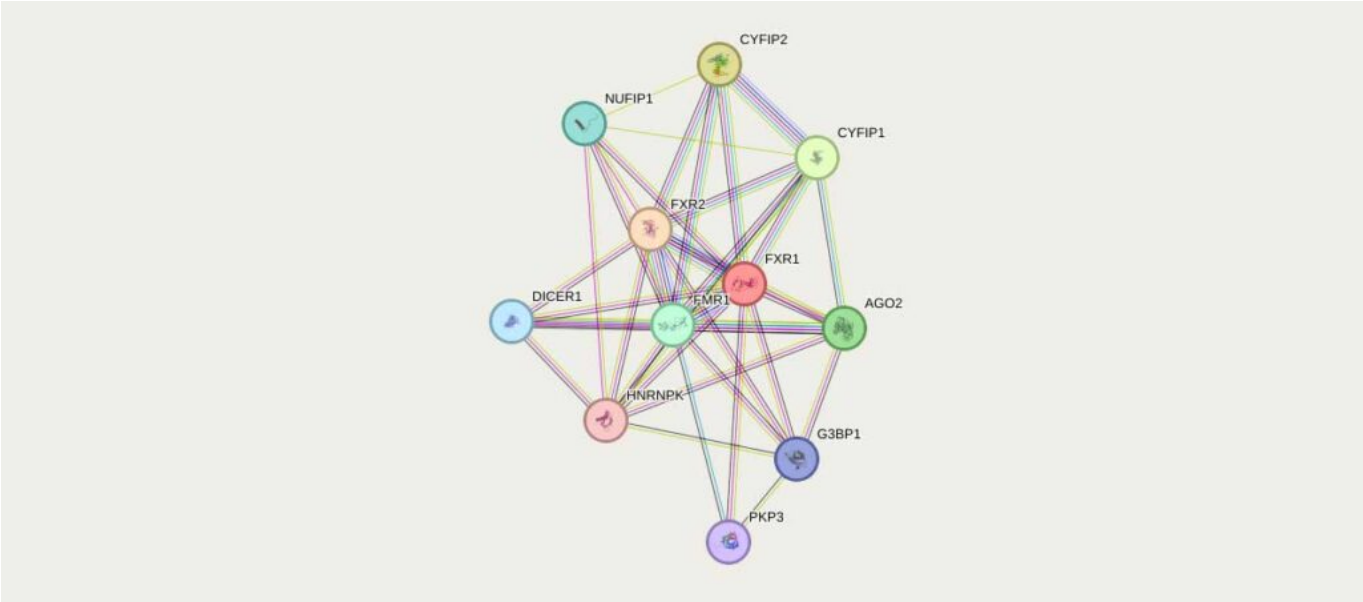
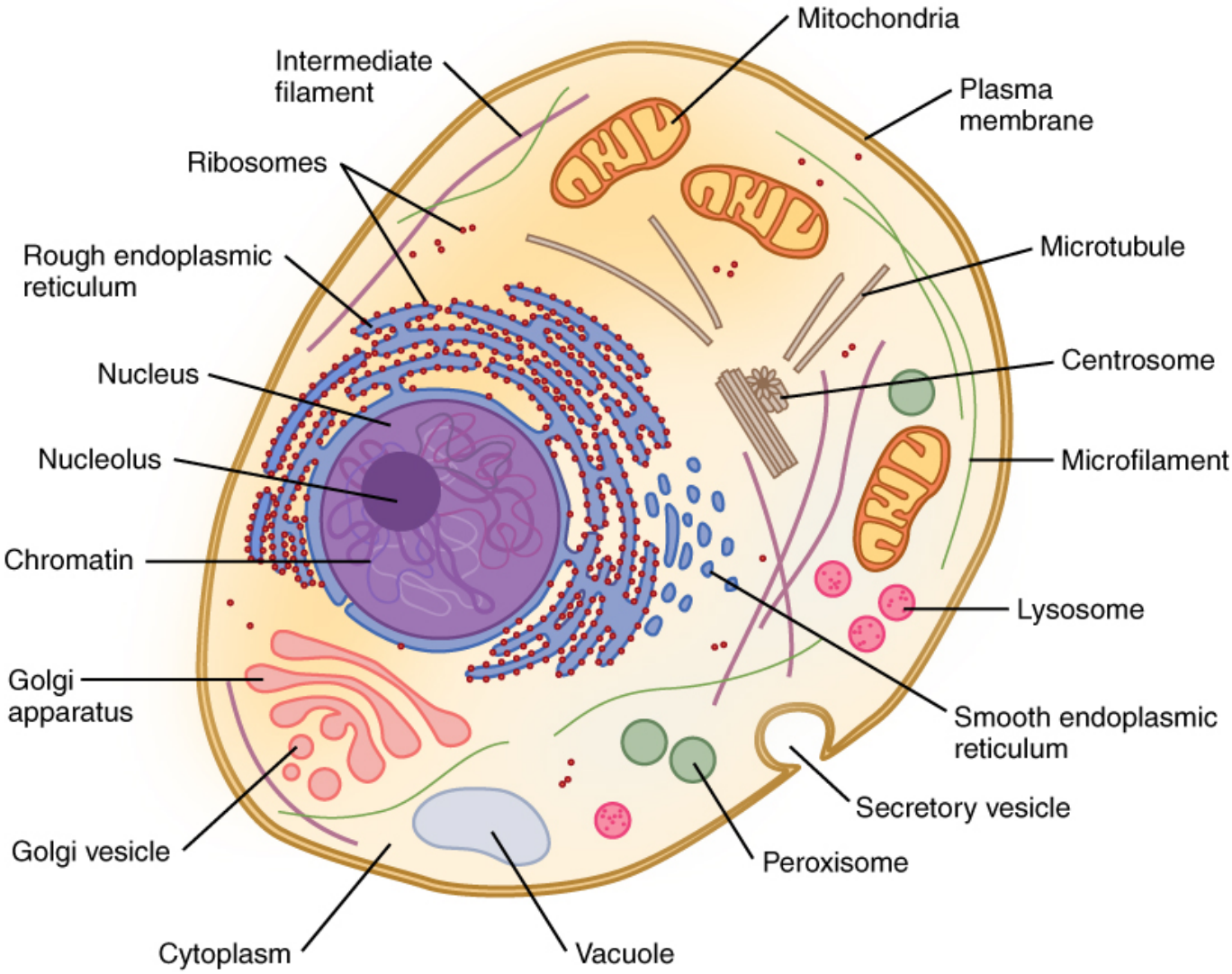


