeted literature review was undertaken to estimate the prevalence of the 10 IRDs included in this study (see Section 3). A targeted search of the PubMed and Cochrane Library databases, using key search terms, identified 34 potential publications for full text review. These were further refined to 14 publications that informed this estimate of the prevalence of these IRDs. Articles were then prioritised for inclusion if they met either of the following criteria:

- The publication provided a population-based prevalence estimate of at least one of the 10 included IRDs.
- The publication utilised national registry data to capture prevalence in a relatively comparable country to the UK.

Given the potential for double-counting where overlapping classification of individual IRDs may occur between different studies, publications providing a more comprehensive list of the included conditions were prioritised.

Publications considered at full text review are detailed in Appendix B.5, along with the prevalence rates reported and the reason for their inclusion or exclusion as an input into this analysis.

B.2. Overall prevalence

Literature providing population prevalence estimates of IRDs (overall or condition-specific) is limited. Recent and nationwide prevalence estimates for IRDs in the UK were not identified. National registry data on the prevalence of five out of the 10 IRDs was identified from a population-based study conducted in Denmark.

This population-based cross-sectional registry study of the prevalence of generalised retinal dystrophy, encompassing a group of hereditary degenerations of the human retina, was based on the Danish Retinitis Pigmentosa Registry (Bertelsen

et al, 2014). The condition-specific overarching population prevalence rates, summarised in Table B.1:, were available from Bertelsen et al (2014) and included in this analysis.

In the absence of other data, a retrospective epidemiologic observational study of Best disease conducted in Denmark was used to inform the estimated population prevalence of Best disease in the UK (Bitner et al, 2012). Based on this article, the overall prevalence of Best disease in Denmark in 2011 was estimated to be 1.5 per 100,000 people (0.0015%).

Table B.1: Overall published population prevalence estimates for RP, LCA/EOSRD, cone-rod dystrophy, choroideremia, Usher syndrome and Best disease

IRD	Source	Prevalence estimate (%)
RP	Bertelsen et al (2014)	0.0154
LCA/EOSRD	Bertelsen et al (2014)	0.0024
Cone-rod dystrophy	Bertelsen et al (2014)	0.0012
Choroideremia	Bertelsen et al (2014)	0.0004
Usher syndrome	Bertelsen et al (2014)	0.0039
Best disease	Bitner et al (2012)	0.0015

Source: Bertelsen et al (2014) and Bitner et al (2012).
To estimate the prevalence of the remaining conditions (cone dystrophy, Stargardt disease, XLRS, and achromatopsia), the relative prevalence of the identified IRDs was derived from published results of the Target 5000 study conducted in Ireland (Dockery et al, 2017).

The Target 5000 research project aims to genetically characterise the estimated 5,000 people in Ireland with IRDs. Findings from approximately 750 persons living with an IRD as part of this target capture next-generation study have been published (Dockery et al, 2017). Based on these results, it was estimated that RP, LCA/EOSRD, cone-rod dystrophy, choroideremia, and Usher syndrome collectively represented 60.6% of the included IRDs. Based on this proportion, and the estimated prevalence for these conditions and Best disease, it was calculated that there were a total of 20,814 prevalent cases in the UK in 2019. This represents an overall population prevalence rate of 0.03%.

Condition-specific prevalence was calculated for the remaining conditions based on a study of the relative prevalence of a subset of IRDs conducted over a 21-year period in a specialised outpatient clinic in Southern France (Bocquet et al, 2013). Based on this study, population prevalence rates were derived using the relative proportion of conditions applied to the overarching prevalence estimate of 0.03%. These derived prevalence rates are summarised in Table B.2:.

Table B.2: Overall derived population prevalence estimates for cone dystrophy, Stargardt disease, XLRS and achromatopsia

IRD	Prevalence estimate (%)
Cone dystrophy	0.0013
Stargardt disease	0.0032
XLRS	0.0007
Achromatopsia	0.0011

Source: Deloitte Access Economics' analysis based on Bertelsen et al (2014), Bitner et al (2012), Dockery et al (2017), and Bocquet et al (2013).

B.3. Age and sex distribution

Our approach to estimating the prevalence of IRDs in the UK utilised age and sex specific rates, where they were available from the literature. Data from the eyeGENE registry, which reports on a sample of 4,635 participants with various IRDs in the United States, was used to inform the sex distribution in the absence of condition-specific literature. It was assumed that the sex distribution of cone dystrophy matched the profile of cone-rod dystrophy reported based on the eyeGENE registry (National Eye Institute, 2019).

