



James at DEBRA UK's Members' Weekend

A message from Ben Merrett, DEBRA UK CEO

DEBRA has worked for over 40 years giving care to the EB Community in tandem with funding research that will enable patients to have the quality of life that we all expect. Research has already passed the discovery phase identifying genes and subtypes that define this appalling condition. Across the world there are now over 20 clinical patient trials assessing improvements that cell and protein therapy, gene editing, skin transplants and other developments may make to improve wound healing, reduce blistering and itch as well as halting extreme levels of pain.

These clinical trials are being funded by commercial organisations as well as a global group of EB organisations.

Repeated intense pain, disfiguration and extensive symptoms experienced by the EB Community are not acceptable and we will not rest in our fight to beat EB.

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Ben Merrett Chief Executive Officer DEBRA UK



Foreword by Professor Jemima Mellerio, DEBRA-funded researcher

In the last four decades since the first DEBRA was founded in the UK, the tremendous headway in our understanding about epidermolysis bullosa (EB) from a basic science perspective has been paralleled by the development of over 50 national DEBRA organisations and a shared international sense of purpose towards common goals: raising awareness, improving the quality of life for individuals and families affected by EB, and funding research to find better treatments and a cure. Reflecting back, it is remarkable how much has been achieved in such a short time, how truly collaborative efforts at achieving these goals has been and how, despite being a rare condition, fundraising for EB has meant that cutting edge research is now making a tangible difference for the EB Community.

How far have we come?

It is less than 30 years since the first EB gene was discovered yet subsequent progress has been rapid, with more than 20 different genes now identified and new discoveries in this arena undoubtedly to follow. Much of the funding behind this work has come directly from DEBRA. Whilst this knowledge is essential for offering an accurate diagnosis and better prognostic information, it has also been the springboard for more recent translational research, to start to develop novel therapies in both preclinical studies and clinical trials in patients. DEBRA has been and remains the major funder of this work, supporting numerous researchers and laboratories worldwide. The grant call system managed by DEBRA International ensures that only the highest quality research is funded, and that duplication of effort is minimised at the same time as exploring a wide range of different approaches, both basic science and clinical. This support has enabled research in many different areas including gene therapy and editing approaches, cell therapies, protein replacement, and drug treatments. Whilst some projects are exploring potential cures for EB, the need for effective therapies to control symptoms, improve quality of life and prevent complications is also recognised as key and equally deserving of DEBRA funding.

An international collaborative effort

Research pursuits in EB are truly international with laboratories and clinical reference centres throughout the world working together to alleviate the suffering of people living with EB. Many of the projects that DEBRA funds bring together expertise from different centres and numerous countries, combining technical know-how, resources such as animal models of EB and patient groups for clinical trials. DEBRA-led international meetings provide a forum for scientists, clinicians, and people living with EB to come together, share knowledge, and enhance collaborative efforts in the EB arena. In supporting the development of best clinical practice guidelines for important areas of EB management and diagnostics, DEBRA combines the best evidence-based information with internationally-guided expert consensus and patient experience to raise standards across the world, in whatever healthcare setting prevails. An international registry for EB patients is another DEBRA-funded initiative offering opportunities for improved knowledge, a better understanding of epidemiology, and targeting for inclusion in future clinical trials.

DEBRA's ongoing impact

The influence that DEBRA has had in facilitating research and improving the well-being of individuals with EB over the last 40 years cannot be overstated. From basic gene discovery to cutting edge clinical trials of novel therapies, vital research into controlling symptoms, and finding better ways to manage devastating complications like cancer, the multimillion Euro investment from DEBRA to the international EB research community has been a remarkable story. It is also a paradigm for organisations supporting other rare genetic skin diseases who look to DEBRA's example to advance research in their own fields. It has been an incredible journey, but one which is not yet complete; striving to find a cure, better ways to treat symptoms and prevent complications requires further work, and ongoing support. However, with DEBRA's strength behind the international collaborative effort, the future looks assured, and progress will continue and reach fruition.

