NEW NAME FOR SRF

LAUNCHING NEW NAME!

RECENT EVENTS

LATEST RESEARCH

LOOKING FORWARD

Cure EB®

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Cure EB is a charity registered in the England and Wales 1158672
When do we want it?  
...uhhh yesterday, but as soon as possible works too. We have decided to make our mission much more obvious and have delayed this newsletter to be able to give you exciting news! We want to Cure EB - so it made sense to change our name to reflect this. We love where we started as the Sohana Research Fund, with our personal story around Sohana’s life. Having raised over £5 million for research in just a few years, it is important to simply reflect what all the hard work is about. Our core aim - to Cure EB. This means for all who have it, whatever the type. It will cut out the ‘What do you do?’ questions and become ‘What is EB and what can be done to cure it?’ The answer to this of course is as much research as possible. We will keep our very personal touch but hope that with slightly more hands on deck we will be more efficient and able to harness more opportunities for funding and therefore research. We still have our costs underwritten so that your donations go to research. We plan to transition slowly, so expect to see a lot of Sohana Research Fund’ still. There will be no costly and massive rebrand but a gentle move away from a name dearly held. 

Last year was very busy with our big Silver Butterfly Dinner at RIBA raising £250,000. We had a great response with sporting events, had lots of fun with EBpop and were very lucky to have people organising cake sales, chocolate challenges, birthday parties that we benefitted from and concerts. We cannot fund research without your support and drive and are deeply grateful for everything you do. 

Research news has been tremendous. With the publishing of an article in Nature describing the treatment of a little Syrian boy with Junctional EB who had skin grafts over 80% of his body, – gene therapy has come of age for EB. It is possible, and the many gene corrective techniques in trial currently point to a time when this condition will be treated. New projects for us include collaborating with US partners to fund a gene editing project in Minnesota with professor Tolar, and joining in to fund EB iPSC Therapeutic Consortium - which involves Colorado, Columbia and Stanford Universities.  

Our UK trials are being written up and work is progressing on gene modifying cells. We have some exciting and hopeful news in the pipeline but it is still too early to report until all the building blocks are in place.  

With Brexit, fundraising will be challenging and we will seek your support in holding Butterfly Brunches, join in runs, cycles, concerts, challenges and anything else we can think of! 

The next few years will be critical. The pace of research is to a big extent determined by what we are able to put in. The more we have - the more we can do - and the sooner we get to really effective treatments and to #cureEB. For the children and adults suffering the pain of EB there is no time to waste. Thank you for your help, 

Sharmila Collins  FOUNDER SRF

What do we want? A cure for EB
PARALLEL LONDON & SRF FAMILY DAY
3rd Sept, Olympic Park London
COLDPLAY
Head Full of Dreams Concerts

BOND WITH RESEARCH
28th February, London Film Museum

BUTTERFLY BRUNCH CLUB
29th November

SILVER BUTTERFLY DINNER
18th May, RIBA
THANK YOU

2017

REDBURN
Charity of the year

BARRY AUSTIN & JAMES COLLINS
London Marathon

VITALITY 10K
London Run

MASHA
Richmond Run

DODINGTON
OPERA

GFI CHARITY DAY

LIBERTY, HELEN, STACEY & MARK
Mc Plan & Site Services
Yorkshire 3 Peaks Challenge

STEVE CORNER &
JOHN COX Half Marathon

CORONA BRASS EVENT

CLAUDE & BEN
RIDENHALGH

TAZIM BLENHEIM
Triathlon

JERSEY COLLEGE
for GIRLS

BIRTHDAYS
Clive Beagles
Jane & Mike Epstein, Pilar Lerma

CHARLOTTE & THEO
Pop Cake Sale

LOEL’s “Lock up
Chocolate Challenge”

THE POPLI KHALATBARI
FOUNDATION

JNANE

Rosetrees Trust

Goldman Sachs
Gives

mariamaria Foundation

The James & Deirdre Dyson
Trust

IGY Foundation

Alta Advisers

RED BURN

theBigGive

GFI

The Tudor Foundation

EXOTIX PARTNERS

IDEA

The George Oliver
Foundation

The Tudor Foundation

The Childwick Trust

The Popli Khalatbari
Foundation

JNANE TAMSNA

REN

The Tudor Foundation

ARLE
There will not be a single answer to curing epidermolysis bullosa. The complexity and the body systems affected will necessitate combination therapy, as in cancer treatment. Research therefore has to be focussed on cell, gene, protein and drug therapies that aim to tackle the damage caused by EB externally and internally and treat the aggressive malignant skin cancer that can develop in early adulthood and is always terminal.

