



Cure EB[®]

ACCELERATING RESEARCH TO END PAINFUL SKIN

SPRING 2022



LATEST UPDATES

RECENT EVENTS

10 YEARS FUNDING RESEARCH

PATRONS DAMIAN LEWIS SEAN BEAN DANIELLE DE NIESE SIR JAMES & LADY DYSON

contact@cure-eb.org 020 3405 8433 www.cure-eb.org f CureEB t @cureEBorg i @cureEB
175-185 Grays Inn Road London WC1X 8UE CureEB is a charity registered in England and Wales 1158672

THANK YOU FOR YOUR HELP

2022

We don't want to exist in 10 more years

It is some time since we ran a print version of the newsletter! Who would have thought that the pandemic would still be with us. We hope that you and your families are well, though I fear many of you will have been touched by it.

In amongst the pandemic years we realised that we have now been fundraising for research for 10 years and it is perhaps time to reflect on what has been achieved with your generosity.

Cure EB (formerly Sohana Research Fund) funded the EBSTEM and ADSTEM clinical trials, mesenchymal stem cells infusions for people with recessive dystrophic epidermolysis bullosa. These trials are the foundation for Mission EB which will see every child under 16 with RDEB being offered infusions of stem cells. It is a pilot project which is testing getting treatments into the rare disease population quickly. The NIHR are putting

in £4.5 million and if successful the treatment will be commissioned by the NHS.

**Cure EB made this happen.
You made this happen.**

What of the future?

Cure EB is focussed on gene editing and gene modification, as well as research to combat the malignant skin cancer that often develops in sufferers of EB. Gene therapies have moved from the realms of science fiction to real trials with exciting results offering proof of potential in EB and many other conditions.

We can see identifiable routes to whole body treatments but if anything the pandemic has taught us what we can achieve with the right level of funding. Treatments and cures are realistic goals now for genetic conditions.

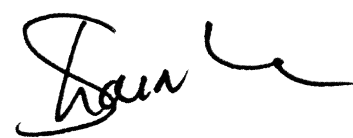
The devastating impact of EB is our motivation, our progress is our hope. What we don't have is time.

Our children with EB suffer incredible pain for years and then die of malignant skin cancer sometimes in their early twenties.

In 10 years from now we do not want Cure EB to need to exist!

Please feel very proud of what you have helped us achieve but please do stay with us to Cure EB.

With love and thanks,



Sharmila Collins
FOUNDER

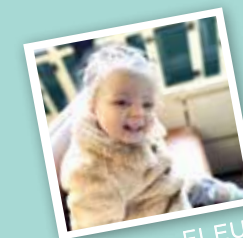


SOHANA 19yrs



FUNDING FREEDOM FROM EB with Damian Lewis
Find us on YouTube Cure EB
<https://www.youtube.com/cureEB>

10 YEARS 



FLEUR



SOHANA

A LOT CAN HAPPEN IN 10 YEARS

50,000 babies were born with EB in the last 10 years

When Sohana was born, we were told there would be nothing to help her. When we started fundraising for research in 2011, there had been one clinical trial for EB. Within a few years we had funded our first clinical trial in children (EBSTEM). We are now funding gene therapy trials and projects that are aiming at whole body treatments. The momentum created around EB research has led to biotech interest and there are currently four Phase Three trials underway. Two products for topical treatment may gain market authorisation this year. For Sohana and all the children still living in pain, with long dressings changes twice daily there is hope but we owe it to them to accelerate progress as fast as possible.



MASON



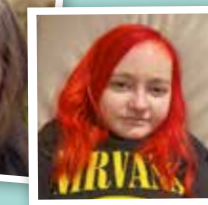
POPPY



GABRIELIUS



HOLLY



LEXIE-MAE



MAISIE





WALK ON THE WILD SIDE

SEPTEMBER 2021

We had a wonderful fundraising event at London Zoo with a research update from Professor John McGrath and a reflection on ten years of Cure EB. With The Jazz Spatz and delicious food it was fabulous to see you in person again after quite a while!



