**From Where (Referral Sources)**

<table>
<thead>
<tr>
<th>For Whom (Populations, listed in order of life stage)</th>
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<tr>
<td>Google / TS Alliance website</td>
<td>Social media sites</td>
<td>Community clinicians</td>
<td>TSC Clinics</td>
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<td>International TSC groups</td>
<td>CDC, NIH websites</td>
<td>Other parents / caregivers</td>
<td>Community Clinicians</td>
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<td>Other advocacy organizations</td>
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<td>Case managers of government benefits</td>
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1. **Parents/caregivers of infants and children (under 18):**
   - a. Diagnosed with TSC
   - b. Undiagnosed

2. **Young adults with TSC (18-26):**
   - a. Semi-independent and independent adults diagnosed with TSC and their family members
   - b. Parents/caregivers of family members of dependent adults diagnosed with TSC
   - c. Undiagnosed

3. **Adults (27+):**
   - a. Semi-independent and independent adults diagnosed with TSC and their family members
   - b. Parents/caregivers/ family members of dependent adults diagnosed with TSC
   - c. Undiagnosed

**Assumptions (Theory of Change):**

- Tuberous sclerosis complex (TSC) is a genetic disorder that causes non-malignant tumors to form in different organs, primarily in the brain, eyes, heart, kidney, skin and lungs, and affects approximately 50,000 individuals in the USA and 1 million worldwide. It is the leading genetic cause of epilepsy and autism.
- TSC is considered a lynchpin disease, meaning that every advance made in our search for answers and a cure may also lead to answers and advances in other more prevalent diseases like epilepsy, autism, and cancer.
- The Tuberous Sclerosis Alliance (TS Alliance) is dedicated to finding a cure for TSC while improving the lives of those affected.

**Strategies/Activities (Program Interventions):**

- **Accelerate Research**
  - Collaborate and expand partnerships with other nonprofits, academic institutes and industry to advance TSC research
  - Advocate for Federal research funding
  - Increase Preclinical Consortium productivity by engaging researchers, improving models, adapting to priorities of members
  - Expand capabilities of centers in the Clinical Research Consortium and fund pilot or add-on studies with pathways to outcomes
  - Increase rate of biosample collection by enrolling individuals anywhere in US
  - Invest research grant funding in postdoctoral trainees and highly novel research ideas
  - Convene targeted workshops on urgent research problems and follow-up with funds

- **Improve Access and Quality of Care**
  - Advocate for state funding for research and/or Centers of Excellence
  - Develop a support hub to navigate access to FDA approved therapies
  - Build evidence for what constitutes high quality care for TSC
  - Expand knowledge of evidence-based, best practices of TSC clinical care among TSC Clinic healthcare professionals and other medical providers involved in the treatment of TSC
  - Expand access to care for people in regions with no TSC Clinic or TSC-knowledgeable healthcare professionals
  - Improve access and delivery of evidence-based, high quality clinical care for patients with TSC, including comprehensive adult care for inpatient and outpatient services

**Leading Indicators:**

- **Accelerate Research**
  - Invest $56 million in research over the next five years to drive projects, workshops, and collaborations
  - Maintain a minimum of $6 million annually for TSCRP
  - Reach 75 mechanisms or combinations tested in Preclinical Consortium with working relationship with 12 industry partners
  - Clinical Research Consortium receives $30 million federal or industry funding for trials
  - Reach 2500 biosamples with serial blood samples on 250 individuals and 150 tissues, with each genetically tested & 12 use requests annually with clinical data captured in the Natural History Database
  - $450K paid annually on research grants, 2/3 for postdocs
  - Annual innovation workshop held with follow-up research funded by innovation fund

