debra

Life with EB in Ireland

An international review of quality-of-life and comparison of access to healthcare resources and government supports

This collaborative report with the University of Limerick's Kemmy Business School was commissioned by Debra. The report was written by Dr Darragh Flannery and Brian Downes from the Department of Economics at the Kemmy Business School, and by Sarah Mullins and Joanna Joyce from Debra.

The authors would like to thank the participants from the DEBRA patient organisations in Spain, Australia, Ireland, the UK, and Austria, for their time and for sharing their expertise, without which this report would not have been possible. For clarity, Debra Ireland is referred to as 'Debra' throughout this report.

We are Debra.

We are here to be a positive force for everyone living with and impacted by Epidermolysis Bullosa (EB). We are here to help in all kinds of everyday ways, to support, command attention, demand and drive research and ensure imperative progress is made.

But above all, we are here to bring bold hope by proving the power of the butterfly effect – that small acts can create the most monumental change.

The butterfly skin charity. Uniting for change for everyone with skin as fragile as a butterfly wing.

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October 2023

Foreword

In Debra, we work hard to ensure that the voices of people living or caring for someone with Epidermolysis Bullosa (EB) are heard by decision-makers. All of our advocacy work is informed by people with EB and based on evidence. It is in this context that we commissioned the University of Limerick's Kemmy Business School to conduct this study on quality-of-life and access to healthcare resources and government supports with a view to identifying areas where access to EB supports and services in Ireland can be improved in the future.

We know that many people living with EB are struggling to access vital services and supports, with people often having to fight relentlessly to get what they need. This report details the significant challenges families face in managing this difficult condition.

As well as identifying gaps that exist, it is heartening to see details of the services that are currently being delivered to people with EB set out in this report, and we would like to take this opportunity to commend the hard work of the dedicated teams in Children's Health Ireland at Crumlin, St. James's Hospital and in the community.

Great progress has certainly been made since Debra was founded in 1988, but there is a lot more to do to ensure that everyone affected by EB in Ireland gets the services and supports they need. We look forward to working with key decision-makers to ensure that services continue to improve in order to meet those needs.

Jimmy Fearon
CEO

Key findings

- People with EB exhibit higher than average frequencies of psychiatric symptoms. Depression has a prevalence of 63.6% among people with EB compared to 10.1% for the general population. Anxiety has a prevalence among people with EB of 45.5% compared to 11.1% for the control sample.
- Quality-of-life for people caring for someone with EB is significantly lower than the control adult population. The average EQ – 5D index for carers is 0.696, 21.4% lower than the average UK adult.
- An analysis of the sparse literature available gives an estimated total economic burden for EB of €130,021 per person, per year. Further primary research into the economics of the condition is needed.
- Relative to those with EB in Australia, Spain, Austria, and the UK, people in Ireland have poorer access to dressing materials with other countries having either national bandage schemes or insurance models that help ensure access to cheap bandaging materials for people living with all severities of EB.
- Access to dental care is an issue across multiple countries, including Ireland, and people with EB can be required to travel long distances to receive care in specialist treatment centres.
- Access to medication was deemed to be good in Spain, Australia, Ireland, the UK, and Austria. People with EB can also access medical equipment across these countries, but such access can depend on the severity of the disease, and it can be harder to access highly specialised equipment.
- Relative to the UK and Austria, Ireland provides a good level of home nursing care to people with more severe EB. However, similar to other countries, challenges remain, such as the lack of a contingency plan when nursing staff are unavailable, and a lack of nurses trained in EB to replace those who move on. Access to respite care is an issue that arose in Ireland, Australia, Spain, Austria, and the UK.
- Given the high psychological burden evidenced for those with EB and those caring for someone with EB, there is relatively poor access to mental health services for those affected by EB in Ireland and in other countries.
- In Spain, Australia, Ireland, the UK, and Austria care is available for people with EB through specialist clinics, though the level of care can vary. These clinics provide access to services such as inpatient and outpatient care, podiatrists, dermatologists, plastic surgeons, physiotherapists, and dentists.
- There is some level of access to physiotherapy and occupational therapy in Spain, Australia, Ireland, the UK, and Austria but in some countries, including Ireland, it can be difficult for people to access these services in the community.

Executive summary

EB is a group of painful genetic blistering conditions that affects the body's largest organ, the skin. People living with EB are missing the essential proteins that bind the skin's layers together, so any minor friction, movement or trauma causes it to break and blister. There are limited treatment options available and no cure for EB, and management focuses on amelioration of the symptoms (Prodinger, et al., 2019).

The aim of this study is to review the impact of EB on quality-of-life and access to healthcare resources and government supports internationally, with a view to identifying areas where access to EB supports and services in Ireland can be improved in the future. In order to achieve this aim, we present:

a) an in-depth literature review of the existing international evidence relating to the quality-of-life and economic burden of EB.

b) a comparative review of access to healthcare resources and government supports across five countries, including Ireland.

From a quality-of-life perspective, the impact of EB is in most cases profoundly negative. However, the impact varies greatly by clinical severity of disease, disease severity as perceived by carers and family members, the skin area involved, level of pain and itch, sex, and age (Kearney, et al., 2020). Several themes emerge from studies that have examined quality-of-life of those living with EB, including physical pain and itchy skin, a sense of being different amongst their peers, a lack of independence due to a heavy reliance on others, challenges with daily activities and physical activity, underdeveloped coping mechanisms, and a higher rate of psychiatric symptoms (Pagliarello & Tabolli, 2010).

The high significance of seeing the self as 'different' for children with EB is replaced with worries about the impact EB may have on employment prospects, social interactions, and relationships for adults living with EB (Williams, et al., 2011; Dures, et al., 2011). Adults living with EB feel further distress about the hereditary, life-long nature of EB, and the fact that there is currently no cure for the illness (Dures, et al., 2011).

The burden of care for parents of children with EB is substantial, but varies in magnitude and intensity, based on the severity of their child's illness. According to Fine et al., (2005), caring for a child with EB curtails employment opportunities and leisure time for parents. The burden strains marital and familial relationships and creates an atmosphere of uncertainty (Fine, et al., 2005). However, the greatest impact for parents is emotional, having to watch their child endure pain, and at times, inflict pain on their child during daily dressing changes (Mauritz, et al., 2021).

As mentioned, the second broad aim of this research is to examine the healthcare resources and government supports offered to people living with EB in Ireland as compared to those in other jurisdictions. Access to healthcare resources for those with rare diseases such as EB has previously been highlighted as an area of concern in Ireland (Keane, 2014). Issues relating to the provision of medical cards and incorporation into the Long-Term Illness (LTI) scheme have also been raised (Keane, 2014). Given this context, a review of the support offered in other countries to those living with EB is warranted and may inform policy regarding the types of resources provided to people living with EB in Ireland.

Using information gathered through a combination of questionnaires and semi-structured interviews with DEBRA patient organisations in Spain, Australia, Ireland, the UK, and Austria, we identified differences in provision relating to multiple types of healthcare resources.

All countries in the sample have relatively similar access to government financial supports and benefits, with most providing some element of care support grants, assistance with bills, grants for home improvements, travel support, and disability allowances. It will be important in the future to consider the adequacy of such provisions considering our review of evidence relating to quality-of-life which suggests that people with EB often face financial challenges.

The financial burden of EB includes ongoing costs for bandages and dressings for wound care. We found disparities in support for access to dressing materials across countries. For example, EB-

specific government dressing schemes are available in Australia and Spain, with the Australian scheme delivering bandages to the homes of people with EB monthly. People in the UK can receive free dressing material delivered to their homes through a pharmacy scheme. Austria has no dedicated scheme, but their insurance model of healthcare provides access to free or cheap bandaging and dressing materials for people living with all severities of EB. However, in Ireland, there is no such devoted scheme, and families may struggle to access dressing materials. People with severe forms of the disease can generally get what they need through the HSE. However, those with moderate and milder forms of the condition, and those on the margin, are often left to pay for bandaging themselves at significant expense.

The management of EB often requires a diverse range of treatment and supports tailored to the person's needs. Our findings suggest that all sample countries have relatively good access to medicines and specialist EB centres. Access to EB-specific dental care was identified as difficult in each of the sample countries, including Ireland. The UK recognises the need for this treatment which is delivered through a combination of specialist centres and free care through the NHS. Access to physiotherapy, occupational therapy, and medical equipment varies across the sample countries. For example, in Ireland, people with less severe forms of the disease, who require specialised equipment for shorter periods, can have difficulty accessing what they need and often rely on Debra for assistance.

Access to mental health care is limited across all countries. In Spain, the Government is seen as not allocating enough resources to the provision of psychological care, and access to long-term care in the UK is difficult. The Irish system is characterised by long waiting times and poor coverage, with people often resorting to paying independently for mental health supports. However, in Australia the roll out of telehealth was used successfully during Covid which increased access to EB specialist counsellors. This demonstrates that barriers to accessing mental health support can be addressed through innovation.

While access to sufficient home nursing care was highlighted as an issue in the UK and Austria, Australia, Ireland, and Spain were identified as having relatively good access to this vital service, offering regular access to home nursing care packages. However, challenges remain in the delivery of these packages, such as the lack of a contingency plan when nursing staff are unavailable, and a lack of nurses trained in EB to replace those who move on. Access to respite care for less severe cases was an issue across multiple countries.

Considering the learning generated by this review of evidence and cross-national comparison of medical and material supports, there are actions that could be taken to improve aspects of quality-of-life for people with EB. The key findings of this study are listed in the opening pages of this report.

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1. Introduction

1.1 Defining the disease

Epidermolysis Bullosa (EB) is a group of hereditary chronic dermatological conditions in which the proteins necessary for the cohesion of the skin are absent (Prodinger, et al., 2019). The disease is characterised by fragility of the skin, and mucosal epithelia in certain cases, that result in non-scarring blisters and erosions caused by minimal mechanical trauma, which can be fatal (Fine & Hitner, 2008). EB has a low incidence rate, estimated in the UK to be 68 per million live births (Petrof, et al., 2022). There are four major types of EB.

Epidermolysis Bullosa Simplex (EBS)

EB Simplex (EBS) is defined as a skin blistering disorder caused by cleavage of keratinocytes within the basal layer of the skin primarily due to mutations in the keratin 5 and keratin 14 genes (Rugg, et al., 2007). EBS is the most common type of EB, but many milder cases can go undiagnosed and more severe cases can be mistaken for Dystrophic or Junctional EB, particularly in the neonatal stage (Rugg, et al., 2007). Incidence of this EB subtype within the US is estimated at 7.87/1,000,000 live births, with a prevalence of 6/1,000,000, and a high likelihood of the condition being underdiagnosed (Monteavaro Mariath, et al., 2020).

Inheritance of this type of EB usually occurs in an autosomal dominant manner, it is rarely inherited in an autosomal recessive manner although this is common in some regions of the world (Has, et al., 2020). Symptoms range widely in levels of clinical severity and are usually extracutaneous in nature. People with severe EBS can experience widespread and acute blistering, with haemorrhagic blisters being a common occurrence, whereas those with milder forms of the condition tend to experience minor blistering of the feet and elbows (So & Teng, 1998). There is a high degree of complexity in the genotype of EBS with mutations found on seven distinct genes, which makes it a more difficult candidate for future gene therapies (Has, et al., 2020).

Junctional Epidermolysis Bullosa (JEB)

Junctional EB (JEB) is an autosomal recessive disease caused by reduced adhesion of the dermis to the epidermis due to deficiencies in one of the following proteins: laminin 332, type XVII collagen integrin $\alpha 6\beta 4$ or integrin $\alpha 3$ (Condrat, et al., 2019). JEB has considerable variation in the severity of symptoms across the two major subtypes, intermediate and severe. Severe JEB is associated with early mortality in infants from age six to twenty-four months, with this subtype making up to 20% of JEB cases in the US (Tran & Cohen, 2022). JEB is characterised by blistering within the plane of cleavage through the lamina lucida at the basement membrane zone of the skin (Tran & Cohen, 2022). JEB is not as common as EBS or dystrophic EB, with an incidence of 1/1,000,000 live births with shorter prevalence rates due to lower life expectancy (Petrof, et al., 2022).

Dystrophic Epidermolysis Bullosa (DEB)

Dystrophic EB (DEB) can be inherited in a recessive or dominant manner, with a high degree of overlap in the phenotypes between both. However, the recessive form of the disease is generally more severe. DEB is characterised by a plane of cleavage beneath the lamina densa in the skin (Fine, 2010). Scarring following blistering of the skin is the hallmark of dystrophic EB. It is also common for severe forms of recessive DEB to have secondary extracutaneous complications (Fine, 2010). DEB has shown to be an ideal candidate for the development of gene therapies, with many research studies currently underway or complete. This is due to both subtypes of the disease being caused by mutations in a single gene, COL7A1, which encodes for the protein collagen VII (Bruckner-Tuderman, 2019).

People with DEB suffer from a variety of symptoms. The skin is often extremely fragile resulting in blistering and open wounds, sometimes throughout the body. Scarring on the hands, feet and elbows is common in this type of EB (Pfendner & Lucky, 2018). Further complications include scarring along the lining of the oesophagus and blistering within the mouth which can make eating and drinking difficult, and an increased risk of

developing Squamous Cell Carcinoma, an aggressive form of skin cancer (Pfendner & Lucky, 2018).

Kindler syndrome

Kindler syndrome is an exceedingly rare subtype of EB, with around 250 affected individuals recorded worldwide since it was first reported (Gkaitatzi, et al., 2019). The condition is caused by a mutation in the FERMT1 gene that encodes for the protein kindlin-1, which plays a critical role in specialised epithelial cells called keratinocytes. Symptoms include gingivitis, skin atrophy, trauma-induced blistering, and skin discolouration (Lai-Cheong & McGrath, 2010).

Current estimates of the overall incidence of EB vary greatly, as seen in Table 1, due to differences in patient capture in the various regions.

Table 1: Prevalence rates of EB

Country	Prevalence of EB
United Kingdom	27.5 per million
Italy	10.1 per million
Netherlands	22.4 per million
Australia	10.3 per million
Northern Ireland	32 per million
Norway	54 per million
United States of America	19.57 per million
Japan	5.1 per million

Source: (Petrof, et al., 2022)

1.2 Aims and objectives of this report

The aim of this study is to review the impact of EB on quality-of-life and access to healthcare resources and government supports internationally, with a view to identifying areas where access to EB supports and services in Ireland can be improved in the future. In order to achieve this aim, we present:

a) an in-depth literature review of the existing international evidence relating to the quality-of-life and economic burden of EB.

b) a comparative review of access to healthcare resources and government supports across five countries, including Ireland.

The latter section was informed by data gathered through a combination of questionnaires and semi-structured interviews with DEBRA patient organisations in Spain, Australia, Ireland, the UK, and Austria.

The quality-of-life burden related to EB is explored first, followed by a review of the literature on the economic burden of EB. A review of the key journals in the area of rare diseases, dermatology and health economics is undertaken to implement this aspect of the project. A review of healthcare resources and government supports offered to people living with EB in Ireland in an international context is then provided using the aforementioned questionnaire and semi-structured interviews approach.

