2 - B: Humans and Connective Tissue Variations

https://www.youtube.com/embed/Q3V1ItCnfas?si=QMEwA5iiHZfnfXpH

Video https://www.youtube.com/watch?v=Q3V1ItCnfas

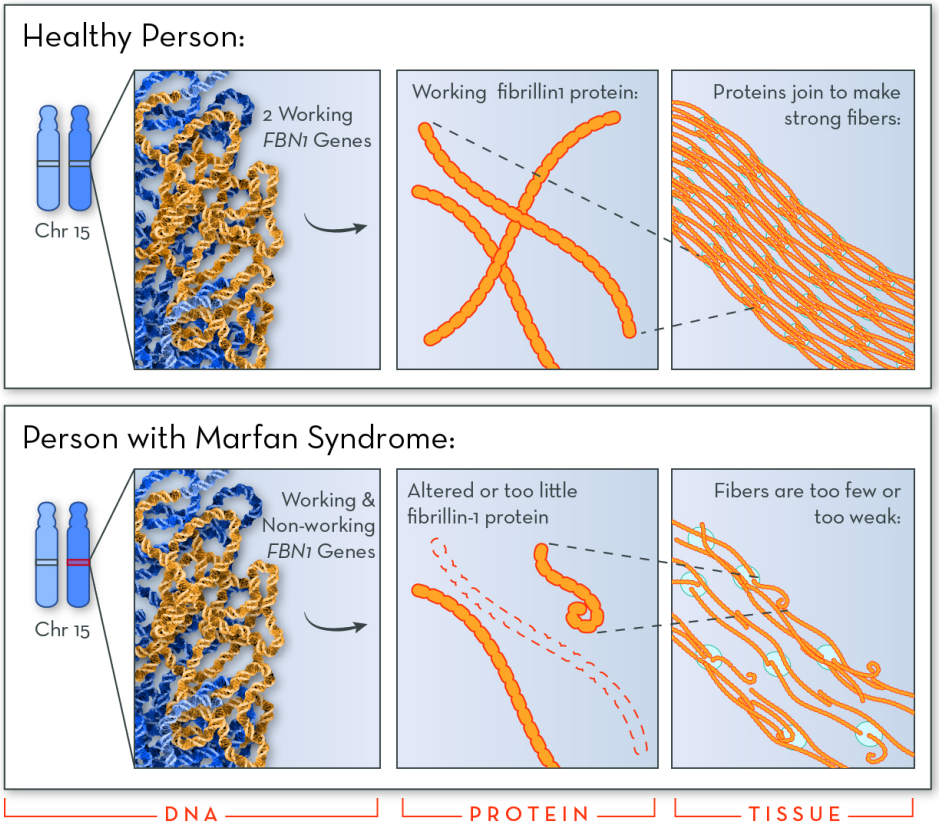
In human beings, variations in connective tissue are grouped into two categories in the current medical model: heritable (genetic) connective tissue diseases (HCTDs) and acquired connective tissue diseases. Acquired connective tissue diseases are currently understood to be autoimmune in nature. That is, they occur when the immune system mistakes various types of connective tissue in different parts of the body for bacteria or viruses that pose a threat to the body.

Some examples of HCDTs:

- Ehlers Danlos Syndromes (Classical, Vascular, and other types) - variations in collagen. People with EDS often have stretchy skin, unstable joints prone to dislocation, gastrointestinal problems, dental problems, near-sightedness, and more.
  - Note: The hypermobile type (hEDS/HSD) has historically been suspected to be related to collagen variations, however in 2024 one study suggested the condition may instead be related to degradation of fibronectin. As of Fall 2026, the genetics of hEDS/HSD have not yet been identified.
- Marfan Syndrome - variations in fibrillin. People with Marfan Syndrome are prone to aneurysm, heart valve problems, dental problems, hernia, retinal detachment, and more.
- Osteogenesis Imperfecta - variations in collagen. People with Osteogenesis Imperfecta (colloquially known as brittle bone disease) may be prone to multiple fractures, have variations in the shape of their bones, be short stature, have hearing loss, and more.
- Loey-Dietz Syndrome - variations in transforming growth factor beta. People with Loey-Dietz Syndrome may have heart valve problems, twisting/winding arteries, loose joints, scoliosis, and more.
- Lipedema - variations in adipose tissue; suspected to be genetic. People with lipedema have fat cells that accumulate in the legs and/or arms. These areas are often painful and hard. That is, the fat of people with lipedema behaves differently to fat in people without lipedema. People with lipedema often have loose joints and immune problems. Many people with lipedema have experienced intense medical fatphobia.

