

GLOSSARY OF TERMS USED IN EB RESEARCH (15Nov23)

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~	Approximately
<	Less than - for example 2 < 5
>	More than - for example 5 > 2
Acute	Acute symptoms began recently and will go away quickly (compare: chronic)
Anchoring fibrils	Sticky protein threads that hold cells together
Allele	Either the maternal (from mother) or paternal (from father) version of a gene - a gene variant
Allogeneic	Transplanted cells or tissue where the donor and recipient are not genetically identical (compare: syngeneic)
Ameliorate	To make better or more tolerable
Amino acid	20 different small molecules called amino acids are linked together to make proteins
Anaemia	Low iron. Reduction of the number of red blood cells or the amount of iron within them
Animal model	An animal that potential treatments can be tested on before trying them out in people
Antibiotic	A substance that stops bacteria from multiplying/spreading - used to treat a bacterial infection
Atopic dermatitis/atopic eczema	Non-contagious, long-term inflammation of the skin
Autochthonous	Occurring in its original place: autochthonous mouse models develop cancer without having cancerous cells transplanted into them
Autograft	Transplanted cells or tissue from one part of the body to another within the same person or animal
Bacteria	Single celled living creatures that can be harmless or cause disease
Basement membrane	Sticky protein layer between outer (epidermis) and inner (dermis) cells of our skin
Basic research	Contributes to understanding/knowledge
Behçet's disease	Disease affecting skin and eyes probably caused by the immune system mistakenly attacking blood vessels
Biochemist	Person specialising in the chemistry that happens inside living creatures
Biofilm	Germs living in a layer of slimy substance they produce to protect themselves
Bisected	Cut in half
Blind trial/study	A trial is single-blind if the patients don't know whether they are getting the test drug or not and double-blind when their doctors don't know either.
Blinding	Blinding is the process of making a clinical trial more accurate by concealing whether the treatment is being received or not (compare: open)
Carcinogenesis	Tumourigenesis. Oncogenesis. The formation of a tumour or cancer. How cells become cancerous.

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Carcinoma	Cancer that starts in epithelial cells
Carrier	A carrier is someone who has inherited a disease-causing genetic change and can pass it on but does not have any symptoms themselves
Cell	Tiny living part of our bodies that carry out many different roles and functions and consist of cytoplasm (goo) inside a cell membrane
Cell line	Cells growing in a laboratory that will continue to multiply indefinitely
Challenge - dechallenge - rechallenge	Trial where patients receive a treatment then stop it then start it again so that changes in symptoms while being treated can be compared to symptoms when untreated
Characterization	Understanding more about something
Chemokine	A type of small protein molecule (cytokine) that attracts immune cells. A chemotactic cytokine.
Chromosome	A very long DNA molecule that can roll/fold up until it is visible under a microscope - we have 23 pairs of chromosomes in each cell
Chronic	Chronic symptoms are long-lasting, ongoing, persistent (compare: acute)
Circulating	Carried around the body in the bloodstream
Clinician	A doctor who works with people
Collagen	Skin protein made from three twisted protein chains - also involved in scars - multiple numbered types are made from different genes eg COL7A1 (DEB) and COL17A1 (JEB)
COL17A1	Gene encoding the alpha 1 chain of type-17 collagen - changes in this gene can cause junctional EB
COL7A1	Gene encoding the alpha 1 chain of type-7 collagen - changes in this gene can cause dystrophic EB
Conceptus	Early embryo
Contagious	Catching. A disease that can be passed from person to person due to a germ (pathogenic microorganism) travelling between them
Conventional	What is normally done or believed
Corticosteroids	Anti-inflammatory medicine. Tablets may delay wound healing and make infections more severe
CRISPR/Cas9	A type of gene editing
Cutaneous	Involving skin
Cytokine	A small protein molecule in the immune system that tells cells how to behave - there are many different types
Cytoplasm	The liquid inside a cell that contains various proteins
C7 expression	Production of collagen-7 protein
<i>De novo</i>	Occurring for the first time or as a new event
DEB	Dystrophic EB - due to changes in gene COL7A1 encoding collagen protein
Deleterious	Harmful
Denaturation	Breaking up the structure of protein molecules
Dermal	Involving the dermis (inner/lower layer of skin)
Dermatologist	Doctor specialising in skin