On the path to a cure we aim to help deliver interim treatments that improve quality of life. For a genetic condition we need to be able to correct or replace the genetic fault that causes the condition and manage the consequences of that fault. To this end we are investing in gene and cell therapy research, using breakthrough gene modification and gene editing techniques to treat the whole body as well as targeting areas such as the surface of the eye.

**RESEARCH PROJECTS**

**FUNDDED BY SOHA**

**EYE TREATMENTS** 2 projects

**LIMBAL STEM CELLS**
For the treatment of eye surface wounds in RDEB
Targeted genomic-medicine based cellular therapy
PROF. JAKUB TOLAR at UNIVERSITY OF MINNESOTA USA

**REGENERATION of the OCULAR SURFACE**
Develop 3D cornea with limbal stem cells derived from iPS in which RDEB mutations have been gene edited using CRISPR/Cas9
PROF. JAKUB TOLAR at UNIVERSITY OF MINNESOTA USA

**LENTICOLLAR CLINICAL TRIAL**
Gene Modified Fibroblasts given by local injection
Taking skin samples from patients with RDEB to produce "patient specific" gene corrected fibroblast cells, injected back into the donor patients skin
PROF. WAHID OASIM & PROF. ADRIAN THRASHER at INSTITUTE OF CHILD HEALTH & GOSH, PROF. JOHN MCGRATH at GUYS HOSPITAL KINGS COLLEGE LONDON

**SKIN CANCER**
RDEB Exome Sequencing Project
- Sequencing study of mutations in RDEB cancer
- Association of cancers with bacterial infections
- Pathways to target for treatment
DR. ANDREW SOUTH at THOMAS JEFFERSON UNIVERSITY USA, DR. RAYMOND CHO at UNIVERSITY OF CALIFORNIA USA

**EB IPS CONSORTIUM**
Collaboration between Colombia, Colorado & Stanford Universities
- Reprogramming patient body cells to IPS cells
- Genome editing to correct genetic mutation
- Validating and sequencing the cell bank
- Generating downstream skin tissue for grafting
_Funded in Collaboration With EBRR and EBMRP, PROF. ANGELA CHRISTIANO at COLOMBIA UNIVERSITY USA, PROF. DENNIS ROOP at UNIVERSITY OF COLORADO USA, PROF. ANTHONY DRO at STANFORD UNIVERSITY USA_
Why Us?

All of your donation goes to research
We are focused on treatment and translational research to correct the genetic defects in EB
We review projects throughout the year which allows researchers to develop projects at any time
We will have one project call for developing priority areas of research and new innovative research towards treatment
We have a flexible approach to project funding which enables easy collaboration with other organisations for peer reviewed projects

ALLOGENEIC DONOR CELLS 3 projects

**EBSTEM Clinical Trial** For Children at GOSH
MSC from unrelated donors who do not have EB given to children with RDEB
Published in Journal of Investigative Dermatology 23/4/15
PROF. JOHN MCGRAITH at GUYS HOSPITAL, KINGS COLLEGE, LONDON

**ADSTEM Clinical Trial** For Adults at Guys
• Safety trial in adults
• Aiming to identify key inflammatory markers
PROF. JOHN MCGRAITH at GUYS HOSPITAL, KINGS COLLEGE, LONDON

**MSC TREATMENT** For Children with RDEB at GOSH
Establishing a cell manufacturing facility at Kings for the treatment of children at GOSH
PROF. JOHN MCGRAITH at GUYS HOSPITAL, KINGS COLLEGE, LONDON, DR. ANNA MARTINEZ, DERMATOLOGY CONSULTANT at GOSH, PROF. FRANCESCO DAZZI at KINGS COLLEGE, LONDON, PROF. PAUL VETS, DIRECTOR BMT UNIT at GOSH

**LENTICOL M** Gene modified MSC
Following on from Lenticol I, aiming to see if Gene Corrected MSC can be given intravenously to improve wound healing
PROF. WASIM QASIM & PROF. ADRIAN THRASHER at INSTITUTE OF CHILD HEALTH & GOSH, PROF. JOHN MCGRAITH at GUYS HOSPITAL, KINGS COLLEGE

**GENE EDITING** Next Generation Genome Sequencing with CRISPR/Cas9
• Regional correction of COL7A1 gene
• Up regulation of COL7A1 protein beyond physiological levels
• Aiming for clinical trial of multiplexed gene and cell therapy for people with RDEB
PROF. JAKUB TOLAR at UNIVERSITY OF MINNESOTA, USA. Project funded in collaboration with EBRP and EBMRF