MARKEN

Professor Jemima Mellerio St John's Institute of Dermatology Guy's & St Thomas' NHS Foundation Trust London United Kingdom





Why DEBRA funds EB research

Epidermolysis bullosa (**EB**) causes constant pain due to unstoppable internal and external blistering. It can be fatal or, at the very least, can cause lifelong disability and pain.

There is currently no cure and very little public funding available – this is why DEBRA has been investing in EB research since 1981, giving hope for treatments to provide better quality of life and, ultimately, a cure(s).

1 in
227people carry a
gene that causes EB1 in
1 in
1 in
17,0001 in
5000,0001 ve births of infants born
with a form of EBPeople worldwide living
with EB

Serving the EB Community

Originally founded in the United Kingdom by a group of parents whose children suffered with the condition, the charity DEBRA has since expanded to more than 50 countries and developed into becoming **the world's leading EB patient advocacy and support network.**

Each DEBRA group functions independently but works with other DEBRA groups around the world to increase the knowledge of, and interest in, EB – including collaborations to systematically fund research into the condition. We also work in partnership with national heath authorities, who will ultimately be responsible for delivering treatments, and other EB organisations that share the goal of reducing suffering.

DEBRA is the leading funder of EB research worldwide amongst patient groups and has invested more than €54 million on 220+ research projects focusing specifically on diagnosis, cures, treatments, and management of EB.

Making a difference

Every project funded by DEBRA has a direct impact on the EB Community. The projects we fund go through a rigorous application process to ensure they are chosen based on selection criteria and weighed against potential outcomes.

Even if a project results in disproving a theory, this achievement means that researchers can shift focus and priorities to other potential breakthroughs. The more projects were able to fund, the sooner we can find a cure(s) and treatments to enhance the quality of life that we all expect. Without DEBRA's investment for more than three decades, the progression of EB knowledge and understanding would not be where it is today.

Many of the DEBRA-funded baseline projects and laboratory studies have since been developed into multi-stage clinical trials for potential therapies or have received further funding (from DEBRA, other EB groups, and commercial organisations) for additional studies.

Investment from other organisations

When DEBRA began funding research in the 1980s, the biotech and pharmaceuticals (biopharma) industry was only just starting to invest in combating diseases and understanding the human body.

Today, biopharmas are investing many millions of Euros in EB research. This surge of interest covers a broad mix of approaches to treatment and could mean a major breakthrough in the treatment and/or management of EB sometime in the next few years.

DEBRA has been responsible – through its researchers – for establishing much of what is now known about EB, providing a solid basis for the development of treatments. With more interest in EB research, DEBRA has been joined in its strategic focus by other charitable organisations (e.g. Cure EB, EBRP, EBMRF, and others), whilst still providing direct care and support to our members.

There are currently 20 clinical trials utilising original genetic understanding. Many academic funded projects are now receiving biopharma support. Dramatic discoveries have been made in bone marrow transplants, skin grafting, gene editing, and cell therapies representing groundbreaking impact.

Repurposing drugs is offering faster routes through regulatory requirements worldwide.

Progressing EB research since 1981

DEBRA continues to be the leading funder of EB research worldwide amongst patient organisations, and our first project was funded in 1981.

Investing in projects focused on specific types of EB or symptoms of the condition to projects evaluating care techniques and technologies, DEBRA funds based on the needs and demands prioritised by the EB Community. We have supported many discoveries in EB, which have then led onto further investigations and trials.

As new technologies and discoveries are made, DEBRA continues to invest more and more into EB research, as can be seen with DEBRA's increased funding over the last decade and which has also led to many millions of Euros of investment by the biopharma industry in the last few years.

Our commitment so far



International research collaboration

Promotional photo of James and Mum, Lesley, for the #FightEB campaign

Our research strategy

EB research is complex. It is important that DEBRA invests strategically in the priorities identified by the EB Community and maintains a balance of research aimed at short-term benefits for patients along with support for long-term research into a potential cure(s).

