

Case study: Dystrophic epidermolysis bullosa

Learning objectives:

- 1.1.1 Define the study of anatomy and physiology and describe the hierarchical organization of the human body into chemicals, cells, tissues, organs and systems.
- 4.1 Describe the structure and functions of the animal cell.
- 4.3 Outline the basic principles of cell processes involved in maintenance and repair: cell respiration, protein synthesis and cell division.
- 5.1 Describe and recognize examples of the major types of human tissues.
- 6.1.1 Define the integumentary system, list its components and describe their roles in homeostasis.
- 6.2.1 Describe the structure and function of the epidermis, dermis and hypodermis.

Consider reviewing the following topics:

Transcription and translation, organelles involved in protein synthesis, primary tissue types, and integumentary system. Consider reviewing the following resources: Gene expression in “Gene expression and the cell cycle” lecture notes, epithelia and connective tissue in “The primary tissue types” lecture notes, epidermis and dermis in “The integumentary system” lecture notes, “Coding and template strands, transcripts, and polypeptides”, “Translation mechanism”, and “Integumentary system” worksheets, and this [video](#).

Introduction

Patient story:

“Mr. and Mrs. Kennedy, you need to understand what to expect,” said Dr. Bjorn, gripping his fountain pen. “Your son has a form of the disease called dystrophic epidermolysis bullosa. Management of this disease involves wound care, pain control, controlling infections, nutritional support, and prevention and treatment of complications. There are no immediate dangers, as people with this disease can live quite long. Another issue is the increased risk of skin cancer. Since the skin falls off easily, the stem cells need to regenerate it often, and when those cells are more active, they tend to accumulate more DNA damage. That increases the risk of cancer. Do you have any questions?” The Kennedy’s hadn’t blinked throughout Dr. Bjorn’s monologue, and suddenly turned to each other. “But what’s the cause of this disease?” asked Mr. Kennedy. “Well, there’s more than one cause, but from what I’ve read in the literature, it’s likely a mutation in a gene that codes for a protein in the basement membrane of his skin.” “Doctor, you’ll need to explain what a “basular membrane” is,” responded Mr. Kennedy. “No, the basement membrane. It’s what keeps your epidermis stuck to your dermis. In general, it’s what keeps the epithelium stuck to the underlying connective tissue. See, there are a bunch of molecules that make up the basement membrane, including proteoglycans, laminins, and collagen. In this case, I believe your son has a dysfunctional collagen type 7 protein. We only find this protein in the basement membrane of stratified squamous epithelia. Because it only affects this type of epithelial tissue, the symptoms are less severe than other types of epidermolysis bullosa,” explained the doctor. “Okay, well, how can we best provide for Johnny? And what kind of support can we expect?” asked Mr. Kennedy. “Caring for Johnny’s going to be a full-time job. His skin will be quite fragile, and the dressings will have to be replaced every couple of days. During that time, it’ll be important to cleanse his skin with a mild soap. Bathing will be one of the more difficult jobs. Salts and a mild disinfectant, like bleach or vinegar, will need to be used to minimize infections. Infections will slow down the healing process, so it’s best to take all necessary precautions to prevent them.

The hospitals have the facilities to change dressings and bathe Johnny, and it's mostly covered by health care. But you'll probably want to alternate between caring for him at home and here. This all depends on your schedules. Now, Johnny will want to run around and be a kid, so it'll be up to you to decide how to best parent him during his younger years." "Thank you, Dr. Bjorn. We'll take it one day at a time, and we'll make regular appointments with you to keep you updated on his health," smiled Mrs. Kennedy. "We'll keep in touch."

Years have passed since Johnny's parents had that conversation with Dr. Bjorn. Johnny's disease has been difficult on his parents, but they're very happy with how things have turned out. Johnny goes to school, has a few hobbies that keep him busy, and regularly sees a therapist to discuss how to best deal with his disease. He's currently at the hospital having his dressings changed. The nurses that care for him have watched Johnny grow up throughout the years, and they've developed a familiar relationship with the boy. Naturally, Johnny feels very comfortable conversing with the staff and usually has no problem striking up a conversation. Recently, Johnny's parents requested a genetic test to confirm the mutation that caused this disease. Johnny is well aware of the gene involved in his disease but has difficulty understanding its implications, so he asks his nurses a lot of questions.