BONJOUR BINKY

'Binky's Time to Fly' is now available in French and around the world in six different languages, helping to spread the word about EB and the hope that we can fix it.



THE TIMES WRITE UP MISSION EB

SEPTEMBER 2021

Gabrielius, age 9, was featured in a Times newspaper article about Mission EB. Mission EB is a pilot treatment project in collaboration with the NIHR founded on our EBSTEM and ADSTEM clinical trial funding. The study is aiming to treat 36 children who have recessive dystrophic epidermolysis bullosa with infusions of stem cells derived from umbilical cord tissue. The potential for a first EB treatment in the UK is truly exciting and with the first patients having already had their infusions we are so grateful for your support that helped get us to this pivotal stage.



SILVER BUTTERFLY DINNER

MAY 2019



EVENTS



WORLD EB CONGRESS

The first global congress on Epidermolysis Bullosa took place on 19-23 January 2020, in London. We were proud to partner with Debra and others to bring together the worlds experts to share knowledge, foster greater research and develop the EB community. Sharmila Collins spoke at a plenary session about forming effective partnerships in EB research as an essential objective if we're going to get better treatments and we look forward to continuing to facilitate those vital partnerships as we hold ever fast to finding a route to cure EB. Sharmila also spoke with Lena Reidl on patient parents perspectives of participating in clinical trials. Over 700 people attended from all around the world.

10 YEARS OF RESEARCH



Cure EB has funded 24 research projects since its inception 10 years ago.

Collaborating with universities, biotechs, charities, research foundations, government organisations and clinicians, Cure EB have built momentum in research, supporting initiatives that will speed up the process towards effective treatments and our ultimate mission - to cure EB.



PROF JOHN MCGRATH



PROF ALAIN HOVNANIAN



PROF DENNIS ROOP

The professors and research teams working on these projects have been dedicating years of their professional lives towards the shared goals of helping children born with EB to live lives free from pain. Here are a few of them from early on in their EB researching careers.

LOCAL THERAPY

EB GRAFT CLINICAL TRIAL

Ex vivo gene therapy
Using grafts of autologous gene corrected skin equivalents
PROF ALAIN HOVNANIAN, MATTHIAS TITEUX INSERM at IMAGINE INSTITUTE PARIS
DR GUSTAVO MELEN, MANUEL RAMIREZ NINO JESUS at UNIVERSITY CHILDREN'S HOSPITAL MADRID

PRE CLINICAL & GENE EDITING TRIAL

Using grafts of autologous gene corrected skin equivalents
PROF LARCHER & PROF MARCELA DEL RIO AT UNIVERSIDAD CARLOS III MADRID SPAIN

FIBRX DERM Development of Topical Human Recombinant Decorin as an Anti-Scarring Therapy for DEB

JEAN TANG AND MARK DE SOUZA FIBRX Derm Inc BERKELEY CALIFORNIA
Co-funded with EBRP EBMRF Debra US and UK

CLINICAL DEVELOPMENT OF TXA127 for EB

Developing an oral anti-fibrotic treatment
In collaboration with CONSTANT PHARMAC, DEBRA AUSTRIA, EBRP & EBMRF

REGENERATION of the OCULAR SURFACE

PROF JAKUB TOLAR at UNIVERSITY OF MINNESOTA

LIMBAL STEM CELLS

For the treatment of eye surface wounds in RDEB
PROF JAKUB TOLAR at UNIVERSITY OF MINNESOTA

LENTICOL F Clinical Trial

Gene Modified Fibroblasts by local injection
PROF WASEEM QASIM & PROF ADRIAN THRASHER at INSTITUTE OF CHILD HEALTH & GOSH
PROF JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE LONDON

SKIN CANCER

RDEB Exome Sequencing Project
DR ANDREW SOUTH at THOMAS JEFFERSON UNIVERSITY USA
DR RAYMOND CHO at UNIVERSITY OF CALIFORNIA USA

NANOCOL

Nanoneedle bandage for topical gene therapy
PROF CHIAPPINI, PROF TOLAR, PROF MCGRATH
KINGS COLLEGE LONDON AND UNIVERSITY OF MINNESOTA

