
The Power of Community: Clinical Trial Experience in Tuberous Sclerosis Complex

Steven L. Roberds, PhD
Chief Scientific Officer
Tuberous Sclerosis Alliance

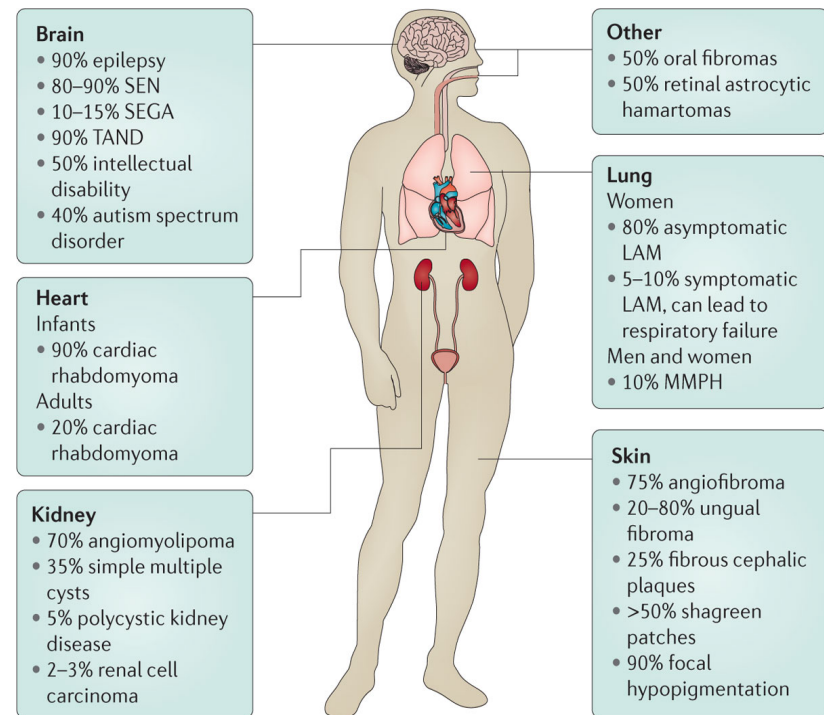


Neurocrine
BIOSCIENCES

*ABCs of Clinical Trials Part 2: What
Rare Disease Families Should Know*

Tuberous Sclerosis Complex (TSC)

