Advances in Research and Treatment:
Junctional and Dystrophic Epidermolysis Bullosa

Supported by an educational grant from CHIESI USA, Inc.

Faculty

Lara Wine Lee, MD, PhD
Associate Professor
Medical University of Southern California
Los Angeles, CA

Joyce M C Teng, MD, PhD
Professor of Dermatology and Pediatrics
Stanford University School of Medicine
Palo Alto, CA

Faculty Disclosures

Lara Wine Lee, MD, PhD

Advisory Board, Consultant, Speakers Bureau: Castle Creek; Eli Lilly; Pfizer; Regeneron; AbbVie; Amryt; Krystal Biotech; Novartis; Pyramid Bioscience; Krystal Biotech; Kimberley Clark; Verrica; **Research:** AbbVie; Amgen; Amryt; Arcutis; Castle Creek; Celgene; Cara; Eli Lilly; Galderma; Incyte Corp; Mayne Pharmaceuticals; MoonLake; Novartis; Pfizer; Regeneron; Sanofi; Target Pharma; Trevi Therapeutics; Timber; UCB

Joyce M C Teng, MD, PhD

Consultant: Abeona; AFT; Amryt (Chiesi); AUCTA; Biocryst; Bridge Bio Castle Creek; Coterie; GLG; Guide Point; Innova; Kamari; KrystalBio; LeoPharm; Lifemax; Menlo Therapeutics; Mitsubishi; NobelPharm; Novartis; Palvella Therapeutics; Pfizer; Pierre Fabre; Regeneron; Syneos; Timber Pharmaceuticals; Topicals, Twi Biotech; **Investigator:** Care within Reach; Castle Creek; EB Research Partner; Gorlin Syndrome Alliance; Innova; LeoPharm; Lifemax; Novartis; Palvella Therapeutics; Pfizer; Regeneron; Timber Pharmaceuticals; Twi Biotech; **Medical Advisor:** BHD Foundation; EverEden Skin Care; FIRST; Gorlin Syndrome Alliance; TSC Alliance; **Loyalty:** GeneReview; UpToDate; Springer Publishing

Disclosures

- The faculty have been informed of their responsibility to disclose to the audience if they will be discussing off-label or investigational use(s) of drugs, products, and/or devices (any use not approved by the U.S. Food and Drug Administration)
  - Applicable CME staff have no relationships to disclose relating to the subject matter of this activity
  - This activity has been independently reviewed for balance

This CME activity includes brand names for participant clarity purposes only.
No product promotion or recommendation should be inferred.

Learning Objectives

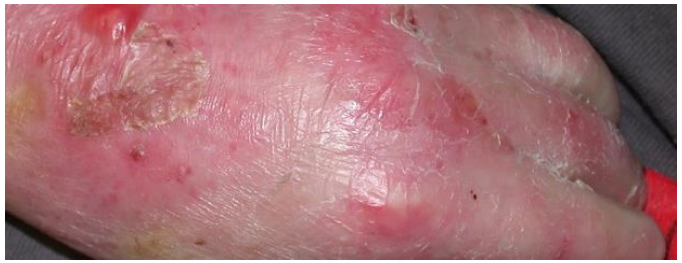
- Examine the genetic mutations and underlying pathophysiology of Junctional Epidermolysis Bullosa (JEB) and Dystrophic Epidermolysis Bullosa (DEB) and recognize their clinical manifestations
- Explain the standard management practices for Epidermolysis Bullosa (EB), including wound care, routine extracutaneous monitoring, and coordination with a multidisciplinary team to address the diverse needs of the patient
- Evaluate current FDA-approved treatments for EB, as well as emerging therapies, understanding their mechanisms of action

JEB = junctional epidermolysis bullosa; DEB = dystrophic epidermolysis bullosa; EB = epidermolysis bullosa.

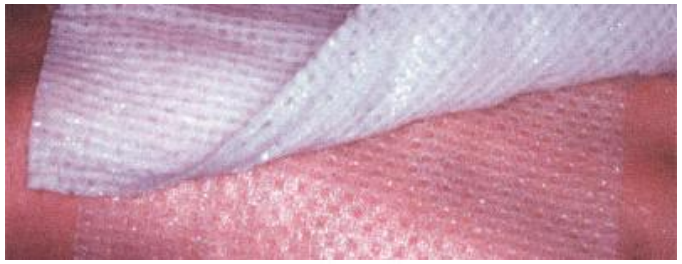
Junctional EB
Genetics and Clinical Features

Lara Wine Lee, MD, PhD

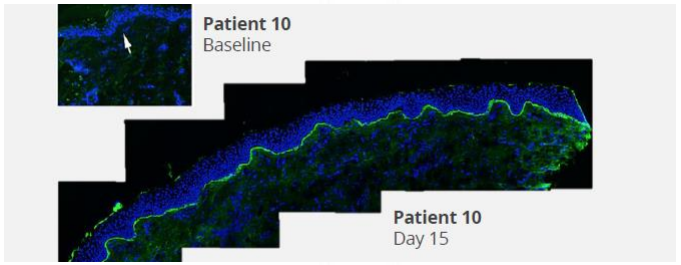
Advances in research have led to growth in our understanding of epidermolysis bullosa and its comorbidities, as well as new therapeutic options



Examine the genetic mutations and underlying pathophysiology of Junctional Epidermolysis Bullosa (JEB) and Dystrophic Epidermolysis Bullosa (DEB) and recognize their clinical manifestations



Explain standard management practices for EB including wound care, routine extracutaneous monitoring, and coordination with a multidisciplinary team to address the diverse needs of the patient

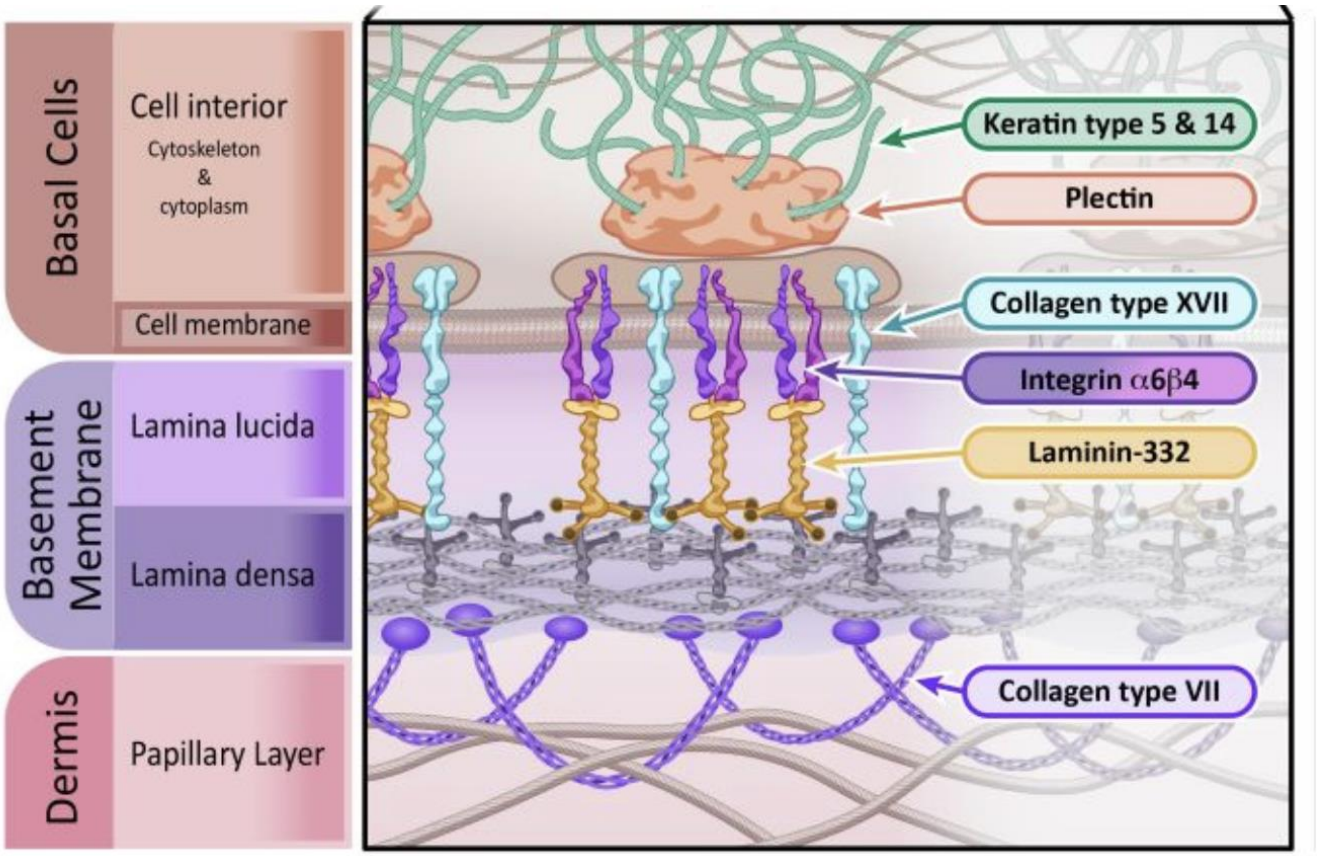


Evaluate current FDA-approved treatments for EB, as well as emerging therapies, understanding their mechanisms of action

What Is Epidermolysis Bullosa (EB)?

- EB is a group of rare inherited disorders that cause extreme fragility of the skin
- EB is caused by a mutation in one of 18 genes
- There are 4 major types
  - Simplex
  - Junctional
  - Dystrophic
  - Kindler Syndrome
- EB affects 1 of every 20,000 births in the U.S. (≈200 children/yr are born with EB)
- People with EB share the lifelong challenge of extremely fragile skin that blisters and tears from minor friction or trauma
- EB affects all genders and racial and ethnic groups equally

EB Types Show a Range of Severity



EB Types	Frequency	Mortality Risk
Junctional EB (JEB)	~ 5%	High
Dystrophic EB (DEB)	~ 20%	
EB Simplex (EBS)	~ 75%	
Kindler EB (KEB)	Very rare	Low

https://www.eurogct.org/epidermolysisbullosa

EB Can Have a Variable Presentation

EB Is a Multifaceted, Multisystem Systemic Condition

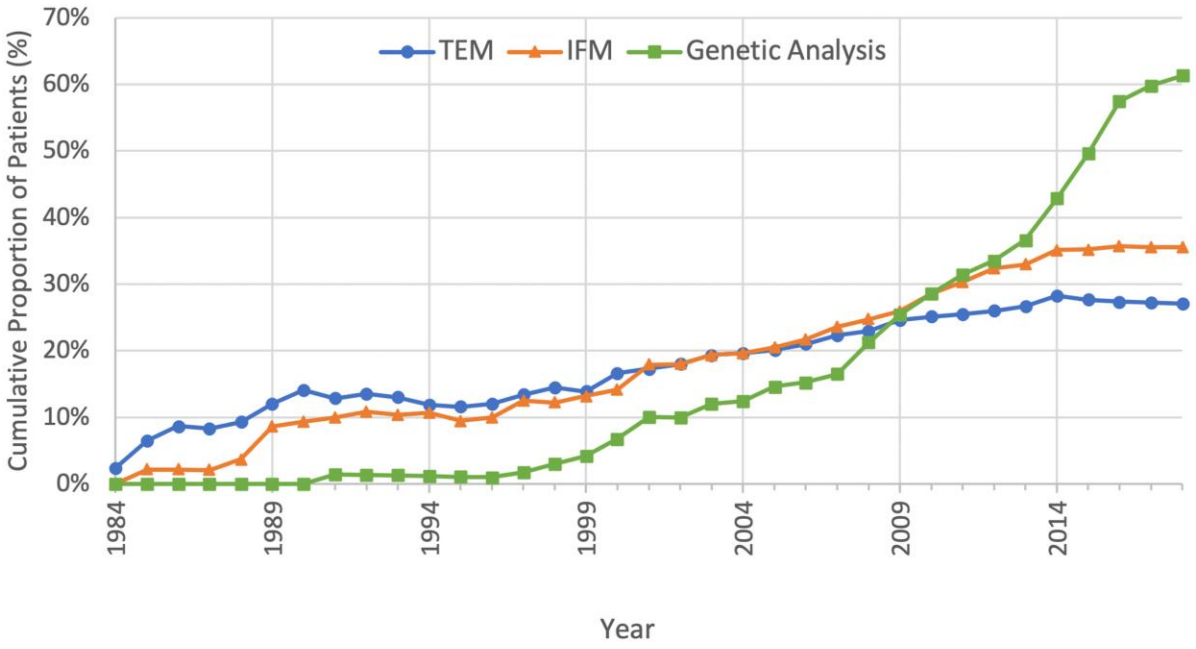
Fig. 1 Schematic representation of the timescale of therapy development for EB

