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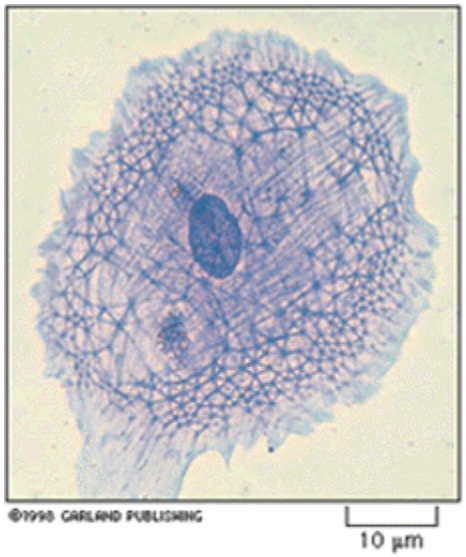
Muscle and Bone



A cultured fibroblast (connective tissue cell) stained with Coomassie blue, a general stain for proteins. Many filamentous structures, which together make up the cytoskeleton, can be seen. The blue central oval is the nucleus. [Reproduced from Alberts et al. (1998) Essential Cell Biology, Garland Publishing Inc., with permission.]

The skeleton provides an anchor point against which muscles, attached via tendons, can exert force. There are a number of diseases that are caused by defects in genes important for the formation and function of muscles, and connective tissues. (Connective tissue is a broad term that includes bones, cartilage and tendons.)

Defects in fibrillin - a connective tissue proteins that is important in making the tissue strong yet flexible - cause Marfan syndrome, while diastrophic dysplasia is caused by a defect in a sulfate transporter found in cartilage.

Two diseases that originate through a defect in the muscle cells themselves are Duchenne muscular dystrophy (DMD) and myotonic dystrophy (DM). DM is another 'dynamic mutation' disease, similar to Huntington disease, that involves the expansion of a nucleotide repeat, this time in a muscle protein kinase gene. DMD involves a defect in the cytoskeletal protein, dystrophin, which is important for maintaining cell structure.

While the gene for Ellis-van Creveld syndrome has been mapped, we await the function of the protein to understand the molecular basis for this disease.

Diseases

Achondroplasia

Amyotrophic lateral sclerosis

Charcot-Marie-Tooth syndrome

Cockayne syndrome

Diastrophic dysplasia

Duchenne muscular dystrophy

Ellis-van Creveld syndrome

Fibrodysplasia ossificans progressiva

Marfan syndrome

Myotonic dystrophy