- **Improve Access and Quality of Care**
  - Grow state initiatives to four states
  - Increase partnerships with specialty pharmacies, payers, industry, private foundations to support at least 200 individuals/families annually by 2023
  - Form task group of key stakeholders (medical professionals, patients/families) to critically evaluate current TSC Clinic designation standards and recommend new organizational structure and requirements to the TS Alliance Board of Directors for approval and implementation by 2020
  - Conduct 7 studies between 2019-2023 focused on advancing evidence-based standards of care for TSC and/or TSC patient/caregiver-reported outcomes, using a combination of patient survey, existing data review, and prospective study designs
  - Develop bi-annual curriculum (e.g. CME/CNE or TSC workshops at professional association meetings) that exposes TSC Clinic Directors and other health care providers to current evidence-based practices for TSC diagnosis, surveillance, and management
  - Participation goal of 136 from TSC Clinics and 80 outside TSC Clinics
  - Develop a telehealth network "TSCNet" to expand access to TSC quality care by 2023
  - Increase TSC clinics providing services to children and adults, inpatient and outpatient services, to 85% by 2023

**Outcomes:**

- **Short Term Research Outcomes**
  - Within 5 years:
    - More clinical trials testing personalized/precision treatments to prevent or eliminate specific TSC manifestations
    - More distinct compounds or interventions advancing toward clinical trials through the Preclinical Consortium
  - **Long Term Research Outcomes**
    - Within 10 years:
      - More clinical trials testing compounds with unique mechanisms of action or gene therapy/editing technologies
      - Neonatal screening with interventions implemented in clinical use to prevent the onset of irreversible, life-altering outcomes

- **Short Term Access/Quality of Care Outcomes**
  - Increase awareness of updated clinical guidelines
  - Lay groundwork for evidence-based treatment standards (including use of telehealth) through new studies and advocacy for improved patient and family care experience

- **Long Term Access/Quality of Care Outcomes**
  - Within 10 years:
    - Expand the number of TSC affected children and adults who have access to high quality care
    - Increase quality of clinical care at TSC Clinics and outside TSC Clinics by better adherence to clinical guidelines
    - Evidence-based treatment standards established, implemented and adopted by recognized TSC Clinics

**Impact:**

As a result of investing directly in TSC research and in infrastructure to support TSC research, identify treatments that improve long-term clinical outcomes and quality of life of individuals affected by TSC and ;the course of the disease for future generations through prenatal diagnosis and preventative treatments.

As a result of expanding access to quality clinical care for TSC, improve short- and long-term clinical outcomes and quality of life of individuals affected by TSC.
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<td>• Create a premiere support services volunteer corps through Clinic Ambassadors, Junior Leaders, Dependent Adult Transition Resource Coordinators, Adult Regional Coordinators, Spanish Support Network, Education Parent Mentors and Community Alliance leadership</td>
<td>• Establish relationships between all Community Alliances and local Arc Chapters where existing by 2023 utilizing Associate Partnership with The Arc US</td>
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<td>• Grow peer to peer support from 20,246 (2017) to 35,867 (10% annually)</td>
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<td>• Expand skills of parents/caregivers and young adults to advocate for and access appropriate educational services to improve quality of life</td>
<td>• Build and maintain working partnerships with 15 national organizations/collaboratives (ARC, Bountiful, CNF, CTF, EF/REN, LAM Foundation, ATS-PAR, ELC, Global Genes, ISAN, NORD, RDCRN, TSC)</td>
<td>• Build and maintain working partnerships with 15 national organizations/collaboratives (ARC, Bountiful, CNF, CTF, EF/REN, LAM Foundation, ATS-PAR, ELC, Global Genes, ISAN, NORD, RDCRN, TSC)</td>
<td>• Increase the Education Parent Mentor program to support school meetings from 74 to 475 by 2023</td>
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<td>• Grow collaborations with other nonprofit organizations internationally, nationally and locally to ensure access to resources, support services, transition tools and information</td>
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<td>• Provide 600 hours of volunteer training per year</td>
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<td>• Expand reach of education and support through use of technology</td>
<td>• Utilize technology or support services app to increase participation by 25% from 2,028 (Community Alliance meetings) attendees (2017) through live virtual meetings monthly in three major time zones, and create a podcast library by 2023</td>
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<td>• Expand to 10 Global Alliances by 2023, growing each countries' constituencies by 15%, increase global TSC Clinics to 18, and develop country-specific strategic plans</td>
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<td>• Increase engagement in educational advocacy and improve tailoring of educational services to needs of individuals with TSC.</td>
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<td>• Increase self-reported social support experienced by individuals and families affected by TSC.</td>
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