

Tuberous Sclerosis Complex

820

TSC patients being treated
at Cincinnati Children's



Patients travel to the TSC
clinic from **33 states** and
12 countries

83

TSC-related publications
by Cincinnati Children's
researchers

The Tuberous Sclerosis Complex (TSC) Clinic at Cincinnati Children's Hospital Medical Center is one of the busiest in the world, providing care for more than 800 children and adults. Established in 1992, the clinic pioneered the concept of offering centralized, comprehensive care for patients with TSC.

HOW WE'RE DIFFERENT

The TSC Clinic provides exceptional patient- and family-centered care. Our experienced, multidisciplinary team specializes in treating TSC disease and its co-morbidities such as developmental delays, cognitive impairment and autism. Our specialists offer immediate therapy recommendations, comprehensive treatment plans and long-term coordination with the patient's primary care physician. Financial and travel support is available for patients and families with limited resources, as well as for international patients.

In 2003, our clinic was the first to conceive of and implement the use of the mTOR inhibitors sirolimus and everolimus to treat manifestations of tuberous sclerosis. Subsequent open-label and placebo-controlled trials led to FDA approval of everolimus first for astrocytomas, and then for angiomyolipomas.

CONDITIONS TREATED

- Angiofibromas
- Behavioral disorders
- Epilepsy
- Subependymal giant cell astrocytoma (SEGA)
- Lymphangiomyomatosis (LAM)
- Renal cystic disease
- Renal, hepatic and adrenal angiomyolipomas (AML)
- Retinal hamartomas
- TSC-associated pancreatic neuroendocrine tumors
- TSC-associated thyroid nodules

Our TSC clinic is the only truly multidisciplinary clinic in the world, with specialists together in one location to maximize coordination of care. We are the only one with a pulmonologist dedicated to the treatment of LAM.



TREATMENT TEAM

Neurology

David Neal Franz,
MD, *Director*

Darcy Krueger,
MD, PhD

Anna Weber Byars,
PhD

Cynthia Tudor,
CPNP

Karen Agricola,
CFNP

Gail McMonigle, RN

Kathryn Loew, RN

Nephrology

Jens Goebel, MD

Stuart Goldstein, MD

Janet Boyle, MD

Cardiology

Timothy Knilans,
MD

Cardiac MRI and Imaging

Michael Taylor, MD

Dentistry

Elizabeth Gosnell,
DMD

Dermatology

Kara Shah, MD

Endocrinology

Laurence Dolan, MD

Genetics

Jennifer Glass,
MS, CGC

Elizabeth Schorry,
MD

Neurosurgery

Charles Stevenson,
MD

Oncology

James Geller, MD

Ophthalmology

Constance West, MD

Ophthalmologic Oncology

James J. Augsburger,
MD

Plastic Surgery

Christopher Gordon,
MD

Psychiatry

Daniel Nelson, MD

Pulmonology

Francis McCormack,
MD

Aarthi Vemana, MD

Radiology

James Leach, MD

Marguerite Care,
MD

Todd Abruzzo, MD

Alexander Towbin,
MD

Alan Brody, MD

John Racadio, MD

Social Work

Linda Wallace,
LISW

TREATMENT APPROACH

The treatment of Tuberous Sclerosis-associated conditions requires highly specialized care from a variety of experts. Our multidisciplinary team includes specialists in neurology, nephrology, cardiology, dentistry, dermatology, endocrinology, genetics, neurosurgery, ophthalmology, plastic surgery, psychiatry, pulmonology, radiology and oncology. Our scheduling system allows patients to see all needed subspecialists and undergo any imaging procedures and diagnostic tests in a single day.

Team members take a holistic approach to patient care, focusing on medical treatment and services that meet each patient's psychosocial, educational and vocational needs. Our social workers involve patients in programs that go well beyond diagnostic studies and medical or surgical interventions. An example is our TSC Summer Camp Program, which helps patients develop friendships, build interpersonal skills and gain confidence.

RESEARCH THAT ENHANCES CLINICAL CARE

Cincinnati Children's is involved in many clinical, translational and basic science research studies aimed at improving care for children and adults with TSC. David Franz, MD, Darcy Krueger, MD, PhD, and Frank McCormack, MD, work closely with researchers at Cincinnati Children's, the University of Cincinnati and other institutions around the world. This collaboration helps the team rapidly adopt evidence-based practices in the care of TSC and better understand the broad range of issues that patients face.

CLINICAL RESEARCH

- Characterization of TSC1 and TSC2 genes (*American Journal of Human Genetics*, 2001)
- Lamotrigine therapy of epilepsy associated with TSC (*Epilepsia*, 2001)
- Use of steroids to manage post-embolization syndrome (*American Journal of Kidney Diseases*, 2002)
- Rapamycin causes regression of astrocytomas in TSC (*Annals of Neurology*, 2006)
- Rapamycin therapy for angiomyolipomas and lymphangiomyomatosis (LAM) in patients with TSC or sporadic LAM (*New England Journal of Medicine*, 2008)
- Use of VEGF-D as a bio-marker for TSC-related lymphangiomyomatosis (*Chest*, 2010)
- Open label trial of everolimus in TSC patients with SEGA (*New England Journal of Medicine*, 2010)
- Randomized placebo-controlled trial of rapamycin to treat TSC patients with LAM (*New England Journal of Medicine*, 2011)
- Efficacy of everolimus for treating SEGA and angiomyolipomas in TSC (Two separate articles, both in *The Lancet*, 2013)
- Efficacy of everolimus for epilepsy associated with TSC (*Annals of Neurology*, *European Journal of Pediatric Neurology*, 2013)

For patient referrals and non-urgent consultation during business hours, contact the program directly at **1-513-636-4222** or **tsclinic@cchmc.org**.