Institution	Project Name	Project Description	Patient Population	Technique	Principal Investigator(s)	Amount Funded
INSERM - Imagine Institute for genetic disease	Multi-omics of Recessive Dystrophic Epidermolysis Bullosa-associated Squamous Cell Carcinoma for targeted anti-tumor therapy.	This lab aims to characterize RDEB squamous cell carcinoma (SCC) tumors to understand the underlying mechanisms of their development. SCC is life-threatening to patients with RDEB. The researchers will use their learnings to identify potential drugs for anti-tumor treatments.	Dystrophic EB (DEB)	Anti-tumor Thearpy	Alain Hovnanian, Helene Ragot	$295,718.53
Northwestern University	Suppressing the Itch of Dystrophic Epidermolysis Bullosa	Itch is one of the most troublesome symptoms of EB causing decreased quality of life as well as well as being detrimental to skin healing. These researchers will study the use of the FDA approved, repurposed drugs dupilumab and a JAK inhibitor (JAKi), ruxolitinib, in clinical trials as well as investigate the mechanisms of their anti-itch properties in an RDEB mouse model and in blood and tissue from treated patients.	EB Simplex (EBS) Dystrophic EB (DEB) Junctional EB (JEB)	Immunotherapy Drug Repurposing	Amy Paller and Ziyou Ren	$236,423
Thomas Jefferson University	Targeting Innate Signaling Pathways in Treatment of Recessive Dystrophic Epidermolysis Bullosa-Associated Squamous Cell Carcinoma	There are no optimal therapies for advanced squamous cell carcinomas (SCCs) that develop in patients with RDEB. Immunotherapy presents a potential approach; however, we lack a thorough understanding of targetable immune pathways in the RDEB-SCC microenvironment. Researchers will leverage their access to patients with RDEB-SCC at the Jefferson Adult Epidermolysis Bullosa Clinic, their expertise in RDEB-SCC carcinogenesis, and anti-tumor immunity to gain a comprehensive understanding of the tumor immune microenvironment in RDEB-SCC and to test the utility of several immunotherapeutic approaches in pre-clinical models.	Dystrophic EB (DEB)	Cancer Research Immunotherapy	Neda Nikbakht and Andrew South	$200,000
INSERM U1163-Imagine Institute for genetic diseases	Developing ex-vivo and in-vivo Base and prime editing strategies to treat Recessive Dystrophic Epidermolysis Bullosa	This project studies more advanced methods of gene editing to be eventually tried as skin organoids and grafts on RDEB patients with five specific, common mutations which have not been previously targeted by other methods (mutations in Exons 3, 74,80,and 105). They will use these novel techniques to gene-correct keratinocytes, fibroblasts and induced pleuripotent stem cells (iPCS's).	Dystrophic EB (DEB)	Gene Therapy Stem Cell Therapy	Araksya Izmiryan, Matthias Titeux, and Alain Hovnanian	$498,431
INSERM - Institut Necker	Strategies for efficient and long-term engraftment of Mesenchymal Stromal Cells for the treatment of Recessive Dystrophic Epidermolysis Bullosa	The objective of this project is to determine the best strategy for efficient and long-term engraftment of bone marrow derived mesenchymal stromal cells (BM-MSC) in the perspective of clinical translation in RDEB patients.	Dystrophic EB (DEB)	Stem Cell Therapy	Alain Hovnanian	$242,694.32
UMass Chan Medical School	Ataluren Treatment in Patients with Epidermolysis Bullosa	Ataluren is an oral medication which works by "read through" of severe mutations (called "nonsense" or "premature termination codons"(PTC)) allowing the gene to produce a normal protein despite the presence of the mutation. It is apparently well-tolerated and has been approved for Duchenne muscular dystrophy in the EU and is being considered by the FDA here. It has also been used in other genetic diseases with PTC mutations such as cystic fibrosis (CF), Miyoshi Myopathy, Hurler syndrome, Carnitine Palmitoyltransferase 1a deficiency, Usher syndrome, and Batten disease. Similar effects have been noted in EB patients using a well-known oral antibiotic, gentamycin, but toxicity to kidneys and hearing may limit its use. At this institution the investigators have noted vast clinical improvement and appearance of the missing protein, LAMB3, in one 11 year old patient with JEB treated for 2 years with Ataluren and would like to undertake a clinical trial of other patients with PTC mutations.	Dystrophic EB (DEB) Junctional EB (JEB)	Gene Therapy Drug Repurposing	Karen Wiss, Sarah Servattalab, and Carolyn Foley	$20,100.00
Stanford University School of Medicine	Impact of COL7A1 gene therapy on SCC recurrence in RDEB skin	BVEC healed areas have show a reduction in fibrosis as well as reduction in erythema and inflammation. These results suggest that collagen VII replacement in DEB skin may not only stop blistering but also may halt, and perhaps reverse fibrosis and inflammation leading to SCC formation. This project hypothesizes that BVEC induced C7 expression in RDEB skin following SCC excision will normalize the invasive tumor microenvironment and reduce tumor recurrence.	Dystrophic EB (DEB)	Gene therapy	Peter Marinkovich	$449,482.00
Stanford University School of Medicine	Targeting collagen VII antibodies in dystrophic epidermolysis bullosa	This proposal focuses on a drug, subcutaneous immunoglobulin (IgG), which the team hypothesizes will synergize with and enhance the effectiveness of gene therapies such as BVEC. This will be the first clinical trial of IgG therapy in DEB patients and the first study which addresses the immune side effects of cutaneous gene therapy.	Dystrophic EB (DEB)	Gene Therapy Immunotherapy	Peter Marinkovich	$330,711.00
The Board Of Trustees Of The Leland Stanford Junior University	Development Of A Non-Invasive "Scanning Biopsy" For Detecting Squamous Cell Cancer In Persons With Dystrophic Epidermolysis Bullosa.	This team aims to develop a "scanning biopsy" using optical imaging of the skin which won't need to be operated on. They're then using machine learning to turn these pictures into detailed images. This technology could enable dermatologists to noninvasively diagnose suspected SCC areas in DEB patients at the bedside.	Dystrophic EB (DEB)	Cancer Research	Kavita Sarin	$784,796.00
