

Hope for children with TSC, a genetic tumor disorder

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Although it is rare, tuberous sclerosis complex (TSC) can be a difficult and frightening diagnosis for parents and children. The genetic disorder causes nonmalignant tumors to form in many different organs, including the brain, eyes, kidneys and heart. If a child's brain is affected by the disease, it can be especially debilitating since it can cause seizures, developmental delays, intellectual disability and autism.

"TSC is not easily diagnosed because the main symptoms are white spots on the skin that can't be seen by the naked eye and [abnormal growth](#) in the heart muscle, which sometimes be found in an ultrasound during pregnancy," said Nikolas Mata-Machado, MD, a [pediatric neurologist](#) at Loyola University Health System and an assistant professor in the departments of neurology and pediatrics at Loyola University Chicago Stritch School of Medicine.

Because the disease can affect several organs, Mata-Machado said it's important for parents to find a center that has a multidisciplinary team.

"It's not just about the brain or heart; it's about how this disease affects many areas of a child's body. I work with pediatric experts in many medical fields, including nephrology, cardiology, pulmonology, hepatology and neuropsychology," Mata-Machado said.

Though there is no cure for TSC, Mata-Machado is one of only a few physicians to use a new drug treatment similar to chemotherapy that causes the tumors to shrink, offering hope to many patients who do not

respond to traditional treatments.

"There are surgical options and also some patients respond well to anti-seizure medications. Unfortunately, I have some patients who do not respond to these medications, some still having up to 40 seizures a day and others are not candidates for surgery. This new treatment gives these patients and their families another chance, a hope," Mata-Machado said.

TSC is a [genetic disorder](#) found in 1 of every 25,000 children. It is estimated that 50,000 people in the U.S. are living with the disorder and possibly more because it is difficult to diagnose. The severity of the disease varies greatly, with some patients able to live independent, healthy lives into adulthood.

"It is important for someone with TSC to be monitored by a physician throughout their life as complications can cause serious medical conditions and even death if left untreated," Mata-Machado said. "Still, with advances in medicine and research we are able to offer [patients](#) more options for living with this disorder."

Provided by Loyola University Health System

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