Figure 2 is a graphic representation of how genetic variations which alter the proteins that make up our connective tissues can produce pain and other problems. In this case we are looking at an example from Marfan Syndrome which is caused by a variant in the FBN1 gene. People with Marfan Syndrome make fibrillin fibrils that cannot lock together well, which causes them to be prone to various health issues. Please note that this graphic problematically labels the person without Marfan Syndrome as “healthy.” It is worth reflecting on how such labelling could be harmful to both people with Marfan Syndrome and people without it.

Figure 2. Fibrillin Protein Encoding in Marfan Syndrome



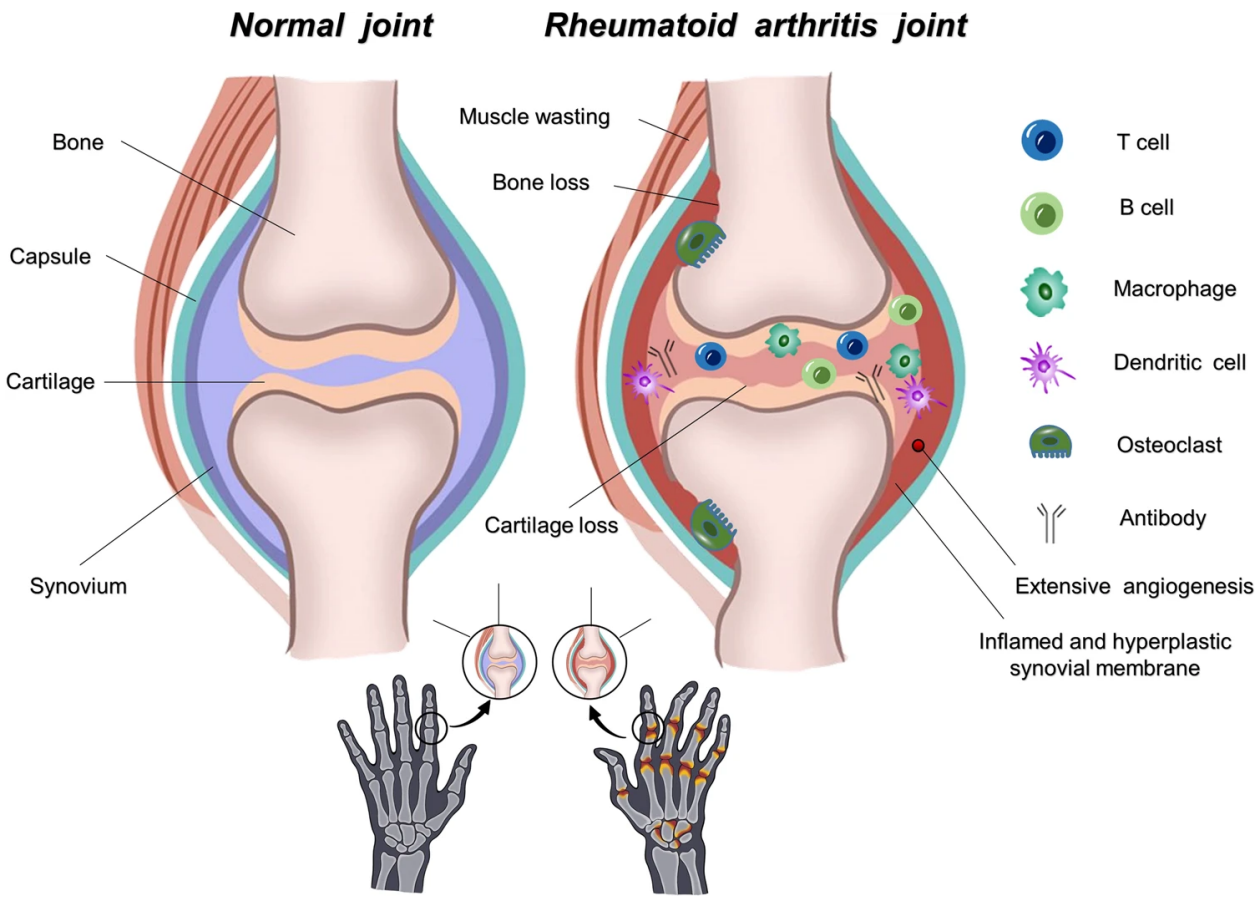
Source: https://learn-genetics.b-cdn.net/genetics/marfan/images/marfan-gene.jpg

Some examples of autoimmune connective tissue diseases:

- Rheumatoid Arthritis - immune system confuses the synovial membrane around the joints for a threat like bacteria or a virus.
- Systemic Lupus Erythematosus - immune system confuses the connective tissue in and around the organs for a threat like bacteria or a virus.
- Scleroderma - immune system creates scar tissue in small blood vessels, eyes, skin, and internal organs.

