CLINICAL TRIALS UPDATE

RECENT EVENTS

LATEST RESEARCH

LOOKING FORWARD
It has been a great year of transition to our new name that everyone seems to love, from researchers to our children with the condition. It gives everyone hope and much needed momentum as there is more work to be done, more trials to start.

That is the wonderful thing though – we are talking about clinical trials. Treatment trials for a genetic incurable condition. Trials happening all over the world aiming to bring symptomatic relief by healing wounds and reducing itch, to trials that are testing gene therapies to vastly improve quality of life by replacing the missing protein. There are trials that are investigator led in University Hospitals, trials being led by the Biotech Industry, and others that are a combination of the two. Much of the research has been funded by organisations such as ours, charity seed funding to do basic research, preclinical work and Phase 1/2 trials. But after that it is necessary to bring in industry for larger scale, multicentre trials and to pave the path to treatments for many. We are seeking ways to enable these transitions and are making good progress.

This year we are funding an exciting trial called EBgrafft which is a phase 1/2 trial of gene corrected skin equivalents for recessive dystrophic epidermolysis bullosa. I feel the need, in the current climate, to point out that the preclinical research was funded by EU scientific research grants and involved collaboration between researchers in France, UK, Germany and Spain. The trial is taking place in France.

Two of our existing trials and projects are currently being written up for publication. We will post updates on our website as they come in. Every research project leads us to further knowledge, and knowledge acquired brings more ideas! Funding towards mesenchymal stromal cells for treatment of children with recessive dystrophic epidermolysis bullosa is now in place and production has started. As a result of this it may be that the NIHR will now conduct a further study around treatment assessment - watch this space…

We had a lovely time with events such as #EBpop and a new Butterfly Run at the Olympic Park. We had a Butterfly Brunch Club speaker lunch in Kensington Gardens and we have our biennial Silver Butterfly Dinner coming up in May! We are hugely grateful to all of you who have run, cycled, baked cakes, had brunches, played golf, climbed, walked or sung to help #CureEB. Without your support we would not make the progress that we are. Raising awareness of EB is one of our greatest ongoing challenges so everything you do to raise the profile of the condition helps immensely. Sohana appeared in a piece called ‘Me and my Butterfly’ skin for BBC 3 which has been recently shared on Snapchat. Millions of people have viewed it.

Later this year we are very excited to share that we have a little book coming out! Here is a little taster…..

Thank you for your help,
EVENTS

2018

Cure EB BUTTERFLY RUN & FAMILY DAY
Olympic Park London in September
EVENTS

2018

HOUSE OF LORDS RESEARCH EVENING
11th May

EB POP
June

BUTTERFLY BRUNCH CLUB
with Mishal Husain on 29th November
The fundraising, administration and governance costs are underwritten so that 100% of your donations goes to research projects.

**2018**
- Total % of income spent on charitable work: 87.7%
- Total governance costs: 1.7%
- Total fundraising costs: 6.2%
- Other costs: 4.4%

**2017**
- Total % of income spent on charitable work: 89.6%
- Total fundraising costs: 8.0%
- Other costs: 2.4%

**2016**
- Total % of income spent on charitable work: 89.3%
- Total fundraising costs: 7.2%
- Other costs: 3.5%

Complete account details available on the Charity Commission website.
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.

Timeline showing the breadth of research projects that CureEB has funded since 2012

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.
Alain Hovnanian was born in 1959 in France. He studied Medicine at the Medical school of Paris XII University from 1977 to 1983. He completed his MD and internships in Paris. In 1993, he identified COL7A1 encoding type VII collagen, as the defective gene for recessive dystrophic epidermolysis bullosa (RDEB) and completed his Ph.D at Paris VII University on the molecular aspects of inherited dystrophic epidermolysis bullosa. In 1993, he joined Prof. Mark Lathrop’s laboratory as a postdoctoral scientist at the Wellcome Trust Centre for Human Genetics at Oxford University, United Kingdom. During his stay in Oxford, he also identified the genes for Darier disease ((ATP2A2) (Nature Genet 1999, 21 :271-277) and for Netherton syndrome (SPINK5) (Nature Genet 2000, 25 :141-2), which are two other rare and severe genetic skin diseases for which functional studies and new treatments are being developed. Since 2009, he has been appointed full professor of Genetics in the department of Genetics at Necker hospital for sick children in Paris. He runs a translational clinic on genetic skin diseases of children and adults aiming at fostering translation of research to new treatments for orphan diseases. He is the director of a new INSERM research laboratory on genetic skin diseases entitled “Genetic skin diseases: from molecular mechanisms to therapies”. His laboratory is one of the founding teams of the new IMAGINE Institute for Genetic diseases which opened in 2014 at Necker hospital in Paris.

I would like to repair an injustice of nature, by fixing the skin of the butterfly children so that they no longer have pain, nor blisters and they can forget that their skin used to hurt before.

What will happen during the EBGRAFT clinical trial?