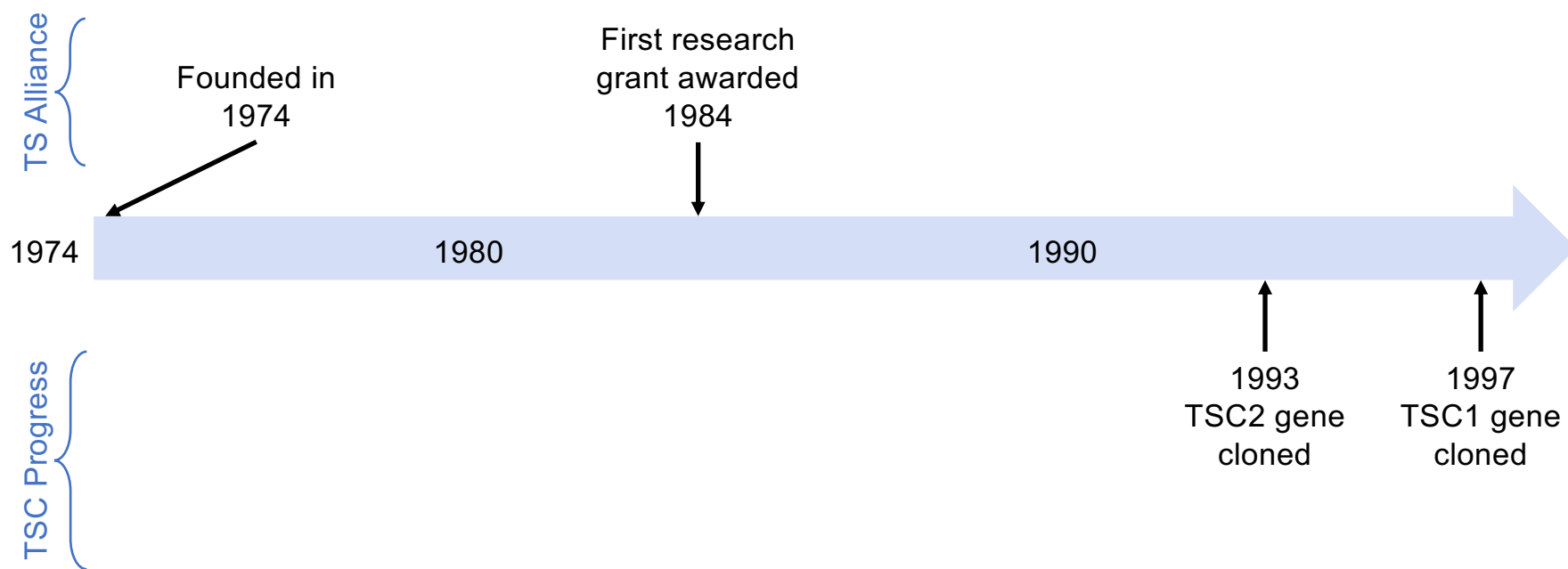
- TSC is a genetic disorder that causes tumors to form in vital organs: primarily the brain, eyes, heart, kidney, liver, lungs and skin.
- Neurological manifestations are often the most devastating.
- TSC affects ~1 in 6,000 live births.
- An estimated 50,000 Americans have TSC.
- No two people are affected the same way, not even identical twins.



Henske, E. P. *et al.* (2016) Tuberous sclerosis complex
Nat. Rev. Dis. Primers doi:10.1038/nrdp.2016.35

Nature Reviews | **Disease Primers**

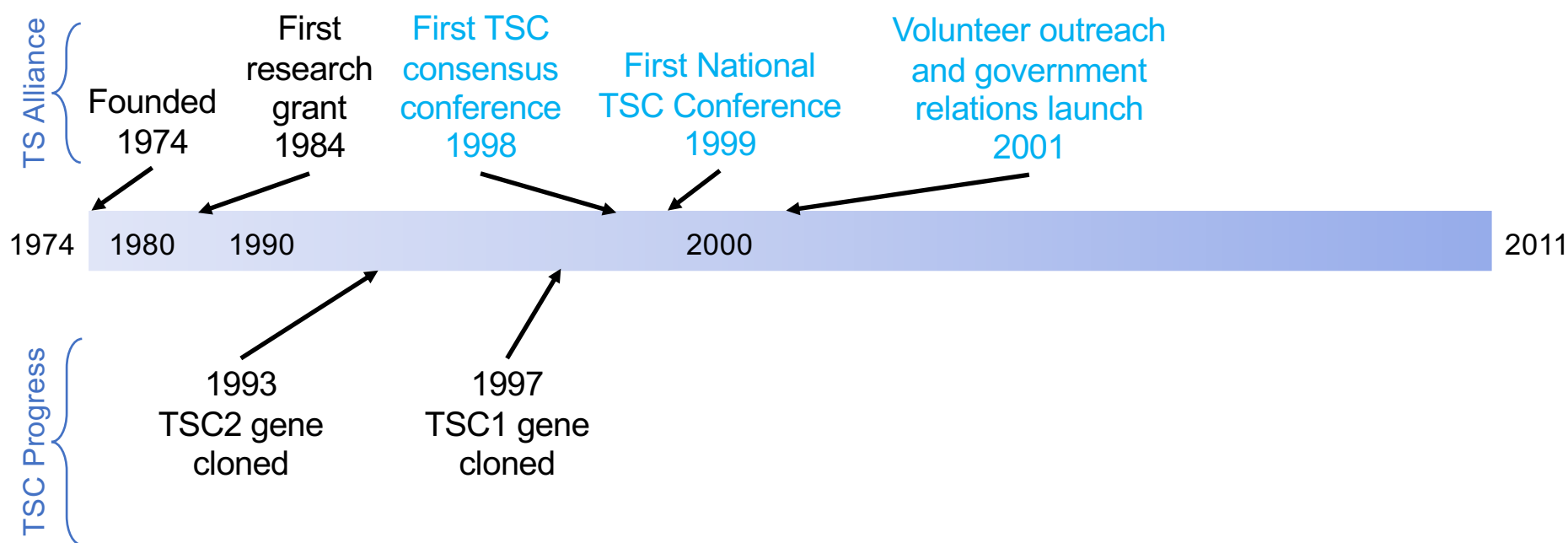
Stimulating Research to Learn the Cause of TSC