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Dermis	Inner/lower layer of our skin
Differentiation	Process by which cells acquire specialised features (eg turning from a stem cell into a skin cell)
DNA	Deoxyribonucleic acid is a long molecule made of linked 'bases' represented as A, C, T or G
Dominant	Dominant inheritance means the genetic change always causes symptoms - there are no unaffected 'carriers' (compare: recessive)
Dominant KRT5/14 EBS-sev	EBS of a severe nature caused by changes to genes encoding keratin-5 or keratin-14 and inherited in a dominant fashion
Drug repurposing	Using drugs that are already working for other patients to treat EB symptoms
Drug treatment scheduling	When to take a drug, how much and how often
Dupilumab	Prescription medicine for dermatitis - works by blocking the action of interleukins 4 and 13
Dysfunction	Not working properly or as expected
Dysregulation	Not being regulated properly or as expected - for example, cancer cells do not stop replicating and die as they should
Dystrophy	Weakening, degeneration, loss of functionality, abnormal development
EB/epidermolysis bullosa	The name comes from 'epiderm' - the outer layer of skin, 'lysis' - the breakdown of cells and 'bullosa' - blisters.
EBA	Epidermolysis bullosa acquisita - acquired autoimmune EB, not inherited but due to mistaken attacks by the immune system
EBS	Epidermolysis bullosa simplex - due to changes in genes KRT5 and KRT14 encoding keratin protein
Efficacy	How well something works - how effective it is
Electroporation	A pulse of electricity applied to cells in a tube to make their outer membrane permeable to DNA. Used to get new genes into a cell.
EMA	European Medicines Agency
Endogenous	Coming from inside the body
Epidermal	Involving the outer/top layer of our skin
Epidermis	Outer/top layer of our skin made mostly of keratinocytes and keratin
Epithelial	Refers to the surface layer of body parts including internal organs and linings
Euthanized	Humanely killed - applied to animals in research
Exogenous	Coming from outside the body
Exon	Part of a gene that codes for part of a protein
Expression	When a protein is actually made from the genetic instructions, the gene is 'switched on' and expressing protein
Extracellular matrix (ECM)	The protein 'mortar' that glues cells together to form a tissue
FDA	US Food and Drug Administration
FERMT1 / KIND1	Kindlin-1 protein is also called FERMT1 and is involved in cell communication (integrin signalling) and holding cells in place within the ECM
Fibril	A small fibre or filament
Fibroblast	Cells that produce collagen following injury

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Fibrocyte	Cell that will develop into a fibroblast
Fibrosis	Scarring; production of scar tissue that is thicker and less stretchy than the original tissue
Gene	Genetic code that can be translated into a protein
Gene editing	Changing the DNA sequence of a gene within a genome
Genetic code	Every three 'letters' of DNA translate into a different amino acid or say stop/start
Genodermatoses	Inherited skin conditions
Genome	All the genetic information in a person or species
Genotype	Type of gene variant(s) of an individual
Heterogeneous/heterogeneity	Showing a degree of difference
Heterozygous	When the versions of a gene inherited from mother and father are different
Homogeneous/(homogenous)	Showing similarity
Homozygous	When the versions of a gene inherited from mother and father are both the same
Hypomorphic	A genetic change that causes only a partial loss of function of the encoded protein
Hypothesis	An idea that can be tested to see if it is correct
Immunological	To do with the immune system
Immunomodulatory	Describing a treatment that changes how our immune system behaves
Immunopathology	Disease due to immune responses
<i>In vitro</i>	Carried out in cells that are grown outside the body in bottles or dishes in a laboratory
<i>In vivo</i>	Carried out inside the body of an animal or person
Inflammation	Increase of blood flow and immune factors to part of the body
Integrin	Type of protein that sticks through the outer membrane of a cell to hold it in place and communicate changes between the outside and inside of the cell
IFN- γ	Interferon gamma: a protein important in the immune system
Interleukin (IL)	Interleukins are types of cytokine (signalling protein) produced by cells - lots of different types are given numbers eg IL-17
Intervention	A treatment that is introduced to try to reduce symptoms
Intraperitoneal (IP)	Injected into the peritoneal cavity - the space surrounding our guts and internal organs
Intravenous (IV)	Injected into a vein
Intron	Part of a gene that doesn't code for part of a protein
Isograft	Transplanted cells or tissue where the donor and recipient are genetically identical
JEB	Junctional EB - due to changes in COL17A1, LAMB3, LAMC2, LAMA3 genes encoding collagen or laminin proteins
kb	kilobases: 1000 'letters' of DNA sequence (ATTCGTCCTGGATCTGATCGGGCTGTC...)