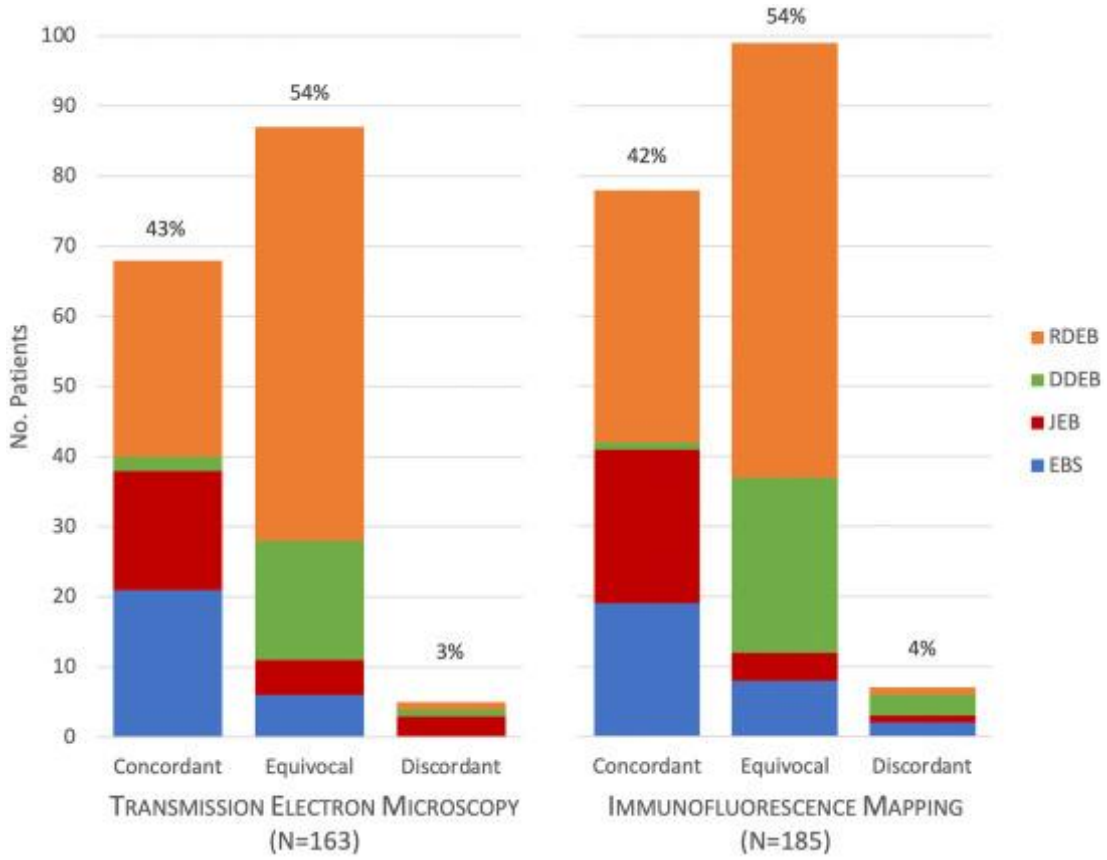
1 day old with erosions at birth, worsening over the first days of life, particularly at friction areas, such as the diaper. Skin peels with minimal manipulation, such as a diaper change.

What Type of EB Does Our Patient Have?

You cannot differentiate types of EB by clinical presentation alone

Diagnostic Accuracy in EB Is Increasing with More Widespread Use of Genetic Testing



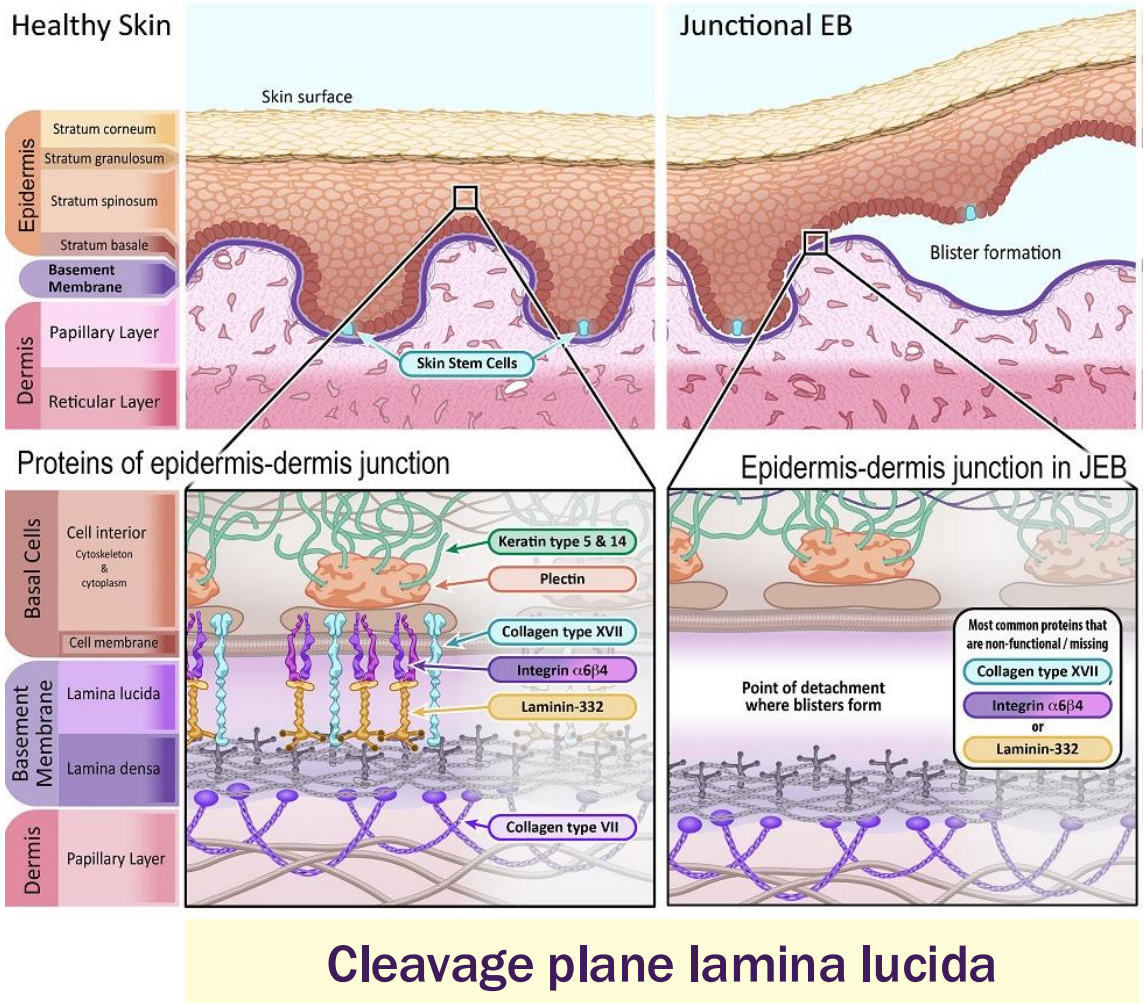


Phillips GS, et al. *JAAD*. 2022;86(5):1063-1071.

Genetic Testing in EB Is the Most Accurate Way To Secure a Diagnosis

CONDITIONS	GENES INVOLVED
Dystrophic EB (DEB)	*COL7A1*
EB Simplex (EBS)	*CD151*, *DST*, *EXPH5*, *KLHL24*, *KRT5*, *KRT14*, *PLEC*
Junctional EB (JEB)	*COL17A1*, *ITGA3*, *ITGA6*, *ITGB4*, *LAMA3*, *LAMB3*, *LAMC2*
Kindler EB (KEB)	*FERMT1* (*KIND1*)
Peeling Skin Syndrome	*CDSN*, *CHST8*, *CSTA*, *SERPINB8*, *TGM5*
Palmoplantar Keratoderma	*DSG1*, *DSP*, *JUP*
Epidermolytic ichthyosis & ED-Skin Fragility Syndrome	*KRT1*, *KRT10*, *PKP1*

Junctional EB



Cleavage plane lamina lucida

Mutations in genes that encode structural components of the hemidesmosomes or anchoring filaments, which provide mechanical integrity across the lamina lucida.

Types

JEB generalized severe (JEB-GS) previously called Herlitz junctional EB

- Laminin 332
- Fatality related to airway involvement, FTT, sepsis

JEB generalized intermediate (JEB-GI previously called Non-Herlitz junctional EB)

- Collagen type XVII (*COL17A1*)
- Laminin-332 (*LAMA3*, *LAMB3* and *LAMC2*)

https://www.eurogct.org/epidermolysisbullosa

Junctional EB

- Widespread blistering
- Granulation tissue, particularly around the nose and mouth
- Granulation tissue of upper airway

Junctional EB

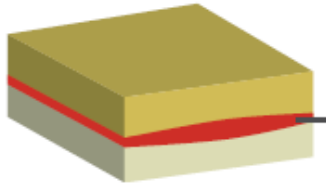
- 50% fatality rate in first year
- Mucosal involvement
  - Laryngeal
  - Respiratory
  - GI
  - GU
- Enamel hypoplasia

Complete nail loss in intermediate JEB

Hypergranulation in intermediate JEB

Enamel hypoplasia

Alopecia in intermediate JEB



5%

Blisters in the basement membrane

Less common that EBS or DEB

INTERMEDIATE JEB
previous names: JEB generalized intermediate, non-Herlitz JEB
affected genes: LAMA3, LAMB3, LAMC2, COL17A1
inheritance patterns: recessive

SEVERE JEB
previous names: JEB generalized severe, Herlitz JEB
affected genes: LAMA3, LAMB3, LAMC2
inheritance patterns: recessive

COMMON SYMPTOMS AND COMPLICATIONS

For full details, visit: www.debra-international.org/junctional-eb

01 02

Harmless skin spots and colour changes can occur

Itching

Inside of the mouth is usually affected

Tooth enamel affected

Eyes are usually affected by blistering

Thinning hair and hair loss

Anaemia (more common in severe JEB)

01

Blistering is widespread but less severe

Nails are usually lost or thick and change shape/ colour

Skin cancer (adulthood)

02

Few blisters (first couple of weeks of life)

Non-healing wounds on buttocks, face, ears, and tips of the fingers and toes.

Inside of the mouth, nose, oesphagus, and windpipe affected. Airway obstruction can be life threatening.