2. International evidence around the quality-of-life burden of EB

2.1 What do we mean by quality-of-life?

Quality-of-life measures the many aspects, social, physical, and psychological, that constitute an individual's wellbeing (Felce & Perry, 1995). It is a multidimensional and abstract construct that is difficult to measure in EB because of the many subtypes and rarity of the disease. There is a low level of quantitative data available when compared with other skin diseases. Combined, these elements mean that the quality-of-life of people living with or caring for someone with EB remain under-researched. This lack of research could affect the level of government funding provided for EB programmes in the future.

Table 2: Conceptualising quality-of-life

Dimension(s)	Domain(s)	Indicator(s)
1. Mortality	Duration of life, death	Years of life lost
2. Functional	Physical	Fitness, Restrictions on activity
	Psychological	Well-being*, Distress**
	Social	Daily role, Ability to work
	Cognitive	Reasoning, Memory, Alertness
3. Health perceptions	Satisfaction	How satisfied is the individual with functioning
	General	Concern, Self-rating
4. Opportunity	Social or Cultural	Support, Level of societal stigma
	Coping	Access to care, Ability to withstand physical or psychological stress
5. Morbidity	Symptoms	Indirectly observable evidence that is subjective in nature
	Signs	Directly observable clinical evidence that is objective in nature
	Physiology	Laboratory measurements/findings, Pathological findings
	Self-reports	Self-reporting by patients on conditions and symptoms

Source: Adapted from (Patrick & Bergner, 1990)

Measuring quality-of-life depends on how well we conceptualise it. Testa & Nackley (1994) and Patrick & Erickson (1993) propose five broad dimensions to quality-of-life, as it pertains to health policy. These five dimensions allow researchers to translate the complex concept into measurable constructs and are described in Table 2. Studying the impact of EB requires the further use of Health-related Quality-of-Life (HRQL), a distinct construct measuring the effect of disease/health conditions on a patient's quality-of-life and those of their carers and families. HRQL instruments have been developed to measure this impact as well as the effect of treatment(s) (Guyatt, et al., 1993). These instruments are sub divided into 'generic' and 'disease-specific'.

General health issues, including disability, distress and functional capacity are addressed using generic HRQL instruments across a range of patient groups (Solans, et al., 2008). A number of EB-related studies, shown in Table 3, utilised such instruments, including The Dermatology Life Quality Index (DLQI), The Children's Dermatology Life Quality Index (CDLQI), The Zarit Caregiving Burden Scale (ZCBS), The Short form of the McGill Pain Questionnaire (SF-MPQ), The Hamilton rating scale for anxiety (HAM-A), The Hamilton rating scale for depression (HAM-D), Skindex-29, SF-36, and The Visual Analogue Scale (VAS). Other studies measure HRQL using semi-structured and psychosocial questionnaires.

Measuring the HRQL for rare diseases, such as EB, can be a difficult task. However, it is increasingly carried out using disease-specific instruments that are more clinically sensitive to a rare disease and have a greater ability to detect change (Lenderking, et al., 2021). Eight studies included in this report utilised an EB-specific HRQL instrument to measure quality-of-life for patients, carers and family members.

In total, 29 studies (Table 3) were reviewed for this study and the results show that the effect of EB on quality-of-life is, in most cases, profoundly negative. However, the impact varies greatly by clinical severity of disease, disease severity as perceived by carers and family, the skin area involved, level of pain and itch, sex, and age (Kearney, et al., 2019). The following sub-sections will detail the impact of the disease on quality-of-life for people living with or caring for someone with EB and their family members.

Table 3: Quality-of-life papers

Year	Author(s)	Country/ Region	Type of Study	Statistical Analysis	Sample Size	Adult Patients	Child Patients	Caregiver	Age Range	QoL Instrument	EB Type(s) Represented
2021	Sangha et al.	Canada	Qualitative participatory research study	No	8	4	4	0	12 to 20	Psychosocial Questionnaire	EBS, DDEB, RDEB
2020	Bruckner et al.	United States	Cross-sectional study	No	156	63	0	93	18 to 70	QoLEB	EBS, DDEB, RDEB, JEB
2020	Togo, et al.	International	Systematic Review	No	745	-	-	-	0 to 86	N/A	EBS, DDEB, RDEB, JEB
2019	Martin et al.	International	Systematic Review	No	-	-	-	-	-	N/A	EBS, DDEB, RDEB, JEB
2019	Kearney et al	Ireland	Cross-sectional study	Yes	14	6	8	10	3 to 67	None	EBS, DEB
2019	Danescu et al	Romania	Cross-sectional study	Yes	50	21	29	0	3 to 60	CDLQI, DLQI, QoLEB	EBS, DDEB, RDEB, JEB, KS
2017	Brun et al.	France	Cross-sectional study	No	57	30	27	0	1 to 85	CDLQI, QoLEB	EBS
2017	Jain, et al.	International	Literature Review	No	949	-	-	-	-	N/A	EBS, DDEB, RDEB, JEB
2017	Kahraman et al.	Turkey	Qualitative participatory research study	Yes	8	0	8	0	3 to 9	ZCBS	Unknown
2017	Fortuna et al.	Mexico	Cross-sectional study	Yes	53 (26 Control)	27	0	0	-	SFMPQ, HAM-A, HAM-D	DDEB, RDEB
2016	Angelis at al.	Europe	Cross-sectional study	Yes	204	121	83	81	-	HRQoL	EBS, DDEB, RDEB, JEB
2016	Jeon et al.	South Korea	Cross-sectional study	Yes	13	-	-	-	-	QoLEB, Skindex-29	RDEB
2016	Medina et al.	Mexico	Cross-sectional study	Yes	18	10	8	0	11 to 59	DLQI	EBS, DEB, JEB
2016	Cestari et al.	Brazil	Cross-sectional study	Yes	57	40	17	0	-	QoLEB	EBS, DDEB, RDEB, JEB
2014	Yuen et al.	Netherlands	Cross-sectional study	Yes	55	55	0	0	19 to 85	QoLEB, Skindex-29, SF-36	EBS, DDEB, RDEB, JEB
2014	Snauwaert, et al.	Netherlands	Cross-sectional study	Yes	40	40	0	0	-	LIS	EBS, DEB, JEB
2014	Eismann et al.	United States	Cross-sectional study	Yes	71	0	71	0	-	QoLEB	EBS, DDEB, RDEB, JEB
2013	Kýrová, et al.	Czech Republic	Cross-sectional study	Yes	43	16	27	-	-	CDLQI, DLQI	EBS, DDEB, RDEB
2013	Grocott et al.	United Kingdom	Qualitative participatory research study	No	-	-	-	-	-	None	Unknown
2012	Adni, et al.	United Kingdom	Phenomenological study	No	6	6	0	0	24 to 67	IPA	DEB, JEB
2011	Williams et al.	United Kingdom	Qualitative participatory research study	Yes	11	0	11	0	10 to 14	IPA	EBS
2011	Dures at al.	United Kingdom	Inductive qualitative approach	Yes	24	24	0	0	21 to 89	Semistructured Interview	EBS, DEB, JEB
2010	Margari et al.	Italy	Cross-sectional study	No	25	11	14	0	1 to 43	DLQI	EBS, RDEB
2010	Pagliarello & Tabolli.	International	Literature Review	No	-	-	-	-	-	N/A	EBS, DDEB, RDEB, JEB
2009	Frew, et al.	Australia	Cross-sectional study	Yes	111	0	111	39	0 to 17	QoLEB	EBS, DDEB, RDEB, JEB
2009	Tabolli et al.	Italy	Cross-sectional study	Yes	125	79	46	28	-	Skindex-29, SF-36	EBS, DDEB, RDEB, JEB, KS
2008	Van Scheppingen et al.	Netherlands	Qualitative participatory research study	No	17	0	11	17	0 to 14	Semistructured Interview	EBS, DDEB, RDEB, JEB
2004	Fine et al.	United States	Cross-sectional study	No	374	234	140	0	-	ADI, VAS	EBS, DDEB, RDEB, JEB
2002	Horn and Tidman	Scotland	Cross-sectional study	No	120	90	30	0	1 to 86	CDLQI, DLQI	EBS, DDEB, RDEB

2.2 Impact of EB on quality-of-life for children

The impact on children living with EB was studied in 17 papers, with most authors concluding that children suffer a greater reduction in quality-of-life due to the disease than adults. A number of themes emerge from the research, including physical pain and itchy skin, a sense of being different amongst their peers, a lack of independence due to a heavy reliance on others, challenges with daily activities and physical activity, underdeveloped coping mechanisms, and a higher rate of psychiatric symptoms.

Pain is a prominent feature of EB that was felt by all participants in the relevant studies. Chronic inflammation, blistering (both external and internal), open wounds, occasional and unpredictable pain caused by mechanical trauma of the skin, pain caused by seasonal changes in the temperature, and the suffering caused by daily time-consuming and extensive dressing changes, negatively affect quality-of-life for children suffering from the disease, as outlined by the studies presented in Table 3. Brun et al., (2017) found that 94% of children above the age of eight and 80% of children below the age of eight suffered pain due to blister formation. The authors also found that 53% of children above the age of eight and 90% of children below the age of eight experienced a high degree of pain due to dressing changes (Brun, et al., 2017). Tabolli et al., (2009) found that 82% of participants experienced pain often due to EB, in the form of sensitive and itchy or irritable skin, a burning feeling or, at times, bleeding. Yuen et al., (2014) found that 84% of participants experienced pain in a similar manner.

In a study by Horn and Tidman (2002), 37.5% of children with DEB were found to experience near constant pain due to “skin scratching” and blister formation. Van Scheppigen et al., (2008), Tabolli et al., (2009), and Horn and Tidman (2002) concluded that children living with EB suffered a greater level of pain than adults, due to a lack of coping and management mechanisms and the pain caused by childhood development. Children with severe subtypes of the disease suffer pain for most of the day and night and are often sleep deprived as a result, while children with milder forms of the disease must deal with unpredictability in the occurrence of pain. Pain can increase swiftly due to changes in temperature and mechanical trauma and patients describe having to take each day as it comes (van Scheppigen, et al., 2008; Tabolli, et al., 2009; Horn and Tidman, 2002).

Children often described a sense of being different from their peer group, locating this ‘difference’ within themselves and having a strong desire to be normal, failing to understand that normality is a relational construct (van Scheppingen, et al., 2008). Those living with EB identified two elements to the sense of self as being different: the visibility of the condition and the fear of contagion (Pagliarello et al., 2010). As EB is a skin disorder, children are often visibly different due to blister or wound formation on the face and hands. The more severe forms of EB require bandaging throughout the body, can cause fusing of the toes and fingers due to scarring, can cause nails to fall off and deformed teeth, and can impair an individual’s ability to walk due to skin contractures which will often result in children having to use wheelchairs (Fine, et al., 2004). Combined, these highly visible physical differences, leave children open to uninvited scrutiny in the form of staring and unwanted comments and questions. These comments can sometimes be unkind, and the children can be subject to bullying and teasing from their peers and the public at large (Sangha, et al., 2021; Williams, et al., 2011).

The reactions of others can make some of the children feel conscious of their condition and more aware in a way that they had not been previously. Adolescence can be a particularly difficult time for children with EB, due to the changing of schools and an increase in the emphasis on physical appearance and attractiveness that occurs at this age (Williams, et al., 2011). For many children, the sense of failing to meet so called normative standards, or western standards of beauty, led them to question whether others would respect their personhood. As a result, adolescent children with EB can feel increased levels of isolation and loneliness, particularly due to the rarity of the condition (Williams, et al., 2011). The second and particularly salient element of the sense of self as being different in children with EB is the fear of others that the disease is contagious (Sangha, et al., 2021; van Scheppingen, et al., 2008). Fear of infection could cause the children’s peer group to distance themselves from the patient leading to further isolation and increased levels of psychiatric symptoms. This fear was often due to a lack of education on the disease.

Frustration caused by an inability to carry out menial daily tasks and physical activities also led to a decrease in the quality-of-life for many participants. In the study by Brun et al., (2017) 52% of children were found to have reduced their level of activity, both general and sports related, due to EB-associated pain. Cestari et al., (2016) found that

difficulties in movement, both within and outside the home, was the factor that contributed most to lowering quality-of-life scores. The results of a study by Eismann et al., (2013) showed that most participants avoided sports, while a little under half needed assistance when bathing and writing. Children with more severe forms of the disease required assistance moving around the home and when eating. Horn & Tidman, (2002) found that an average of 75% of children had great difficulty in participating in sporting activities, with more severe forms having a higher degree of difficulty. Kýrová et al., (2013) also found that children with RDEB have great difficulty in playing sports, and often avoided exercise due to pain and feelings of embarrassment.

There was an extensive study carried out by Tabolli et al., (2009), involving 46 children with various subtypes of EB. Researchers investigated the impact of EB on self-care, mobility, and daily activities. They found that 59% of children had difficulty with mobility, compared to 33% of adults, 82% of children had difficulty with self-care, compared to 29% of adults, and 76% of children had difficulty in carrying out daily tasks, compared to 53% of adults with the disease. Tabolli et al., (2009), also found that children and adolescents with limited physical abilities, and ability to carry out menial daily tasks, have a corresponding reduction in their level of independence which can hamper their ability to make friends and socialise and lead to feelings of isolation and frustration, which contribute greatly to reducing their quality-of-life.

Many studies have found a higher-than-average frequency of psychiatric symptoms in children with EB. Horn & Tidman (2002) and Kýrová et al., (2013) found that children living with EB had feelings of embarrassment and sadness, and a number of participants suffered heightened levels of bullying. The authors also found that children had difficulties in forming and maintaining friendships. A study by Brun et al., (2017) confirmed these results, finding that 32% of participants stated that their friendships had been affected by the disease and 56% of children felt higher levels of sadness. This research also noted that only 10% of participants consistently followed up with a psychologist. Tabolli et al., (2009) reported that 41% of children had feelings of anxiety and depression. Medina et al., (2016), concluded that children had higher levels of stress than adults, with such feelings decreasing over time with improvements in coping mechanisms. It is clear that EB provokes numerous psychiatric and psychosocial problems, as well as physical problems, that impact on quality-of-life for children suffering from the disease. Above average psychiatric symptoms have been found to be present in this cohort, including anxiety, depression, and behavioural disturbances.

2.3 Impact of EB on quality-of-life for adults

The impact on adults living with EB was studied in 18 of the 29 papers included for review. EB greatly reduces quality-of-life for adults living with the disease, compromising employment prospects, relationships, the pursuit of education, and home life. A number of research papers found that the impact of the disease is greater for women than for men, but this difference is only minor with Yuen et al., (2014) noting there was no statistical significance to the finding. Adults, like children, suffer from daily limitations resulting from the pain of blisters and open wounds. There remain issues with physical mobility, carrying out daily activities, and psychological complications. However, there is a reduction in the importance of feeling 'normal' when compared to children and adolescents, although there is still distress caused by discrimination as a result of the visibility of the disease (Yuen, et al., 2014). The high significance of the desire to feel 'normal' is replaced with worries over the impact the disease may have on employment prospects, social interactions, and relationships. Adults feel further distress over the hereditary, life-long nature of EB, and the fact that there is currently no cure for the illness (Martin, et al., 2019).