Table B.3: Sources used to estimate age and sex specific prevalence rates by IRD

IRD	Source	Male (%)	Female (%)
RP	Bundey and Crews, 1984 (UK), condition-specific	52.3	47.7
LCA/EOSRD	Haim et al, 1992 (Denmark), condition-specific	55.0	45.0
Cone-rod dystrophy	eyeGENE registry, 2019 (US), condition-specific	61.2	38.8
Choroideremia	eyeGENE registry, 2019 (US), condition-specific	84.4	15.6
Usher syndrome	Sadeghi et al, 2004 (Sweden), condition-specific	46.8	53.2
Best disease	eyeGENE registry, 2019 (US), condition-specific	45.3	54.7
Cone dystrophy	eyeGENE registry, 2019 (US), condition-specific (cone-rod dystrophy)	61.2	38.8

IRD	Source	Male (%)	Female (%)
Stargardt disease	eyeGENE registry, 2019 (US), condition-specific	42.3	57.7
XLRS	eyeGENE registry, 2019 (US), condition-specific	95.3	4.7
Achromatopsia	eyeGENE registry, 2019 (US), condition-specific	44.8	55.2

Source: Deloitte Access Economics' analysis based on Bundey and Crews (1984), Haim et al (1992), eyeGENE registry (National Eye Institute, 2019), and Sadeghi et al (2004).

B.4. Results

Overall prevalence of IRDs in the UK disaggregated by age and sex is summarised in Table B.4: and shown in Chart 5.2. The prevalence of IRDs increases progressively with age until the 60-69 year age group, after which prevalence gradually declines. Overall prevalence is slightly higher in males (52.4%) compared to females (47.6%).

Table B.4: Prevalence of IRDs in the UK (2019) by age and sex

	Male		Female		Total	
Age group	**Rate (%)**	**Cases**	**Rate (%)**	**Cases**	**Rate (%)**	**Cases**
0-9	0.0111	460	0.0103	406	0.0107	866
10-19	0.0260	1,013	0.0241	892	0.0251	1,904
20-29	0.0310	1,376	0.0286	1,212	0.0299	2,588
30-39	0.0323	1,435	0.0290	1,295	0.0306	2,730
40-49	0.0403	1,676	0.0364	1,546	0.0383	3,223
50-59	0.0430	1,916	0.0385	1,771	0.0407	3,686
60-69	0.0460	1,597	0.0391	1,431	0.0425	3,029
70-79	0.0361	964	0.0285	850	0.0321	1,814
80+	0.0345	470	0.0252	504	0.0290	974
Total	**0.0330**	**10,906**	**0.0293**	**9,908**	**0.0311**	**20,814**

Source: Deloitte Access Economics analysis.
Note: Components may not sum to totals due to rounding.

B.5. Summary of sources informing overall prevalence estimates by condition

Table B.5: Summary of sources informing overall prevalence estimates of RP

Study	Country	Reason	Prevalence estimate (%)
Bertelsen et al (2014)	Denmark	Included	0.0154
Bundey and Crews (1984)	UK	Excluded – age of study; outpatient setting	0.0206
Grøndahl (1987)	Norway	Excluded – age of study	0.0227
Peterlin et al (1992)	Slovenia	Excluded – age of study; outpatient setting; generalisability of study population	0.0166
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0237

Source: Deloitte Access Economics research.

Table B.6: Summary of sources informing overall prevalence estimates of LCA/EOSRD

Study	Country	Reason	Prevalence estimate (%)
Bertelsen et al (2014)	Denmark	Included	0.0024
Haim et al (1992)	Denmark	Excluded – age of study	0.0018

Source: Deloitte Access Economics research.

Table B.7: Summary of sources informing overall prevalence estimates of code-rod dystrophy

Study	Country	Reason	Prevalence estimate (%)
Bertelsen et al (2014)	Denmark	Included	0.0012
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0045

Source: Deloitte Access Economics research.

Table B.8: Summary of sources informing overall prevalence estimates of choroideremia

Study	Country	Reason	Prevalence estimate (%)
Bertelsen et al (2014)	Denmark	Included	0.0004
Haim et al (1992)	Denmark	Excluded – age of study	0.0004
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0006

Source: Deloitte Access Economics research.

Table B.9: Summary of sources informing overall prevalence estimates of Usher syndrome

Study	Country	Reason	Prevalence estimate (%)
Bertelsen et al (2014)	Denmark	Included	0.0039
Grøndahl (1987)	Norway	Excluded – age of study	0.0036
Haim et al (1992)	Denmark	Excluded – age of study	0.0031

Study	Country	Reason	Prevalence estimate (%)
Hope et al (1997)	UK	Excluded – age of study; outpatient setting	0.0062
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0015
Rosenberg et al (1997)	Denmark	Excluded – age of study	0.0050
Spandau et al (2002)	Germany	Excluded – age of study; outpatient setting	0.0062
Sadeghi et al (2004)	Sweden	Excluded – age of study	0.0033

Source: Deloitte Access Economics research.

Table B.10: Summary of sources informing overall prevalence estimates of Best disease

Study	Country	Reason	Prevalence estimate (%)
Bitner et al (2012)	Denmark	Included	0.0015
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0044

Source: Deloitte Access Economics research.