Nails lost (first few months of life)

Hoarseness and high-pitched wheezing

Life expectancy is unfortunately very limited (few weeks to a couple of years)

Clinical Spectrum of Junctional EB

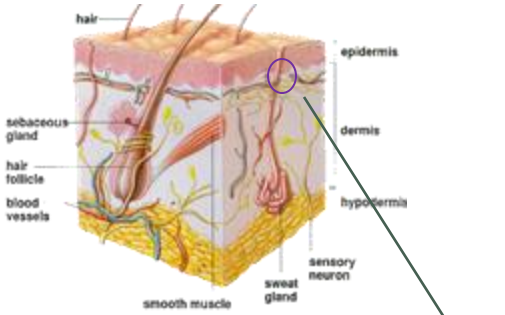
Br J Dermatol, Volume 183, Issue 4, 1 October 2020, Pages 614–627, https://doi.org/10.1111/bjd.18921

Rare JEB Subtypes
Autosomal Recessive Inheritance

Type	Affected gene (protein)	Clinical symptoms
JEB with pyloric atresia	ITGA6 (integrin α6) ITGB4 (integrin α6β4)	• Extensive. severe skin fragility at borth. deformity of structures such as the ears and nose. • Severe phenotypes can present with rudimentary ears. Nail dystrophy and loss. • GI atresia is evident within the first days-week of life. • Lethal within the first few weeks of life despite surgical correction of pyloric atresia. • Milder, non-lethal forms have less severe skin and nail involvement but with frequent genitourinary tract involvement.
JEB localized	COL17A1; LAMA3, LAMB3, LAMC2; ITGB4; ITGA3 (integrin α3 subunit)	• Limited cutaneous fragility and blistering, often only acral. • Variable nail dystrophy and mucosal involvement. • Variable dental enamel defects. Normal hair.
JEB inversa	LAMA3, LAMB3, LAMC2	• Onset of blistering from birth in flexural sites. Atrophic scarring. • Dental enamel abnormalities. Variable nail loss.
JEB late onset	COL17A1	• Onset of skin fragility in childhood, starting acrally. Progressive fragility with age. • Healing with skin atrophy and loss of dermatoglyphs. • Scarring leading to flexion contractures of the fingers and reduction of mouth opening may occur with age. • Variable dental enamel and nail involvement.
JEB-laryngo-onycho-cutaneous (LOC) syndrome	LAMA3A (laminin α3A)	• Fragility from birth . • granulation tissue (much more than JEB severe). Predilection for the face and neck. • Nail dystrophy and loss with granulation tissue of the nail beds. • Conjunctival and eyelid granulation tissue leading to symblepharon, scarring and impaired vision. • Laryngeal granulation tissue with respiratory obstruction which can be lethal. • Anaemia due to bleeding from over granulating wounds.
JEB with interstitial lung disease and nephrotic syndrome	ITGA3	• Variable cutaneous features with absence or presence of skin fragility from infancy. • Nails may be dystrophic and hair may be sparse. • Interstitial lung disease and nephrotic syndrome predominate the phenotype and can be diagnosed soon after birth. • Death in infancy or early childhood is the norm.

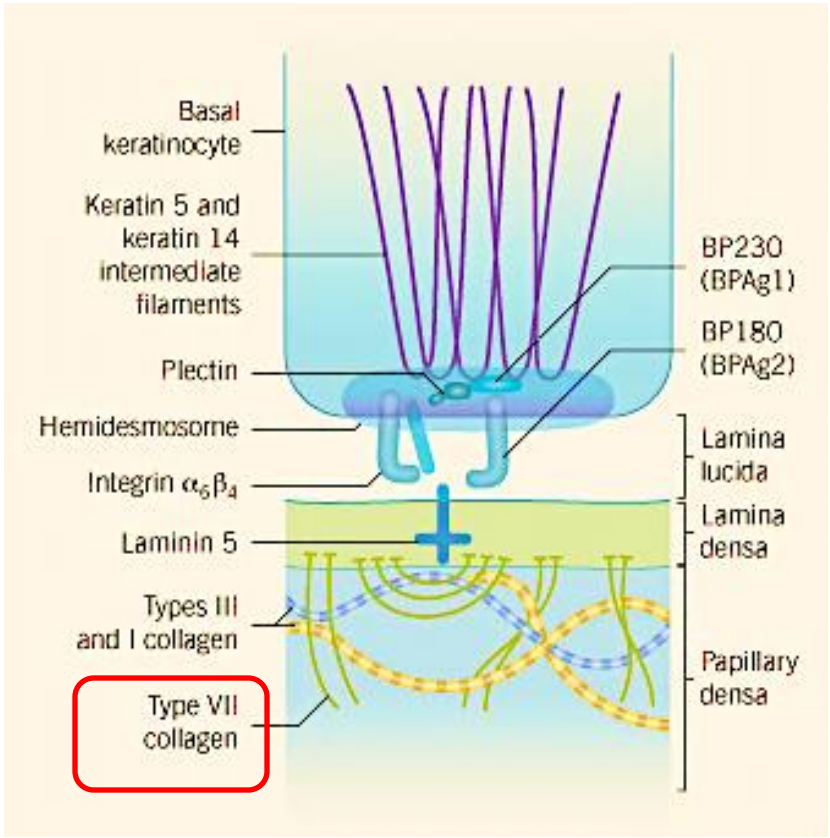
Dystrophic EB
Genetics and Clinical Features

Joyce MC Teng, MD PhD

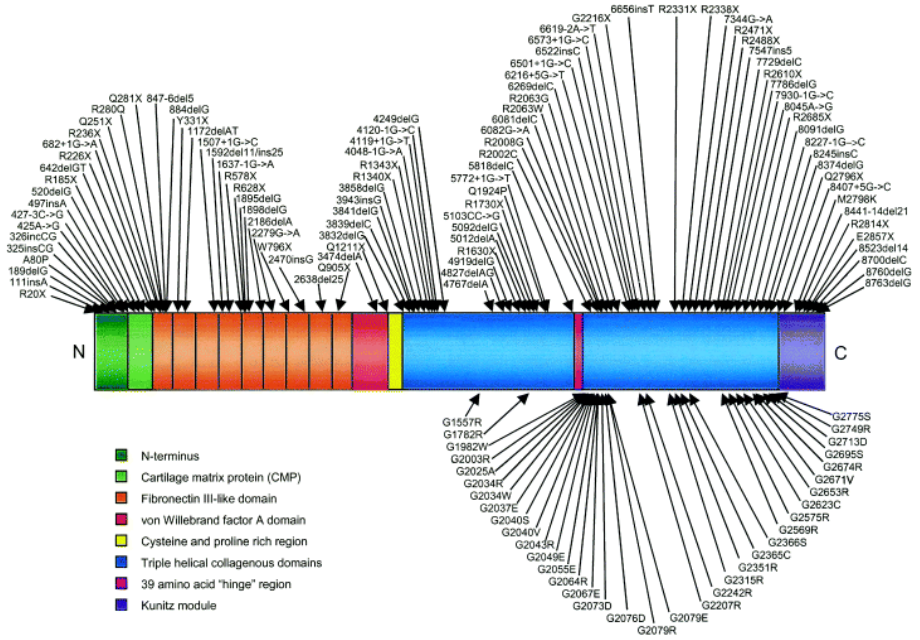


DDEB

RDEB



Human type VII Collagen (Col7A1) Gene



31,132bp at 3p21. 118exons (largest number yet),
8.9kb ORF, 2944 AA, ~300kDa prot.

Unito et al. Journal of Investigative Dermatology (2002) **7**, 6–16

Type	Clinical Manifestations
Dominant dystrophic 	Onset at birth to early infancy Blistering on dorsal hands, elbows, knees, lower legs Milia associated with scarring scar-like lesions, especially on the trunk 80% have nail dystrophy
Recessive dystrophic 	Onset at birth Widespread blistering, scarring, milia Deformities: digital fusion (mitten), joint contractures Severe involvement of mucous membranes, nails Growth retardation, poor nutrition Anemia Mottled, carious teeth Predisposition to squamous cell carcinoma

Dominant Dystrophic EB (DDEB)

- Autosomal dominant
- 3/1,000,000 live births
- Involvement of skin, nails, oral mucosa
- Milder phenotype than RDEB
- Extracutaneous complications rare

- May present in infancy or early childhood
- Flaccid blisters or erosions
- Heal with milia, hypertrophic or atrophic scarring
- Nail dystrophy
- Improve with age

Recessive Dystrophic EB

Dystrophic EB

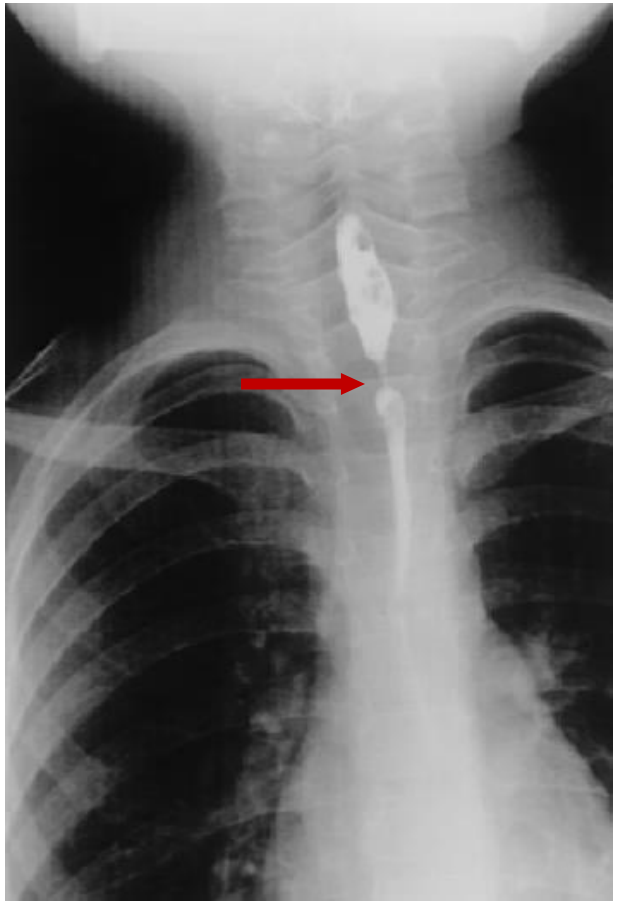
Minor subtypes

- Pretibial (DDEB-Pt)
- Pruriginosa (DDEB-Pr)
- DDEB, nails only
- Bullous dermolysis of the newborn (DEB-BDN)
- Inversa (RDEB-I)
- Centripetalis (RDEB-Ce)
- DEB, autosomal dominant/ autosomal heterozygote

RDEB: Extracutaneous Involvement

- Ocular involvement
- GI: Esophagus, small intestine, anus
  - Strictures
  - Constipation
- GU system, vagina may be involved
- Hematologic: Multifactorial anemia
- Cardiac: Cardiomyopathy
- Growth retardation
- Musculoskeletal: Osteoporosis (wheelchair bound)
- Renal: Glomerulonephritis, IgA nephropathy
- Neurologic: Chronic pain
- Psychosocial: Depression

Esophageal Stricture



Management of EB

- Perinatal care
- Dermatologic care
  - Minimize hyperhidrosis
  - Protective dressing
  - Prevent infection
- Nutrition (protein, Fe, Ca, Vit D, Zn, Se supplement)
- Ophthalmology
- Dental care
- Gastrointestinal care (esophageal stricture)
  - G Tube
  - Dilatation
- Hematology
- Pain/pruritus management
- Orthopedic reconstruction (Osteoprosis)
- PT/OT
- Cancer surveillance
- Social work

Risk of Cancer in EB Patients

- **SCCs** arose primarily in **RDEB**, beginning in adolescence
  - Less frequently, in junctional EB (JEB)

- **SCC deaths** occurred only in **RDEB**, with cumulative risks of 38.7%, 70.0%, and 78.7% by ages 35, 45, and 55, respectively.