Research has identified up to 20 genes and more than 30 subtypes with four main groupings of junctional, dystrophic, simplex, and Kindler. Our assessment is that each subtype of EB will need different treatments and a range of therapies will be required to help each patient.

Key aims of DEBRA research

1.

Improve our understanding of the biology and genetics of all forms of EB as better understanding can lead to new approaches to diagnose and treat EB. 2.

Work towards the development of therapies (including possible gene, cell, drug, or protein therapies) for **possible treatment(s)** of EB.



Understand the nature of wound healing and the development of skin cancer in EB, and seek to develop better treatments and **prevention strategies** of these symptoms.

4.

Support clinical-care research to improve the management of EB through symptom relief.



How we select projects

To help advise which projects DEBRA should fund, we have formed a medical and scientific advisory panel (**MSAP**) made up of senior EB researchers and clinicians who, jointly, reflect the breadth of EB research.

It is the MSAP's role to oversee DEBRA's centralised peer-review process of all research grant applications and to advise on research grant progress. Additionally, they ensure scientific quality, innovation, feasibility, value for money, as well as potential benefit to people living with EB.

In addition, we have in recent years run expert panel roundtable meetings to identify research priorities and draw global experts into the field of EB.





The role of the DEBRA International MSAP

It is the role of the DEBRA International MSAP, a group of EB dermatologists and pure scientists from many disciplines, to consider research applications from both scientific and medical standpoints.

The concept of peer review of applications, particularly in scientific realms, has been well established for many years. This process ensures feedback is provided by experts working in that field.

It is important this process is rigorous, fair, and meets the highest standards, not to advise on funding but to ensure scientific rigour and suitability from the applications.

The ability to see the wider picture has always been to the fore, aiming to change the lives of people with EB and lead to better understanding of the condition.

Professor Adrian Heagerty MSAP, Acting Chair

Research that directly impacts the EB Community

Fazeel campaigning to #FightEB at a DEBRA UK Members' Weekend

Our ongoing projects



YEAR STARTED	INSTITUTION(S)	PROJECT	RESEARCHER(S)	AMOUNT AWARDED
2013	St Thomas' Hospital, UK University of Dundee, UK	Natural History and Clinical Endpoints Study in EB	Professor Jemima Mellerio Doctor Anna Martinez Mrs Elizabeth Pillay	€487,203
2015	Guy's Hospital, UK	A prospective phase I study of lentiviral- mediated COL7A1 gene-corrected autologous fibroblast therapy in adults with RDEB (LENTICOL-F)	Professor John McGrath Professor Jemima Mellerio Doctor Waseem Qasim Doctor Wei-Li Di Professor Adrian Fletcher	€629,143
2015	University of Dundee, UK	Development of Novel Gene Technology for Treating EBS	Doctor Peter van den Akker (<i>DEBRA Clinical Research</i> <i>Fellow</i>) Doctor Aileen Sandilands	€473,744
2016	University of Freiburg, DE	REFLECT (symptom-RElieF with Losartan – EB Clinical Trial): A dual- center prospective phase II trial to establish safety, tolerability and efficacy of losartan in children with RDEB	Professor Leena Bruckner Tuderman	€498,670
2016	Stanford University, USA	CRISPR-mediated therapy of DEB	Doctor Marius Wernig	€287,925
2017	Stanford University, USA	A phase 2 clinical trial of type VII collagen gene therapy for RDEB	Professor Peter Marinkovich	€433,112
2017	University of Dundee, UK	EBS Genotyping database	Professor WH Irwin McLean	€28,560
2017	Netherlands Cancer Institute, NL	High-content screening for new therapies for associated with Muscular Dystrophy (EBS-MD)	Professor Arnoud Sonnenberg	€125,875
2017	Thomas Jefferson University, USA University Hospital Salzburg, AT St Thomas' Hospital, UK	"First in EB" Phase II trial of Rigosertib for RDEB SCC	Doctor Andrew South Professor Johann Bauer Professor Jemima Mellerio	€502,058