University of South Australia	Development of topical anti-granzyme K therapy for the treatment of itch, inflammation and skin damage in epidermolysis bullosa	This research has identified granzyme K as a target protein that contributes to chronic itch induction, impaired wound healing, inflammation, and fibrosis in skin. Granzyme K is abundant in EB compared to healthy skin and is secreted by mast cells, which are elevated in EB skin and have important roles in itch induction. They have also identified a granzyme K inhibitory drug called bikunin which when delivered topically to itchy skin, reduces scratching by >80% and dramatically decreases scratch-mediated skin damage. This proposal is to further develop topical bikunin as treatment for EB.	EB Simplex (EBS) Dystrophic EB (DEB) Junctional EB (JEB) Kindler Syndrome	Drug research	Chris Turner	$257,841.00

The Trustees of Columbia University in the City of New York	Wearable engineered physiological dermal extracellular matrix (wEPDEX) to treat mitten-like deformities and anatomically-complex wounds	This project focuses on using a new technology called Wearable 3D Skin, which has continuous 3D tissues mimicking the skin's properties. The initial aim is to develop a wearable skin technology called wEPDEX, using advanced techniques and testing its effectiveness in regenerating skin in the lab and in mice. The ultimate goal is to create two commercial products within four years: wEPDEX-Glove for treating specific skin issues in patients with mitten-like deformities and wEPDEX-Joint for complex areas like knees and elbows. These wearable products could significantly improve wound healing and reduce inflammation in patients, offering a personalized and regenerative approach.	Dystrophic EB (DEB)	Stem Cell Therapy Protein Therapy	Hasan Abaci	$161,757.00
University of Southern California	Enhancing Readthrough Therapy for RDEB and JEB: The Synergistic Potential of CC-90009 and Gentamicin	This team has previously showed that gentamicin induced PTC readthrough and C7 and laminin 332 production in RDEB and JEB patients, respectively. However, gentamicin-associated toxicities at elevated doses limit long-term use. In this study, they aim to investigate the potential of a new compound called CC-90009 to enhance the positive effects of gentamicin in promoting protein production while minimizing harmful effects. The goal is to see if the combination of CC-90009 and gentamicin can reverse abnormal cell characteristics and become part of the skin's structure.	Dystrophic EB (DEB) Junctional EB (JEB)	Drug research	Mei Chen	$243,600.00
University Of Chicago	Development of a Novel RNA Replicon Vector for Treatment of EB	This study aims to address limitations associated with viral-based gene therapy for RDEB. Viral vectors can sometimes trigger immune responses and potential genotoxicity. Instead, the researchers propose using RNA-based therapeutics, an alternative approach with its own challenge of a short half-life. To overcome this, they've developed a continuous directed evolution platform leveraging skin keratinocytes to engineer RNA vectors for prolonged expression in the skin. Through this platform, they've identified a promising Sindbis replicon that significantly enhances gene expression. The proposal involves a preclinical study to assess the therapeutic efficacy of this engineered replicon vector for RDEB gene therapy.	Dystrophic EB (DEB)	Gene Therapy	Xiaoyang Wu	$500,000.00
Thomas Jefferson University	Epidermolysis Bullosa Community Cell Bank	This lab has developed multiple cell populations and cell lines from over 30 patients which they have been providing to researchers over the past 10 years. Previous funding over 12 months has established a homogenous cell bank aimed at providing the EB research community a point of reference for therapy development. This proposal will establish a web-based portal for requesting cell lines and for access to pooled, de-identified, data utilizing the current bank of cell populations and generated by the South Lab and other investigators.	EB Simplex (EBS) Dystrophic EB (DEB) Junctional EB (JEB) Kindler Syndrome	Stem Cell Therapy Gene Therapy Protein Therapy Immunotherapy Drug Research Cancer Research Data	Andrew South	$44,800.00
University of South Australia	Development of a cell-free therapy for the treatment of epidermolysis bullosa wounds.	This project aims to develop a novel thermo-responsive biodegradable gel that will be administered directly into wounds, where it will conform to cover the opening and promote healing responses. This gel will contain bioactive agents produced by human gingival fibroblasts that they have identified to stimulate healing with minimal scarring via the promotion of angiogenesis and inhibition of pro-inflammatory responses.	EB Simplex (EBS) Dystrophic EB (DEB) Junctional EB (JEB) Kindler Syndrome	Stem Cell Therapy	Allison Cowin	$217,922.00
Board Of Regents Of The University Of Wisconsin System	Microphysiological systems to evaluate new therapies for epidermolysis bullosa	In this project, the team proposes the use of microphysiological systems (MPS) to study EB Simplex. MPS are advanced in vitro platforms that allow researchers to mimic complex biological structures such as the architecture of human skin. They will use this MPS technology to generate a human-derived skin construct (skin organoid) to evaluate new genome editing therapies for EB Simplex.	EB Simplex (EBS)	Gene therapy	Jose Ayuso	$335,144.00
Stanford University	National EB Registry: AWS Portal (RENEWAL)	Continued funding for Stanford's role in EBRP's Curator platform	EB Simplex (EBS) Dystrophic EB (DEB) Junctional EB (JEB)	Gene Therapy Drug Research Data	Jean Tang	$372,697.00