



One patient at a time, Mayo Clinic takes on the world's rarest diseases

VIDEO	AUDIO
	"Genetic therapies have really come to a place that they can address so many different types of genetic disease, that now this is a reasonable path."
Margot Cousin, Ph.D. Center for Individualized Medicine Mayo Clinic	"My name is Margo Cousin. I'm a clinical and translational scientist. I'm assistant professor of medical genetics, and the director of our N-of-1 therapeutics program in the Center for Individualized Medicine."
	"I study ultrarare genetic diseases. I've really been trained in genetics, and the overarching goal of the work is to get patients who are in desperate need of therapies to therapeutic opportunities that might exist."
	"We often get cells from our patients — so we'll get a small, tiny, little skin sample, and we can bring those into the lab. And since they're from that patient, every one of those cells has that same genetic alteration that's causing that patient's disease. And we can take those cells and actually turn them into something like a neuron. Then we can strategize and test therapies to try to overcome that problem."
	"We need to understand not only what causes the disease, right, finding that genetic alteration that's positive ... but then we need to know how that gene or protein is broken."
	"It's been remarkable to see patients receiving these custom N-of-1 or first-in-human genetic medicines ... and we're seeing early signs of potential benefit. And it really gives us confidence that the knowledge we're standing on to develop these therapies and bring them to patients is fairly solid ground."

	“And we're really hopeful that we can continue to expand and treat more.”
	“Collaborations in rare disease research are key to success. And you can't do it without partnering with your families, right, because they know this disease better than anyone, and we need to understand how to measure it clinically or how to study it in the lab. So their voices are incredibly important to build the process to find the right answers.”
	“So I'm hoping that every time we solve one problem for one patient or one scientific question, that we didn't just answer that, but we also improved the system. We improved the infrastructure, and we improved the speed and efficiency and rigor with which we can do the science.”
	“But precision medicine is coming right in a wave that's going to be helpful for common diseases as well. And I see this very much in understanding the genetic contributors to all types of disease and having the therapeutic strategies to intervene. So the same techniques we're using for these severe ultrarare patients are going to be amenable to common disease. And this is really building the knowledge base to support that.”