YEAR STARTED	INSTITUTION(S)	PROJECT	RESEARCHER(S)	AMOUNT AWARDED
2018	Istituto Dermopatico dell'Immacolata, IT	Anti-Fibrotic Therapeutic potential of Histone Deacetylase Inhibitors (HDACi) for RDEB	Doctor Daniele Castiglia	€160,000
2018	University of Birmingham, UK	Characterisation of the skin microbiome and investigation of neutrophil function in EB patients	Professor Iain Chapple Doctor Sarah Kuehne Doctor Josefine Hirschfeld Doctor Melissa M Grant Professor Adrian Heagerty	€334,807
2018	University Hospital of Cologne, DE	Exploring innate immunity in wound healing complications in RDEB patients	Professor Sabine Eming Doctor Dimitra Kiritsi Doctor Alexander Nyström	€194,500
2018	Association pour le Développement des Sciences Médicales, FR	Targeting the tumor niche in EBD skin cancer; Molecular and cellular mechanisms of skin cancer expansion govern by the extracellular matrix in EBD skin cancer	Doctor Cedric Gaggioli	€64,700
2018	Université Paris Descartes - Sorbonne Paris Cité, FR	CRISPR/Cas9-based editing to treat RDEB	Professor Alain Hovnanian	€132,600
2018	Universitair Medisch Centrum Groningen, NL	Substantiating the effectiveness of a sublingual phyto-cannabinoid based oil for the treatment of pain and pruritus (itch) in EB	Professor Marcel Jonkman Mister Nicholas Schrader Professor André P. Wolff	€177,200
2018	University of Leipzig, DE	A compound-based therapy approach for EBS	Professor Thomas Magin	€253,100
2018	Guy's Hospital, UK	Evaluation of the early efficacy of intravenously administered allogeneic mesenchymal stromal cells on itching in adults with EB Pruriginosa	Professor John McGrath Professor Jemima Mellerio	€537,149
2018	University of Freiburg, DE	Understanding the modifiers of growth in generalised JEB and DEB	Doctor Antonia Reimer	€90,600
2019	University of Colorado, USA	Optimizing gene editing strategies for RDEB	Professor Ganna Bilousova	€289,575
2019	Pontificia Universidad Católica de Chile, CL	Neutrophic factors to prevent the development of neuropathic pain in RDEB	Professor Margarita Calvo	€92,480
2019	University of Glasgow, UK	Mechanisms of TGF-beta mediated tumour promotion in RDEB cSCC	Professor Gareth Inman Doctor Peter Bailey Doctor Karen Blythe Doctor Andrew South	€90,405
2019	King's College Hospital, UK	Spray-on gene therapy for RDEB: Preclinical studies of lentiviral-mediated COL7A1-supplemented epidermal stem cell and CD39 + CD26 - fibroblast spray-on therapy	Doctor Su Lwin	€193,165
2019	Thomas Jefferson University, USA	Implementation of non-invasive, next generation sequencing-based early prenatal diagnosis for EB	Professor Jouni Uitto	€155,441
2019	University of New South Wales, AU	Establishment of a non-invasive protocol for early detection of malignant carcinoma in RDEB	Associate Professor Albert Mellick	€25,200
2019	University of South Australia, AU	Safe and Intelligent Dressings for the Treatment of Infected Blister Wounds	Doctor Zlatko Kopecki	€25,200
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¹ Includes a small amount of designated funds for joint projects with other EB organisations; all amounts not originating in EUR have been estimated with spot exchange rates based on year averages, commitment dates, or project start dates.