The objective of EBGRAFT is to provide a safe, efficient and permanent treatment of chronic wounds in 3 adult patients with RDEB. The 3 participants have optimal clinical, molecular and immunological characteristics best suited for the trial. The clinical protocol is illustrated here.

Transplantation of genetically corrected skin will provide an essential and permanent treatment of chronic and large skin wounds because gene-corrected skin equivalents have the potential to definitely cure the treated area. Therefore, treated areas will locally reverse the disease phenotype. They will prevent the formation of blisters, skin erosions and inflammation, retraction and skin cancer. They will reduce itching, pain and infections. For these reasons, this project has a strong potential to bring clinical improvement to RDEB patients and to represent a major progress in the treatment of this devastating disease.

Effective treatment of RDEB will have also strong, positive knock-on effects on the quality of life of patients and their families. Restoring skin cohesion over the most vulnerable areas such as the hands, forearms, feet and legs to prevent the recurrence of skin lesions and skin cancers should result in a significant reduction in the functional and systemic complications of RDEB.

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Global progress supported by Cure EB

PROF JOHN McGRATH

PROFESSOR OF MOLECULAR DERMATOLOGY
King’s College London, Guy’s and St Thomas NHS Foundation Trust London, Mary Dunhill Chair in Cutaneous Medicine

Clinical trials funded by Cure EB have shown that intravenous infusions of bone marrow cells called mesenchymal stromal cells (MSCs) can improve the skin of both children and adults with RDEB.

The EBSTEM and ADSTEM trials took MSCs from unrelated people and gave them to people with RDEB. These cell infusions led to better wound healing, reduced pain and itch, and improved quality of life — all of which lasted for several months. These are the first trials of intravenous MSCs in people with RDEB and the results provide encouragement for getting these cells into regular clinical care until such a time as better treatments that look to cure EB are available.

Cure EB is also supporting a gene therapy trial called LENTICOL-F. This work involves growing some skin cells called fibroblasts from someone with RDEB, adding back the missing gene (type VII collagen), and then injecting back that persons own genetically modified fibroblasts into the skin. The goal is to try to prove this form of gene therapy is safe and to build a platform for future gene therapy that might be given intravenously to benefit the whole body.

Cure EB is also supporting research to develop gene editing of RDEB cells. This is a new technique that can silence or repair defective genes, the aim being to restore type VII collagen that works normally in cells and the skin. Gene editing work is not yet in clinical trials for RDEB, but the studies funded by Cure EB are building the pre-clinical data needed to make this a reality. Cure EB is also supporting other clinical research that aims to improve management of the complications of RDEB, such as sore eyes and also skin cancer.

Over the last 3 years, there have been clinical trials taking place around the world that are trying to improve the lives of people living with EB, and Cure EB is delighted to be at the forefront of this work. Cure EB funds work by several researchers in many countries, and is always looking to support the best studies that show potential in helping people with EB, and in striving towards that ultimate goal of curing EB.
THANK YOU TO EVERYONE WHO HELPPED RAISE FUNDS IN 2018

The James & Deirdre Dyson Trust

Alta Advisers

IGY Foundation

PRUDENTIAL RIDE LONDON
Raised over £9,800

GFI CHARITY DAY

THE BIG GIVE 2018
Raised £83,984

The Royal Parks Half Marathon
Raised over £900

BLENHEIM PALACE TRIATHLON
Raised over £1,350

REDBURN
Charity of the year

REBECCA GREENWOOD

FELICITE DU JEU & XAVIER PERKINS

AIMEE SEDDON & FRIENDS

LONDON MARATHON 2018
Our runners raised over £18,500

ROYAL PARKS HALF MARATHON
 Raised over £900

THE BIG GIVE 2018
Raised £83,984
THANK YOU TO EVERYONE WHO HELPED RAISE FUNDS IN 2018

GARY WALDOCK
CYCLING 50KM
Raised over £1,300

WALL IN IT TOGETHER
CAROLINE, ELOISE, FELICITY
REBECCA & KATE
Raised over £3,300

JENNY HAMILTON
Golf Day raising over £7,000

FRIDA D’SILVA
VITALITY LONDON
Raised over £600

DODDINGTON OPERA

KIRAN & RUBEN
Raised over £700

SYCAMORE SUPERSTARS
Raised over £1,600

ST CHRISTOPHERS ‘DEN’ DAY

THE POPLI KHALATBARI FOUNDATION
JNANE TAMSNA
Rosetrees Trust
The Tudor Foundation
The Childwick Trust

THANK YOU
EVENTS

2019

Cure EB®

28 APRIL

LONDON MARATHON

16 MAY

Silver Butterfly Dinner

ORGANISED BY CURE EB

22 SEPTEMBER

BUTTERFLY RUN

EB AWARENESS WEEK 2019

25 - 31 OCTOBER

butterfly speaker lunch

NOVEMBER

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 DELATHAUWER  TAZIM HALL  PROFESSOR DAVID KELSELL

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

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