

GLOSSARY OF TERMS USED IN EB RESEARCH (15Nov23)

To search for a word in this glossary, hold down CTRL and press F. This will open a search box. Type in the word and press enter and you will be taken to the word in this document.

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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 |
| Autochthonus | 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. |
| Carcinoma | Cancer that starts in epithelial cells |

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| 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 |
| 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) |

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| 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 |
| 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 |

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| 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 (ATTCCGTCCTGGATCTGATCGGGCTGTC...) |
| 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. |

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| 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 |
| 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 |

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| 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 |
| 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 |

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| 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 |
| 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 |

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| 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 |