Projects leading to advances In treatments

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Evaluating the long-term impact of RDEB

A team of DEBRA-funded researchers across England is evaluating the long-term impact of RDEB in efforts to better understand the progression of the condition. The project, led by Professor Jemima Mellerio in London, is helping to identify and define relevant clinical endpoints that could be measurable target outcomes for EB clinical trials.

Clinical data are now being collected in a consistent way and the information stored on an electronic database to monitor and review many aspects of an RDEB patient's life: demographic details, growth and development information, clinical treatments and therapies, and more.

In obtaining these data, this study will ultimately help identify meaningful endpoints to form future clinical trials needed for all types of EB.

€487,203

Understanding how EB impacts development

EB affects more than just the skin. In many cases EB manifests in other organs, which can lead to poor growth and anaemia, meaning EB patients do not always reach certain development milestones (e.g. puberty).

A study led by Doctor Antonia Reimer at the University of Freiburg is looking at the long-term outcome ("natural history") of EB – particularly within JEB and DEB – to determine if poor growth in children is due to the underlying gene or a variety of factors.

Gaining a better understanding of how EB impacts development will help clinicians develop new treatment therapies that could improve a patient's quality of life.

€90,600

Developing a 'smart dressing' for wounds

A groundbreaking study in 2011 by Dr. Wenxin Wang at the National University of Ireland has led to further research and potential development of an advanced 'smart dressing' by a leading pharmaceutical company for people living with RDEB.

The first of its kind, the dressing would not only provide closure and cover for wounds but also release the critical collagen gene (COL7A1) absent in people living with RDEB. Starting as a liquid at room temperature, the solution would be applied and turn into a gel to cover and seal the wound, which could then be covered by bandages and wrapped.

This innovative hydrogel could be applied to wounds of all sizes and for sensitive applications and has the potential to be used as an effective therapy for EB.

€71,056









Reducing suffering from pain and itch

Cannabinoid based medicines (CBM) have been studied for many years. DEBRA is currently funding a study using CBMs, which aims to gain insight in addressing the widespread EB-related symptoms of pain and itch.

This project is based in the Netherlands and led by Mr. Nicholas Schräder and Professor André P. Wolff. It is hoped this study will help determine if CBMs could be a potential treatment to help alleviate or reduce pain and itch for those living with EB.

There is now significant investment by biopharmas in CBM research in EB, including the development of clinical trials that could lead to new treatments for the condition.

€177,200 *Professor Marcel Jonkman sadly passed away mid-study



Limiting excess scarring

Understanding ways to repurpose an existing drug to help treat symptoms of EB – specifically scarring – is the central focus of several studies by Professor Leena Bruckner-Tuderman (University of Freiburg in Germany), one project of which continues investigations from a previous DEBRA-funded project in evaluating the use of the drug Losartan.

Losartan, and other similar agents already used in treatment or in development for other conditions, has been known to prevent excess scarring and fibrosis as a secondary effect.

If found to have the same results in humans as in the murine model, the agents may have the potential to reduce inflammation and help limit the growth of mitten deformities and joint stiffness. This area of medicine is being further explored by the biopharma industry.

€498,670



Stopping cancer from spreading

Following on from a previous DEBRA-funded study, Dr. Andrew South (United States) and Professors Johann Bauer (Austria) and Jemima Mellerio (United Kingdom) began a Phase II trial to test the safety of Rigosertib, a drug which has the potential to stop the growth of EB-related skin cancer – one of the current research priorities set by the EB Community.

Rigosertib also has wider clinical reach and is being used in a number of trials for other cancers. The purpose of this study in EB is to test if the drug can be tolerated by EB patients and administered without significant impact to daily routines.

Researchers are working alongside the biopharma industry to see if this drug could potentially alter the course of treatment and prognosis of EB patients diagnosed with skin cancer.

€578,117

Manipulating molecules to treat EBS

Following on from a previous DEBRA grant, Professors WH Irwin McLean (University of Dundee) and Adrian Heagerty (University of Birmingham) have investigated ways to change the DNA and RNA (the body's genetic code and method of passing on good genes and mutated genes) in cells.