**EDITING for EB**
A diversity of gene editing approaches using CRISPR/Cas9 on a range of mutations
PROF. WASIM QASIM & DR. ANASTASIA PETROVA at INSTITUTE OF CHILD HEALTH & UCL, MCGRAITH at GUYS HOSPITAL, KINGS COLLEGE

**GENE EDITING**
TALEN based approach
Developing individualised treatment options for RDEB
PROF. JAKUB TOLAR at UNIVERSITY OF MINNESOTA, USA
MSC cells for RDEB treatment at Great Ormond Street Hospital
Manufacturing facility at Kings College, London
Professor John McGrath, Dr Anna Martinez, Professor Francesco Dazzi, Professor Paul Veys
Signed project £190,000 to Kings College London
Pending final costings from Great Ormond Street Hospital

Next generation genome sequencing
Professor Jakub Tolar, Professor of Paediatrics, University of Minnesota
Aiming at correcting the errors on the gene responsible for causing RDEB and up-regulating the missing protein beyond normal levels to address the suboptimal production
$1,000,000
Being funded in collaboration with EBRP and EBMRF

EB iPS Consortium
Professor Antony Oro Stanford University, Professor Dennis Roop University of Colorado Anshutz Medical Campus, Dr Angela Christiano Columbia University Medical Center
iPS stands for "induced pluripotent stem" cells. These are reprogrammed from adult cells and could become any other cell in the body. Under this protocol, cells from an individual with EB will be modified using a non-viral gene editing method to produce iPS cells with a corrected gene encoding the collagen VII protein. These gene-corrected iPS cells can then be differentiated into other cells such as skin cells known as keratinocytes. These cells can then be given back to an individual with EB.
$850,000 Being funded in collaboration with EBRP and EBMRF

Gene Editing for EB
Professor Waseem Qasim and Dr Anastasia Petrova
Evaluating the gene editing approaches using CRISPR/Cas 9 system for both recessive and dominant forms of dystrophic EB
£298,799
ACCELERATING RESEARCH TO END PAINFUL SKIN

IN THE PIPELINE

Continuation of gene editing projects in the UK and US with a focus on getting to clinical trials both locally and whole body

Continuation of gene modified stem cell work aiming for clinical trials if preclinical research is supportive

Continuation of funding towards mesenchymal stromal cell production at Kings College London and treatments at Great Ormond Street Hospital

FOR FUTURE FUNDING

Generation of gene modified skin grafts and clinical trial

RNA project

RDEB squamous cell carcinoma is a particularly aggressive malignant skin cancer. It has extremely high mortality and young adults with severe RDEB are at huge risk of developing it. We aim to pursue projects researching treatment options

Gene modification and gene editing approaches for Junctional EB

New project exploring novel treatment approaches for both local and systemic treatments including delivery mechanisms, skin substitutes and protein therapy

Dedicated GMP facilities for gene editing, gene modification and EB gene and cell therapy treatment suite

Lectureships in Dermatology, PhD studentships & Clinical Research fellowships

ADVANCES AT ANSCHUTZ MEDICAL CAMPUS
UNIVERSITY OF COLORADO PUBLIC RELEASE APRIL 2018

Colorado University Anschutz科学家 awarded $3,8 million Dept of Defense grant

String of major recent grants underlines promise of new potential treatments for Epidermolysis Bullosa and other chronic skin wounds

AURORA, Colorado. (April 12, 2018) - Scientists from the Gates Center for Regenerative Medicine at the University of Colorado School of Medicine are part of a consortium awarded $3.8 million from the U.S. Department of Defense to move discoveries in stem cell-created skin grafts into the manufacturing stage, bringing further hope to victims of debilitating inherited skin diseases.

The major grant for the Epidermolysis Bullosa (EB) iPS Cell Consortium, which includes research teams from the University of Colorado Anschutz Medical Campus, Stanford University School of Medicine and Columbia University Medical Center, will move production of stem cells into the Gates Biomanufacturing Facility at CU Anschutz.

The $3.8 million grant follows recent awards for the same investigators by the 21st Century Cures Act and the California Institute of Regenerative Medicine, boosting research that could not only benefit EB sufferers, but also countless patients with severe chronic skin wounds. In February, CU Anschutz’s EB researchers reported a more efficient approach to reprogramming a patient’s diseased skin cells into stem cells, raising hopes for future clinical trials and potential cures. The results were published in Nature Communications.

In announcing the new grant, Department of Defense (DoD) reviewers issued one of the most emphatic research endorsements possible, saying, “This study is based on the strongest cutting-edge scientific rationale in the field of wound care and dermatology. It is also a collaborative effort among top physician-scientists, scientists, health care providers, epidermolysis bullosa patients, families, and charities’.

One evaluator wrote: ‘The proposed research has the highest probability of success of bringing gene-corrected tissue to patients in the hospital’.