POTENTIAL GENE THERAPY FOR AIRWAY DISEASE In Junctional EB

Epithelial cell & gene therapy
For amelioration of respiratory symptoms in children with junctional EB
DR BUTLER UCL, GREAT ORMOND STREET, INSTITUTE OF CHILD HEALTH, PAOLA DE COPPI, NEONATAL SPECIALIST UCL

SPIN THERAPEUTICS RNA derm for EB simplex

Funded In Collaboration With EBRP and EBMRF
DR AARON SATO, BERKELEY CALIFORNIA

BRANCA BUNUS

Developing a non-viral gene therapy for RDEB
PROFESSOR WENXIN WANG AT NOVA UCD, DUBLIN, IRELAND

ALLOGENEIC DONOR CELLS 3 projects

EBSTEM Clinical Trial

For Children at GOSH
PROF JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE LONDON

ADSTEM Clinical Trial

For Adults at Guys
PROF JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE LONDON

MISSION EB

Cord MSC for treating children with EB in the UK GOSH/Birmingham Childrens Hospital
DR ANNA MARTINEZ, PAUL VEYS, MARK LOWDELL, PROF JOHN MCGRATH Co-funding with NIHR

LENTICOL M

Gene modified MSC
PROF WASEEM QASIM & PROF ADRIAN THRASHER at INSTITUTE OF CHILD HEALTH & GOSH, PROF JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE

GENE EDITING

Next Generation Genome Sequencing with CRISPR/Cas9
PROF JAKUB TOLAR at UNIVERSITY OF MINNESOTA
Project funded in collaboration with EBRP & EBMRF

GENE EDITING

TALen based approach
PROF JAKUB TOLAR at UNIVERSITY OF MINNESOTA USA

GENE EDITING for EB

PROF WASEEM QASIM & DR ANASTASIA PETROVA at INSTITUTE OF CHILD HEALTH & UCL, PROF JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE LONDON

EB IPS CONSORTIUM

Collaboration between Colombia, Colorado & Stanford Universities
Funded In Collaboration With EBRP and EBMRF
PROF ANGELA CHRISTIANO at COLOMBIA UNIVERSITY USA, PROF DENNIS ROOP at UNIVERSITY OF COLORADO USA, PROF ANTHONY ORO at STANFORD UNIVERSITY USA

EB IPS Developing a therapeutic approach for delivering stem cells systemically to treat fragile internal epithelia in RDEB

DENNIS ROOP, GANNA BILOUSOVA, ANNA BRUCKNER at UNIVERSITY OF COLORADO

EB IPS Testing an alternative approach to delivering IPSC derived keratinocytes and fibroblasts

DENNIS ROOP, GANNA BILOUSOVA, ANNA BRUCKNER at UNIVERSITY OF COLORADO

SYSTEMIC THERAPY



MISSION EB

Pilot treatment project in collaboration with the NIHR founded on our EBSTEM and ADSTEM clinical trial funding

The study is aiming to treat 36 children who have Recessive Dystrophic EB (RDEB) with infusions of Mesenchymal stem cells derived from umbilical cord tissue. It is being conducted at Great Ormond Street Hospital (GOSH) and Birmingham Children's Hospital (BCH), led by Dr Anna Martinez (Consultant Paediatric Dermatologist).

The trial has been made possible thanks to over £4.5 million of funding from the National Institute for Health Research (NIHR) in partnership with NHS England and NHS Improvement as well as a grant from Cure EB, with The University of Sheffield Clinical Trials Research Unit (CTRU) overseeing the study.