Adults experience similar levels of acute pain, chronic pain, and itch to children, although they are generally better equipped to deal with the pain they experience. Fortuna et al., (2017) included within their study a control sample of adults without EB. They found a significant difference in the level of pain experienced by the two groups, with four characteristics of pain having a p-value less than 0.05. These characteristics were shooting pain, splitting skin, tender skin and throbbing pain. Severe throbbing and shooting pain were felt by 44.4% of people with EB, while only 15.4% of the control sample felt mostly mild throbbing pain and 19.4% felt shooting pain. Splitting pain was felt by 37% of people with EB compared to only 3.8% of the control sample. Tender skin was experienced by 59.3% of people with EB compared to 19.2% of the control sample who mainly experienced mild pain. Horn & Tidman, (2002) found that the majority of adult participants suffered from frequent pain and itch, especially those with the

RDEB subtype of the disease. These findings were mirrored by Kýrová et al., (2013), and Medina et al., (2016) while Margari et al., (2010) found that 84% of participants felt pain on a frequent basis.

In a study by Jeon et al., (2016) researchers found that women felt more pain that men, but men were more affected by symptoms such as itching, burning sensations, irritation, and tenderness. Yuen et al., (2014) gives a detailed account of the pain suffered by adults with EB, with 84% of participants frequently suffering from pain. A total of 51% of adults felt skin sensitivity often or all the time, while 31% felt a burning sensation on their skin often or all the time. A further 33% of participants experienced itching often or all the time, and 20% of adults experienced cutaneous bleeding often or all of the time. The study by Tabolli et al., (2009) gives similar findings with 56% of adults experiencing skin sensitivity, but with a greater number of participants experiencing skin itch and burning at 51% and 40% respectively. Pain is the most debilitating symptom of EB and causes the sharpest reduction in quality-of-life for adults with EB. Pain is identified in adult participants just as much as in children, with differences in its management (Tabolli, et al., 2009).

Adults with EB also experienced frustration caused by an inability to carry out daily tasks and physical activities which served to lower quality-of-life. Of all the daily activities reviewed by the papers, eating was reviewed the least, with Yuen et al., (2014) reporting that 20% of adults found eating difficult. The authors also reported that a similar number (19%) required assistance bathing, with 40% of participants finding doing the weekly shop difficult, and 24% experiencing chronic fatigue due to their skin condition. Tabolli et al., (2009) found a similar level of chronic fatigue and mobility problems in adults, at 31% and 43% respectively. Kýrová et al., (2013) reported that adults had increased difficulty shopping, as well as difficulties socialising and engaging in leisure activities. Findings from the study by Horn & Tidman (2002) once again show an increased difficulty in shopping for adults with the disease. The authors also found that 23% of participants had great difficulty engaging in sporting and leisure activities. Difficulty with shopping is also evident in the study by Brun et al., (2017).

The literature gives clear evidence that what are menial and mundane everyday tasks for healthy individuals are often strenuous and painful for adults living with EB. People are deprived of their independence, often having to rely on carers, family members and friends to assist them with shopping, bathing, and bandage dressing. Chronic fatigue and difficulty sleeping due to pain and itch were also found to impact negatively on quality-of-life for adults (Jeon, et al., 2016). The combined influence of pain, sleep deprivation, and difficulties in carrying out physical activities for adults with EB reduce their social mobility and compromise their ability to progress in education and seek stable employment (Kearney, et al., 2019).

Adults with EB exhibit higher than average frequencies of psychiatric symptoms, including depression, increased anxiety, and paranoia. Margari et al., (2010) reported that 80% of adult participants suffered from increased levels of psychiatric symptoms, compared to 79% of child participants. In the same study, depression had a prevalence of 63.6% amongst people with EB compared to 10.1% for the general population. Anxiety had a much greater prevalence amongst people with EB at 45.5% compared to 11.1% for the control sample. Interestingly, Margari et al., (2010) also found there is a similar level of social phobia amongst people with EB and the general population at 2.1% and 2.2% respectively. EB compromised the ability of people to form stable emotional relationships, with 73% of adults being unmarried or having no emotional relationship. A total of 87% of this subgroup stated that they would never marry, but only a small fraction (27%) had feelings of anxiety or fear about their lack of sexual or emotional relationships. On a positive note, 91% of participants stated that they expected to realise their expectations in the romantic sphere (Margari, et al., 2010). Yuen et al., (2014) also found that 51% of adult participants experienced depression and anxiety.

Research carried out by Brun et al., (2017) found that 87% of adults experienced frequent frustration due to EB Simplex and that a smaller number of participants had feelings of shame (27%), depression (17%), and anxiety (40%). A large portion of the adult participants in this study (47%) reported that the disease placed a high strain on their family relationships. The study by Tabolli et al., (2010) reinforces these findings with a large percentage (51%) of participants reporting feelings of anxiety and depression, and 27% of participants reporting anxiety and difficulty in their sexual and romantic relationships.

A final study by Yuen et al., (2014) found similar levels of frustration (71%) amongst participants to Brun et al., (2017) and a similar level (22%) of anxiety when engaging in romantic and sexual relationships. Encouragingly, most adults develop mechanisms of coping with and managing the

psychological impacts of the disease over time. In the study by Margari et al., (2010), the majority of participants (56%) had strategies to deal with EB, with 27% emphasising the value of family support, and 56% stressing the importance of social services and access to adequate healthcare to alleviate pain and discomfort, and increase psychological health.

2.4 Impact on quality-of-life across disease severity

The variation in subtypes of EB has a corresponding variation in disease severity, which can affect quality-of-life in differing ways. Researchers that included multiple subtypes of the disease reported that the subtypes with the lowest overall quality-of-life are the most severe forms of EB, JEB and RDEB (Danescu, et al., 2019; Fine, et al., 2004), and the frequency of pain was greatest in people with JEB and RDEB. Pain became more pronounced in adulthood for people with these subtypes, which corresponded with an increase in the surface area of skin affected (Fine, et al., 2004). People with EBS were found to be severely impacted by pain, while people with Dominant DEB (DDEB), Recessive DEB (RDEB) and JEB were extremely severely impacted by pain.

Taken collectively, the findings of the various studies demonstrate that people suffering from RDEB and JEB need a great deal of assistance to complete the most basic of daily tasks, with severe limitations on social interactions, physical mobility, maintaining or attaining romantic relationships, and restrictions on their independence. There is an even distribution of psychiatric symptoms, regardless of the disease subtype. Horn & Tidman (2002) found that children with EBS had a greater difficulty in forming friendships, while patients with DDEB and RDEB were bullied more often and had greater feelings of embarrassment respectively. Children with milder forms of EBS were found to have similar levels of embarrassment and feelings of being different to the more severe forms of EB.

2.5 Quality-of-life for those caring for someone with EB

The burden of care for parents of children with EB is substantial but varies in magnitude and intensity based on the severity of their child's illness (Kahraman, et al., 2017). The impact on quality-of-life for carers of children with EB was studied in six papers (Angelis, et al., 2016; Bruckner, et al., 2020; Kahraman, et al., 2017; Kearney, et al., 2019; Martin, et al., 2019; van Scheppingen, et al., 2008). Caring for a child with EB curtails employment opportunities and leisure time for parents. The burden also strains marital and familial relationships and creates an atmosphere of uncertainty (van Scheppingen, et al., 2008). However, the greatest impact for parents is emotional, having to watch their child endure pain, and at times, inflict pain on their child during daily dressing changes (van Scheppingen, et al., 2008).

The burden of care is often greatest for female carers, with Angelis et al., (2016) finding that 86.4% of their primary caregiver sample was female. Mothers and female relatives of patients take up traditional roles within the family and, as a result, unemployment and underemployment disproportionality affect this cohort. Male carers, including fathers and male relatives, are more likely to take up employment and focus on administrative and organisational tasks, such as paying hospital bills, transportation, and seeking government grants (Angelis, et al., 2016). There are a number of themes that impact the lives of primary carers that emerge from the research: feelings of uncertainty surrounding their loved one's illness, always being on duty, family problems, and restrictions to leisure time and employment (Angelis, et al., 2016; Bruckner, et al., 2020; Kahraman, et al., 2017; Kearney, et al., 2019; Martin, et al., 2019; van Scheppingen, et al., 2008;).

2.51 Impact of care on parents/primary carers

EB is an unpredictable disease, and as a result, carers never know when their child/loved-one will have a particularly bad day. Blisters can develop unexpectedly, people can have falls and develop infections from open wounds, and physical deteriorations can prevent people with EB from walking or carrying out basic daily tasks. This unpredictability is discussed in the literature and greatly troubles carers, who are often prevented from engaging in any spontaneous activities such as family gatherings or socialising with friends (van Scheppingen, et al., 2008). This contributes to a lower quality-of-life for carers, with Angelis et al., (2016) finding scores for carers were significantly lower than the control adult population. The average EQ – 5D index for carers in the study was 0.696, 21.4% lower than the average UK adult. Similar results were found for the EQ – 5D VAS scores with carers having an average score of 73.1, while the average UK adult had a score of 86.6,

significantly higher than the result for carers (Angelis, et al., 2016).

Uncertainty over short-term issues is combined with uncertainty/unpredictability as to the future prospects of their loved-one's health. Fear over the future remains even with a clear diagnosis of their loved-ones disease subtype. Parents and carers of people with severe EB have anxiety over their loved-one's mortality, as their condition worsens. Van Sheppingen et al., (2008) found that parents of children affected by severe forms of the disease had high levels of anxiety over how long they would have with their children and intense guilt when they sometimes felt that their child would be better off if "it was all over with".

The quality-of-life of primary carers is further decreased due the mental strain of having to watch their loved-one endure pain and sometimes having to inflict pain as part of their role as a carer (Kearney, et al., 2019). Parents of children with EB of all severities are greatly afflicted by having so few tools available to them to relieve the suffering of their children. They are often distressed when having to cause their child pain when teaching them to walk and taking care of open wounds and blisters (Fine, et al., 2004). Daily bandaging affects carers greatly due to the length of time they must cause pain and discomfort to their loved-one (Kearney, et al., 2019). Carers regularly play multiple roles as nurses, administrators, and parents. Playing so many roles inevitably leads to a feeling of never being off duty.

Bandaging is the most time-consuming activity, but there are numerous other activities that require the attention of carers (van Scheppingen, et al., 2008). They include the prevention of the fusing of fingers and toes through the splinting and stretching of hands and feet, food and medicine preparation, bathing, assisting with going to the bathroom, mobility, playing games and social activities, and assisting with other basic activities such as blowing of the nose (Eismann, et al., 2014; Fine et al., 2004).

Time-consuming care greatly restricts the ability of parents to spend time with and focus attention on their other children. As a result, parents have feelings of guilt that can increase the burden of care (Kahraman, et al., 2017). Furthermore, most parents of children with EB, of all degrees of severity, experience a strain on their relationship as a result of the disease. In the study by van Scheppingen et al., (2009) there were four divorces amongst the eleven families in the sample. There is a sense of isolation and distance within relationships as a result of not being able to pay enough attention to each other and often discussing their worries over the illness on occasions that they do get to spend time together (van Scheppingen, et al., 2008).

2.52 Impact of disease severity on quality-of-life for carers

Disease severity has a clear impact on quality-of-life for carers. The major issue is time spent caring for their loved one. Parents of severely affected children, such as those with DEB, may have to spend two to three hours daily changing bandages (Grocott, et al., 2013). Bandages often cover much of the body which increases time spent on dressing them and increases costs for carers (Bruckner, et al., 2020). Severely affected children routinely need assistance with basic activities such as using the bathroom and parents can be on-duty for 16 hours a day (van Scheppingen, et al., 2008). However, parents of children of all disease severities share common problems. Parents struggle with the visibility of the disease, which can lead to their children being bullied or excluded (van Scheppingen, et al., 2008). The burden of care restricts leisure time and reduces employment opportunities (Bruckner, et al., 2020). Finally, there is often strain on relationships as other children within the family can feel neglected and there is less time to be spent with partners (Kahraman, et al., 2017). It is clear that EB has a negative impact on carers, regardless of disease severity.

Table 4: Experiences of children with EB

Primary Theme	Concern	Experience	Study
Enduring physical pain and itch	Itchy skin	*"I think that the itch is so awful that you can't even begin to understand. (…) I wish I could just stop it, but I can't."*	van Scheppingen et al. (2008)
		"It's almost always there, wherever I am."	
		"Even in your sleep you're all tensed up, telling yourself, 'Don't scratch!"	
	Pain from mechanical trauma	*"When you're walking in the city, you just aren't comfortable walking. I can't imagine walking without pain."*	
	Pain from wound care	*"Because the dressings stick to the wounds and they have come off. That hurts a lot."*	
Feelings of being different and social anxiety	Being different	*"Sometimes I find it hard that I always have to explain it to everybody (…) Because you have this idea, 'I'm just normal', and 'I just want to be somebody without a skin disease'."*	van Scheppingen et al. (2008)
		"When people, like, have to describe me, they say 'that girl with the skin disease.' (…) I would rather have people see me as myself instead of 'that girl with the skin disease'."	
		"I guess it (other people staring) is because I'm different … I can't walk properly like the other people. And I can't do things like the other people can."	Williams et al. (2011)
	Social interactions	*"(I feel) sort of left out."*	van Scheppingen et al. (2008)
		"In fact you do get involved in stuff, but you just have to be careful all the time."	
Loss of independence	Mobility	*"I would just like to run free for once. I can't do that anymore because of my feet."*	van Scheppingen et al. (2008)
	Basic activities	*"(I don't like) when I drop things all the time and then I can't pick them up."*	
		"(…) cos I can't do anything, do like all the things that other people can do."	Williams et al. (2011)
The visible nature of the disease	Visibility	*"I find it kind of hard that when I hurt myself you can see I have this disease."*	van Scheppingen et al. (2008)
		"The most annoying thing is when people look at you and they don't say a word. Then you know that they're just staring all the time."	
		"I just get crazy with all that staring."	