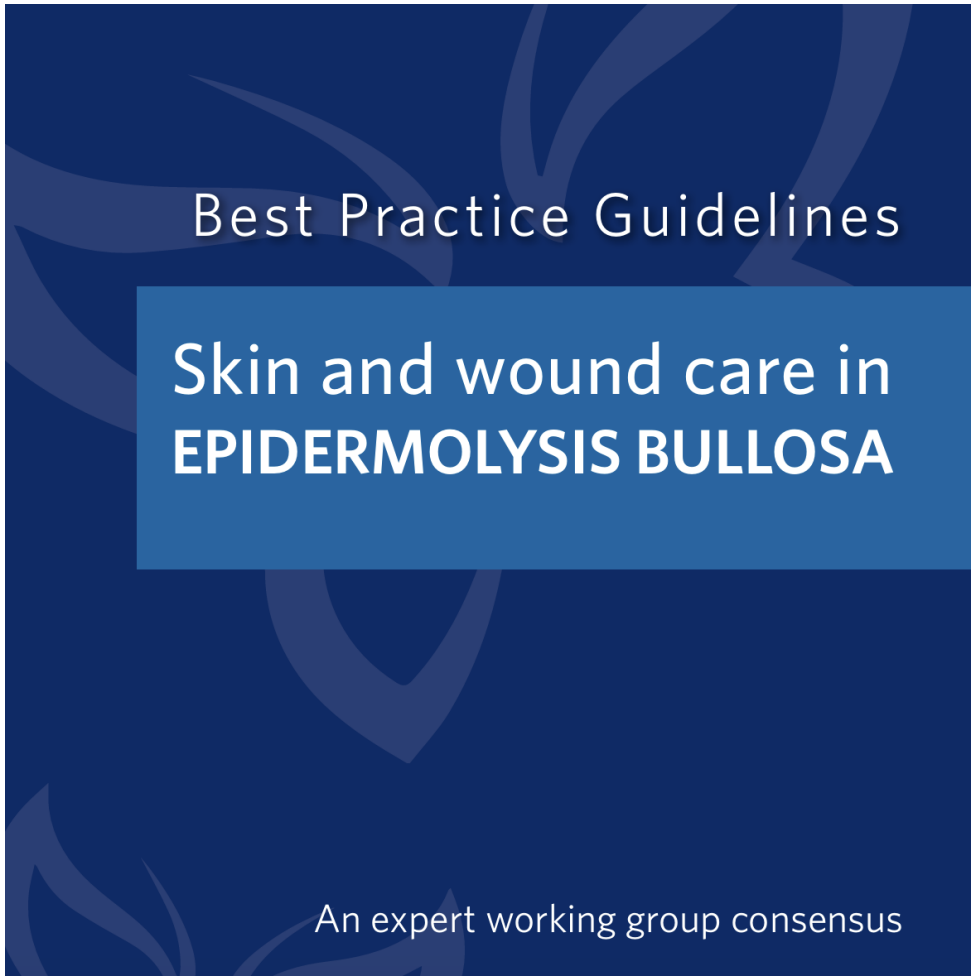
Fine JD, et al. *J Am Acad Dermatol*. 2009;60(2):203-211.

EB Wound Care

Lara Wine Lee, MD, PhD

Elements of EB Wound Care

- Cleansers
- Emollients
- Antimicrobial treatment/prevention
- Wound dressings
- Retention dressings/garments
- Foot care



https://www.debra-international.org/skin-and-wound-care-in-eb-cpg

Wound Care

Images from DEBRA

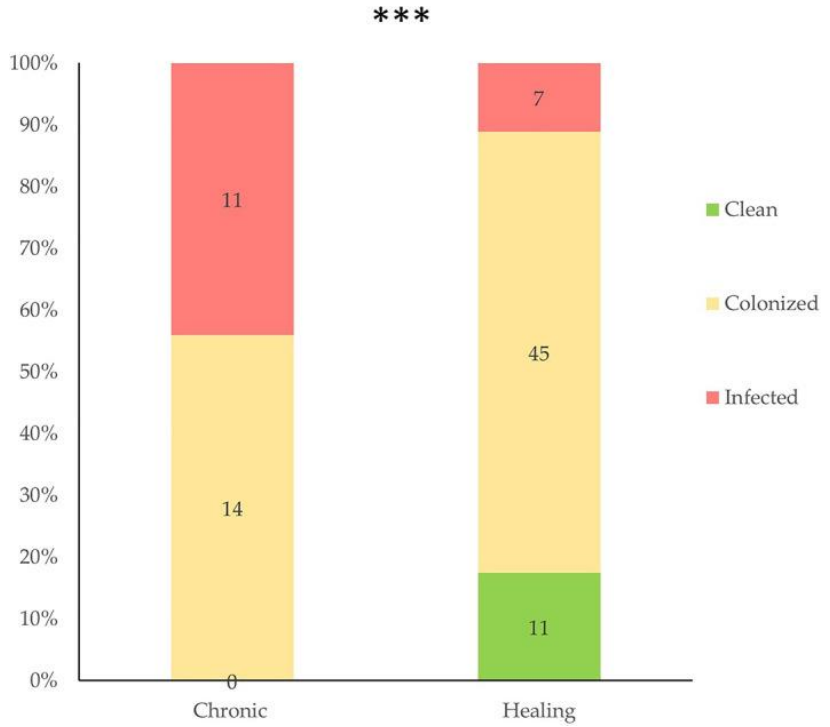
- First layer
  - Contact layer
  - Hydrogel
- Second layer
  - Foams
  - Non-adherent gauze
  - Impregnated gauze
- Retention layer
  - Conforming stretch gauze
  - Tubular elastic net
  - Tubifast
  - Self adherent elastic wraps

Cleansing and Bathing in EB

- Gentle cleansers
- Saline-based sprays for adherent dressings
- Saltwater baths reduce pain
  - 2lbs of salt to 40-gallon tub/half filled bathtub
  - 5 tsp of salt per gallon of bath water used
  - Salt is safe for use with babies and infants
    - Pool salt is inexpensive; table salt may be used
  - Rinsing with clear water after bathing is not necessary
  - Reduces pain, pain medication use, odor, discharge

Infection in EB

- Most common organisms found in EB
  - *Staphylococcus aureus*
    - 23/30 pts had MSSA
    - 2/30 pts had MRSA
  - *Streptococcus* species
  - Diptheroids
  - *Pseudomonas aeruginosa*
  - *Candida albicans*
- 93 (48.4%) wound cultures grew more than 1 type of bacteria
- 26 (13.5%) cultures grew 3 different bacteria
- 14 (7.3%) cultures grew 4 or more bacteria



Fuentes I, et al. *Int Wound J*. 2023;20(3):774-783.

Infection Control

- Bleach or vinegar may be added to bathwater in order to lower the amount of bacteria on the skin
- They may be alternated
- They should not be used together in the same bathwater

Bleach Bath

- ¼ cup bleach in ½ bathtub of water
- Mix 1-2 tsp bleach in 1 gallon of water for compresses
- General recommendations (no established clinical guidelines)
    - 2-4 times/wk, 5-15 min
    - Rinse with water after dilute bleach; otherwise, itching can worsen

Vinegar Baths

- White Vinegar 1:20
    - Useful against gram-negative bacteria, such as *pseudomonas*
    - Useful against yeast
    - Excellent bactericidal activity against *Pseudomonas aeruginosa, Pseudomonas vulgaris, Acinetobacter baumanni*

El Hachem M, et al. *Orphanet J Rare Dis*. 2014;9:76.

Wound Care Resources

Stanford Children's Health | Lucile Packard Children's Hospital Stanford

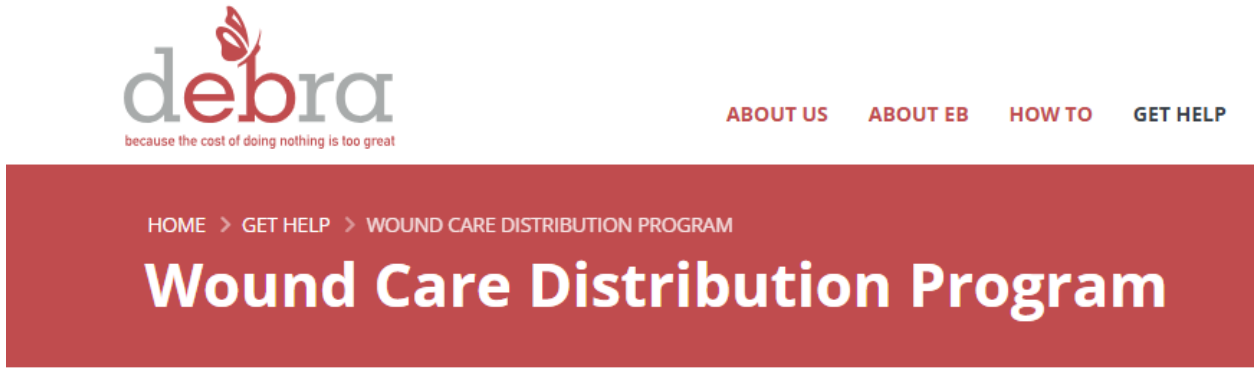
Stanford MEDICINE

Epidermolysis Bullosa Clinic Care Videos and Guides

Wound Care Supplies are often difficult to obtain in adequate quantities

DEBRA EB Nurse

To connect with the debra of America's EB Nurse, please complete the Participant Request Form below. For additional questions, email nurse@debra.org or call 833-DEBRAUS (833-332-7287).



AdaptHealth Patient Care Solutions

Phone: 1-855-532-5463
Email: EBline@adapthealth.com
Website: www.ebadvocate.org

CCS Medical

EB Representative: Jason Clark
Phone: (General) 800-690-1255; (Direct Line) 601-331-8074
Email: Jason.Clark@ccsmed.com
Website: ccsmed.com/products/wound-care-supplies

Connect (CHC Solutions, Inc.)

EB Representative: Nicole Shankle, Director of Special Programs
Phone: 1-888-248-1975
Email: connect@chcsolutions.com
Website: www.connectchc.com
Facebook & Instagram
States Served: AL, DE, KY, MD, NJ, OH, PA, VA, WV

Direct Medical

EB Representatives: Edward Brown and Karen Pauls
Phone: 1-800-659-8037 or 713 869 5585 (direct)
Email: directmedicalinc@gmail.com
Website: www.directmedicalinc.com
Facebook
States Served: CO, GA, IA, NE, NM, OK, OR, SD, TX

Other

Edgepark Medical Supplies: 800-321-0591

FDA-Approved Treatments

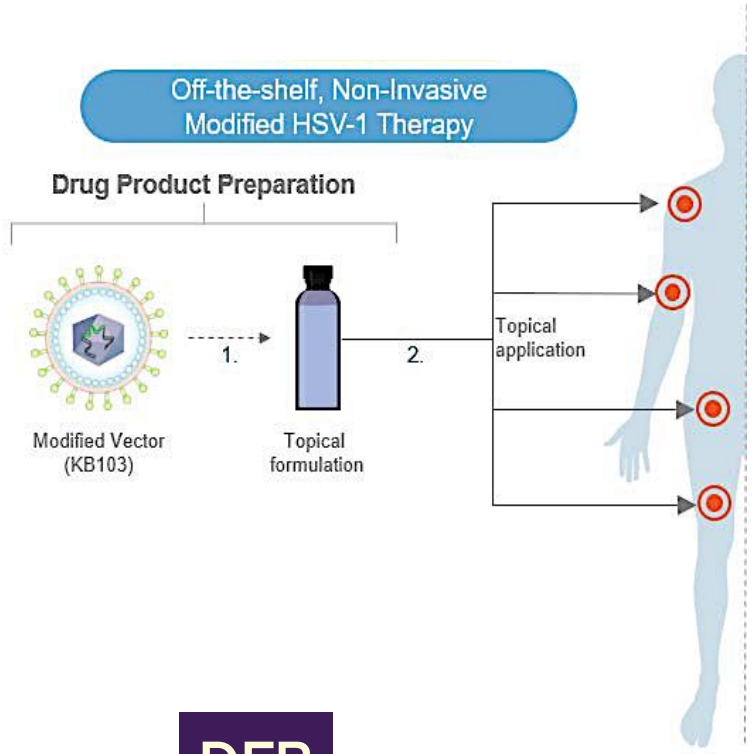
Joyce M C Teng, MD, PhD

Wound Care Considerations in EB: Every Patient is Different

- Age
- Characteristics
  - Location
  - Area of body
  - Placement of gastrostomy tube
  - Acute vs chronic: How long have the wounds been there?
  - Infection
  - Size
  - Depth
- Severity
  - EB subtype
  - Body surface area involvement
- Long term sequalae: Complications?

Recent FDA-Approved Therapy for DEB >6mo

Gene Therapy (Vyjuvek)



DEB

Filsuvez® (birch triterpenes) (Oleogel-S10)

TOPICAL GEL FORMULATION WITH
MULTIPLE BIRCH TRITERPENES

A non-aqueous gel with thixotropic properties containing birch triterpenes from *Betulae* cortex consisting of betulin, betulinic acid, lupeol, oleanolic acid, and erythrodiol

DEB JEB

Filsuvez

Indication

FILSUVEZ is indicated for the treatment of wounds associated with dystrophic and junctional epidermolysis bullosa (EB) in adult and pediatric patients 6 months of age and older.