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KEB	Kindler EB/Kindler syndrome - due to changes in KIND1/FERMT1 gene encoding Kindlin-1 protein
Keratin	Protein making up the top layer of our skin - multiple numbered types are made from different genes eg KRT5 or KRT14 (EBS)
Keratinocyte	A cell in the top layer of our skin - also called a squamous cell - that produces the protein keratin
Kinase inhibitor	Kinases are enzymes (type of protein) that change how other proteins work. Kinase inhibitors are small molecules that interfere with this.
KIND1 / FERMT1	Gene encoding Kindlin-1 protein. Changes to this gene can result in KEB
Knockout/loss of function	A genetic change that completely stops a working protein from being produced
KRT5	Gene encoding type-5 keratin (keratin-5)
KRT14	Gene encoding type-14 keratin (keratin-14)
LAMA3	Gene encoding laminin protein alpha-3 chain
LAMB3	Gene encoding laminin protein beta-3 chain
LAMC2	Gene encoding laminin protein gamma-2 chain
Laminin	A protein made from three protein chains twisted together that helps to glue cells together
LNP	Lipid-based nanoparticle: tiny fat ball that can contain and deliver therapeutic substances safely into cells
Macrophage	White blood cells that engulf germs or dead/damaged/cancerous cells and cause immune responses
Malnutrition	Illness from getting too many or too few nutrients
Mean	The average value in a group when all values are added then divided by the number of individuals in the group
Mechanism	How something happens
Median	The value that falls halfway between the smallest and largest value in a group of measurements
Metabolome	All the small-molecule chemicals found within a biological sample
Metastasis	Cancer spreading to other parts of the body
Microbiome	All the microorganisms living on or in a person (or any specific defined place)
Microenvironment	The proteins and molecular interactions in or around cells
Microflora	Microflora
Microorganism	Living creature only visible under a microscope - bacteria, viruses, yeast etc
Mode	The most common (most frequently occurring) value in a group of measurements
Model	An animal or laboratory-grown cells that can be used to test treatments before trying them out in people
Monoclonal antibody (mAb)	Antibodies to one specific protein - created in a laboratory cell line rather than extracted from blood (compare: polyclonal antibody)
Monocyte	White blood cells that can turn into macrophages, fibrocytes and other cell types
Monogenic	Due to a change in one gene only - a 'single gene disorder'
Morbidity	Proportion of people suffering from disease in a population or proportion of people receiving treatment who develop side effects

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Mortality	Proportion of deaths in a population
Mucositis	Sore/inflamed mouth/gut
Mucous membrane/mucosa	Thin skin that covers the inside surface of parts of the body such as the nose and mouth and produces mucus to protect them
Multiomics	Using computers to analyse genomes, transcriptomes, microbiomes or other '-omes'
Murine	Involving mice or rats
Mutation	A change to the genetic code
Myofibroblast	Cells found in wound healing, scarring and tumours
Neonatologist	Doctor specialising in caring for new babies
Neutrophil	White blood cells that destroy bacteria as part of an immune response and are present in pus
Non-integrating vector	Gene therapy that doesn't become part of the existing chromosome
Nucleus	Compartment within a cell that contains the genetic code (chromosomes)
Oncogenesis	Carcinogenesis. Oncogenesis. The formation of a tumour or cancer. How cells become cancerous.
Open trial/study	Everyone knows who is receiving the treatment that is being trialled (compare: blind)
Paediatrician	Doctor specialising in caring for children
Paediatric dermatologist	Doctor specialising in caring for children's skin
Pathogen	'Germ'. A microorganism (bacteria, virus, parasite, fungus) that causes disease
Pathogenesis	The process of a disease developing
Paucity	Not enough of something
PCR	Polymerase Chain Reaction: a way of easily creating many copies of a specific part of a gene
Phage display	A method for studying which molecules stick to which other molecules
Phenotype	Observable characteristics based on genotype (which must be established by genetic testing)
Physiological	Normal bodily function
Placebo	A treatment that looks and feels like real but doesn't contain the active drug or involve the actual process being tested in a clinical trial
Plasmid	Mini chromosome; genes that exist separately to chromosomes
Prebiotic	Promoting the growth of beneficial microorganisms
Pre-clinical	Research before patients are involved
Precision prevention drug intervention studies	Studies on drugs to prevent specific symptoms from occurring
Preclinical	Studies that happen before any testing in humans is done
Prevalence	How frequently a disease or symptom occurs in a population in a defined time period