Table 5: Experiences of adults with EB

Primary Theme	Concern	Experience	Study
Enduring physical pain	Pain from wound care	*"That pain dressing – when I feel really bad I stay in bed. It just takes the pain away."* *"… It is excruciating when the dressings keep coming on and off and (there is) unbearable pain."*	Grocott et al. (2013)
	Inadequate dressings	*"He also has a fear of his skin being damaged or delayed healing due to inadequate dressing provision. He added 'I felt like I had reverted back 10 years to the previous unsatisfactory wound dressings I had to cope with.' 'It's like telling a diabetic patient that they can't have insulin anymore'."*	Downe (2017)
	Fear of pain	*"(…) Luke, (having) suffered hours of painful dressings changes, (…) since childhood, (…) talked about the fear of going back to the days when his dressings were painful to apply and remove."*	
Loss of independence	Reliance on others for aid with wound care	*"I am 25 this year, and I have never done a dressing by myself!"* *"(…) My mum has trained my little sister now to do my dressings."* *"My mum and I usually start to do my dressings at around 7pm after I'm back from college. (…) It often takes up to 3 hours."*	Grocott et al. (2013)
Economic burden	Concerns over the financing of bandages	*"I have been told my local GP practice gets an extra £2,000 a year to cover my additional needs. When you think the dressings cost £10, and you have 18 on at any one time, three times a week, that £2,000 soon gets blown out of the window (…) So if I add to what I call my shopping list at the GP, they don't like adding to it, because they think I am costing them enough already. I have arguments all the time."*	Grocott et al. (2013)

Table 6: Experiences of parents/carers

Primary Theme	Concern	Experience	Study
Loved Ones Enduring Physical and Psychological Pain	Pain from mechanical trauma	*"Lilly was at the age kids start to crawl, so at that point her face was always battered."* *"I would stand him up, but then he would start to cry really hard. With your first child, of course, you find that a terrible thing. You don't want to hurt him."*	van Scheppingen et al. (2008)
	Inflicting pain during dressing changes	*"I found it very hard to hurt my own child. You really have to shut yourself off completely."* *"There is always yelling and crying and screaming: 'I don't want this and it hurts so much.' Yes, that just stabs you in the heart."* *"You know that you are going to be hurting your child for an hour or an hour and a half. That runs totally contrary to your feelings. You want to protect your child from this and make sure they don't have to go through it."*	van Scheppingen et al. (2009)
	Psychological pain from bullying	*"The saddest thing for me was when she came home from school, and there had been another confrontation, and someone had said something bad about it again. That was what I found the most difficult thing."*	
Uncertain Nature of the Disease	Uncertainty over pain	*"Just when you think things are all right, another blister develops. At the most unexpected times, really. Well, that's EB."*	van Scheppingen et al. (2008)
	Uncertainty of future health	*"You never know how bad it's going to be (…) You always carry that fear with you: will she get worse or will it just stay like this?"* *"How long do we have."* *"You always have to choose the lesser of two evils. You can never say that if you do this, then you will solve that and that. It is always palliative, with several other problems appearing afterwards. Actually, it's just never ending."*	

Table continues…

Table 6 continued

Primary Theme	Concern	Experience	Study
Always being 'on duty'	Organisation	*"Every time there was so much to arrange that you actually needed a secretary for a day and a half to get everything set up."*	van Scheppingen et al. (2008)
	Burden of care	*"I can't leave Alex alone for longer than half an hour. He can't go to the toilet himself; he can't pour himself a drink. You're always worried that he will fall or that he will hurt himself."* *"It just exhausts you. You live with it twenty-four hours a day. It is different from being at work; you can't close the door behind you."*	
Strain on Familial Relationships and Restrictions to Leisure Time	Family problems	*"I often feel guilty about that. Then I think that she's (child without EB) just going to have to wait her turn. Always."* *"When we (parents) sit down in the evening, we only talk about our worries and about the things that have to be done."*	van Scheppingen et al. (2008)
	Restrictions on planning activities and leisure time	*"You could never make an appointment, and, if you did, nine times out of ten you had to cancel. Either James had just fallen, or something had happened so that he was completely exhausted and needed to sleep. And people just don't understand that; they think you worry too much."* *"Planning is actually very difficult. There are a lot of things that you would like to do, which you can't. We can't go to the beach. The wind hurts him. When it's too hot, you can't go out, when it's too cold, you can't go out, because either way his skin breaks open. You are very restricted."*	van Scheppingen et al. (2008)

Table continues...

Table 6 continued

Primary Theme	Concern	Experience	Study
Ignorance of the Public and Lack of Skills of Healthcare Professionals	Ignorance of general public	*"At first, when we walked with Lily through the village, people were inclined to say something about her, like, 'Oh, how sad.' And there were people who asked questions like, 'Has she fallen down the stairs?' or 'Have you been hitting her?' or 'Does she have chickenpox?' That kind of remark. Well, at first they are very hard to deal with."* *"When John is wearing his sweater and his trousers and all, then you don't see anything. At the most all you see is that his hands are battered, that's all. And then people think that it is not that bad. They don't think about his feet, his knees, his elbows all being bandaged up at that same time."*	van Scheppingen et al. (2008)
	Lack of awareness/ skills of healthcare professionals	*"In a hospital, nobody knew what EB was. Can you imagine, in a hospital? They had never heard of it. Not many hospitals seem to know much about it."* *"At a certain point we were able to get home care. That meant that every three days there was a different nurse on the doorstep, who we had to show how to do John's dressings. (…) And when they started, you thought 'I could have done that four times faster. Then John could go back to sleep.' That cost us so much more time and energy than it gave us. So then you just stop."*	van Scheppingen et al. (2008)

3. International evidence on the economic burden of EB

3.1 Economic burden of rare diseases

Rare diseases are associated with a high economic burden as a result of a lack of effective treatments and a lack of research into discovering effective cures (Angelis, et al., 2015). Calculating the cost of illness for a rare disease is a systematic process that quantifies the economic burden on the individual, their carers and families, and on society (Angelis, et al., 2015). The economic burden consists of direct costs and indirect cost of disease, with the latter being more difficult to measure, especially for rare diseases such as EB (Angelis, et al., 2016). Studying the cost of an illness allows policymakers to prioritise and justify healthcare spending and allocate resources more effectively. Informed budgetary decisions on the economic burdens of disease improve the lives of patients and their carers while also reducing public spending (Jo, 2014). The following subsections of the report will examine the direct economic costs, indirect economic costs, and overall economic burden of EB.

3.2 Cost of illness – direct healthcare costs

Direct costs are occasioned explicitly by the disease and include medical treatment and diagnosis, rehabilitation, home health care, outpatient clinical care, and terminal care (Lee, et al., 2019). Estimating the cost of an illness to society requires the inclusion of additional direct costs including health care training, research expenditures and administration costs to support the treatment of the disease. A complete account of direct costs should also include direct costs borne by carers and patients that are not covered in national health accounts (Carter, et al., 2019). These include transportation costs incurred for travel to health care providers, and additional household expenditures due to the disease such as alterations of property, special diets, special clothing, etc (Carter, et al., 2019). The economic burden of rare diseases is often under researched, with most of the available research on EB focusing on the science of the disease and its impact on quality-of-life, as set out in Section 2 of this report. Therefore, estimating the direct costs of the disease relies on a small number of economic studies.

3.21 Direct healthcare costs of EB

The direct costs of illness for EB were examined in four papers, with a further four papers estimating the expense of bandages and dressings. In each of the papers, significant direct healthcare costs were reported, particularly for people suffering from more severe forms of the disease. Direct costs were found to be high across all studies, with the greatest expense coming from wound and drug costs, along with hospitalisations.

Wound care is the most significant direct cost across all studies, with variations by disease severity and region. A total of eight studies included the cost of bandaging, with four focusing exclusively on this expense. Most of the studies focused on more severe forms of the disease, particularly RDEB, which incur greater bandaging costs as people often have to wear bandages across most of their bodies. However, Flannery et al., (2020) included a range of disease severities in their sample, examining two milder forms of the disease, EBS and a moderate form of RDEB. EBS was found to have a wound care and drug cost of €17,732 per annum, while the cost for moderate RDEB was €5,986. These costs were significantly lower than those for the severe forms of EB, which had an average wound care and drug cost of €59,910, with a maximum cost of €89,780 and a minimum cost of €27,252.

A case study in the UK of a patient with severe RDEB by Downe (2017) found a similar wound care

and drug cost of €74,702.86. The study gave a detailed analysis of dressing costs for an adult with severe RDEB, including mepitol, PolyMem roll, medihoney, octenilin irrigation, tubifast, and Slinky Bandages. This analysis shows that the complex nature of the dressings required for people with RDEB is one of the reasons for such high expenditure on wound care. Kirkorian et al., (2013) found average wound care costs of €71,689 for infants and children living with RDEB in the United States, with variations based on age and disease severity. In another UK based study, Mellerio (2017) found average wound care costs of €42,017 for people living with RDEB. Jeon et al (2016) found that wound care costs were greater than medical costs, with total direct costs of €6,768. These studies show the significant direct cost of wound care and drugs for patients living with EB, and particularly for those suffering from severe forms of the disease.

Hospitalisations and medical visits represent the greatest direct cost after wound care and drug costs. Flannery et al., (2020) found a great deal of variety in the cost of hospitalisations, with the milder forms of the disease incurring no cost for overnight hospitalisations, while the more severe forms had an average cost of €41,731 with a maximum of €56,910 and a minimum of €28,455. Similarly, Angelis et al., (2016) found a high degree of variation in the cost of hospitalisations across eight sample countries. In the Swedish sample, there was no expense for hospitalisations which can be explained by the exclusively mild forms of EBS within the patient sample. In contrast, the average cost of hospitalisations for the Italian sample was €5,420. The higher cost of hospitalisations correlates with the higher proportion of people with severe RDEB within the sample. Average costs for medical visits were €1,670 in the study by Angelis et al., (2016) with visits to day clinics representing a significant cost for people with severe, moderate, and mild forms of the disease and their carers.

The studies by Angelis et al., (2016) and Flannery et al., (2020) list additional direct costs associated with EB. In the study by Flannery et al., (2020), these costs are listed as 'other' and are €1,764 on average. These other costs included physiotherapy, occupational therapy and visits to general practitioners. Transportation for healthcare needs, social healthcare services and professional care are additional direct costs included in the study by Angelis et al., (2016). The combined average for these additional costs is €1,718, with the greatest expense being social health services or formal health carers, while the least expensive additional direct cost was transportation. Combined, these additional direct costs impose a significant economic burden on people living with or caring for someone with EB. Angelis et al., (2016) and Flannery et al., (2020) include additional direct costs, which average €1,741, and include visits to general practitioners, social health services, transportation for healthcare, and professional care such as physiotherapy and occupational therapy.

Estimating the total direct costs for EB is difficult due to the severe lack of research, small sample sizes, and a focus on more severe forms of the disease. The most in-depth studies were carried out by Angelis et al., (2016) and Flannery et al., (2020) with the latter finding an average cost of €73,029 in Ireland. However, these studies have limitations. For example. Flannery et al., (2020) gives a detailed breakdown of direct cost by disease severity including milder forms of the disease but utilised a small sample size. Angelis et al., (2016) included a much greater sample size, but fails to give a breakdown of cost by disease severity. Future research should include as many subtypes and severities of the disease as possible with increased sample size.

3.3 Cost of illness – indirect costs

Indirect costs result from an output loss due to illness, and the informal care of persons with chronic or terminal diseases (Hanly & Sheerin, 2017). Output losses occur from a loss, or cessation of productivity due to morbidity or mortality. Productivity losses are measured in forgone wages due to the inability to engage in or maintain employment and forced early retirement due to illness (Hanly & Sheerin, 2017). Additional productivity losses result from visits by carers and patients to physicians, other healthcare professionals, and visits to hospitalised persons that lead to ancillary forgone wages. Further reductions in productivity such as fewer opportunities for promotion and educational advancement due to disease can generate supplementary indirect costs (Angelis et al., 2016). Informal care is the second form of indirect cost and is often absent from research studies or is underestimated. Van den Berg, et al., (2004) define informal care as "a nonmarket composite commodity consisting of heterogeneous parts produced by one or more members of the social environment of the care recipient as a result of the care demands of the care recipient".

Heterogeneous parts include support with mobility, housekeeping, administrative tasks, personal care, and assistance with socialising. Informal carers do not receive a full market wage for their activities (van den Berg, et al., 2004).

3.31 Indirect costs of EB

Indirect costs of EB are researched in only two relevant studies, by Angelis et al., (2016) and Mellerio et al., (2017). The lack of research makes estimating total indirect costs for EB difficult. Angelis et al., (2016) gives the most detailed account of indirect costs, while Mellerio et al., (2017) gives an estimation of informal care. The latter study provides an average estimate of €25,959.48 for time spent dressing wounds by informal carers, incorporating a variety of disease severities and subtypes.

Angelis et al., (2016) include early retirement, productivity loss, and informal care in their valuation of indirect disease cost. The study values early retirement at €1,892, productivity loss at €369, and informal care at €21,710, with a great deal of variation across the countries within the sample, and no breakdown by disease severity. Total indirect costs of EB range from €582 in Sweden to €37,971 in Spain from Angelis et al., (2016).

Table 7: Estimated economic costs of EB across countries

Year	Study	Country/ Region	Sample	Direct Costs (€)	Indirect Costs (€)	Total Costs (€)	Disease subtype(s)
2022	Authors Estimate	Ireland	-	73,029	56,992	130,021	EBS, RDEB
2020	Flannery et al	Ireland	5	73,029	-	71,421	EBS, RDEB
2020	Gorell et al	United States	249	11,040	-	11,040	EBS. RDEB, JEB
2019	García-Romero et al	Mexico	1	27,834.22	-	27,834.22	RDEB
2017	Downe	UK	1	74,702.86	-	74,702.86	RDEB
2017	Mellerio et al	UK	52	42,017.00	25,959.48	67,976	RDEB
2016	Angelis et al	Bulgaria	8	3,906	13,765	17,671	JEB, RDEB, DDEB
		France	37	8,097	6,834	14,931	EBS, JEB, RDEB
		Germany	15	11,474	34,642	46,116	EBS, JEB, RDEB
		Hungary	6	1,271	8,538	9,809	RDEB, DDEB
		Italy	35	13,122	36,111	49,233	EBS, JEB, RDEB
		Spain	54	5,166	37,971	43,137	EBS, JEB, RDEB
		Sweden	6	8,927	582	9,509	EBS
		UK	43	4,907	14,851	19,758	EBS, JEB, RDEB
2016	Joen et al	South Korea	13	6,767.71	-	6,767.71	RDEB
2014	Kirkorkian et al	United States	-	71,689.51	-	71,689.51	RDEB
2014	Pagliarello	International	-	33,120	-	33,120	–
2012	Grocott	UK	-	26,302.15	-	26,302.15	–

Note: Indirect costs include informal care costs, productivity loss, and early retirement. Direct costs include wound care costs, drug costs, cost of overnight hospitalisations, day clinic visits, GP visits, and additional primary care costs.

The papers by Hanly & Sheerin, (2017) and Angelis et al., (2016) were used to estimate Irish informal care costs per person, per year for this report. Angelis et al., (2016) used an average of 68.5 hours per week for informal care. This figure was combined with the hourly wage of €16 for informal care from the study by Hanly & Sheerin, (2017) to give an estimated Irish informal care cost of €56,992 per year. While this figure is substantial, it is most likely low due to the much greater hours of informal care needed by people with more severe forms of EB, which can be as great as 16 hours per day. The figure of €56,992 is markedly higher than the average informal care cost for dementia of €30,836, reported by Carter et al., (2019).