Table B.11: Summary of sources informing overall prevalence estimates of cone dystrophy

Study	Country	Reason	Prevalence estimate (%)
Dockery et al (2017); Bocquet et al (2013)	Ireland; France	Included (derived from distribution of IRDs)	0.001
Puech et al (1991)	France	Excluded – age of study	0.0044

Source: Deloitte Access Economics research.

Table B.12: Summary of sources informing overall prevalence estimates of Stargardt disease

Study	Country	Reason	Prevalence estimate (%)
Dockery et al (2017); Bocquet et al (2013)	Ireland; France	Included (derived from distribution of IRDs)	0.003
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0116
Orphanet (2019)	International	Excluded – primary sources and methods not reported	0.0100

Source: Deloitte Access Economics research.

Table B.13: Summary of sources informing overall prevalence estimates of XLRS

Study	Country	Reason	Prevalence estimate (%)
Dockery et al (2017); Bocquet et al (2013)	Ireland; France	Included (derived from distribution of IRDs)	0.001
Puech et al (1991)	France	Excluded – age of study; outpatient setting	0.0036
Orphanet (2019)	International	Excluded – primary sources and methods not reported	0.0050

Source: Deloitte Access Economics research.

Table B.14: Summary of sources informing overall prevalence estimates of achromatopsia

Study	Country	Reason	Prevalence estimate (%)
Dockery et al (2017); Bocquet et al (2013)	Ireland; France	Included (derived from distribution of IRDs)	0.001
Orphanet (2019)	International	Excluded – primary sources and methods not reported	0.0027

Source: Deloitte Access Economics research.

Appendix C Additional information on other costs

C.1. Descriptions of welfare payment types

C.1.1. Disability Living Allowance (DLA)/Personal Independence Payment (PIP)

The DLA/PIP provides support for the extra costs of living with a long-term health condition or disability, for those between the age of 16 and 64[8]. The rate received depends on how the condition affects individuals, rather than the condition itself. Eligibility requires individuals to have had difficulties with daily living or getting around (or both) for at least three months. A needs assessment and determination of rate of payment is regularly reviewed (UK Department of Work and Pensions, 2019a; 2019b).

There are two components of the DLA:

- Daily living/care award: if unable to perform some daily activities, an individual may receive support ranging from £58.70 - £87.65
- Mobility award: if normal aids are not sufficient to enable walking, an individual may receive support ranging from £23.20 - £61.20 (UK Department of Work and Pensions, 2019a; 2019b).

C.1.2. Attendance Allowance

The Attendance Allowance offers financial support to those with a severe disability that necessitates support from another person. The payment is only available to those who have reached the state pension age, have a severe mental or physical disability, require help caring for yourself and have

[8] The DLA is an older form of the PIP that is only available to those who were born prior to 8 April 1948 and to those under 16. As such, people receiving the DLA fall outside this age range.

needed this help for at least six months (UK Department of Work and Pensions, 2019c).

Individuals can receive one of two rates depending on the level of support that they require:

- Lowest rate: those requiring frequent help or constant daytime supervision, or night supervision can receive £58.70
- Highest rate: those requiring help or supervision throughout both day and night can receive £87.65 (UK Department of Work and Pensions, 2019c).

C.1.3. Enhanced Disability Premium

The Enhanced Disability Premium is a welfare payment for people with a more serious disability that is offered in addition to the standard disability premium or income-related ESA, and either a higher rate independence payment or higher rate DLA/PIP. Recipients must also be under the pension credit age. Individuals receive a weekly payment of £16.80 while couples are entitled to £24.10 (UK Department of Work and Pensions, 2019d).

C.1.4. Severe Disability Premium

The Severe Disability Premium is similar to the Enhanced Disability Premium but is provided to those with the most severe disabilities. Individuals receive a weekly payment of £65.85 while couples are entitled to £131.70 (UK Department of Work and Pensions, 2019d).

C.1.5. Employment and Support Allowance (ESA)

The ESA provides support to people who are having difficulty finding work because of their long-term medical condition or a disability. ESA is only available for those people less than state pension age (UK Department of Work and Pensions, 2019e).

The rate of payment that individuals receive depends on both their age and the impact that their disability has on their ability to work. Individuals 25 years or older in the work-related activity group (able to engage in some form of employment) may receive up £73.10 while those below 25 years may receive up to £57.90 per week. Similarly, those in a support group can receive up to £111.65 per week (UK Department of Work and Pensions, 2019e).

C.1.6. Carer's Allowance

The Carer's Allowance provides support for people caring for someone with a significant illness or disability for at least 35 hours per week. In addition to the time requirement, the person being cared for must also receive an allowance such as PIP. Rates of payment are fixed at £64.60 per week for all eligible carers (UK Department of Work and Pensions, 2019f).

Limitation of our work

General use restriction

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