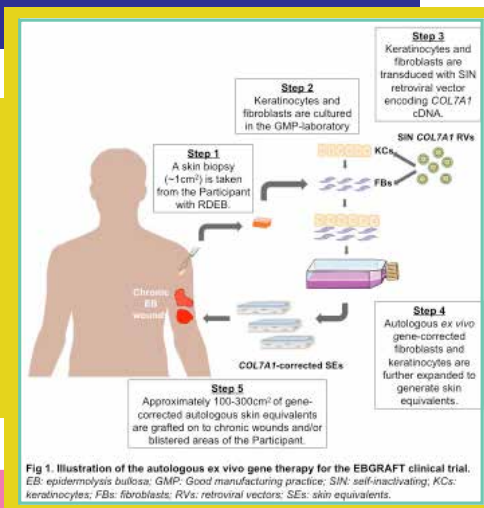
It is hoped that this treatment will offer reduced skin inflammation, reduced blistering and better wound healing and if successful will be commissioned as a treatment by the NHS.

EB Graft Trial

Patient cell derived gene corrected skin equivalent grafts

This trial started in adults in Paris, after significant Covid related delays. The first patient has been grafted and we await the next stages.

PROF ALAIN HOVNANIAN, DR MATTHIAS TITEUX Paris



Nanocol

A challenge of gene therapy is to find effective methods of delivering a functional copy of the gene that is missing

Nanoscale needles so small that they can inject substances into cells have been developed and tested, including on cells from EB patients, and researchers plan to create a bandage decorated with these needles that can be applied onto an EB patient's wounds. Once inside the cells, the functional version of the gene starts producing healthy collagen, enabling the EB sufferer's skin to heal.

DR CIRO CHIAPPINI & PROFESSOR JOHN MCGRATH Kings College London
PROFESSOR JAKUB TOLAR Dean of University of Minnesota

Gene Editing for EB

Preclinical towards Clinical Trial

To carry out the necessary pre-clinical studies for a Phase I / II clinical trial of gene edited skin equivalent grafts. Gene editing offers the possibility of permanent genetic correction of defective cells.

LARCHER ET AL Madrid

Spray-on skin cells

As part of our iPSC consortium funding, this project seeks to show successful regeneration of skin from gene-modified skin cells which are being delivered via a 'spray-on' system developed by Avita Medical. Early results have been encouraging.

PROF DENNIS ROOP & GANNA BILOUSOVA
Gates Center for Regenerative Medicine University of Colorado School of Medicine

<https://www.globenewswire.com/news-release/2022/01/06/2362375/0/en/AVITA-Medical-Establishes-Proof-of-Concept-for-Novel-Treatments-Using-Genetically-Modified-Skin-Cells.html>

Combined respiratory epithelial cell and gene therapy

The work here aims to deliver laboratory-based tools to model Junctional EB airway disease, as well as to use lentiviral gene editing tools to correct EB-affected airway lining. This proposal is a proof-of-principal project demonstrating the feasibility of combining lentiviral gene editing with airway epithelial stem cell therapeutics.

DR COLIN BUTLER Great Ormond Street Hospital

COMPLETED 2020

Gene Editing for EB

Investigation of CRISPR/Cas9 gene and base editing strategies for the COL7A1 gene mutation hotspots common within the UK paediatric population.

PROF WASEEM QASIM, DR ANASTASIA PETROVA, PHD STUDENT GAETANO NASO Institute of Child Health, UCL London

PUBLISHED 2020



LENTICOL M

'Human Mesenchymal Stromal Cell engineered to express collagen VII can restore anchoring fibrils in Recessive Dystrophic Epidermolysis Bullosa Skin Graft Chimeras'

Lenticol M Mesenchymal Stromal Cells engineered to express collagen VII for the treatment of Recessive Dystrophic Epidermolysis Bullosa.

TRIAL COMPLETED AND RESULTS PUBLISHED IN JANUARY 2020
VOL 140 ISSUE 1, PAGES 121-131. E6 <https://doi.org/10.1016/j.jid.2019.05.031>

IN SUMMARY Human MSC cells engineered to overexpress collagen V11 might provide therapeutic benefit when injected into sites of localized blistering. Intravenous infusions did not home to sites of wounding in mice and further work around this area is needed.

PUBLISHED 2020



ADSTEM Clinical Trial

'Do donor mesenchymal stromal cells have a disease modifying effect in adults?'

Published in the Journal of the American Academy of Dermatology
VOLUME 83, ISSUE 2, AUGUST 2020, PAGES 447-454

Phase 1/11 open-label trial of intravenous allogenic mesenchymal stromal cell therapy in adults with Recessive Dystrophic Epidermolysis Bullosa.