3.4 Economic burden of EB

EB generates a considerable economic burden for those living with the disease, their carers and society. The analysis of the sparse literature available gives an estimated total economic burden of €130,021 per person, per year. Carter et al., (2019) found a similar economic burden for dementia of €122,252 per person. The economic burden of EB presented here is significant, but the true economic burden of this disease is likely greater. Estimates for productivity loss and early retirement are quite low considering the life-long nature of the illness as opposed to a disease such as dementia.

The economic burden is shared between high direct and indirect costs, and future interventions and decisions on policy by government and other relevant authorities must take account of these findings. Further primary research into the economics of the condition, combined with a greater understanding of its impact on quality-of-life, will support more informed budgetary decisions that better allocate the State's resources to alleviate the suffering of people and their families, while reducing the strain on the State's finances.

4. Overview of access to healthcare resources and government supports

4.1 Introduction

The review in Sections 2 and 3 show the excessively high quality-of-life and economic burdens that people with EB, their carers and families endure. Inadequate access to care including bandages and dressing materials, dental care, medications, and necessary medical treatments may be a source of these burdens (Martin et al., 2019). From a policy perspective, access to health resources such as medical cards for those with rare diseases has previously been highlighted as an area of concern by Keane (2014). The recent *Model of Care for Rare Diseases* report (National Clinical Programme for Rare Diseases, 2019) also drew attention to such issues and recommended that efforts be made "to evaluate the need for discretionary decision for provision of medical cards for rare disease patients with chronic diseases to include an assessment of the burden of the medical condition" (p. 49).

Furthermore, the issue of including rare diseases in the Long-Term Illness (LTI) scheme in Ireland has also featured in the policy debate with patient groups calling for "all those living with serious and chronic rare disease(s) to be given access to the LTI" (Rare Disease Taskforce, 2020, p.12). The issue of access to medical cards was also noted with a call for "all those living with a serious and/or chronic rare disease should be given a Medical Card, without means testing" (Rare Disease Taskforce, 2020, p. 12).

In relation to EB, there is currently no evidence surrounding the differences in access to healthcare resources between Ireland and other countries. This raises the important question as to whether any differences exist. Such a review may help inform policy regarding the types of resources provided to people living with EB. In order to explore this issue, this qualitative research has three specific aims.

- **Aim 1:** liaise with DEBRA patient organisations in Spain, Australia, Ireland, the UK, and Austria to consider the **healthcare resources** available to people with EB in these countries.
- **Aim 2:** liaise with DEBRA patient organisations in Spain, Australia, Ireland, the UK, and Austria to consider **government supports and benefits** available to people with EB in these countries.
- **Aim 3:** Identify issues with access to healthcare resources and state supports that warrant further consideration in policy and practice.

The following section will clearly define the research methodology, before the key findings are summarised. The findings of our review are then described in more detail in the remaining sections. These findings are split into two broad themes: access to care and access to government supports and benefits. To help provide institutional context, the health systems of each participant country are detailed in the appendix, along with the list of participants involved in this research.

4.2 Methodology

Given the research aims described above, qualitative research methodology was utilised through a combination of questionnaire-based responses and semi-structured interviews. Given their expertise in supporting people living with EB and their interaction with the health/social system in their respective countries, the participants in this study were drawn from DEBRA family support teams from five countries: Spain, Australia, Ireland, the UK, and Austria. The participants were given a list of questions two to three days prior to the interview with a total of fifteen questions included. Nine of these focused on access to healthcare, while the remaining six focused on access to government supports (see Appendix A for the full list of questions).

The questions were initially broad and open-ended, becoming more focused on EB as the questionnaire progressed. Interviews took place over Microsoft Teams with the semi-structured nature allowing the conversation to stay relevant to the topic, but also having a degree of flexibility to the opinions of the respondents. Interviews lasted for two hours on average. Some respondents required a brief follow-up interview to clarify details on questions they may not have had answers to during the initial interview. Questionnaires were circulated after the interviews to clarify any specific points that could not be answered by participants.

To analyse the information gathered, a thematic analysis (TA) approach was employed. Such an analysis can help to provide new insights into poorly understood and complex issues, such as access to healthcare and government supports for people with rare diseases. More formally, TA is used to organise and interpret qualitative data to create a narrative understanding of the participant's subjective experiences and opinions (Sundler, et al., 2019). The primary step in TA is to read and re-read transcripts/information gathered in order to become familiar with the data, with the researcher keeping the research question at the forefront (Braun & Clarke, 2021). The next step is to search for themes within the data gathered from the interviews and questionnaires. These are then presented by referencing information gathered with a relevant narrative constructed (Braun & Clarke, 2021).

4.3 Summary of findings

As outlined above, individuals from DEBRA family support teams in five countries (Spain, Australia, Ireland, the UK, and Austria) participated in the study contributing their insights from practice regarding access to resources such as medical care, medications, bandages and dressing material, home nursing care, and medical equipment for people with EB. Here we outline an extended summary of the important themes that emerged from this process.

Access to bandages

The first theme that emerged from the interviews was access to dressing materials. DEBRA family support teams highlighted the importance of getting regular and reasonably priced bandages and dressing materials. The economic burden is simply too great for families to bear without support from government. Ireland performed poorly in access to bandaging, with no devoted scheme and many families struggling to access dressing materials. People with severe forms of the condition can generally get what they need through the Health Service Executive (HSE). However, those with moderate and milder forms of EB, and those on the margin, are often left to pay for bandaging themselves at significant expense. Without a devoted scheme, high medical costs can also prevent people from purchasing more expensive materials that may work better than cheaper alternatives.

The countries with the best access to dressing materials within our sample are Australia, the UK, and Spain. There are EB–specific government dressing schemes available in both Australia and Spain, with the Australian scheme delivering bandages to the homes of people with EB on a monthly basis. Under both schemes, people with a diagnosis of EB can access dressing materials: "*Everyone with a diagnosis of EB can get the dressings they need … it doesn't matter which type*" (Australian Respondent). In the UK, people can receive free dressing materials delivered to their homes through a private pharmacy scheme. These EB-specific schemes greatly reduce, or remove, the financial burden, but also reduce the bureaucratic stress associated with purchasing monthly dressing materials in countries with no specific scheme. While Austria has no devoted scheme, their insurance model of healthcare guarantees free or cheap access to bandaging and dressing materials for people with all severities of EB.

Access to dental care

Access to dental care also quickly emerged as a theme in this review. It was noted in multiple countries that dentists are often apprehensive about treating people with a rare condition that they are unlikely to have come across in their practice. People with EB will frequently be forced to travel long distances to receive care in specialist treatment centres, even in countries with good access to dental care. In Spain, "*some regions will cover the travel costs, others will not, and this causes inequity*" (Spanish Respondent). The UK is the only sample country to have good access to dental care for people with EB, with specialist clinics across the UK, and good access to local dentists through the National Health Service (NHS). However, it should be noted that the level of access has been declining in recent years due to private dental clinics refusing to take on additional public patients.

In the case of Australia, specialist dental care is mainly located in the states of South Australia and Victoria, while in Spain there is one dental specialist located in the city of Valencia. This distance creates a disparity in access to dental care. In Ireland, specialists are available in Dublin, while Salzburg is the site of the specialist centre in Austria. The central locations of Dublin and Salzburg reduce the disparity in care associated with distance but can still require people to undertake long return journeys or overnight stays to receive treatment. DEBRA support teams in Austria have prioritised the training of dental specialists, particularly in universities, through conferences and training seminars while there is also effort to improve training in Spain and Australia.

Access to medication and medical equipment

The third theme that emerged from the interview process was access to medication and medical equipment. Access to medication was deemed to be good across all sample countries. There was access to the necessary drugs at reasonable prices, with only a prescription fee in most sample countries and a payment ceiling in the case of Ireland and the UK. People can generally access the medication that works best for them. In Ireland, costs have been managed in recent years with families now paying a maximum of €80 per month under the Drugs Payment Scheme. Access to medicines is an issue that may emerge in the future, as new EB treatments are in development, with a number at the stage of regulatory approval and market access. Access to medical equipment is generally good across most countries, but access can depend on the severity of the disease and it can be harder to access highly specialised equipment.

Access to home nursing care

A fourth theme to emerge from the review was access to home nursing care. People with moderate and severe forms of EB need access to home nursing care but in many cases, due to a lack of resources, parents are forced to provide complex care themselves. To be effective, care must be regular and reliable which requires significant resources in the form of nurses and funding. The level of access varies across the sample countries. Access is an issue in the UK, and Austria, while Ireland, Australia and Spain have relatively good access to home nursing care. In Australia, families on the National Disability Insurance Scheme (NDIS) scheme[1] *"can access home-help nurses two to three times a week"* for moderate and severe cases, and families in rural areas "*can self-manage their [care] package*" (Australian Respondent).

While people can access a State-funded home nursing care package in Ireland, challenges remain, such as the lack of a contingency plan when nursing staff are unavailable, and a lack of nurses trained in EB to replace those who move on. In the UK, there is a high threshold to receive home nursing care leaving many along the margin without care, while in Austria there is severely limited access to home nursing care. Across all countries, families can struggle to access respite care.

Access to mental health care

Access to mental health care emerged as the fifth theme. EB greatly impacts mental health "*because EB just appears in the life of a family… and suddenly life changes*" (Australian respondent). Parents are forced to care for a condition they did not know existed prior to that moment. They are forced to inflict "*hours of severe pain*" on their children during dressing changes, which can lead to the development of a "*complex relationship*" (Australian respondent). People with EB experience pain and discomfort, and the feeling of being different, which is particularly difficult during adolescence. Combined, these factors make receiving psychological care particularly important for people with EB, but also for their carers and families.

Access to mental health care is limited across all countries. In Spain, the Government is seen as not allocating enough resources to the provision of psychological care, and access to long term care in the UK is difficult. The Irish system is characterised by long waiting times and poor coverage, with people often resorting to paying independently for mental health supports. However, in Australia the roll out of telehealth was used successfully during Covid which increased access to EB specialist counsellors. This demonstrates that barriers to accessing mental health support can be addressed through innovation.

1. The National Disability Insurance Scheme (NDIS) was introduced in 2013 and provides assistance to people with a disability via support packages that are tailored to individual needs.

Access to Physiotherapy and Occupational Therapy

Access to physiotherapy and occupational therapy emerged as another theme in this review. While some level of access is available in all countries, access to these services in the community can be difficult. In Ireland, paediatric care is available in the specialist EB clinic, but this means that families must travel to Dublin for appointments.

Access to Specialist EB Centres

The final theme to emerge was access to specialist EB centres. All countries provide care for people with EB through specialist clinics, though in larger countries such as Australia this care is not provided equitably on a national basis due to the distances involved. Such specialist clinics provide access to services such as inpatient and outpatient care, podiatrists, dermatologists, plastic surgeons, physiotherapists, and dentists.

5. Review of access to healthcare resources across countries

This section will give a more detailed comparison of the five sample countries in terms of access to care. Specifically, it will examine access to bandaging and dressing materials, dental care, medication, medical equipment, home nursing care, physiotherapy and occupational therapy, and specialist EB centres. As noted in the previous section, this is based on the insights of DEBRA family support teams in these five countries.

5.1 Access to bandages and dressing materials

Spain

People with EB in Spain have had free access to bandages and dressing materials since 2015. To access these materials, people attend a dermatologist, normally as a child, who will make a list of all supplies they will need. These materials can then be accessed locally, often from the nearest hospital pharmacy or health centre. DEBRA Spain advocated extensively for this scheme, as prior to its implementation, families were spending €1,000 to €3,000 per month on health supplies which was unsustainable. Within the current scheme, there are some challenges when trying to change to newer bandages and materials, or in accessing materials that work better for the individual.

Australia

In Australia, a federally funded National EB Dressing Scheme (NEBDS) has been in place since 2009. Under the scheme, a monthly supply of approved dressings is delivered to a person's home. People living with or caring for someone with EB only pay the cost of a prescription fee on a concessional card, which is currently $6.80 (€4.72). A dermatology review of the person's condition is required every six months or annually depending on the severity of their EB and dressing list. An extensive list of dressings is approved for subsidised supply to eligible applicants[2] under the NEBDS. People or their carers can order dressings up to the amount of their standard order, as prescribed by their dermatologist.

Ireland

Ireland has no devoted EB dressing scheme, with people receiving differing degrees of support from the State. Bandages are available through the HSE depending on the severity of the person's condition. Those with severe forms of EB can access the necessary dressing material with little difficulty, with the HSE funding the cost, which can be up to €10,000 per month per patient. People with less severe forms of EB often do not have access to bandages, seamless garments for infants and other dressing materials, and are left to fund these medical expenses themselves. If they pass a means test, they can receive some support for the necessary dressing supplies. The majority of people with EBS, for example, must pay for their own bandages and dressing materials as they do not qualify for a medical card, or their symptoms are not considered severe enough. This results in those patients on the margin falling short of the criteria necessary for full support.

UK

There is a de-facto bandage scheme specifically for EB in the UK, through Bullen Healthcare. Although it is not a government scheme, it is

2. For the purposes of the Scheme, an initial clinical diagnosis is required by an Approved Healthcare Professional with a list of Approved Healthcare Professionals available from the NEBDS Administrator. The clinical diagnosis is followed by a skin biopsy with immunofluoresecence mapping, transmission electron microscopy or genetic testing within six months of application. In the case of newborns, confirmatory diagnosis must be provided within three months of application.

funded through the NHS. The scheme is free of charge for low-income households and pensioners, or through the prescription prepayment certificate (PPC) which most people with EB qualify for. This caps expenses at £108.10 (€126.32) for bandages and dressing materials. Bullen Healthcare maintains a consistent stockpile of medical supplies that are commonly prescribed to people with EB and has a dedicated team for supporting these people. The prescription is delivered to the person's home monthly, with used packaging being removed at no additional cost. People with EB can even receive their supply of bandages in other European countries if they are travelling. All necessary bandaging supplies are granted through a prescription from the EB clinic in hospitals, or through their local GP. Other pharmacies can also offer prescription delivery services for people with EB, although the person or their carer will have to set this up with their local pharmacy if they do not wish to use the specialist service from Bullen Healthcare.

Austria

In Austria, people with EB are generally reimbursed by health insurance for a wide range of materials, although in certain cases a self-retention fee must be paid. Generally, there is not much difficulty obtaining the dressing materials needed. Prescriptions for dressing materials can be provided by a physician and obtained through a low-cost prescription fee. However, health insurance companies do not generally cover the cost of all available materials and this can become a problem in individual cases, for example, in the case of particularly difficult wounds. This may necessitate longer discussions with the insurance provider and the ambulance team of the 'EB Haus' provides support with these discussions. If it is not possible to obtain reimbursement from the health insurance company, DEBRA Austria supports the families and assumes the costs once certain criteria are fulfilled.

