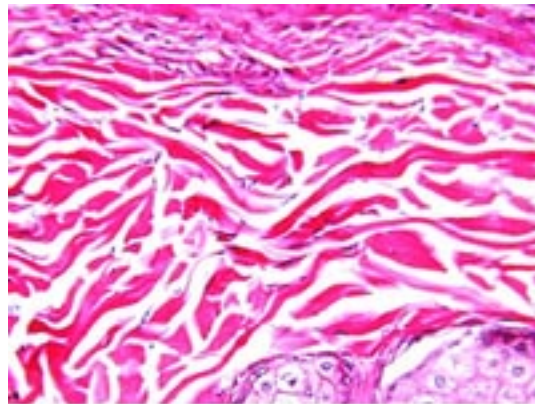




## Skin and Connective Tissue



The dermis, shown above, is a layer of cells - mostly fibroblasts - that lies below the epidermis. Together these two layers make up the skin. The dermis is responsible for thermoregulation and the supply of nutrients to the epidermis. The fibroblasts secrete substances such as collagen and elastin, which supports and confers elasticity to the skin. The dermis is the tough connective tissue from which leather is made. (Reproduced from the website of David Hawkins, Ph.D, Exercise Biology Program, UC Davis, with permission.)

The skin is the largest organ in the body — both in weight and in surface area — and separates the body's internal environment from the external environment. The skin has many diverse roles. It acts as a channel of communication with the outside world; protects the body from water loss; uses specialized pigment cells, called melanocytes, to protect the body from ultraviolet radiation; participates in calcium homeostasis by contributing to the body's supply of vitamin D; and helps regulate body temperature and metabolism.

Elastic tissues such as the skin require a strong and resilient structural framework. This framework is called the extracellular matrix, or connective tissue. The orientation of the connective tissues — adipose (fat cells), cartilage, bone, tendons, and ligaments — found beneath the skin are also key for tissue appearance and function. All connective tissue is composed of three major classes of biomolecules: structural proteins (collagen and elastin), specialized proteins (fibrillin, fibronectin, and laminin), and proteoglycans.

Some skin and connective tissue diseases, such as those discussed in this section of genes and disease, are due strictly to genetic inheritance, while others do not have specific gene abnormalities as their sole cause. Many features of skin and connective tissue disorders overlap with each other, and with other disorders, even though they have unique genetic causes.

## Diseases

Male pattern baldness

Diastrophic dysplasia

Ellis-van Creveld syndrome

Marfan syndrome

Malignant melanoma

Menkes syndrome

Porphyria