

So now we have a growing list of autism genes. And that has in turn allowed us to now really start to think about what is the underlying cellular mechanism? Why does the autism brain show a brain that has a normal shape and size but just doesn't work well? And how can we utilize our understanding of mechanisms to try to develop better treatments. And that is illustrated in this video. So if we can run the video I can walk you through how many of these autism genes work. And they have to do with the communication of one neuron with another. So there's a neuron here shown in blue, the myelin of the axon is shown in yellow, and the neurons communicate to one another. They carry electrical signals and then the electrical signal from one cell is transmitted to the other by a chemical signal at a synapse. And these synapses are critical points of communication of one cell with another and they actually change in response to learning. And this shows a close up of those synapses. And so for example there are mutations in genes that connect neurons to one another through the synapse, or mutations in genes that encode the proteins inside the spines, as they're called, that regulate the receiving of those signals. When you look at the proteins of these synapses they're littered with proteins which mutations have been found to be associated with autism spectrum disorders. So neurexin, and neuroligin are encoded by genes subjected to autism mutations. Shank3 is one of the most commonly mutated genes in autism spectrum disorders, GRIN2B also as well. And we know also that when these synapses are not properly managed that they tend to shrink so that the electrical signals from one cell are not properly conducted by the chemical signals through the synapse to the second cell. And also, there are other autism mutations that seem to act in the nucleus of neurons, but their action in the nucleus seems to be to regulate the expression of these genes and hence proteins that also regulate the health and the maintenance of the synapses.