Knowledge from this study is now being applied by a biopharma to create a potential treatment, which could lead to a potential cure for EBS and other forms of EB

€227,509





Learning from twins with EB

Doctors Giovanna Zambruno, Daniele Castiglia, and Teresa Odorisio at the Istituto Dermopatico dell'Immacolata in Italy aimed to determine why a set of identical twins, both living with RDEB, had different levels of severity and symptoms of the condition. They suspected that additional genes might be a factor in how RDEB presents itself in an individual.

Through their research, data indicated that a specific protein (TGF-beta) might play an important role. They were also able to identify that decorin (DCN) protein plays a role in adjusting TGF-beta levels.

This project has led to ongoing biopharma research on both proteins in hopes of learning more about EB and potential treatments.

€102,703

Growing new skin as a treatment option

A group of researchers in Austria, Italy, and Germany (including Professors Johann Bauer and Michele De Luca) have collaborated on a number of projects in which they have developed a new method of growing human skin and established it could be a safe and efficient way to treat JEB patients.

The knowledge gained from these projects has helped to treat a child in Germany suffering from JEB (transplanting 80% of the total body surface).

Similar trials using gene edited skin are happening across the United States of America and similar successes could mean a major change in EB treatments, which has caught the attention of biopharmas.

€260,000





Assessing and improving wound healing in skin grafts

Following a successful Phase I study, Professor Peter Marinkovich in the United States of America is evaluating the long-term effectiveness and durability of wound healing in skin grafts in RDEB patients.

The project focuses on using a targeted gene therapy approach in skin graft treatment, and the team is collaborating with a biopharma organisation to further develop a large-scale clinical trial.

If success continues, the outcomes could ultimately transform patient care and the approach to treatment for people living with RDEB.

€433,112



Advancing research in gene therapy

DEBRA-funded researchers across France, Germany, Switzerland, and the United Kingdom collaborated in multiple preclinical studies investigating the use of gene therapy (introducing normal genes into a patient's cells to replace or correct the genes affected by EB) as a treatment option for patients with RDEB.

Their research has led to a breakthrough demonstrating the success of gene grafting as a technique that could be applied as a treatment option for people living with RDEB.

In addition, these projects have led to further research (including human trials) in this area, receiving a nearly €5 million European Commission contribution, and helped to establish a gene therapy programme.

€703,323



Progressing a potential breakthrough with academic and commercial partnerships

In 2010, DEBRA funded projects with Professor John McGrath (Guy's Hospital) and Doctor Gabriela Petrof (King's College Hospital) investigating the first use and understanding of fibroblasts – a cell in the body that produces collagen – injections as a method of treatment for RDEB patients.

Their investigations resulted in better understanding of the cells, as well as determining fibroblast injections could help speed up the wound healing process. Further studies have shown repeated success and a leading biopharma is trialling treatments with gene-edited fibroblasts.

If success rates continue, this breakthrough could help form a range of treatment options for patients suffering from RDEB.

€226,577



Enhancing quality of life

There are key regular procedures (e.g. throat dilation, gastrostomy and cancer surgeries, dental treatments, dietary advice, and others) currently available to help improve the quality of life for people living with EB.

In addition, as technology is advancing, more and more products and tools are also being developed to help manage symptoms caused by EB.

DEBRA works with its members and major manufacturers to develop innovative products that could have a lasting impact for the international EB Community, and it is hoped that these products could greatly improve the quality of life for people suffering from EB.



Restoring a sense of 'normality'

When patients are unable to use their muscles on a regular basis, the muscles become weak, which is called muscle atrophy. EB patients can develop muscle atrophy if they are unable to use their hands due to severe blistering.

Linking with the local EB Community and working with DEBRA Chile, students at the Universidad del Desarrollo were able to design a glove prototype with a system of magnets and a metal plate that enables users to perform routine tasks such as eating, brushing teeth, writing and drawing, and holding the phone.