5.2 Access to dental care

Spain

It is difficult for people with EB in Spain to access dental care as dentists are reluctant to treat people with EB for fear of causing harm. Dentists will rarely come across the condition during their careers and, due to the severity of EB on gums and teeth, they will often choose to refer people to other healthcare professionals. There is only one dentist that specialises in EB in Spain, located in the city of Valencia. People may have to travel hours and stay overnight to receive dental care, incurring high economic costs and impacting on their quality-of-life. Dental care is available for free through the public health system, but this is only for basic dental care. Spain is divided into seventeen autonomous regions, and the region an individual lives in determines the dental care available to them. Most dental interventions tend to be covered from the ages of six to fifteen. DEBRA Spain is working to improve knowledge of the condition amongst the profession through medical seminars and training events.

Australia

Australia has two specialist dental teams in Sydney and Melbourne that people with EB can access through the public health system. DEBRA Australia is currently organising the training of a specialist team in Brisbane. It remains a challenging area, due to the size of the country. People living in New South Wales and Victoria have good access to the specialist teams, while people living in Western Australia have to fly across the country to receive specialist dental treatment. This disparity in the level of dental care is also seen in rural areas of the country, where it can take hours to reach the nearest airport. DEBRA Australia is funding training for dentists to try and improve the gap in knowledge and expertise.

Ireland

In Ireland, there are two specialist EB clinics located in Dublin, namely Children's Health Ireland at Crumlin and St. James's Hospital, that provide dental care to people with severe forms of EB. People with less severe forms of the condition can face significant issues in accessing care. People with medical cards can face long waiting times for non-specialist care, while those without medical cards must bear the financial burden of accessing private dental care.

UK

The UK has one of the best standards of dental care in the world, with every resident, in theory, having access to subsidised dental care through the NHS. A significant number of dentists in the UK will provide a mix of private and NHS services. In recent times the number of dentists catering for NHS patients has reduced. Many are closing their NHS lists to new patients or becoming completely private, with people having to pay commercial

rates to avail of their services. Free dental care is only available to a certain cohort of the population: children, students under the age of 19 who are in full-time education, pregnant women, and women who have had a baby in the last 12 months, lower income households, and those on the personal independence payment. Those who fall outside of this group will have to contribute to the cost of treatment, which is capped to maintain affordability. People with milder forms of EB can access dental care through their local dentist if they are willing to treat this condition, while people with more severe forms of EB can access dental care for free through specialist centres in London, the midlands and Scotland.

Austria

Although Austria has a very high standard of dental care, access to specialist and complex dental care is often difficult for people with EB. Most citizens have access to free basic dental care through the social security system. People can choose their own dentist once they are contracted dentists with the Austrian public health system. Complex dental treatments such as bridges, orthodontics and implants are not covered by social security, but part of the cost of these procedures may be reimbursed by health insurance. People with EB are in many cases classified by the health insurance as a "special medical case", and therefore receive a higher reimbursement for dental treatment. There are only a limited number of dentists in Austria who can treat the dental symptoms of EB. The 'EB Haus' in Salzburg, which is heavily subsidised by DEBRA Austria, collaborates with a specialist dental clinic to provide good dental care for people with EB, particularly children.

5.3 Access to medication

Spain

In Spain people with EB have access to most medications free of charge. The beneficiary of the medication and their carer receive medication free of charge, with occasional auxiliary charges for specialised medications and treatments such as ointments, but these charges are usually very low. People with EB experience hidden costs when purchasing specialised moisturisers and shampoos that are not considered medications by the healthcare system but are vital in treating EB.

Australia

Medications are provided via the Pharmaceutical Benefits Scheme (PBS) for people with EB in Australia. The scheme provides affordable access to necessary medicines, with medications being subsidised by the Federal system. People may qualify for a concessional card due to low income or age, but if not, they pay a maximum of $42.50 (€29.50) for prescriptions. If they qualify for the concessional card, they pay a maximum of $6.80 (€4.72) per item. The Government pays the remainder of the cost in both cases. There is a safety net threshold for people at $244.80 (€169.93) per year for concession card holders and $1,457.10 (€1,011.46) per year for all other patients. People with EB who have a concessional card and cross the threshold of $244.80 (€169.93) in a year will therefore pay nothing for medications for the remainder of that year. If people with EB who do not have a concessional card pass the threshold of $1,457.10 (€1,011.46) in a year, they pay the concessional card rate of $6.80 (€4.72) per item for the remainder of that year.

Ireland

In Ireland, people with a medical card pay a small fee of €1.50 per item dispensed by a pharmacist up to a maximum of €15 per month, but will only qualify for a medical card if they pass a means test, require full time care, or on the grounds of hardship. A discretionary medical card is issued on the grounds of hardship where a person has excessive medical expenses. Anyone who spends more than €80 per month on prescription medications can qualify for the Drugs Payment Scheme. This ensures that no person will pay more than €80 per month and this is not means tested. People with EB currently do not qualify to receive free medications under the long-term illness scheme (LTI).

UK

The NHS will provide all necessary medications for people with EB, with varying costs depending on the person's circumstances. Children, the elderly, and those on low incomes can receive all prescribed medications from the NHS for free. For people with EB outside of this category, prescription costs are limited to £9.35 (€10.93) per item if they do not have an NHS 'prescription passport'. The NHS prescription passport limits the price an individual will pay if they require regular prescription refills. A three-month passport will cost a person with EB £30.25 (€35.35) and saves them money if they require three or more items over

three months; while a twelve-month passport will cost a person with EB £108.10 (€126.32) and will save them money if they require more than 11 prescribed items in a year.

Austria

Medication prescribed by a doctor in Austria can be purchased at pharmacies for a prescription fee of €6.85. In many cases, doctors only have the option of prescribing certain products or generics by prescription. If special medications are needed or better tolerated, they must be purchased at the regular purchase price. There is a possibility of getting a prescription fee exemption under certain conditions and upon application. When deciding on an application, an individual's disability certificate is not taken into consideration and the decision is based on income level alone. There is also a prescription fee upper limit for people with health insurance. This means that an insured person is only required to pay a prescription fee until he or she reaches an amount equal to 2% of his or her annual net income, based on the current calendar year. After that, he or she is exempt from the prescription fee for the rest of the calendar year.

5.4 Access to medical equipment

Spain

In Spain, there are different prosthetics and orthotics catalogues where people can see the necessary steps to take to get any item contained in the catalogue, such as manual and electric wheelchairs, and upper and lower limb prostheses. They will be able to get these products for free or by paying a minimum amount of the total price, with the remainder being financed by the national health system. Anything that does not appear in the catalogue is not dispensed and people must purchase it themselves. It can be difficult to access specialised equipment such as anti-decubitus cushions.

Australia

People in Australia who require specialised wheelchairs normally qualify for support through the National Disability Insurance Scheme (NDIS). If unsuccessful in qualifying, people can access mobility equipment via a hospital programme or through DEBRA Australia funding. People who require lifting equipment would normally qualify for the NDIS. Access can vary across the different territorial health systems.

Ireland

In Ireland, access to specialised equipment is based on the severity of the disease. People with more severe forms of EB, such as RDEB, can generally access what they need for free. While there are some gaps, most equipment can be accessed through the occupational therapist as part of the EB team in the specialised clinics. People with less severe forms of the disease, who require specialised equipment for shorter periods can have trouble accessing what they need and often rely on Debra for assistance.

UK

Medical equipment, such as electric wheelchairs and bed lifts, are generally available for people with EB in the UK. However, highly specialised equipment, such as wheelchairs with rise and fall features, may not be available and DEBRA UK will subsidise the cost. People with EB mostly receive the equipment through the hospitals.

Austria

The social insurance agency will cover part of the costs for medical aids and appliances if a medical prescription is available, but there is no legal entitlement to this. Decisions are made on a case-by-case basis as to whether a medical device will be subsidised or not, and to what extent. Due to this, access to medical equipment is more difficult in Austria than access to medication. Not every medical device or aid is covered by health insurance, and sometimes only a part of the cost will be reimbursed. For example, a normal electric wheelchair is easy to obtain, but if a special version is needed, it can be more difficult to get. A person with EB or their carer will need to justify exactly why a certain model or device is required, and a doctor's recommendation is also required. This can be very time-consuming process.

5.5 Access to home nursing care

Spain

People with more severe forms of EB can access regular home nursing care for two to four times a week in Spain, funded through the Spanish National Healthcare System (SNS). Some nurses are trained to treat EB, but this varies by region, with many nurses receiving training from the parents themselves. The ability to request to have the same nurse, due to the importance of having a

relationship with this carer, also varies by region. In most cases, there will be two nurses that make repeated visits to allow some degree of trust and bonding between the nursing team and the person with EB and their family. Treating children with EB who are in intense pain, crying and sometimes screaming, takes a psychological toll on the nurses. Nurses are encouraged to take turns in treating people with EB to reduce this psychological burden.

Australia

Australian in-home EB nurses are trained to support families with care needs. Some are funded by the NDIS, while DEBRA funds those families who cannot access care through NDIS. Nurses will provide care in the home two to three times a week. Nurses are trained to care for people with EB by the hospital EB nurse or by the family. DEBRA advocates for families to help to get them on to the NDIS. Families on the NDIS can have their package managed in the following ways: (1) managed by a service provider; (2) self-managed; or (3) hybrid setup (service provider and partially self-managed).

Ireland

Home nursing care packages are usually approved by the HSE and sent out to tender with a private nursing agency who sign an agreement to provide this package of care. Currently care packages are fulfilled for those who need them, but unfortunately there is no contingency plan when nursing staff are unavailable, which can ultimately leave the patient with no care. Additionally, if a nurse leaves their employment, there are no nurses trained in EB-care to replace them immediately, leading to long periods where a person has no care, and a family member has no choice but to take on the home nursing care role. As an adult with EB gets older, home nursing care may become inadequate, as their needs become greater and often this is not recognised due to a lack of understanding/awareness of EB. A large amount of advocacy work is carried out by Debra due to this.

UK

To access home nursing care in the UK, a carers' assessment or care needs assessment is carried out by the local authority social services. A care needs assessment evaluates the needs of the person requiring care, while a carers' assessment evaluates the needs of the parents/carer. The relevant authorities decide whether home nursing care is justified. Only the most severe cases will receive regular care, such as patients with a tracheostomy who require overnight monitoring. EB nurses will make one or two visits to train parents on how to bandage correctly. The main request from parents is to get access to respite care. However, the threshold can be quite high, meaning that parents must fight to get the help they need.

Austria

In Austria, people with EB are not entitled to home care nursing under statutory social insurance, and there is unfortunately very little experience or suitably qualified staff to provide this service. If necessary, or in individual cases, the team at the EB Haus in Salzburg support the training of EB nurses. However, it is possible for a person with EB or a parent to apply for assistance with the financial expenses of home care nursing through the respective federal state. If successful in their application, a part of the home care nursing cost will then be reimbursed with a remaining fee known as 'self-retention' being financed by the person or their family.

5.6 Access to mental health care

Spain

In Spain, the public health system covers a wide range of treatments but falls short in other areas such as access to psychological care, where "*you may get access to care once a month, or once every three months*" (Spanish Respondent). Health officials and relevant stakeholders agree that access to psychological care needs to be improved, but it is a question of resources. DEBRA Spain identified improved access to psychological care as one of the most effective ways of improving the lives of people with EB and their carers.

Australia

In Australia, DEBRA has focused on training counsellors to care for people with EB so they will have a deeper understanding of the condition. Again, psychological care is believed to be neglected when it comes to funding, but there is variation across the states and territories. Consistent, long-term care is often difficult to access, especially for those in isolated rural areas. The onset of Covid caused an increase in the number of people with EB and carers wanting to access mental health care, with DEBRA improving

access through the use of telehealth, "*which worked quite well*" (Australian Respondent). This allowed people to receive care from a professional counsellor online, and was funded partly by Medicare, and partly by DEBRA Australia.

Ireland

In Ireland, accessing regular mental health care through the public health system is exceptionally difficult, with long waiting times, and low numbers of counsellors available. People with EB and their families can access a grant for 12 counselling sessions through Debra but are often left to pay independently for longer-term care.

UK

In the UK, the issue once again is resource allocation, with psychological care often being a low priority during funding decisions. Again, long-term care is difficult to access, and waiting times can be long.

Austria

Findings from Austria suggest that the barrier to access is not an issue of cost, but a cultural issue, "*with families not wanting to admit they are having troubles*" (Austrian Respondent). There is a national scheme in place and people can access care. Those who have lived with the condition for some time are less likely to access care than "*new families with young children who are more open to assistance*" (Austrian Respondent).

5.7 Access to physiotherapy and occupational therapy

Spain

The health system in Spain has a shortage of physiotherapists and occupational therapists which can make it difficult for people with EB to access regular treatment outside of the centres in Madrid and Barcelona. Furthermore, due to the regional nature of the Spanish healthcare system, with differing approaches across the territories, access to regular care can vary by region, with some people having easier access than others. Physiotherapy can be accessed both publicly and privately. It can be accessed free of charge through a medical referral, usually as a result of muscle pain caused by an accident or some specific situation, for example, for rehabilitation. Currently, EB is not a cause of special consideration to obtain this free access. There is an exception for children aged six years or younger, who can access regular physiotherapy through early care if their GP recommends it. Children aged six years or younger can also receive occupational therapy in early care centres. Normally there are no waiting times, but from six years onwards such care can be more difficult to access. While people continue to have free access, they no longer receive a full complement of care and may face long waiting times.

Australia

The level of access in Australia is similar to that in Spain. People can access therapy via the public health system to an extent, but this greatly depends on the patients' location. There is excellent access to physiotherapy and occupational therapy in New South Wales, Queensland, and Victoria. However, people with EB have poorer access in the remaining regions of the Australian Capital Territory, Tasmania, South Australia, Western Australia, and the Northern Territory.

Ireland

The Irish system is struggling to meet the needs of people with EB for physiotherapy and occupational therapy. Service provision again depends on the severity of the disease. Paediatric care is available in the specialist EB clinic, but this means that families must travel to Dublin for appointments. Adults living with EB are limited in their access to care.

UK

There is good access to occupational therapy and physiotherapy for people with EB in the UK. Hospital services provide timely care to people at no expense, with people with EB gaining access through their GP. Specialised care for EB can be accessed through one of the five EB clinics in the UK. However, community services can be difficult to access. This creates a certain disparity of care for those living in more remote areas, but this level of disparity is lower than in most of the selected countries for this study.

Austria

In Austria, access to physiotherapy depends on where the person with is living. It can be hard to find adequate physiotherapists who feel confident enough to work with someone with EB. For physiotherapy, a referral from the family doctor is needed followed by a decision from the health insurer. Part of the costs will be reimbursed with the balance depending on the respective health insurance, and the income of the family/person with EB. Again, there is some regional disparity in accessing occupational therapy due to it being harder to find a therapist in certain regions that feels confident enough to work with someone with EB patient. Part of the costs will be reimbursed with the amount depending on the respective health insurer.