ABCs of Clinical Trials Part 2: What Rare Disease Families Should Know

Slide 12

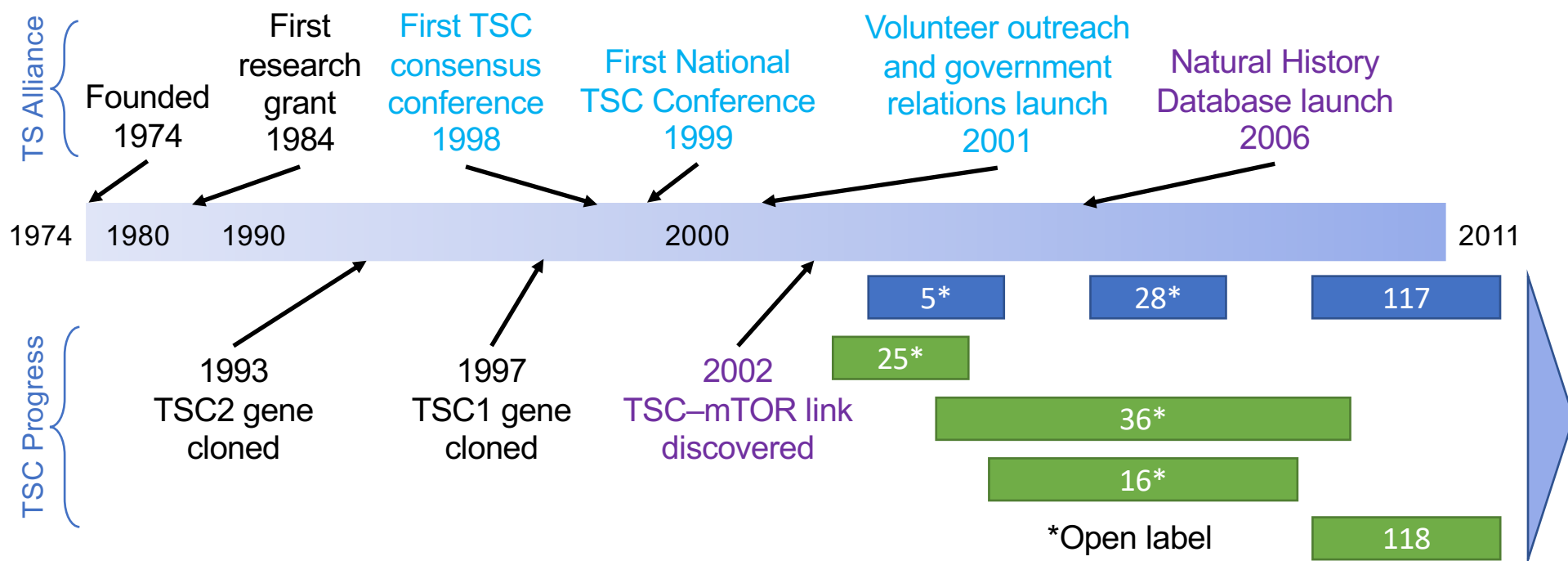
Preparing and Empowering the Community



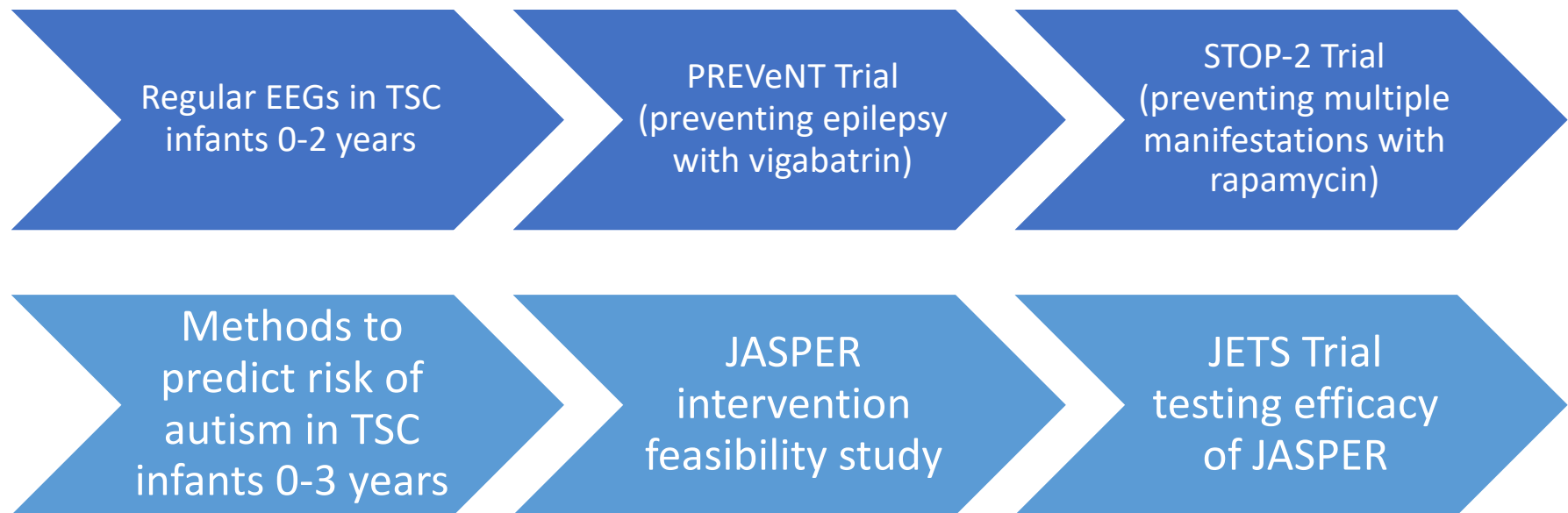
ABCs of Clinical Trials Part 2: What Rare Disease Families Should Know

Slide 4

A Breakthrough Leads to Many Clinical Trials



Biomarkers or Validation Studies Often Come First



Some Takeaway Messages

- A united, collaborative ecosystem of patients, researchers, and clinicians sets the stage for success
- Clinical trials are part of a process – expect them to evolve:
 - Natural history or pilot study provides key data to design a good trial
 - Feasibility study (often open-label) tests the trial design
 - Pivotal study must be well controlled and rigorously conducted
- Well informed peer-to-peer advocacy coordinated through patient advocacy organizations can be the biggest contributor to successful clinical trial enrollment



Neurocrine
BIOSCIENCES

***ABCs of Clinical Trials Part 2: What
Rare Disease Families Should Know***

Some Tools

- TS Alliance's Basic Introduction to Clinical Trials
 - <https://www.tsalliance.org/individuals-families/tsc-clinical-trials/>
- Clinical Trials Transformation Initiative
 - Home page <https://www.ctti-clinicaltrials.org/>
 - Effective engagement with patient groups around clinical trials
<https://www.ctti-clinicaltrials.org/files/pgctrecs.pdf>



Neurocrine
BIOSCIENCES

***ABCs of Clinical Trials Part 2: What
Rare Disease Families Should Know***