The utility of the design means users can fasten magnetic objects (up to 2.2 pounds in weight) in a variety of ways for ease of use. The magnetic glove is now available to patients worldwide and is used in 21 countries.



Pain-reducing dressing adhesive

Blisters and wounds are a common symptom amongst all forms of EB. Working with DEBRA members, a leading dressings manufacturer was able to develop an adhesive material that reduced the amount of pain and stress associated with dressing changes.

This silicone-based adhesion is purposefully designed to help improve the healing process through not further damaging the skin whilst bandages are being removed, as well as protecting new tissue and intact skin to support natural healing.

This type of adhesion is now available on multiple dressing products and there have been numerous studies confirming its effectiveness.



Protecting EB patients with new innovations

Researchers at King's College London have collaborated with clinicians, engineers, a knitwear manufacturer, digital data capture company, and EB patients and parents to develop a unique hand therapy device.

This prototype was specially designed for EB patients to protect against blisters, reduce the number of dressing changes, as well as promote better hand hygiene.

This DEBRA-funded study has laid the foundation for further research to design and develop additional hand therapy devices, as well as helped in the development of the DEBRA-funded Hand Surgery and Rehabilitation Therapy Clinical Practice Guideline for people living with EB.

Leading EB centres around the world



UK diagnostics centre instrumental in prenatal diagnosis of EB

Diagnosis of EB, not possible 40 years ago, is vital in understanding how to treat the condition.

DEBRA funding helped establish the UK-based National Diagnostic Epidermolysis Bullosa Laboratory at St Thomas' Hospital in London.

This centre provides skin biopsy and gene mutation analysis for people living with EB (and other related skin conditions) and is credited with developing the first DNA-based prenatal diagnosis of EB. This development, along with further progress of IVF techniques for people living with EB, helps ensure parental choice.

This centre has been a model for similar diagnostic facilities now open in other countries.



EB Haus providing specialist EB care, research, and training

Established in 2005, the EB Haus is a unique, purpose-built facility that has become a central hub of activities for the EB Community in Austria. Founded by DEBRA and affiliated with the General Hospital Salzburg, the EB Haus combines care, research, and training to improve the quality of life for people living with EB.

The collaborations taking place at the EB Haus allow doctors and therapists to offer the best possible medical treatments, an opportunity for researchers to develop techniques and liaise directly with both patients and clinicians, as well as provide training and education about EB.

This model is a unique, all-in-one offering that has the potential to be repeated in other countries to improve quality of care, as well as advance research and understanding of EB.



Rare Diseases Centre transforming patient care

The development of the Rare Diseases Centre at St Thomas' Hospital has transformed patient care for people living with EB.

Patients can now be seen in the same facility by a variety of clinical specialists all on the same day – a welcomed change by members of the EB Community.

The centre also houses facilities for the clinical team to work more closely with the patient's local health services and to share experiences with colleagues around the world.

To make this development possible, DEBRA invested more than €280,000 into the project – the single largest external injection of funds.

Investing in the future

DEBRA UK Member, Freddie, enjoying a day out

Biopharma investment in EB research

The biopharma industry is investing many millions of Euros into EB research. With more than 20 clinical trials, over 15 biopharmas actively engaged, and around five EB organisations regularly funding new projects, we are far more confident than ever before that we will beat EB.

The global bioparma companies working in this area are exploring a whole range of treatment options (including drugs, infusions, injections, skin grafts, and topical treatments) using some of the most modern technologies (e.g. gene editing).

Three of the more studied areas for clinical trials are drug and cell therapies, as well as topical treatments. This focus could be due to their less invasive approach for patients and ease of access – often existing formulas and approved products in the market being repurposed to treat symptoms of EB.

Crucially, biopharmas are helping to identify measureable endpoints and assess the enormous cost that EB inflicts on individuals, families, and everyone impacted by the condition.