5.8 Access to specialist EB centres

Spain

There are three centres in Spain, one in Madrid and two in Barcelona.

- Hospital Universitario La Paz in Madrid for paediatric and adult patients.
- Hospital San Juan de Déu, Barcelona for paediatric EB care.
- Hospital Clínic de Barcelona for adult EB care.

Specialists with experience treating EB are practicing in these centres. They include nurses, surgeons, dermatologists, oncologists, paediatricians, and physiotherapists. Specialist dental care is provided in the city of Valencia.

Australia

Australia does not have equitable care on a national basis because of the size of the country, but the situation is slowly improving. There are no medical centres specifically for people with EB, but in some states, there are EB clinics with a multidisciplinary team meeting held on a monthly to quarterly basis.

There are a total of eight states/territories in Australia, all governed by state governments.

- Victoria (VIC) and New South Wales (NSW) – EB clinics are held at a major hospital for children and adults.
- Queensland EB clinics are held at a major paediatric hospital, and they are working on providing an adult/transition EB nurse at a different hospital.
- Australia Capital Territory / Tasmania / South Australia / Western Australia / Northern Territory – there are no dedicated EB clinics.

Ireland

Specialised care for EB patients is available at two centres in Dublin.

- The Butterfly Clinic in Children's Health Ireland at Crumlin (CHI) for paediatric care.
- The EB clinic in the dermatology department in St James's Hospital for adult care.

There is a specialised multidisciplinary team and specialist nursing available at both centres. The paediatric centre in CHI also offers some home nursing visits if medically required for children with severe forms of the disease. It is important to note that there have been no adult outpatient clinics since December 2019, but a new consultant was appointed in June 2023 to fill this care gap.

UK

There are five specialist centres in the UK – four in England and one in Scotland.

- London:
  - Rare Diseases Centre, St Thomas' Hospital for adults with EB.
  - Great Ormond Street Hospital for children with EB.
- Midlands:
  - Department of Dermatology, Solihull Hospital for adults with EB.
  - Dermatology Department, Birmingham Children's Hospital for children with EB.
- Scotland:
  - Queen Elizabeth Hospital and the Royal Hospital for Children in Glasgow.

The specialist clinics provide access to inpatient and outpatient care, podiatrists, dermatologists, plastic surgeons, physiotherapists, and dentists.

Austria

Austria has one major specialist clinic in the centre of the country.

- 'EB Haus' at the University Clinic of Dermatology on the site of the General Hospital in Salzburg.

EB Haus is a major centre of expertise, with many patients travelling from surrounding countries, including Germany, Hungary, and Switzerland, to access highly specialised care at the facility. The centre has four main units: the outpatient clinic, the research centre, the academy, and the study centre. The central location of the facility gives good access to people from all regions of the country. The EB Haus is predominantly financially supported by DEBRA Austria.

6. Consideration of state benefits

The fourth broad area of the research examines access to additional financial benefits for people living with EB and their carers across the sample countries. This section examines the five participant countries in terms of the number of grants/ benefits available, but also the value of these supports. The following section reviews access to carers allowance, domiciliary allowance, disability allowance/pension, carers benefit, utility support payments, personal independence payments, education assistance, travel assistance, access to early retirement, rental assistance, nursing assistance payments, medical expenses tax grants (or equivalent), and access to mobility grants. This review is based upon information gathered from DEBRA patient organisations and research of the individual tax/benefit systems of the respective countries.

The table below compiles the results. Domiciliary Care Allowance, available in all countries apart from Australia, is given to carers who live with the person receiving care, and the payment often ends after childhood, being replaced by a disability allowance/pension/surcharge. In all countries, a disability allowance/pension is available.

Reducing working hours for carers, or leaving work temporarily, is permitted in the sample countries, with the exception of Austria. Payments made in these cases are labelled as 'carer's benefit' and ensure the security of employment and income. Utility support payments are available in four countries, in the form of grants for electricity, gas, or both. The UK has a personal independence payment for people with a disability. Education and travel assistance schemes are available in most countries. Early retirement, rental assistance, nursing home payments, and mobility grants are also included. Finally, there are two tax breaks, or equivalent, included in the table. Medical expenses tax breaks are available in all sample countries, while Ireland is the only state to include an incapacitated child tax grant of €3,300.

6.1 Comparative overview of available benefits

The aim of this section was to explore the perspectives of practitioners from five countries sampled regarding access to state benefits in order to reflect on areas where there is scope to consider more effective levels and forms of supports in Ireland.

Apart from Austria, in all countries a form of in-work allowance or carer's benefit supplements the wages of parents or carers, or in the case of the UK provides a form of financial support for carers leave. A carer's allowance is also evident in the five countries on a scale, depending on the level of care provided. Ireland and Australia provide similar rates of payment, but in Austria the level of support provided is much higher.

Domiciliary allowance is also available for parents in all countries sampled except for Australia. Spain has the lowest allowance rate set at a standard rate. However, all other countries have scales of payment allocated dependant on needs or scale of disability. It is worth noting that in the UK disabled or incapacitated persons can also avail of a grant that will allow them to purchase their own nursing care.

Other measures aimed at alleviating financial strain and promoting social inclusion are evident in this comparison. All countries except for the UK provide an allowance towards utilities. Different aspects of assistance with education are covered in Ireland, Austria and the UK, while no assistance with education is provided in Spain or Australia.

Financial relief for families and individuals with EB also takes the form of tax relief and additional financial support measures. Early retirement on health grounds is supported by all countries sampled except for Ireland. People with disabilities can access housing supports including rental support in Ireland, Australia and the UK, but not in Spain or Austria. Ireland provides a housing adaption grant which is of particular benefit to

people who may find their mobility restricted by conditions such as EB. A mobility grant is available in Austria and the UK which supports the adaption of vehicles.

The burden of medical expenses is addressed through various measures across the five countries sampled. This ranges from vat waiver on disability aids in the UK, to prescription fee waiver in Austria, and a disability income scheme in Australia, followed by a reduction in income tax in Spain.

In Ireland there is a tax relief scheme on medical expenses you pay for yourself.

The UK provides the widest range of supports as identified by stakeholders, followed by Ireland, and then Austria. Ireland performs relatively well in comparison to other countries, but the learning from the literature suggests there is much to be improved on given the quality-of-life and economic burden of EB.

Table 8: Access to government supports for people with EB across five countries

Support/Benefit	Ireland	Spain	Australia	Austria	United Kingdom
Domiciliary Allowance	€154.75 – €309.50/month	€80/month	–	€269.90 – €321/month – Increased Family Allowance	€125.66 – €806.43/month – depends on the level of care dependant needs.
Disability Allowance	€832/month	€122.83 – €737.45/month	€1,154.84/month – if criteria are met	€176.03– €968.10/month – Disability Surcharge	€312 – €393.51/month – adjustment takes place after three-month assessment increasing to between €393.51 – €600.99/month.
Personal Independence Payment	–	–	–	–	€115.91 – €743.82/month – includes the daily living component and mobility component.
Utility Support Payment	€35/month for electricity or gas. Free TV Licence.	Can get assistance with water and electricity bills.	Electricity subsidy up to €221.93/year.	Some support funds are available.	–
Education Assistance	Schools will give individual assistance. DARE Scheme for third level education.	–	–	School Travel Grant	Education authority assesses the needs of students with EB and may provide additional items such as laptops.
Travel Assistance	People can access the Free Travel Scheme if they are in receipt of Disability Allowance or Carer's Allowance. The Free Travel Scheme allows travel, free of charge, on all public transport owned by the State. Free travel is also available on some private services.	Free/low-cost public transport card.	Taxi Subsidy Scheme – subsidises half of the cost of the fare with a max payment of €20.81 per trip.	Discount tickets are offered by the Austrian Federal Railways for people with a disability. Freeway Vignette Parking Permit.	Concessionary bus travel pass gives free local bus travel for people with a disability. Disabled Rail card gives a 2/3 reduction in fare.
Carer's Benefit (% of wage/ temporary work leave payments)	Paid to people who leave work or reduce hours to care for a person full time. €900 – €1,088/month for up to 2 years. Carers Leave: keeps job secure for up to 2 years.	Reduction in working day 50% – 99% (depending on severity of dependants' illness) – State pays amount corresponding to the percentage of reduced working hours.	€188.64/month to supplement income for carers who can remain in employment.	–	Unpaid time off while leaving to care for dependant – job must remain secure while on leave.
Carer's Allowance	€896 – €1,240/month – caring for a single person.	€150 – €350/month	€1,258.04/month (max) – must care for a min of 25hours per week.	€162.50 – €1,745.10/month – depending on hourly extent of care (65hrs/month at level 1, and 180hrs/month at level 7).	€330.28/month – must care for a min of 35hours per week.
Tax Breaks & Additional Supports	Ireland	Spain	Australia	Austria	United Kingdom
Retirement/Pension	–	Early Retirement for State Pension.	Early Retirement permitted.	Early Retirement permitted.	May retire early on health grounds.
Rental Assistance	Housing Adaption Grant Rent Supplement	–	Assistance with Rent if on Disability Pension.	–	Can receive direct housing payments.
Nursing Assistance Payments	–	–	–	–	Payments to allow choice of nurse for patients.
Medical Expenses Tax Grant	You can claim tax relief on medical expenses you pay for yourself.	Reductions in Physical Income Tax.	National Disability Income Scheme	Prescription Fee Waiver.	No VAT charged on disability items.
Mobility Grant	–	–	–	Subsidy for the purchase of car if necessary – includes alterations.	Provides new vehicle with necessary alterations.
Other	Incapacitated Child Tax Grant – €3,300	–	–	–	–

6.2 Review of state benefits across countries

Spain

The allowance available in Spain is lower in absolute terms than those in other countries, but people still have good access to government support. Spain's care allowance, or Non-Professional Caregiver Benefit, has a value of between €150 and €350 per month. The payment varies depending on the severity of the disease, with three categories of disability in Spain. A Carer's Benefit also exists which enables those caring for minors affected by severe EB to reduce their working day by up to 50% without a salary reduction. The State is responsible for paying any gap in income from the reduced working hours.

Domiciliary care allowance/benefit for a dependent child with a disability is approximately €1,000/year and is not compatible with a non-contributory disability pension. The payment is valid for children until the age of 18 except for those with very severe forms of illness/disability. Spain's Disability Allowance or non-contributory pension for disability is only between €1,474/year and €8,849.40/year depending on other circumstances (for example, level of disability, help from a third person.), and can be requested from the age of 18. People with a severe disability can apply for early retirement in Spain.

People can also access free or low-cost travel cards on buses and discounts on train journeys. There are other discounts available from private entities (supply of electricity, water, transport) if the person has been granted the need for third party help (grade 1, grade 2 or grade 3). Finally, there are tax exemptions consisting of a reduction in the annual IRPF (Physical Income Tax). The amount will depend on other family circumstances (single-parent family, large family, percentage of disability). Individuals must have a disability of 33% or greater[3].

3. The percentage comes from a report carried out by an evaluation team. The Evaluation and Orientation teams decide the degree of disability, and are made up of a doctor, a psychologist, and a social worker, who carry out specific actions to establish the diagnosis, evaluation, and description of the disability. A percentage score is generated from a data gathering exercise related to evaluating the disability and the degree of it with a score of 33% necessary for the disability to be recognised as such.

Australia

The Carer's Allowance is a maximum of $2,074.04 (€1,258.04) per month, paid in two fortnightly instalments of $1,037.02 (€629.02). There is a replacement of income working on a sliding scale, with a minimum time spent caring for the person of 25 hours per week. The Disability Allowance/ Pension is $1903.90 (€1,154.84) per month, and there is no Domiciliary Care Allowance. Carer's Benefit is $311 (€188.64) per month, and this supplements income for carers who can remain in employment. Electricity subsidies are available with a value of $365.88 (€221.93) per year. While there is a free public transport scheme, the taxi subsidy scheme subsidises half of the cost of the fare with a maximum of $34.31 (€20.81) per journey. Early retirement is permitted, and people with a disability can get rental assistance. The National Disability Income Scheme is equivalent to a medical expenses tax break and funds necessary home renovations.

Ireland

Carer's Allowance is a weekly social welfare payment to people who are caring for a person who needs support because of their age, disability, or illness (including mental illness). The payment is means tested with an income disregard of €350 for a single person and €750 for a couple, and a capital disregard of €50,000. The conditions for qualifying for the payment include successfully passing a means test, being over the age of 18, providing full-time care to a person who is not living in a hospital setting, and not being in employment for more than 18.5 hours per week. The allowance is a minimum of €896, and a maximum of €1,240 per month when caring for a single person.

Domiciliary Care Allowance is not means tested and is based not on the type of disability, but rather the severity of the disease. The rate varies from €154.75 per month to €309.50 per month. Disability Allowance is a weekly allowance that can be claimed from the age of 16 and replaces the Domiciliary Care Allowance. The rate is €832 per month, with weekly payments of €208. To qualify, the claimant must have an injury, disease or physical or mental disability that has continued for at least one year or is expected to continue for at least one year, be substantially restricted because of the disability from doing work that would be suitable for a person of their age, experience, and qualifications, and pass a means test (which does not consider parents' income).

Carer's Benefit is paid to people who leave work or reduce their hours to care for a person in need of full-time care. It is not means tested, but you must have enough social insurance (PRSI) contributions and the benefit is taxable. You are entitled to receive Carer's Benefit for a maximum of 2 years (104 weeks) for each person that you are caring for, and the rate is between €900 and €1,088 per month. Carer's Leave allows you to leave your employment for up to 104 weeks to provide full-time care to a person in need of full-time care. The leave is unpaid, but your job must be kept for you to return to at the end of your carer's leave.

Finally, there are two main tax breaks that apply to carers of people with more severe forms of EB: the incapacitated child tax credit and the medical expenses tax credit. The incapacitated child tax credit has a value of €3,300 per year and involves claiming tax relief on medical expenses families have paid for themselves. People can claim tax back only if they cannot recover the expenses from any other source, for example, from a health authority. Families cannot claim tax back for amounts already received or due to be received from: a public or local authority, for example, the HSE, an insurance policy, or any other source, for example, compensation.

Austria

The Care Allowance in Austria varies significantly based on the person's need for care, measured in terms of hourly care requirements. There are seven levels of care allowance with rates varying from €162.50 to €1,745.10 per month. The Domiciliary Care Allowance or increased family allowance is granted upon application to families caring for a child with a degree of at least 50% disability or greater[4], and ranges from €269.90 to €321 per month depending on the age of the child. The Disability Allowance or disability surcharge is €176.03 per month in addition to any social welfare payment the person may be receiving.

A disabled person's card shows the degree of disability of a person and allows the individual with a disability to get free access to certain services and facilities. There are a number of travel supports available, including the free school travel grant, discounts on buses and Austrian Federal Railways, the parking permit, and the freeway vignette, which allows a person with a disability to use toll roads free of charge.