Warnings & Precautions

Local hypersensitivity and skin reactions have been reported in patients treated with FILSUVEZ, including urticaria and dermatitis. If signs or symptoms of hypersensitivity occur, discontinue use immediately and initiate appropriate therapy.

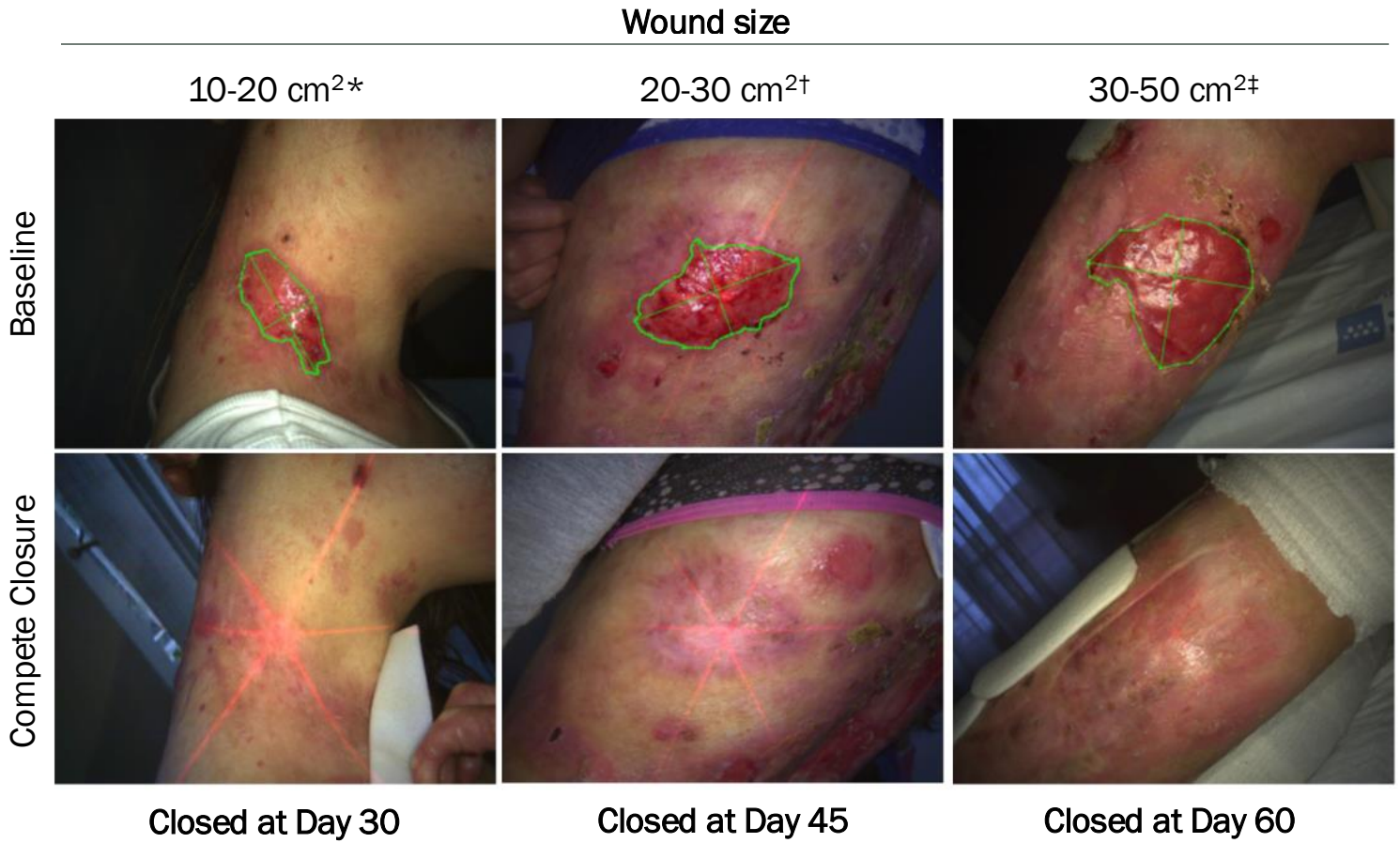
Adverse Reactions

The most commonly reported adverse reaction in clinical trials was pruritus and pain at the wound application site (7.3%).

Patient Counseling Information: Please refer to Prescribing Information for administration instructions.

To report SUSPECTED ADVERSE REACTIONS, contact Amryt Pharmaceuticals DAC at 1-855-303-2347 or FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

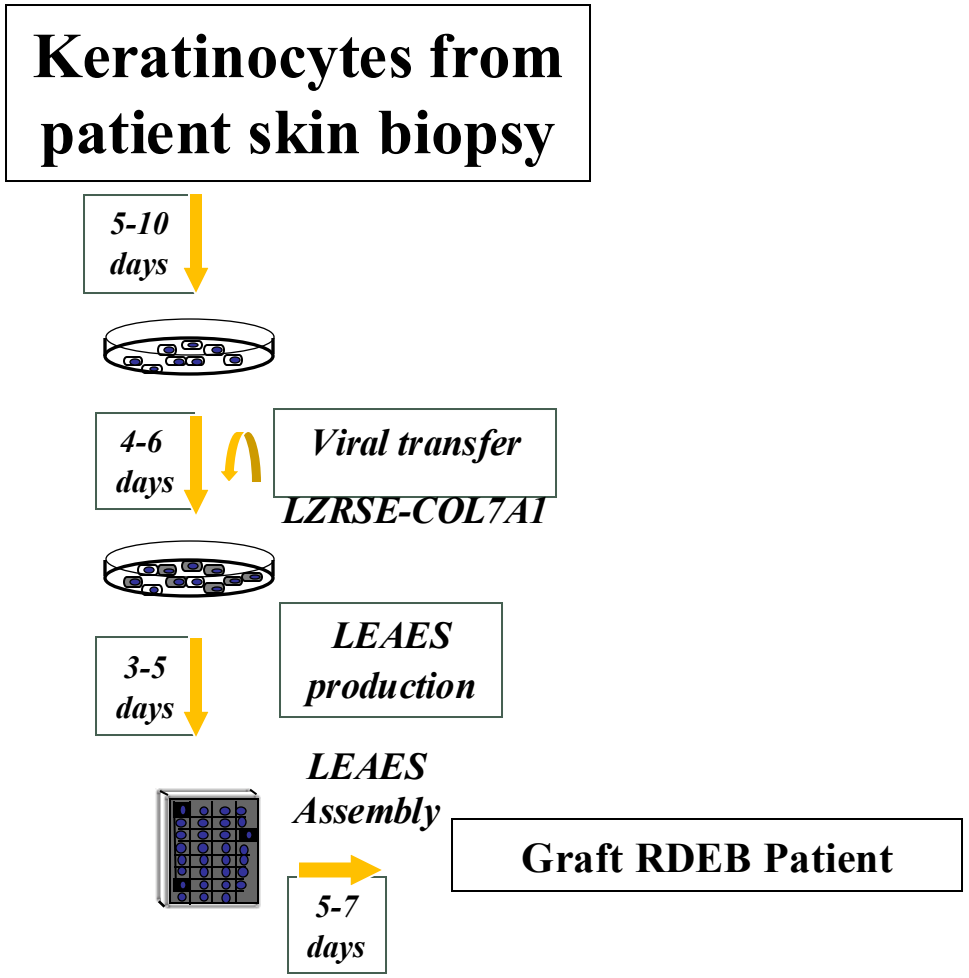
How Target Wounds Were Measured in EASE



*Target wound size 10.0 cm^2, wound age 25 days, neck, right lateral (RDEB generalized severe, 14yo)
†Target wound size 20.5 cm^2, wound age 180 days, left back, lumbar region (RDEB generalized intermediate, 23yo)
‡Target wound size 30.3 cm^2, wound age 40 days, lower left leg, lateral (RDEB generalized severe, 18yo)

RDEB = recessive dystrophic epidermolysis bullosa.
Kern JS, et al. *Br J Dermatol*, 2023;188:12-21.

Genetically Correct Autologous Skin Graft



ARTICLE

doi:10.1038/nature24487

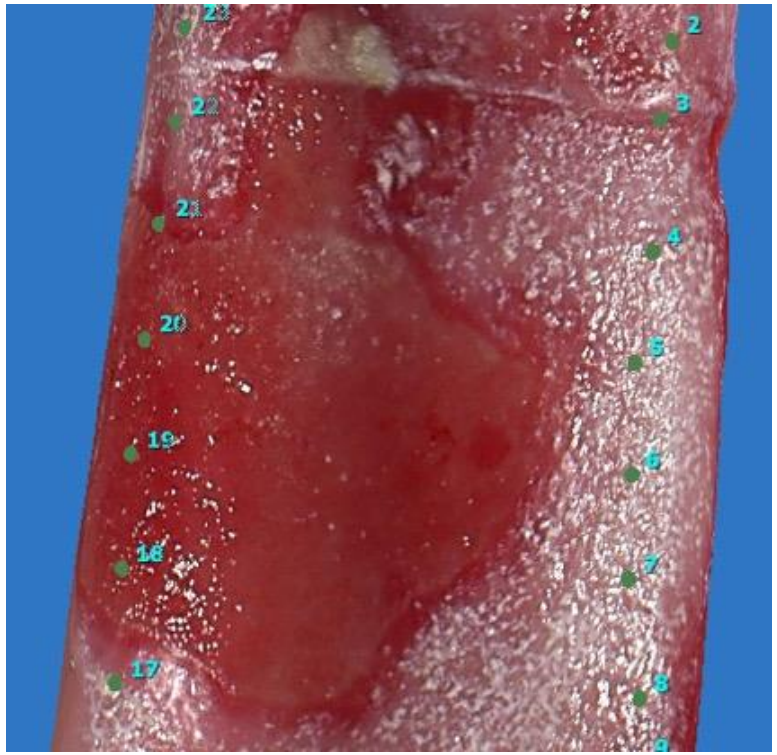
Regeneration of the entire human epidermis using transgenic stem cells

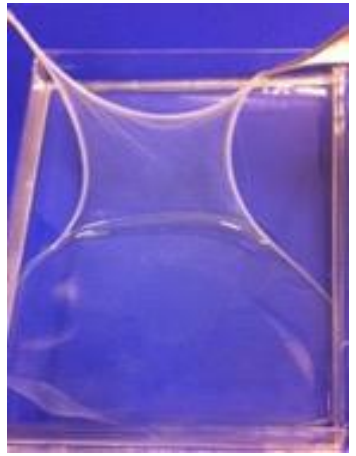
Tobias Hirsch[1]*, Tobias Rothoeft[2]*, Norbert Teig[2]*, Johann W. Bauer[3]*, Graziella Pellegrini[4,5]*, Laura De Rosa[5]*, Davide Scaglione[6], Julia Reichelt[3], Alfred Klausegger[3], Daniela Kneisz[3], Oriana Romano[7], Alessia Secone Seconetti[5], Roberta Contin[5], Elena Enzo[5], Irena Jurman[8], Sonia Carulli[9], Frank Jacobsen[1], Thomas Luecke[10], Marcus Lehnhardt[1], Meike Fischer[2], Maximilian Kueckelhaus[1], Daniela Quaglino[7], Michele Morgante[8], Silvio Bicciato[7], Sergio Bondanza[9] & Michele De Luca[5]

Hirsch, et al. *Nature*. 2017.

