Mayo Clinic uses DNA screening to prevent life-threatening reactions during anesthesia

A rare genetic condition can turn routine surgery into a life-threatening emergency. Now, doctors at Mayo Clinic are using DNA screening to identify that hidden risk before a patient ever enters the operating room — including in one of their own anesthesiologists.	ANCHOR INTRO
Inside this Mayo Clinic operating room, Dr. Timothy Curry and Dr. Adam Jacob monitor every heartbeat and breath — adjusting anesthesia and guiding patients through surgery one moment at a time.	
"We're always monitoring a patient's electrocardiogram, their oxygen saturation, their blood pressure, their temperature."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
As anesthesiologists, they prepare for every scenario.	
"It's extremely rare to have a problem in anesthesia. It's an incredibly safe experience, and you have incredible clinicians that are watching over you every minute while you're under anesthesia."	TIMOTHY CURRY M.D., Ph.D. ANESTHESIOLOGY Mayo Clinic
Yet one condition can transform a routine procedure into a medical emergency. It is called malignant hyperthermia. Certain anesthesia drugs can trigger it. The vulnerability is written into a person's DNA.	
"The way we see it is that people develop really high temperatures. We start to see evidence that their body is becoming acidotic, which is a result of the uncontrolled metabolism."	TIMOTHY CURRY M.D., Ph.D. ANESTHESIOLOGY Mayo Clinic

Surgical teams train for malignant hyperthermia. A lifesaving antidote is stocked in every location where anesthesia is administered.	
Without treatment, it can be fatal. An estimated 1 in 2,000 to 3,000 people carries this genetic risk — though actual reactions during surgery are far less common. Dr. Curry set out to determine whether those rare but dangerous emergencies could be prevented. He examined genetic sequencing data from more than 150,000 Mayo Clinic research participants.	
“We wanted to really understand how common is it for people who might have these genes in our population?"	TIMOTHY CURRY M.D., Ph.D. ANESTHESIOLOGY Mayo Clinic
He identified 1 more than 130 people with genetic variants linked to malignant hyperthermia susceptibility. Most were unaware. Each participant was notified and offered genetic counseling.	
“Once we identified those individuals, we then went back to our other research databases to try to understand how many of them might have had anesthesia, at least at Mayo Clinic.”	TIMOTHY CURRY M.D., Ph.D. ANESTHESIOLOGY Mayo Clinic
The goal was to understand how often this genetic risk leads to complications during surgery.	
Among the carriers was someone Dr. Curry knows well.	

"One of those individuals was someone I trained with in anesthesia. He's a colleague here at Mayo Clinic."	TIMOTHY CURRY M.D., Ph.D. ANESTHESIOLOGY Mayo Clinic
That colleague is Dr. Jacob.	
"Which is terribly ironic."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
The irony is hard to miss. An anesthesiologist trained to respond to this rare crisis was carrying the genetic risk himself.	
"I've had surgery a couple of times, and I fortunately have never experienced a reaction."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
The reaction is only triggered by certain anesthesia medications. Those medications remain essential in emergencies, including trauma cases or when doctors must quickly secure a patient's airway. Because the condition is inherited, Dr. Jacob had his two teenage sons tested.	
"And it turns out, they are both carriers of the same mutation that I have."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
Their medical records were updated to guide future anesthesia care. They now wear medical ID bracelets.	
"It's a very manageable condition — easily avoidable — as long as you know. Knowledge is power."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
Mayo Clinic researchers are also using this approach to uncover gene variants linked to hereditary cancers, heart disease and other inherited conditions. In many cases, early detection leads to screening and targeted care.	

Genetic testing for malignant hyperthermia is now incorporated into Mayo Clinic's presurgery care.	
For Dr. Jacob and his family, a hidden risk can now be anticipated and prevented.,	
"I have a greater understanding or greater appreciation for doing what I can every day for every patient, as though, you know what? Maybe they're a carrier too."	ADAM JACOB M.D. ANESTHESIOLOGY Mayo Clinic
For the Mayo Clinic News Network, I'm Susan Murphy.	