

Tuberous sclerosis complex the focus of new therapies and continued research



Tuberous sclerosis complex (TSC) is an often unrecognized or misdiagnosed genetic disorder that causes benign tumors to develop in multiple organ systems and frequently causes epilepsy. Research being done at UCLA and elsewhere is yielding new understanding of the condition and, in some cases, new treatments.

The UCLA Tuberous Sclerosis Complex Program has been designated by the Tuberous Sclerosis Alliance — the TSC national advocacy and research support organization — as one of a handful of TSC programs nationwide with the resources and expertise to address the complexities of this disease and the multiple medical needs of TSC patients. UCLA's program includes both clinical care and research and is one of the largest in the Western United States.

No two cases alike

Affecting more than 50,000 people in the United States and over 1 million worldwide, TSC symptoms are unpredictable and vary significantly from person to person. Some individuals experience only minor organ involvement and are cognitively normal while others are more severely affected with progressive, multi-system tumors; intractable or drug-resistant epilepsy and intellectual disability.

TSC studies advance understanding of other diseases

“Tuberous sclerosis complex is a ‘linchpin’ disease or one in which the genetic pathway is the same as that affecting other diseases and disorders. TSC research is creating breakthroughs in how we treat epilepsy, autism and cancer,” says Joyce Wu, MD, professor of pediatric neurology, director of the UCLA Tuberous Sclerosis Complex Program and a founding member of the Tuberous Sclerosis Complex Clinical Research Consortium.

“Every research-hour spent on unlocking the cure to TSC improves our understanding of TSC and so many other diseases as well,” says Dr. Wu. “The drug everolimus has been groundbreaking in shrinking brain and kidney tumors associated with TSC and is currently in clinical trials to improve epilepsy and neurocognition in TSC even though it is not a seizure or behavioral medication, per se. If we can understand how this medication improves epilepsy or cognition in TSC, the next stage may be to expand to non-TSC epilepsy and neurocognitive impairments.”

With its broad and varied progression and presentation, diagnosis has been an ongoing challenge and genetic testing does not detect all cases of TSC. Updated guidelines by the International Tuberous Sclerosis Complex Consensus Conference published in October 2013, however, have provided gold-standard recommendations for diagnosis, surveillance and management of TSC.

The new guidelines call for a diagnosis of TSC with the presence of two major features, or one major feature and two minor features of the disease. Major features include multiple angiofibromas (raised red facial tumors), tubers in the brain and cardiac rhabdomyoma or non-cancerous heart tumor. Minor features are kidney cysts, “confetti” skin lesions, dental pitting and retinal growths.

TSC care

Although there currently is no cure for tuberous sclerosis, UCLA physicians offer personalized care to help each patient manage the disease. The neurological manifestations of TSC are often the most debilitating aspects of the disorder. UCLA program physicians coordinate care of pediatric patients or consult with specialists in cardiology, dermatology, genetics, interventional radiology, nephrology, neurology, ophthalmology and pulmonary medicine to provide a wide range of treatment options. Adult patient care is currently limited to TSC diagnosis and ongoing testing guidelines, with the goal of helping primary-care physicians and subspecialists manage the various aspects of care.

UCLA physicians have been leaders in the development of some of the therapies and techniques adopted by TSC centers globally.

Medical therapy for TSC tumors

- Everolimus (Afinitor) — shrinks brain and kidney tumors associated with TSC
- Sirolimus/rapamycin (Rapamune) — cousin to everolimus that reduces the size of angiofibromas, which affect more than 90 percent of TSC patients

Imaging techniques used with epilepsy surgery

- Magnetoencephalography (MEG) — non-invasive method localizes sources of epileptic brain activity
- Functional MRI (fMRI) — highly specialized pre-surgical technique for mapping sensation, movement and language areas of the brain
- Intraoperative MRI (iMRI) — UCLA was one of the first hospitals in the world to offer this method pre-surgery and during surgery to enhance structural brain imaging

Alternative therapies for epilepsy

- Vagus Nerve Stimulator (VNS) — minimally invasive surgical alternative for intractable epilepsy
- Ketogenic Diet — high-fat, low-carbohydrate diet to improve seizure control

Participating Physicians

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