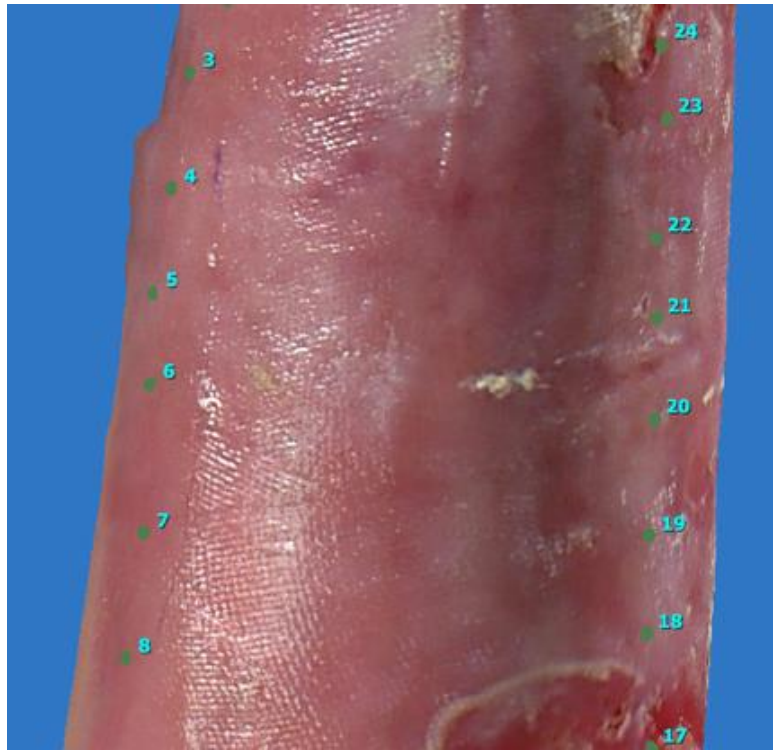
Results: 21 of 24 Wounds Healed after 6mos

Baseline





6mos

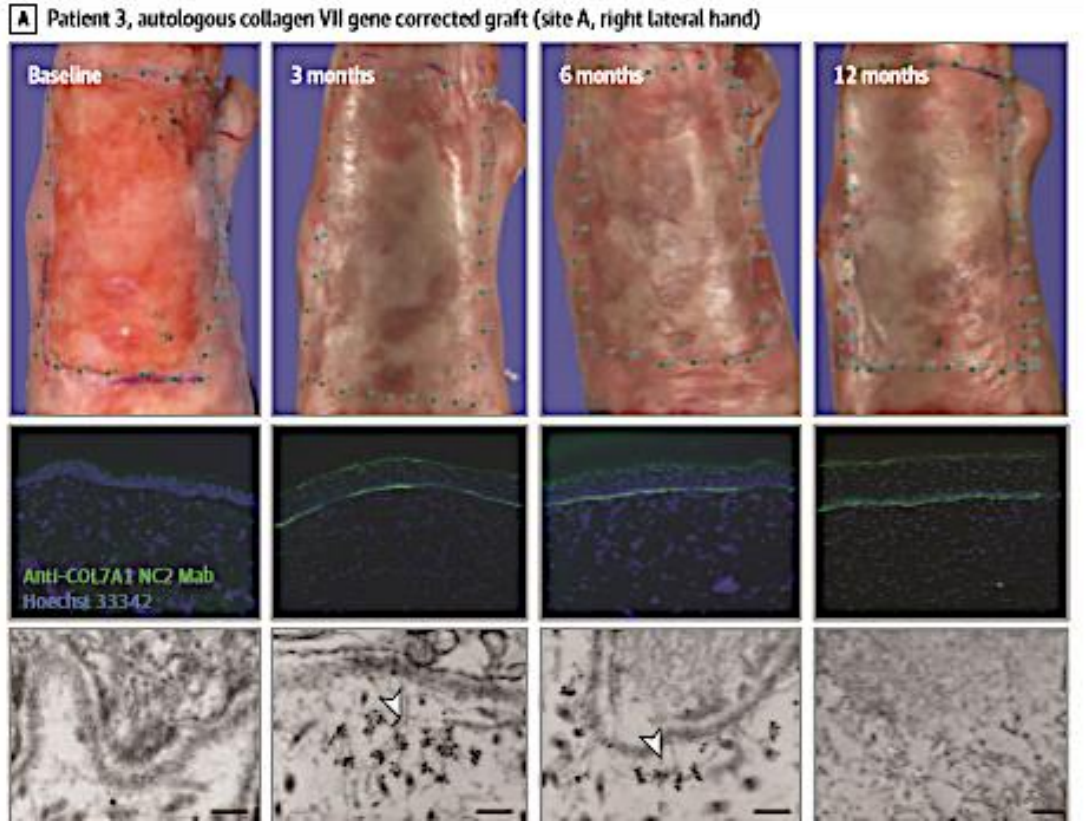


JAMA | Preliminary Communication

Safety and Wound Outcomes Following Genetically Corrected Autologous Epidermal Grafts in Patients With Recessive Dystrophic Epidermolysis Bullosa

Zurab Siprashvili, PhD; Ngon T. Nguyen, BS; Emily S. Gorell, MS; Kylie Loutit, BS; Phuong Khuu, MD; Louise K. Furukawa, MD; H. Peter Lorenz, MD; Thomas H. Leung, MD, PhD; Douglas R. Keene, BS; Kerri E. Rieger, MD, PhD; Paul Khavari, MD, PhD; Alfred T. Lane, MD, MA; Jean Y. Tang, MD, PhD; M. Peter Marinkovich, MD

Figure 2. Clinical and Microscopic Analysis of Autologous Collagen VII Gene-Corrected Grafts in Patients 3 and 4 and Untreated Wounds From Patient 4



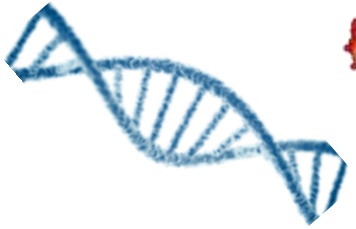
Siprashvili, et al. *JAMA*. 2017.

Pending FDA Approval in April

Abeona Receives FDA Regenerative Medicine Advanced Therapy Designation for EB-101 Gene Therapy in Epidermolysis Bullosa

Gene Therapy and Pharmacological Approaches for EB

DNA

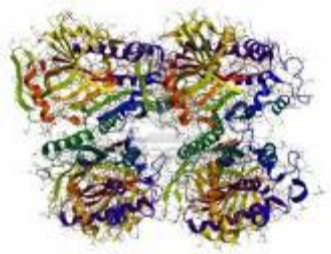


1. cDNA addition
2. Gene editing

RNA

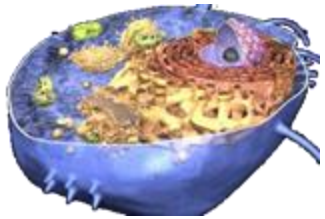
3. Exon skipping
4. Transplicing
5. siRNA

Protein



6. Protein replacement

Cells



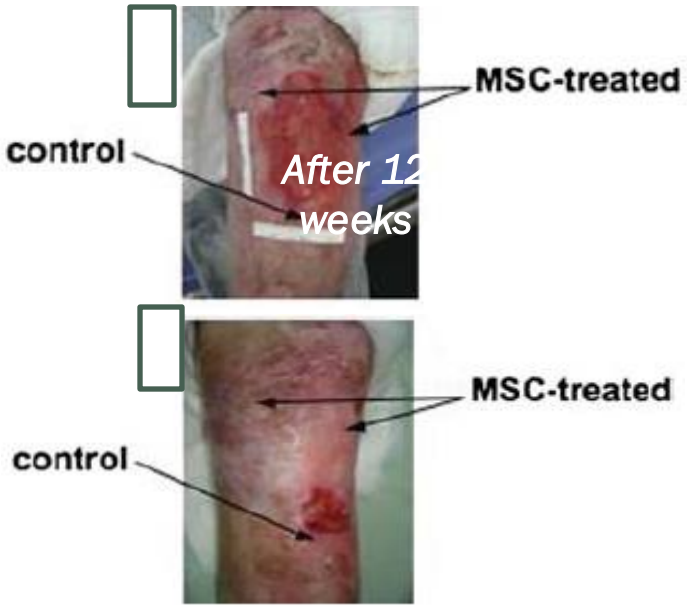
7. Fibroblast injection
8. BM transplantation
9. MSC injection
10. Revertant cells
11. iPSC

Drugs

12. Diacerin
13. Gentamicin
14. Losartan
15. rhHMGB1
16. Apremilast
17. Oleagel

Allogeneic BM-MSC Lead to Transient Clinical Improvement of RDEB

Intradermal injections



→ *injection of 0.5.10⁶ hBM-MSC/site*

➢ *4 months*

Conget, et al. *Cytotherapy.* 2010.

Intravenous infusions

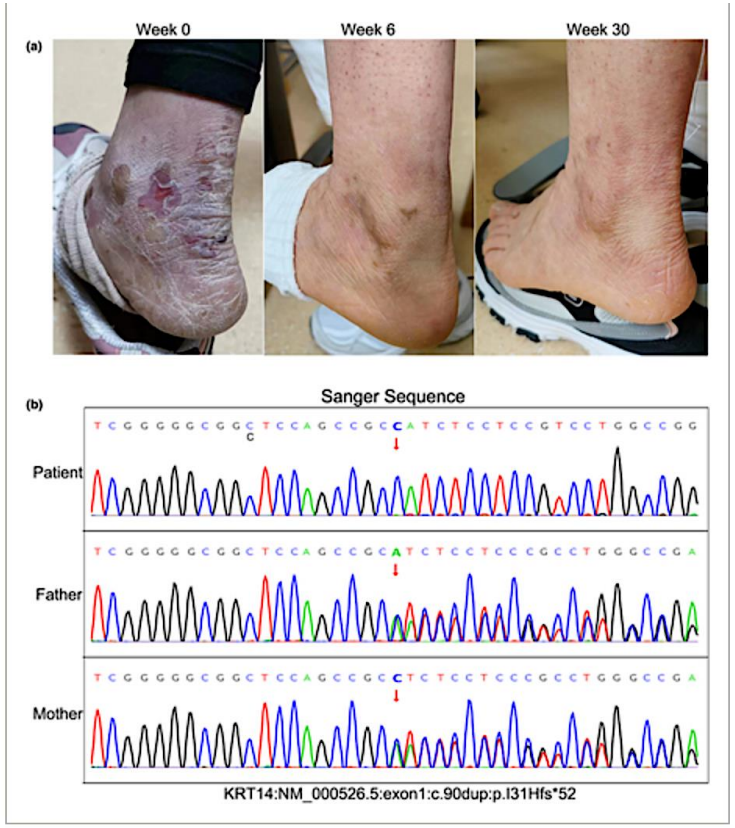
→ *3 infusions of 3x10⁶ hBM-MSC/kg*

➢ *6 months*

Petrof, et al. BJD. 2015

Treatment of epidermolysis bullosa simplex with dupilumab

Sun X, et al. *J Dermatol.* 2024;51(4):e131-e132.