The consortium is funded by the U.S.-based EB Research Partnership (EBRP), EB Medical Research Foundation (EBMRF) & the Sohana Research Fund from Great Britain.

Read the full release: https://www.eurekalert.org/pub_releases/2018-04/uoca-cas041218.php
Mason’s morning routine starts when he gets up at 7am. We will do any dressings that need doing, then feed him through his gastrostomy. Mason loves school and hardly has any time off, even if he’s feeling sore or tired. Being with his friends takes his mind off EB and he never wants to miss school.

Mason has roughly 3 to 4 visits to GOSH and 3 to 4 visits to our local Poole Hospital a year unless he has surgery, then we are at Great Ormond Street for weeks at a time. His hand surgery was 13 weeks in total, week after week.

Mason’s most vulnerable areas are his throat after coughing up the lining of his oesophagus and his hands where he’s slowly losing the use of his thumbs, also his fingers are seriously contracting so we are waiting for more surgery dates.

Mason’s a very sociable little boy but, because he is ‘nil by mouth’, eating out can be quite difficult. Even though he loves going out we feel guilty that he cannot eat, but he always tries something and he has his iPad to keep him amused sometimes too. It’s never ever stopped Mason wanting to go out as he is such a people person and loves being out and about with friends and family. He even sits with his friends at school lunch times and just chats with them without a lunch.

Mason’s often asked what’s wrong with him and I always step in and take over with an answer because he doesn’t like having to explain it. He sees himself as normal though he lives with a condition. If Mason hears the word ‘normal’ his response is ‘What is normal?’ which is such a true saying.

I still explain what EB is regularly, especially if we are in a different town to where we live (where he is a celebrity having been in so many charity campaigns) or if we are on holiday. I usually explain it to people that he has a fragile skin condition, or he has poorly skin or he has skin like a butterfly’s wing - but he’s a happy little boy who lives life to the max and he’s our world.

Mason has great interactions with his friends, peers and family but his condition stops him from doing so much, which he does get frustrated about. It never gets easier, but he also has learnt to accept it to a degree. As parents, it’s heartbreaking to see how much Mason can’t do, but we do our best and so does the school, to adapt everything to get him involved in some way.

Mason is totally football crazy, goes to every AFCB Bournemouth football match and belongs to a local football team where they have made him manager as he is unable to play. He also loves his Playstation where he plays with all his friends online, and they chat through headphones and join in games together.

By Kerry White, Mum of Mason
MY STORY

BY KATE GEE

So why am I running the London Marathon this year?

My daughter Poppy is now 6 years old and has never experienced a day in her life without pain.

EB leaves little time for play. Poppy misses out on so much of the childhood experiences that most of us take for granted.

This is life with EB everyday, there’s no break and you can’t take a holiday away from it. We desperately want to be able to change it, not just for Poppy but for the 5000 people living with EB.

I have committed to doing this run which, if I’m honest, I didn’t think was going to be as tough as I am finding the training! “I used to be fit” I keep telling myself, “so I’m sure it won’t be too difficult to get back in shape”. I thought I would enjoy the personal time I have had to schedule. How wrong I was! Every session is a fight to the finish and it doesn’t seem to be getting easier as my mind goes in to overdrive.

But what will keep me motivated is that all this pain I’m going through is nothing compared to what a child with EB has to go through on a daily basis. Come April 22nd I will make it over that finish line no matter what for my beautiful, brave and very funny little girl who through everything always manages a smile.

TO DONATE: virginmoneygiving.com/KateGee2

GENE THERAPY SUCCESS IN EB


A seven year old boy with severe Junctional EB has been given new genetically modified skin that covers 80% of his body. After nearly two years the new skin appears completely normal. His life has been transformed and his skin remains strong. This incredible work, which is the product of painstaking research by Professor de Luca over many years, proves that gene therapy can work in Epidermolysis Bullosa.

The challenge is to replicate these results in other types of EB and progress to treatments that address internal as well as external damage.
## Forthcoming Events 2018

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
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<tr>
<td>22 April</td>
<td>VLM Marathon</td>
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<td>11 May</td>
<td>Cure EB Research Update</td>
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<td>June</td>
<td>EBpop</td>
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<td>15 July</td>
<td>Virgin Sport British 10k</td>
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<td>28/29 July</td>
<td>Prudential Ride London</td>
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<td>Sept</td>
<td>Cure EB Family Day</td>
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<tr>
<td>14 Oct</td>
<td>Royal Parks Half Marathon</td>
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<td>Nov</td>
<td>Butterfly Brunch Club</td>
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**May 2019**  
Cure EB Silver Butterfly Dinner at RIBA