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Primary	The first or initial event
Prime editing	Gene editing technique to change the DNA sequence of a gene
Probiotic	Containing beneficial microorganisms
Proteasome	Bundle of proteins, including proteases, that break up broken or unwanted proteins
Proteome	All the proteins in a cell or tissue
Protein	Molecule made from chains of 20 different amino acids (building blocks of proteins) - proteins have numerous different functions
Protracted	Lasting a long time; longer than expected or usual
Pruritis	Itch
Psoriasis	A non-contagious skin condition where skin cells multiply quickly and create itchy patches and the immune system attacks healthy skin cells
Recapitulate	To repeat. An animal model repeats the process of (recapitulates) disease development so potential treatments can be tested.
Recessive	Recessive inheritance means a person may inherit a genetic change but not have any symptoms
Regimen	A prescribed course of treatment
Resection	Cutting out tissue or part of an organ surgically
Ribosome	Proteins and RNA molecules that clump together inside a cell to create a protein-making factory
RNA	Ribonucleic acid - like DNA but folds up and sticks to itself rather than making a double helix and has the letters A, C, G and U (instead of T)
RNA - miRNA	micro RNA molecules are short pieces of RNA that stick to matching sequences in message RNA and target it for destruction rather than translation into a protein
RNA - mRNA	messenger RNA is a copy of the gene sequence from a chromosome in a cell nucleus that travels out into the cytoplasm where a ribosome 'reads' it and creates a protein
RNA - rRNA	ribosomal RNA molecules are part of a ribosome and help to build a protein from a messenger RNA
RNA - tRNA	transfer RNA molecules stick to an amino acid and match it to the messenger RNA code to build a protein chain
Sanger sequencing	The original method for determining the DNA sequence of short lengths of a gene
Sepsis	The immune system's reaction to a serious infection affecting the whole body.
Significant	Changes in symptoms after a treatment in a trial are calculated as being statistically significant if they are unlikely to be due to chance
Spatial transcriptomics	Measurement of gene activity and where the activity is occurring in a sample
Squamous	Thin, flattened or scale-like. Squamous cells forming the top layer of our skin are keratinocytes which have become flattened and full of the protein keratin
Statin	Medicine that reduces production of cholesterol and other substances.
Stroma	Supporting tissues
Synbiotic	A mixture of live microorganisms (probiotics) and substances to promote the growth of beneficial microorganisms (prebiotics)
Syngeneic	Genetically identical (compare to allogeneic)
Systemic	Affecting the whole body

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T cells	T cells are immune cells that help fight 'remembered' germs and can be 'killer' T cells (CD8+) or 'helper' T cells (CD4+)
TGFβ1	Transforming growth factor beta: a well-studied protein with many functions
Th17 cell	Immune cells called helper T cells that make a specific protein called interleukin-17
Tissue	Piece or part of an organ or a grouping of similar types of cell
Topical	Affecting only part of the body
Tractable	Easy to work with, influence or control
Transcription	The process of making an RNA copy (messenger RNA) of a gene sequence - happens in a cell nucleus - part of gene expression
Transcriptome	All the messenger RNA transcripts in a cell or tissue
Transcriptomics	Measurement and comparison of all gene activity (which genes are actively producing proteins)
Transgenic	An organism whose genome (DNA) has been artificially altered
Translation	The process of 'reading' a messenger RNA and creating a protein chain - happens in cell cytoplasm - part of gene expression
Translational research	Moving basic science into actual practical applications/treatments
Trophoblast	Part of the placenta
Tumourigenesis	Carcinogenesis. Oncogenesis. The formation of a tumour or cancer. How cells become cancerous.
Ulceration	Open wound on the skin or surface of an organ
Viability	Related to survival
Viral vector	Gene therapy delivered using a virus
Virus	An infective agent that can insert genetic code into cells and use them to replicate itself
Whole exome sequencing (WES)	Sequencing all the DNA of an individual that encodes proteins (the exons) but not non-coding regions that may have other functions
Wild type	The original, unchanged genetic type

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