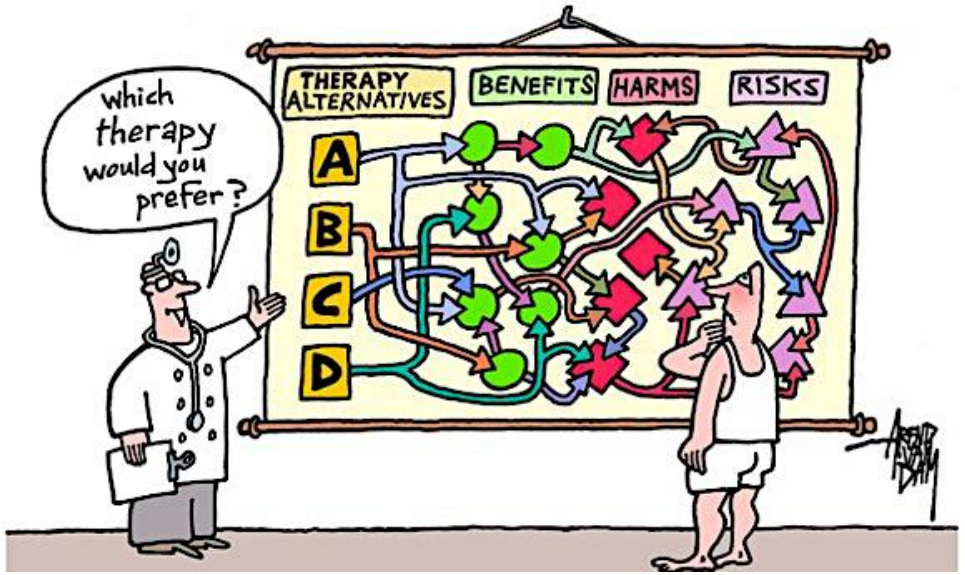


RDEB Treated with Dupixent

Combination/Comprehensive Therapies Will Probably Be Needed in the Future

- **Gene therapy**
- **Protein therapy**
- **Cell therapy**

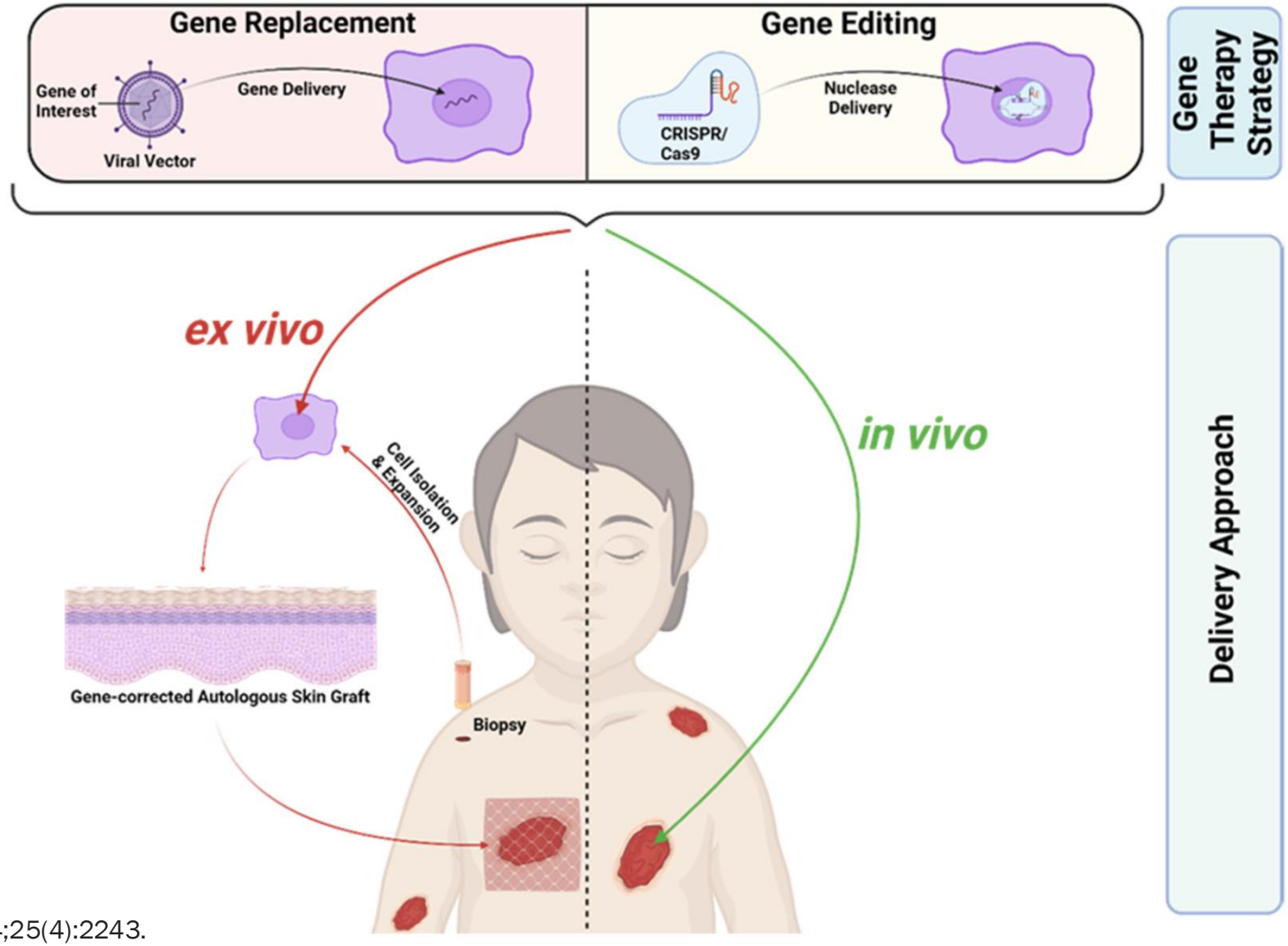
- Repurpose drugs to reduce impact of the faulty protein or manage disease aspects, such as inflammation
  - Botox
  - Dupixent
  - Dapsone
  - Apremilast
  - Sirolimus
  - Erlotinib?



Emerging Therapies

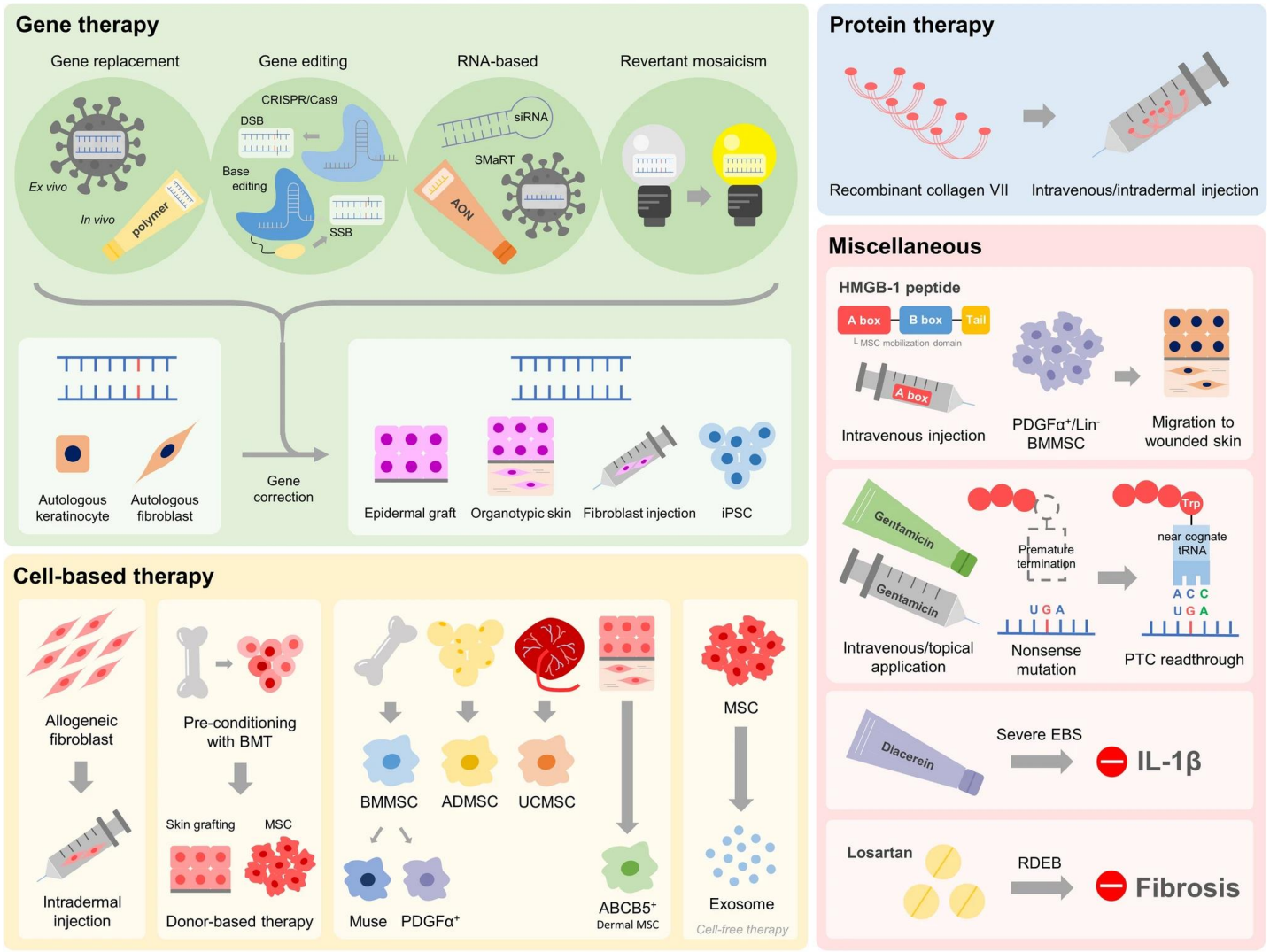
Lara Wine Lee, MD, PhD

Emerging Therapeutics in EB



Bischof J, et al. *Int J Mol Sci*. 2024;25(4):2243.

Therapeutic Pipeline



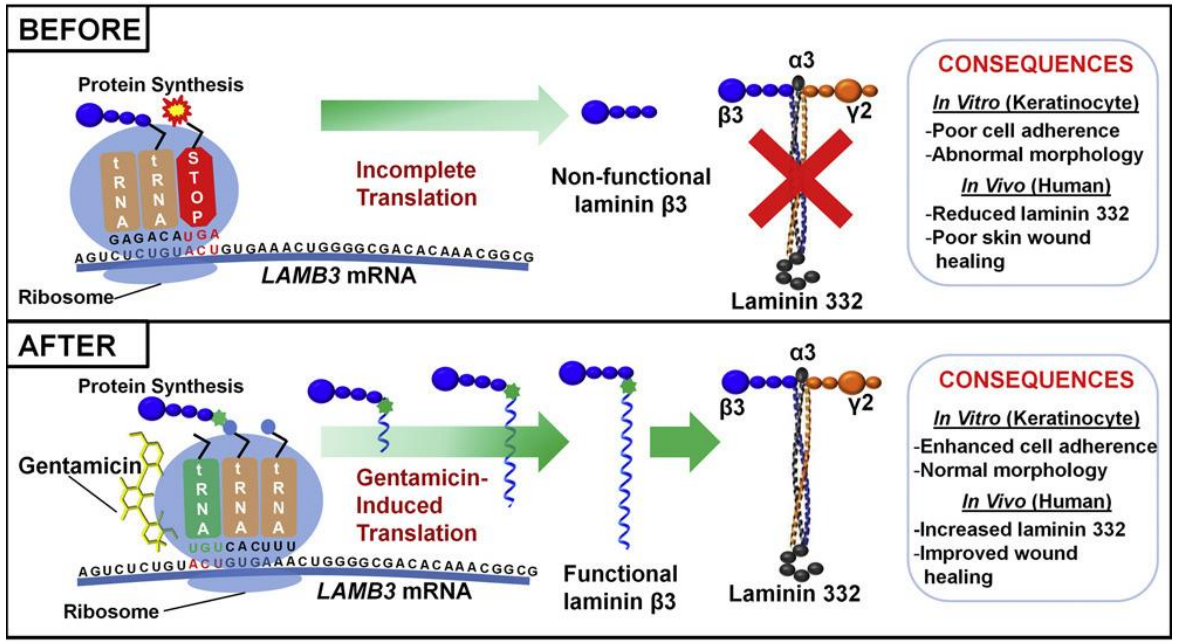
Hou PC, et al. *Am J Clin Dermatol.* 2021;22:801–817.

