Moving TSC Research Forward in the Face of COVID-19

April 6, 2020
The Tuberous Sclerosis Alliance’s Mission

The TS Alliance is dedicated to finding a cure for tuberous sclerosis complex, while improving the lives of those affected.
Accelerating research and drug development through collaborative projects
Ongoing work to impact clinical care

- PREVeNT nearly fully enrolled
- RDCRN: received 5-year renewal by NIH with TS Alliance co-funding
  - Research Coordinators
  - Clinical Fellowship
- STOP-2 beginning early 2020
- TACERN autism biomarker study readout expected in 2020
- Working group updating TSC Clinic standards
Clinical Research

Clinical impact by industry partners

- Supplemental New Drug Application under consideration by FDA for Epidiolex® (cannabidiol) to treat seizures associated with TSC
- Two companies planning clinical studies in TSC based upon:
  - Studies with TSC human biosamples
  - Preclinical Consortium partnership
Preclinical Consortium seeks to stimulate more clinical trials

What is the Preclinical Consortium?

- Public-Private partnership among industry, academia, and TS Alliance designed to stimulate innovation and new therapies for TSC

What we do?

- Evaluate (screen) compounds in established cell or animal models for stopping or reversing TSC manifestations, including tumors and epilepsy
Preclinical Research Consortium

Continue to engage consortium members!

Preclinical Consortium Members Meeting
April 22-23

Expanding Industry Membership
- New member joined today!
- In discussions with another two companies
- Reaching out to new companies to stimulate interest

Currently have 6 industry Preclinical Consortium Members – all with a focus to improve the lives of those affected by TSC

Discuss TSC research strategy, evaluate research models and data

Bringing proprietary drugs and clinical development expertise!
Research continues...

Some research has been slowed due to COVID-19

- One study for a company partner delayed due to availability of research materials
- TAND model development delayed at PsychoGenics – but still developing plans and protocols with PsychoGenics and Consortium members
- LAM model development delayed at Porsolt – protocol is complete

Some compound testing is moving forward

- Finishing a study at the Van Andel Institute to evaluate the effectiveness of 2 compounds for inhibiting tumor growth
- 5 compounds being tested for company partner in epilepsy at PsychoGenics
- Study plans being discussed for as many as 27 experiments testing compounds
What is it?

- The TSC Natural History Database (NHD) launched in 2006 to collect clinical information about children and adults with TSC over their lifetime to better understand the disease.

- We use a web-based platform called StudyTRAX to enter information about different conditions affecting people with TSC such as:
  - Epilepsy and infantile spasms
  - Angiofibroma
  - Renal angiomyolipoma
  - Subependymal giant cell astrocytoma (SEGA)
  - Rare conditions such as pancreatic neuroendocrine tumor
TSC Natural History Database (NHD)

Highlights

- 18 TSC Clinics and TS Alliance
- Clinical information collected on more than 2,200 individuals with TSC.
- More than 70 requests for data since 2009 with 10 peer-reviewed publications.
- The TS Alliance cheek swab collection at the 2018 World TSC Conference.
Moving research forward

- Research staff continue to update NHD records while adhering to “Stay at Home” order.
- TS Alliance sent reminders to WC participants for medical records.
- Follow up to March 20, 2020 COVID-19 Town Hall: Collect clinical data to help determine whether use of mTOR inhibitors in TSC is a risk factor for developing COVID-19.
- Share clinical data associated with DNA and other biosamples for TSC researchers to better characterize the disease.
What is a biosample?

A sample of biological material such as blood or a piece of tissue taken during a surgery or a biopsy.

Why does this matter?

Research with biosamples and clinical data could:

- Lead to biomarkers that enable preventative interventions
- Identify additional targets for drug development
- Enable the prediction of an individual's risk for developing a specific TSC manifestation
- Help answer why some cell types develop into tumors while other cells do not
- Predict an individual's likelihood of responding to a specific drug
The TS Alliance aims to collect 437 additional samples by the end of 2020

Biosamples collected vs. target 2016-2020

- The TS Alliance has partnered with Phlebotek, a mobile phlebotomy service, to permit blood collections at participants’ homes.
- The TS Alliance can accept other samples, such as tissues collected during surgeries.
- The TS Alliance aims to collect medical records from participants to connect their sample in the Natural History Database.
How can you help?

- Complete the consent process over the phone with Gabrielle or Jo Anne to submit a biosample later this year
- Send medical records to the TS Alliance so we can connect information to the Natural History Database
- Spread the word with our flyer
- Consider donating remnant tissue from a biopsy/surgery

Contact us at biosample@tsalliance.org for more information.
What is TSC Academy?

- An interactive **online educational platform** to support our community and advocates understanding of TSC
- Can complete on desktop or smartphone/tablet
- Currently, **three courses** are available:
  1. Primer – TSC 101
  2. Introduction to Research and Funding
  3. The Advocate’s Role

Visit: [www.tscacademy.org](http://www.tscacademy.org) to register and enroll!
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Government advocacy

Tuberous Sclerosis Complex Research Program (TSCRP)

- $89 million in cumulative funding since 2002, $6 million in FY 2020
- $8 million appropriations request for FY 2021
- Bipartisan Dear Colleague Letter
  - House – Sponsored by Representatives Loebsack (D-IA) and Mullin (R-OK)
  - Senate – Sponsored by Senators Cramer (R-ND) and Tester (D-MT)
- 216 House signers and 35 Senate signers
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How can you help?

- Call and write your representatives
- Hold a virtual meeting with your elected officials
- Connect with your local Community Alliance
- Engage with your offices around virtual events like TSC Global Awareness Day on May 15

Contact us at grc@tsalliance.org for more information.
Researchers continue working hard

- Grants are like small businesses, dependent on productivity and revenue
- Dean and Zoë have talked with our grant recipients
- Labs remain open with restricted access and physical distancing
- Many critical research activities can be done outside the lab: analyzing data, writing, planning, etc.
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Questions?

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