CONCLUSION MSC infusion is safe in RDEB adults and can have clinical benefits for at least two months, particularly in the reduction of itch.



LIFE WITH EB



POPPY

Big areas of concern are Poppy's hands and feet and her oesophagus. Hands dexterity is worsening as she uses them more at school. The skin becomes damaged and blistered. On repair the skin is definitely tighter causing contortion. Grip is considerably weakened.

Her feet have been the most vulnerable area since birth. The skin on the left foot is so fragile it breaks down regularly. This results in difficulty walking. We are keen to keep her as mobile as possible for as long as possible.

Her throat has been a real struggle of late. We have kept a dilatation at bay until now. This has been so hard to see as she loves food so much and it definitely brings comfort when she's having a hard time. Eyes are an ongoing problem too. Abrasions have been less frequent but are just awful for her when they do happen. 4-5 days without sight spent in the dark.

Daily life is consumed with dressing changes, preparation for dressing changes, preparation for meal times. There is little time outside school for play during the week. Every hour has to be factored in around dressings. Play dates have to be structured and the routine scheduled around them. There is a huge impact on family life especially for siblings who very quickly have to learn patience and understand that a lot of mummy/daddy time has to be given to the child with EB.

BY KATE GEE MUM OF POPPY

THANK YOU

TO OUR SUPPORTERS

IGY
Foundation

The James & Deirdre Dyson
Trust

Goldman
Sachs
Gives

mariamarina
FOUNDATION

W
Garfield Weston
FOUNDATION

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The Tudor Foundation

Rosetrees Trust

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The Childwick Trust

THE POPLI KHALATBARI
FOUNDATION

EXOTIX
PARTNERS

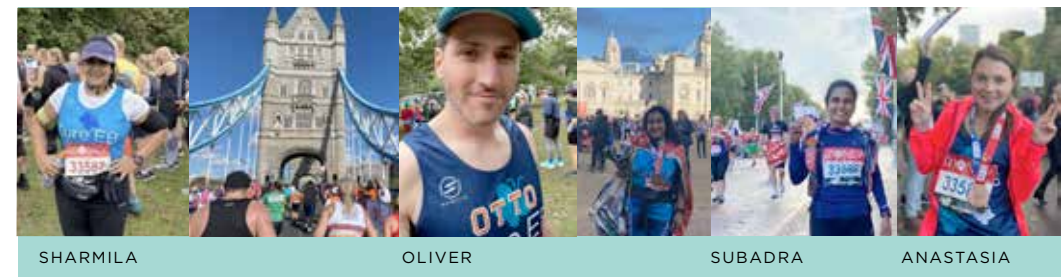
REN
GLORIA BIO PICTURE SCIENCE

GFI

THANK YOU



ACCELERATING RESEARCH TO END PAINFUL SKIN



ESSEX COURT CHAMBERS
3,000 MILES FOR MARCH CHALLENGE



DUNCAN WALES
SOLO COASTAL RUN



MARATHON MUM
14 MARATHONS SO FAR



BGC CHARITY DAY



bgc

The Phoebe
Research Fund
A child's fight for a future

CLAIRE ADAMS
100 MILE WALK



DODDINGTON
OPERA



IN MEMORY OF
KEITH TOMKINSON
GEORGE ASHCROFT



CHRIS GEE
GOLF DAY



AIMEE SEDDON
EXHIBITION



ARNOLD HOUSE SCHOOL
CHARITY DAY





EVENTS



2022



100% OF DONATIONS GO TO FUND RESEARCH FIGHT FOR A LIFE FREE OF PAIN. TO END EB. RESEARCH THE CURE

TRUSTEES SHARMILA NIKAPOTA JAMES COLLINS MICHAEL DE LATHAUWER TAZIM HALL DUNCAN WALES PROFESSOR DAVID KELSELL

PATRONS DAMIAN LEWIS SEAN BEAN DANIELLE DE NIESE SIR JAMES & LADY DYSON

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