Fragile X-Related Proteins

Proteins 101

Each cell works somewhat like a factory.

- The external plasma membrane functions as the exterior walls of the factory.
- The nucleus stores the genetic blueprints in the form of DNA.
- The machinery (ribosomes) for assembling the products (proteins) is attached to the shop floor (endoplasmic reticulum).
- The Golgi concentrates the proteins to export them.
- The lysosome is used for waste disposal and recycling.



The body makes about 50,000 different kinds of protein. These proteins serve in two major roles. Some of them make up part of the structure of our bodies. Others are enzymes. An enzyme is a protein that works like a tool. It helps a particular chemical reaction take place.

For example, there is a particular enzyme that breaks apart starch into sugar molecules; you use it in your small intestine to digest your carbohydrates. Another enzyme takes a sugar molecule and adds a phosphate onto it to make it unstable. Then a series of about 20 more enzymes each process the unstable sugar molecule in a disassembly line that pulls the sugar molecule apart and releases energy.

Proteins are made out of 20 different amino acids like leucine, lysine, tryptophan,

etc. When you eat foods that contain proteins, the first thing your digestive system does is to break them down into amino acids. Then you assemble those amino acids into your own proteins.

In a way, it's like words. You can make all kinds of words out of 26 letters. You could take a magazine, cut apart all of the letters and reassemble them into your own words. The proteins you make are assembled out of the parts from the proteins you digested from your food.

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Proteins and Fragile X

Fragile X syndrome is caused because people with the disease don't make a particular protein called FMRP (Fragile X messenger ribonucleoprotein 1). The problem is not lack of protein, it is this specific protein that is missing, not proteins in general.

We're sometimes asked if proteins can somehow be added or ingested. But even if someone was injected with lots of FMRP, it would not help.

FMRP needs to be present in the right cells at the right time in the right amount.

We are just beginning to learn which cells normally make FMRP. We have some clues about what the function of FMRP is but we have much to learn.

The synthesis of a protein such as FMRP begins in the nucleus of the cell when the DNA receives a request for that specific information. It is a bit like someone going to a library and selecting a particular book. In response to the request, the DNA opens up and a copy of the coded information is transcribed, much like someone photocopies a chapter of a book. That copy of the DNA is called messenger RNA, or mRNA.

The mRNA leaves the nucleus and goes out to the main part of the cell, the cytoplasm. There the coded mRNA is translated on ribosomes with the help of transfer RNA and a protein is assembled out of amino acids.

The protein takes on a particular shape that allows it to perform very specific tasks. While we know the tasks of some of the proteins that humans make, we only have hints about the role of FMRP.

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