4. Similar to Spain above, this is determined from a report carried out by an evaluation team including a doctor, a psychologist, and a social worker.

People with high medication needs and low income can apply for prescription fee relief. An account of prescription fees paid is maintained for each insured person. Once the exemption is calculated in the system, it is reported to the physician via the e-card system. The physician notes the exemption on the prescription and no prescription fee is payable. Chronically ill people are exempt from prescription fees if they have an income of no more than €1,073.02 per month as a single person with increased medication needs and no more than €1,608.82 per month as a married couple with increased medication needs. These income limits are increased by €143.97 per dependent child. Finally, a mobility grant subsidises the purchase of a car, if necessary, and any required alterations.

Most people with EB in Austria are entitled to a disability certificate. This can lead to additional tax benefits in the context of "extraordinary burdens in case of disability" in the employee tax assessment. Expenses for one's own disability or that of their partner can be claimed as extraordinary burdens for tax reduction in the employee tax assessment. The individual will need proof of their disability (or the disability of their partner), either in the form of a certificate from a public health officer or by means of a disability passport. Expenses for aids that are not regularly incurred, as well as costs for doctor's treatments, can then be claimed for by the individual or their partner. Some examples of such expenses include doctor's visits, hospital stays, spa therapies and medication costs.

In addition to this, physically disabled individuals, who are unable to use public transport due to their disability and who own a motor vehicle, can claim a lump sum allowance of a maximum of €190 a month to assist with the use of their vehicle and any modifications it may require. If a physically disabled person does not have their own motor vehicle, actual costs for taxi journeys up to a maximum of €153 can be claimed per month.

UK

Carer's Allowance is not means tested and carers can avail of the allowance if they care for a person with EB for 35 hours or more a week. The allowance has a value of £69.70 (€82.57) per week or £278.80 (€330.28) per month. The Domiciliary Allowance has a value of between £24.45 (€29.00) and £156.90 (€186.10) a week and depends on the level of care the child needs. A personal independence payment assists those with long term disabilities to cover the additional costs associated with illness. There are two parts to the payment: the daily living component and

the mobility component. The value of the payment ranges from £97.80 (€115.91) to £627.60 (€743.82) per month.

Disability Allowance/Contributory Employment and Support Allowance is a premium on social welfare payments for those with disabilities that limit their ability to work. Initially, the payment will be either £61.05 (€72) or £77.00 (€90.81) per week and after a three-month assessment, the rate will be increased to £77.00 (€90.81) or £117.60 (€138.69) per week. Unpaid carers leave is available to keep employment secure. Finally, the Motability Scheme provides people with EB with a new vehicle with all necessary alterations (for example, hand pedals). To be eligible to join the Motability Scheme, a person with EB must receive one of these disability allowances: Enhanced Rate of the Mobility Component of Personal Independence Payment (PIP) or Higher Rate Mobility Component of the Disability Living Allowance (DLA).

7. Conclusion

People affected by EB face significant challenges in managing this complex condition. Our aim in this report was to undertake an in-depth literature review of the existing international evidence relating to the quality-of-life and economic burden of EB. We then presented a comparative review of the healthcare resources and government supports offered to people living with EB in Ireland, and four other developed countries.

In Section 2 we outlined the findings of the review of international evidence, considering the impact on the social, physical, and psychological variables that affect the quality-of-life for children, and adults, across the disease severity continuum. This review revealed a number of key themes. Children with EB are coping with physical pain and itchy skin, a sense of being different amongst their peers, a lack of independence due to a heavy reliance on others, challenges with daily activities and physical activity, underdeveloped coping mechanisms, and a higher rate of psychiatric symptoms (Section 2.2).

EB also greatly reduces quality-of-life for adults living with the disease, compromising daily activities, employment prospects, relationships, the pursuit of education, and home life. The independence of adults with EB can be severely constrained and many have a reliance on others to help them manage their condition and daily tasks. Adults experience similar levels of acute pain, chronic pain, and itch to children, although they are generally better equipped to deal with the pain they experience. Nonetheless they also bear the burden of a higher rate of psychiatric disorders such as depression and anxiety (Section 2.3). For both of these cohorts, the burden of pain, and the level of dependence increased due to the severity of disease associated with different subtypes, such as RDEB and JEB (Section 2.4).

In Section 2.5 we also reviewed the impact on the quality-of-life of carers and parents and considered the severity of the disease on their experience of care, their circumstances, and their lives. The burden of care for parents and carers of children with EB is substantial but varies in magnitude and intensity based on the severity of their child's illness. It can impact on employment, leisure time, and relationships. This is because caring for someone with EB is time consuming involving the co-ordination of appointments and treatments. However, the greatest impact is emotional due to the impact of watching their child endure pain, and at times having to inflict it during wound care and dressing changes. Parents and carers often experience mental strain and distress when providing care for their loved one.

In Section 3 we outlined the available evidence on the economic burden of EB, considering both the direct and indirect costs of this illness. EB generates a considerable economic burden for those living with the disease, their carers and society. In each of the papers we reviewed, significant direct healthcare costs were reported, particularly for people suffering from more severe forms of the disease. Direct costs were found to be high across all studies, with the greatest expense coming from wound and drug costs, along with hospitalisations, with wound care emerging as the most significant direct cost, followed by drug costs with variations by disease severity. Hospitalisations and medical visits represent the greatest direct cost after wound care and drug costs, subject to the same variation by severity of disease. Costs incurred also include visits to general practitioners, social health services, transportation for healthcare, and professional care such as physiotherapy and occupational therapy.

In Section 3.3 we consider the indirect costs of EB and note that these costs are incurred due to a lack of or limitation to participation in economic activity, particularly for carers who take on the burden of supporting access to treatments and 'informal' care. These indirect costs require more research so that we can understand the full impact of caring for someone with EB.

In Sections 4 and 5 we present an overview of access to healthcare resources across different countries, including access to bandages and dressing materials, dental care, medication and equipment, nursing supports and auxiliary treatments.

While our comparative review of access to healthcare resources shows that provisions are relatively comparable, gaps in services exist across all the sample countries included in this study.

While the study demonstrates a number of areas where Ireland is performing well in terms of the delivery of EB services, such as access to specialist EB centres, the study also identifies a number of gaps. These include issues around access to affordable bandaging, access to respite care, access to mental health supports, access to physiotherapy and occupational therapy in the community, and access to medical equipment.

In Section 6 we present a review of state benefits and supports, including income supplementation for carers both in work and in the home, financial supports for the care of incapacitated children, and disability payments. We also reviewed the provision of tax relief, and social inclusion supports such as educational grants, and subsidised travel. While the scope of provision may be comparable, there is room to consider the adequacy of the rate of provision relative to need across the spectrum of EB, as the discussion around the direct and indirect costs of this condition outlined in Section 3, and impact on quality-of-life in Section 2, have demonstrated.

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Appendix A: Interview questionnaire

Country 'X' questionnaire

The aim of the questionnaires is to compare and contrast the standard of care that patients with Epidermolysis Bullosa receive across five separate countries.

There are two broad themes to this, with the first being access to healthcare, and medical equipment and supplies. The second theme is access to other supports, such as monetary assistance, and assistance with education.

Theme 1: Access to healthcare, and medical equipment and supplies

Question 1: What type of health system do you have in country X?

Examples of Healthcare Models:

- The Beveridge Model: single-payer National Health Service.
  - Example Countries: UK, Spain, Cuba.
  - Description: The Government acts as the single-payer, eliminating competition in the market and generally keeping prices low. Funding health care through income taxes allows for health care to be free at the point of service – after an appointment or operation, the patient does not have to pay any out-of-pocket fees because of their contribution through taxes. Central Tenant = Healthcare is a human right.
- The Bismarck Model: social health insurance model
  - Example Countries: Germany, Belgium, Japan, Switzerland
  - Description: A more decentralized form of healthcare than the Beveridge Model. Employers and employees fund health insurance in this model – those who are employed have access to "sickness funds" created by compulsory payroll dedications. Health providers are generally private institutions, though the Social Health Insurance funds are considered public. In some countries, there is a single insurer (France, Korea); other countries may have multiple, competing insurers (Germany, Czech Republic) or multiple, non-competing insurers (Japan). Regardless of the number of insurers, the Government tightly controls prices while insurers do not make a profit. These measures allow for the Government to exercise a similar amount of control over prices for health services seen in the Beveridge model.
- The National Health Insurance Model: single-payer national health insurance
  - Example Countries: Canada, Taiwan, South Korea, Australia?
  - Description: The National Health Insurance model incorporates aspects of both the Bismarck and Beveridge models. Like the Beveridge model, the Government acts as the single payer for medical procedures, and like the Bismarck model, providers are private. The universal insurance does not make a profit or deny claims. The balance between public insurance and private practice allows hospitals to maintain independence while also reducing internal complications with insurance policies. Financial barriers to treatment are generally low, and patients usually are able to choose their healthcare providers. Like the Beveridge model, this system covers most procedures regardless of income level.
- The Out-of-Pocket Model: market-driven health care
  - Example Countries: China, the United States?
  - Description: patients must pay for their procedures out-of-pocket. Without enough money, the poor are unable to afford appropriate health care.

Question 2: Do patients have access to all necessary medical treatment, and specialist care regardless of income level?

These include:

- Occupational Therapists
- Dermatologists or Pediatric dermatologists
- Dental Care
- Psychologists

Question 3: Can patients access all necessary supplies and equipment?

These include:

- Bandages, including EB specific dressings
- Wheelchairs
- Medication
- Lifting equipment

Question 4: Do patients and their families have access to necessary home help?

Question 5: How often do they have access to this care?

Question 6: Is this home help specialized? Are nurses trained to treat EB?

Question 7: Are there clinics/medical centers specifically for EB patients?

Question 8: Are there health schemes available specifically for EB/rare disease patients? E.g. home delivery of bandages

Question 9: What are the greatest issues for patients in country X trying to access healthcare?

Theme 2: Benefits, Grants and Additional Supports

Question 10: What, if any, benefits can patients with EB receive from the state? E.g. Tax breaks, grants for home redesign, living allowance.

Question 11: Are there any specific benefits for EB patients only?

Question 12: What, if any, benefits can patients with EB receive from the State? E.g. Carers benefit, Tax breaks.

Question 13: What supports/benefits are (country X) patients and their families missing?

Question 14: Are there any other programs for EB patients and their families, such as assistance with schooling?

Appendix B: Comparing health systems

Comparing access to care for patients with EB across the selected countries requires an account of the health systems in each of the participant countries. This information will allow for a comparison of access to healthcare and government supports for EB patients, their carers, and families across countries with different healthcare systems and social support systems.

Healthcare models

The Beveridge Model

Single-payer national health service. The Government acts as the single payer, eliminating competition in the market and generally keeping prices low. Funding health care through income taxes allows for health care to be free at the point of service – after an appointment or operation, the patient does not have to pay any out-of-pocket fees because of their contribution through taxes. The central tenant is that healthcare is a human right.

The National Health Insurance Model

Single-payer national health insurance. The National Health Insurance model incorporates aspects of both the Bismarck and Beveridge models. Like the Beveridge model, the Government acts as the single payer for medical procedures, and like the Bismarck model, providers are private. The universal insurance does not make a profit or deny claims. The balance between public insurance and private practice allows hospitals to maintain independence while also reducing internal complications with insurance policies. Financial barriers to treatment are generally low, and patients usually are able to choose their healthcare providers. Like the Beveridge model, this system covers most procedures regardless of income level.

The Bismarck Model

Social health insurance model. Regardless of the number of insurers, the Government tightly controls prices while insurers do not make a profit. These measures allow for the Government to exercise a similar amount of control over prices for health services seen in the Beveridge model. A more decentralised form of healthcare than the Beveridge Model; employers and employees fund health insurance in this model.

Spain

The SNS covers most health expenses/materials for the population in Spain, with most medical treatments available free of charge through the public health system which is funded through taxation. There is a shortage of resources and manpower with high waiting times for specialists (up to six months), although the waiting times are comparatively low when compared to Ireland. The major issue is the disparity in care that patients across the 17 autonomous communities of Spain receive. The SNS is decentralised in nature and therefore regional authorities are in charge of providing care which leads to a disparity for patients due to mismanagement, or differing views on how funding should be allocated.

The United Kingdom

The National Health System (NHS) of the United Kingdom (a Beveridge Model) is free for all UK residents including ex-pats. Healthcare in the UK is directly funded through taxation rather than health insurance. Individuals can also access private healthcare if they wish to incur additional costs. The constituent countries of the UK have their own bodies, NHS England, Scotland, Wales, and Northern Ireland, with some structural differences between them. The NHS commissions primary care services, such as dentists, doctors and pharmacists, with additional responsibility for secondary care including rehabilitative care, emergency care, community health services, planned hospital care, mental health care, and disability care. Most services are provided for

free, with some minor charges for certain services including charges for dental treatment, ranging from £22.70 (€26.87) to £269.30 (€318.76), and charges for prescriptions of £9 (€10.65) per item. Low earners and individuals aged 60 or over receive exemptions are large reductions in these expenses.

Austria

The Austrian healthcare system is an example of the Bismarck health model. All citizens, residents, refugees, and citizens from the EEA have access to care. There is a catalogue of different treatments which are free under this system, with a small number of additional treatments that patients will have to pay a portion of the cost.

Australia

The National Health Insurance Model is present in Australia with single-payer national health insurance. Financial barriers to treatment are generally low, and patients are usually able to choose their healthcare providers. The system covers most procedures regardless of income level. Patients have access to necessary care in public hospitals. However, there is a huge disparity between regions due to the size of the country.

There are eight territories, each with its own separate health system. New South Wales and Victoria have the greatest access to EB-specific healthcare.

Ireland

Two-tier healthcare system: Irish citizens are technically entitled to receive public healthcare but there are significant variations in accessibility depending on where the patient lives, and their level of income. The bulk of the funding for the healthcare service comes from taxpayers, with the remaining funds coming from voluntary health insurance and additional charges. According to Burke et al, 2018, Ireland is the only western country not to have universal care, with approx. 60% of the population, who are above the income threshold, paying out of pocket to receive care. Those with the lowest income receive primary and hospital care through a medical card, but often suffer long waiting times for treatment and diagnosis. It is for this reason that the country's healthcare system is described as a two-tier system. EB patients with middle or higher incomes can be left to bear the brunt of the cost of treatment where they do not pay voluntary health insurance, or their insurance does not provide adequate care, under this system.

Appendix C: list of participants

Country	Date of Interview	Participants
Spain	Tuesday 22 June 2022	Dr Nuria Tarrats, Ms Evanina Morcillo Makow
Australia	Thursday 24 June 2022	Ms Sue McKenna
Austria	Thursday 28 July 2022	Ms Sandra Eder Ms Lena RiedlMs Sabine Wittman
United Kingdom	Tuesday 2 August 2022	Dr Helen Weaver
Ireland	Monday 20 June 2022	Ms Deirdre Callis Ms Eve O'Donnell

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