

GET INVOLVED IN RESEARCH

I would like to introduce myself as the new Research Manager of DEBRA Ireland. I am very excited to be part of the DEBRA Ireland team and the wider DEBRA network. With my background in chemistry and skin research, I hope to make a valuable contribution to DEBRA and will work hard to find key research that will meet your needs. I have joined the organisation at a good time where the world of EB research is expanding significantly and many small wins are being made in the journey for a treatment and cure. Research is being performed in many areas, from investigating wound healing and pain management to finding and fixing genes and proteins that affect EB. I am very much looking forward to getting involved in the research into treatments for EB, meeting you to understand your needs in relation to research and will continuously keep you updated on developments through the DEBRA Ireland website and Research Newsletter.



Dr. Sinéad Hickey (sinead.hickey@debraireland.org; 014126924)
Research Manager



HAVE YOUR SAY

We encourage you to get involved in research into EB. Your help is vital in developing treatments for all forms of EB. You can get involved in many ways including:

Facebook - [DEBRA International Research Involvement Network](#)

- ✓ Targeted at **people with EB** who want to learn more and get involved in EB research.
- ✓ Participate in assessments and questionnaires.
- ✓ Get up to date information on current research.

Contact me - Sinead.hickey@debraireland.org

- ✓ What would you like to see in our research newsletter?
- ✓ What would you like to see on our website?
- ✓ What type of research would you like to see investment in?

Trials and studies - see information on this page

Outreach Nurse Study

As you know, having a nurse on the road was a big focus for DEBRA Ireland and now that Suzi is in place, we need to make sure we get it right. The only way we can know is getting feedback from EB families and EB medical professionals. Before Christmas, you got some information as to how to take part to help create the best EB outreach nursing programme for your family and all families living with EB.

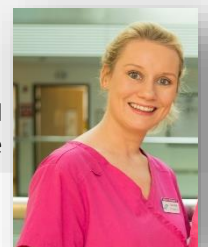
The kind of things we need to know are:

- What you think you need most from the EB outreach nurse role?
- How does that compare with what you do get?
- What would you like to see?

Your opinion is really important to us, so if you need any more information, please contact me.



Suzi O'Neill
EB Outreach Nurse



Clinical Trials

We encourage you to become familiar with the clinical trials that are currently being recruited for. Examples of current trials include:

- Castle Creek Pharma. Phase III Clinical Trial to test the safety and effectiveness of an ointment which contains a medicine called diacerein – Location St Thomas's Hospital, London (travel expenses from Ireland covered)
- Amryt Pharma EASE Phase III Clinical Trial to study the safety and effectiveness of a gel called Oleogel-S10 in EB. Location – St James Hospital; OLHSC Crumlin



In addition to these trials many other companies such as Abeona Therapeutics and Krystal Biotech are involved in research into various treatments for EB. More information on clinical trials can be found on the DEBRA International Research Involvement Network, DEBRA websites or through your consultant.*

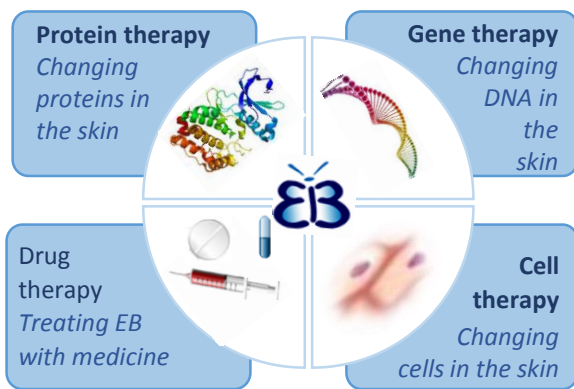
RESEARCH into EB

Research into EB happens in many different ways, for example, investigation into:

- ❖ Causes of EB and why different patients suffer in different ways.
- ❖ Treatments for the various types of EB.

Research starts in the lab using test tubes and then progresses in different stages until a treatment is ready to be trialled in patients. Some treatments are very specific at targeting certain DNA and therefore can only be trialled on a small number of patients, others are more general and can be trialled in a larger group.

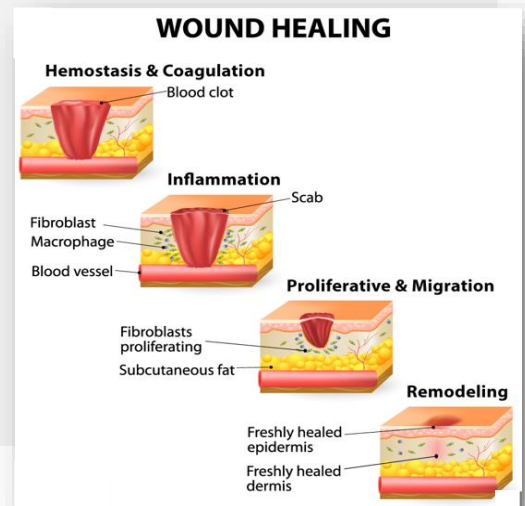
Treatment types



An example of a recent breakthrough in EB treatment is the work of De Luca and his team of scientists who made a fully functional skin for a child suffering from Junctional Epidermolysis Bullosa. More information on this breakthrough is attached.

Exciting New Project

DEBRA Ireland is entering into an exciting new project investigating the role of certain components of the skin in wound healing and why they act differently in the skin of those who suffer from EB compared with those that don't. Professor Sabine Eming of the University of Cologne, Germany will undertake the research over the next 3 years. Professor Eming is a well-known scientist working in the area of wound healing. DEBRA Ireland is delighted to fund this project along with DEBRA UK and hopes it will yield promising results in the understanding of wound healing. We will keep you updated on the progress of this projects and others DEBRA Ireland will be involved in through the DEBRA Ireland website and Research Newsletters.



EB Researchers Break New Ground With Skin Graft Trial

An international team of researchers has broken new ground with a pioneering skin grafting procedure, reconstructing a fully functional outer skin – or epidermis – for a child suffering from a severe, and often fatal, form of Epidermolysis Bullosa (EB) – a genetic skin condition that causes constant pain due to unstoppable internal and external blistering.

Researcher Michele De Luca and colleagues reconstructed skin covering approximately 80% of the total body surface area for a seven-year-old child suffering from Junctional EB. Junctional EB is caused by mutations (or mistakes) in the genetic code that creates the protein *laminin-332*, which binds the layers of the skin together. This is the first time grafting such a large area of fully functioning skin on a person suffering from EB has been trialed.

Junctional EB has a severe negative impact on the quality of life. Forty percent of people suffering with Junctional EB die before adolescence. Those who survive develop chronic wounds, infections and tissue damage. People suffering from the condition also face an extremely high risk of developing an aggressive form of skin cancer.

During this experimental treatment, skin cells were taken from a non-blistering area on the patient's body, genetically modified to contain the corrected form of *LAMB3* – one of the genes responsible for the creation of the protein *laminin-332*. The corrected skin cells were then grown into grafts in the laboratory and transplanted back onto the patient in three separate procedures. Crucially, during the 21 month period following the study, the transplanted skin remained robust and did not suffer from blistering during this time.



Earlier research has shown that transplantation of gene corrected skin cells could create functional skin on a smaller scale. This new research confirms that the transplanted skin is being maintained by specific groups of epidermal stem cells, which are capable of renewing both in the laboratory and in the person's skin without apparent deterioration.