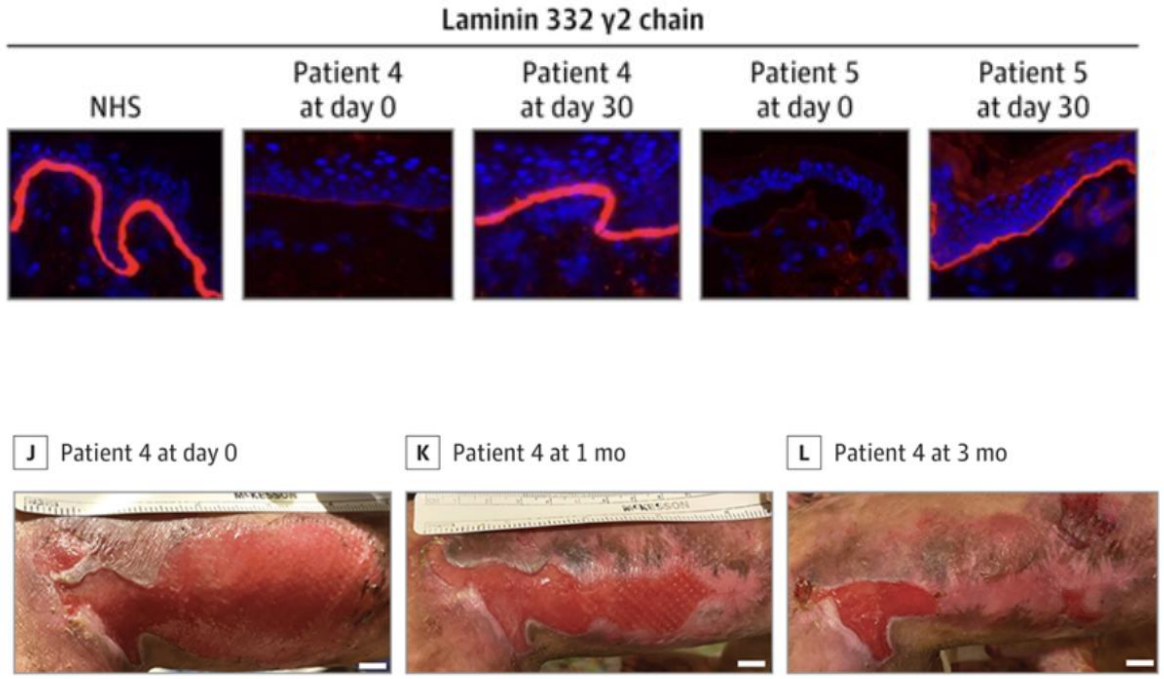
Therapeutic Potential in JEB

EB Type	Gene	Strategy	Phase	Status	Sponsor	NCT ID
JEB	LAMB3	Transplantation of autologous gene-corrected epidermal sheets	Phase I/II	Completed	University of Modena and Reggio Emilia, Modena, Italy	Not listed [39]
JEB	LAMB3	Transplantation of autologous gene-corrected epidermal sheets	Phase I/II	Completed	University of Modena and Reggio Emilia, Modena, Italy	Not listed [40]
JEB	LAMB3	Transplantation of autologous gene-corrected epidermal sheets	Phase I/II	Completed	University Hospital of the Paracelsus Medical University	Case study, not listed [41]
JEB	LAMB3	Transplantation of autologous gene-corrected epidermal sheets	Phase II/III	Recruiting	Holostem Terapie Avanzate s.r.l., Modena, Italy	NCT05111600
JEB	COL17A1	Transplantation of autologous gene-corrected epidermal sheets	Phase I/II	Terminated (no patient ongoing/completed)	Holostem Terapie Avanzate s.r.l., Modena, Italy	NCT03490331

Bischof J, et al. *Int J Mol Sci*. 2024;25(4):2243.

Aminoglycosides Induce Readthrough of PTC



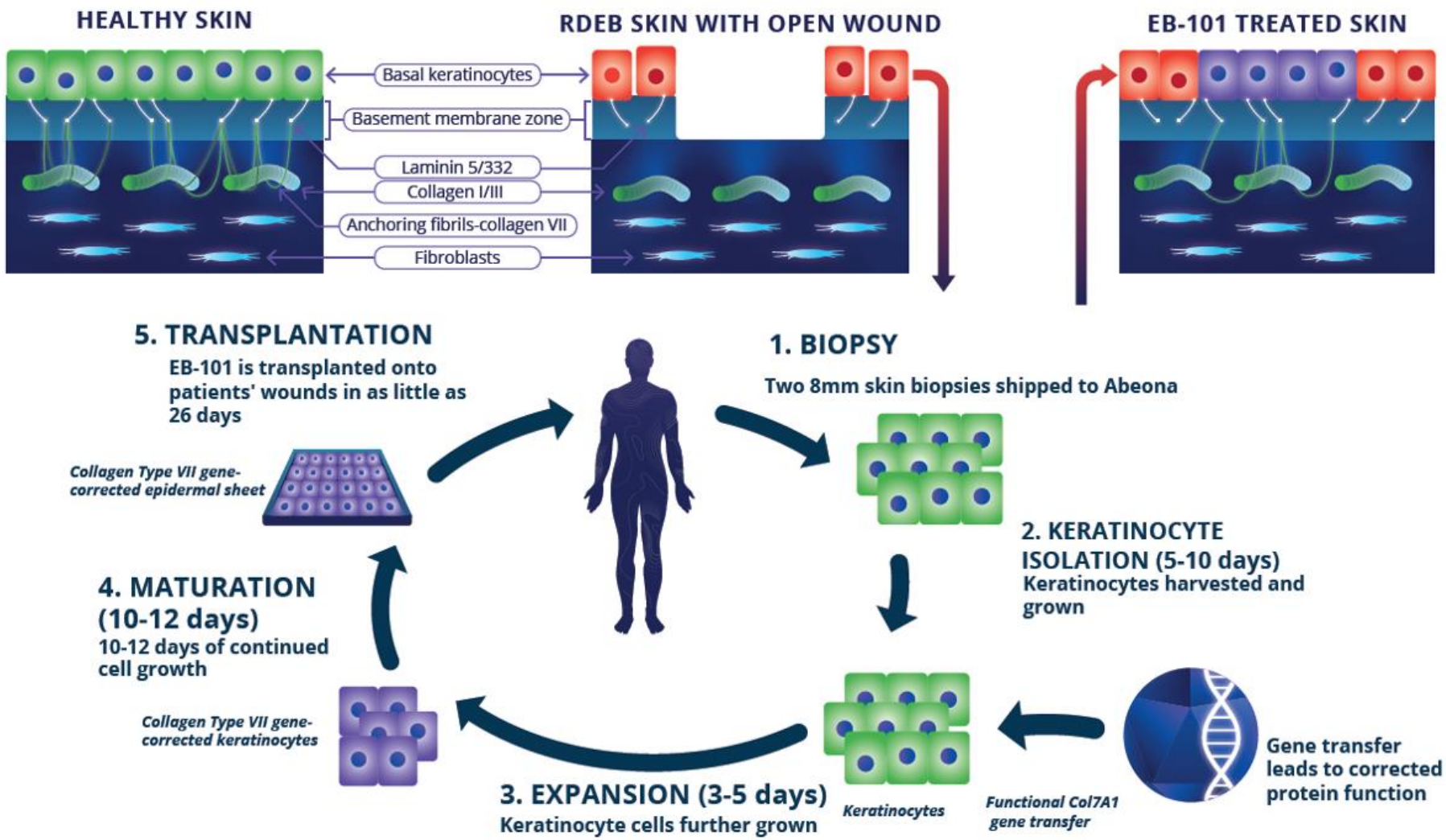


Mosallaei D, et al. *JAMA Dermatol.* 2022;158(4):366–374. Kwong A, et al. *Molecular Therapy*. 2020;28(5):1327-1338.

Therapeutic Potential in DEB

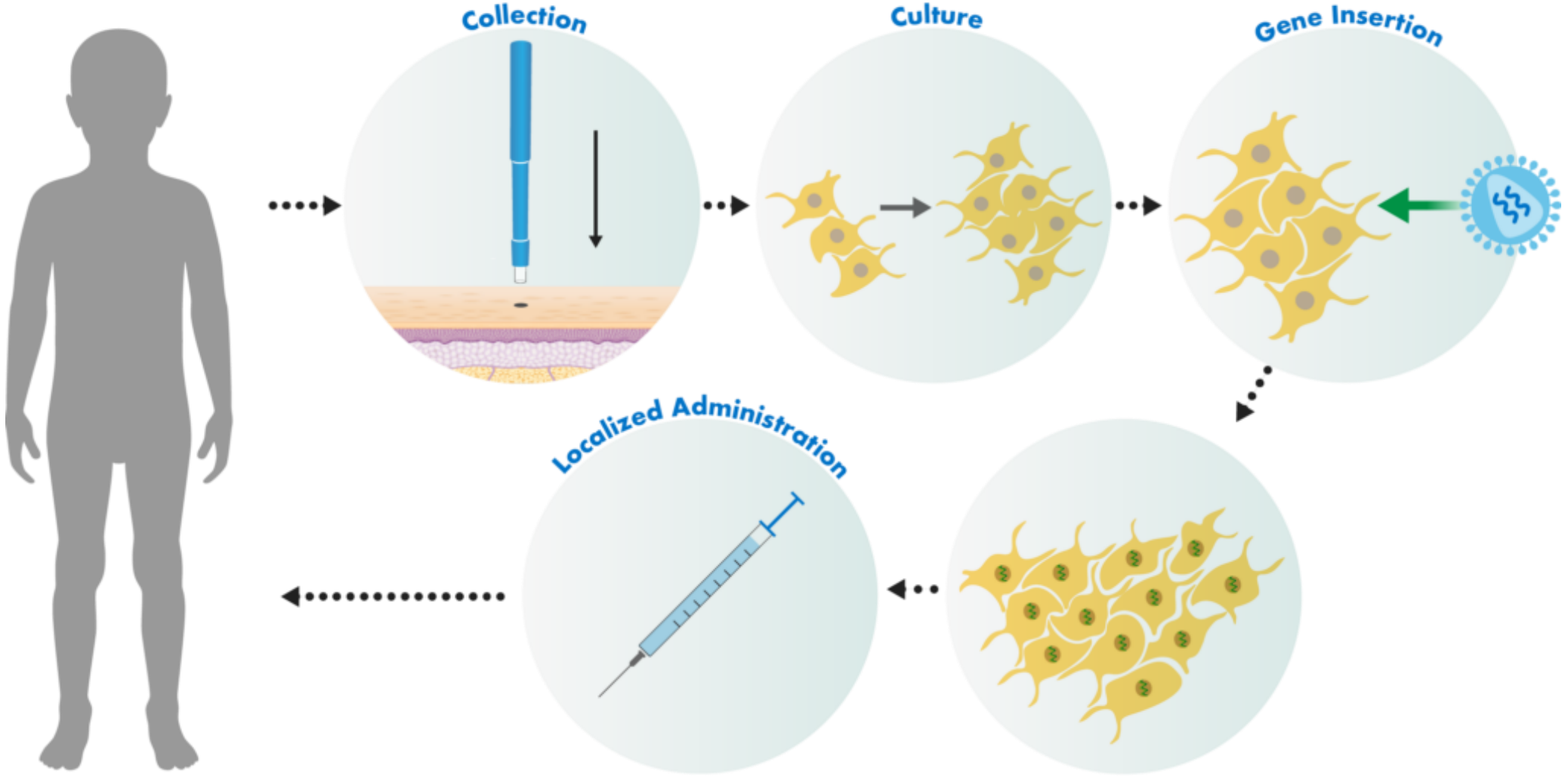
EB Type	Gene	Strategy	Phase	Status	Sponsor	NCT ID
RDEB	COL7A1	Transplantation of autologous gene-corrected epidermal sheets	Phase I/II	Completed	Abeona Therapeutics, Inc., Cleveland, OH, USA	NCT01263379
RDEB	COL7A1	Transplantation of autologous gene-corrected epidermal sheets	Phase III	Completed	Abeona Therapeutics, Inc., Cleveland, OH, USA	NCT04227106
RDEB	COL7A1	Transplantation of autologous gene-corrected epidermal sheets	Phase III	Recruiting	Abeona Therapeutics, Inc., Cleveland, OH, USA	NCT05725018
RDEB	COL7A1	Intradermal injection of autologous gene-corrected fibroblasts	Phase I	Completed	King's College, London, UK	NCT02493816
RDEB	COL7A1	Intradermal injection of autologous gene-corrected fibroblasts	Phase III	Active, not recruiting	Castle Creek Biosciences, LLC., Exton, PA, USA	NCT04213261
RDEB	COL7A1	Transplantation of autologous gene-corrected keratinocyte sheets	Phase I/II	Active, not recruiting	Institut National de la Santé Et de la Recherche Médicale, Paris, France	NCT04186650

Gene-Corrected Epidermal Sheets



https://www.abeonatherapeutics.com/clinical-trials/rdeb

Intradermal Gene-Corrected Fibroblasts



https://castlecreekbio.com/technology/autologous-fibroblast-technology/

Epidermolysis Bullosa Resources

debra
for people whose skin doesn't work we do

CAMP DISCOVERY
AMERICAN ACADEMY OF DERMATOLOGY

Childhood without limits
Camp Wonder