Figure 3 is a graphic representation of how the immune system affects connective tissue in Rheumatoid Arthritis causing pain and mobility problems.

Figure 3 Rheumatoid Arthritis - Immune System confuses



Source: https://media.springernature.com/full/springer-static/image/art%3A10.1038%2Fs41392-023-01331-9/MediaObjects/41392_2023_1331_Fig3_HTML.png?as=webp

Sex, Gender, Race, Size and Connective Tissue Variations

HCTDs are generally not sex-linked genetic conditions, which means they should occur at the same rate across all sexes, however HCTDs that cause joint hypermobility seem to often (but not always) manifest more severely in people assigned female. In the case of hypermobile EDS (hEDS), a type of HCTD, there is some speculation that progesterone and/or estrogen may play a role in this. After all, hormones affect not just our reproductive systems but many body systems because reproduction is connected to those other systems. For example, the hormone relaxin causes muscles and connective tissues to relax during pregnancy. This is thought to facilitate both gestation and birth, but it can lead to pelvic girdle pain during pregnancy. It should also be noted that there is some emerging evidence that hEDS has a higher prevalence among transgender, non-binary, and other gender-diverse persons.

In addition, acquired/autoimmune connective tissue diseases tend to affect people assigned female (78%) at much higher rates than those assigned male. The reasons for this are complex and have to do with interactions between genetics, the immune system, and the natural and social environment.

The difference in prevalence and severity by sex have some complicated effects for people with variations of connective tissue. In the case of HCTDs, particularly the most common type hEDS/HSD, the assumption that the condition is milder among those assigned male can lead to dismissive attitudes about pain from doctors and others in the community towards people assigned male. Some people face attitudes like “Aren’t you man enough to tough it out?” On the other hand, medical misogyny diminishes and dismisses conditions that are perceived to affect those assigned female. This phenomenon affects both HCTDs and acquired/autoimmune connective tissue diseases. For example, in the United Kingdom the average time to diagnosis for Lupus, an acquired connective tissue variation, is 6-7 years. Researchers Harwood et al. (2025) suggest diagnostic overshadowing is responsible for much of this delay. Their data is filled with stories from people assigned female reporting being dismissed as complaining or told their symptoms are all in their heads.

Race is not genetically determined, but is instead a social construct which imbues certain biological traits (skin color, etc) with social/political meaning [link to Renae’s module] that can have very real medical consequences. Let’s return to the example of Lupus. Black women are diagnosed with Lupus at three times the rate of white women. Black women also experience higher severity of Lupus symptoms. These differences do not trace to racial differences of biology or physiology, but instead to social, economic, and environmental consequences of structural racism, such as chronic racial stress, poverty, exposure to certain environmental toxins, among many other things.

Race-based disparities are also seen among people with heritable connective tissue diseases, too. However, the problem here is underdiagnosis, which is also the result of a complex set of factors which include: the perception that people of color are more likely to be “naturally” hypermobile (more flexible/athletic) and thus less likely to be negatively impacted by these variations; racist beliefs that black people, in particular, feel pain to a lesser degree; a lack of medical documentation of the particular ways these conditions manifest in skin with higher levels of melanin; and structural factors that produce racial disparities in insurance coverage and general access to healthcare for black and indigenous people of color.

In addition, many people in larger bodies experience disparities in relation to variations of connective tissue [Link to weight science module]. Fat people with autoimmune connective tissue diseases are often dismissed by doctors and told that weight loss is the answer to their pain, despite the fact that many thin people develop these diseases too. Fat people with heritable variations of connective tissue experience similar dismissive and shaming attitudes, but also experience the misperception that only tall/thin people can have HCTDs, in part because many people with Marfan Syndrome are tall and slender. While that may be true of Marfan it is inaccurate to generalize the Marfan phenotype to all HCTDs. This is often the result of inadequate training in medical school related to HCTDs. This can lead to very long delays in diagnosis for fat people with variations of connective tissue, which can worsen health outcomes and mobility issues. It should also be noted that racism, sexism, and fatphobia are deeply interlinked and thus likely to most negatively affect fat, black, women and femmes.

Sex, race, and body size have complicated intersections with variations of connective tissue that weave together what is going on within individual bodies and what is going on in society. These issues are an excellent example of how impossible it is to disentangle the biological from the social. This is one reason the reductive model of science and medicine is problematic. More problems with the reductive model are outlined in the next section.

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