Advancing research

With major commercial organisations taking interest in EB research, these companies are supportive of DEBRAfunded projects, as they help to evaluate the risks in further studies beyond the preclinical stage.

Many of the clinical trials taking place today can be linked back directly to the initial studies funded by DEBRA. Our projects cover a variety of research areas and key learning from these projects helps shape the direction of EB research. It is from this learning that biopharmas can form their investment strategy of clinical trials aimed at further investigations and potential discoveries.



Diacerein ointment reduces blister number in EBS patients

Patients suffering with EBS may have mutations in one of two genes that are responsible for the stability within skin cells, which can cause skin blisters. Once blisters form, the body may trigger an inflammatory response making the skin even more sensitive and prone to blistering.

Researchers in Austria, led by Doctor Verena Wally at EB Haus, formulated an ointment containing the anti-inflammatory drug diacerein to help treat patients suffering from EBS. In the patient clinical trial, the ointment showed promising results in reducing blisters and no side effects that were assumed to be related to the ointment were observed.

Further investigations for this project have been picked up by Castle Creek Pharmaceuticals with hopes of bringing the product to market.

DEBRA invests in a future free of EB

Isla and Andy taking a break at a DEBRA UK Members' event

Other ways we support the EB Community

DEBRA supports the entire EB Community – from providing care and support to those living with the condition to funding groundbreaking research and offering training grants to EB professionals. Until there is a cure(s), DEBRA continues to work towards a future free of EB.

Visit our website to learn more about all the ways in which DEBRA supports the EB Community and how you or your organisation can get involved and make a difference.

www.debra-international.org



Advocating for the EB Community

Prioritising research

Through the development of the Research Involvement Network (RIN), DEBRA works directly with the international EB Community to ensure that "lived experience" of patients and family members is taken into consideration in the process of progressing EB research.

A survey is shared amongst the international EB Community every three to four years. We continuously engage with our international EB community to understand their research priorities, which are currently itch, cancer, and wound healing.

Making change happen

In 2018, DEBRA of America held a meeting with the US Food and Drug Administration (FDA).

Both patients and clinical experts attended, and several members spoke in efforts to educate the regulators about daily life with EB and the importance of effective treatments.

This historical event, a first ever in EB, provided valuable insights into the symptoms, effects, and real-life impact of the condition that could help guide the FDA in determining the regulatory pathway of all EB research and therapy development.

The FDA has international recognition and it is hoped that this will also pave the way for potential treatments to be approved and made accessible in other countries around the world.



Pictured: Scott, DEBRA UK Member, speaking at the FDA meeting

Developing EB clinical practice guidelines

Clinical practice guidelines (CPGs) are a set of recommendations for clinical care based on evidence from medical science and, where no formal evidence exists, expert opinion.

DEBRA is one of the few patient organisations in the world leading the development of CPGs. CPGs can help shape future research and, vice versa, the CPG priorities might be reset based on groundbreaking research.

Until there is a cure, there is probably no greater way to positively impact the lives of people living with EB. For more information about CPGs, download your copy of our CPG Fact Sheet:

www.debra-international.org/clinical-guidelines

Initiating a global EB patient registry

Both the scientific and medical communities recognise the need for a global EB patient registry in order to better understand trends and commonalities amongst patients living with the condition.

DEBRA has pledged more than €140,000 to develop and maintain an international EB registry, which is aimed to be a network of national EB registries that can share and report data on the condition.

This registry will allow EB professionals to:



Bringing together the world's leading experts in EB

EB World Congress is an independent, global congress on EB bringing together the world's leading experts in EB research, clinical management, and the EB Community. This is a culmination of DEBRA International organising research conferences over the last 20 years.

This pooling of state-of-the-art knowledge across the key stakeholders not only gives guidance to the professionals involved in EB, but also helps in the development of strategies for research, regulatory, funding, and healthcare communities.

Please visit www.ebworldcongress.org for more information.





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