

The TSC Patient Experience

A study of the real-world experience of patients and caregivers living with tuberous sclerosis complex.

Includes Input From More Than 950 TSC Patients and Caregivers



Introduction

Thanks to the participation of more than 950 patients and caregivers affected by tuberous sclerosis complex (TSC), we have compiled this overview of commonly reported experiences of those living with the disease. We hope that the knowledge of your peers' experiences with TSC aids in your own understanding of the disease and how it may be managed. Armed with this broad understanding of the possible realities of living with TSC, we hope that you will feel better able to make informed decisions regarding medical care for yourself or the person you care for.

It's important to remember that the content in this report is not intended to be a substitute for professional medical advice, diagnosis, or treatment. Always seek the advice of your physician or other qualified health provider with any questions you may have regarding a medical condition.

We sincerely thank you for taking the time to share your experience with many other individuals affected by TSC, and hope you take comfort in knowing that there is a community of people working alongside you to learn more about this disease.

Study Highlights

- ✓ Most of the survey respondents reported symptoms beginning within the first year of the patient's life
- ✓ 49% of the patients were diagnosed at 3 years of age and younger
- ✓ 48% of patients had genetic testing done to confirm their TSC diagnosis
- ✓ 32% of patients were diagnosed with a different or general seizure disorder prior to receiving a confirmed TSC diagnosis
- ✓ Over 20% reported not being satisfied at all with the current quality of life
- ✓ Approximately 60% reported low to moderate degrees of seizure control
- ✓ 43% said they were starting to pursue or actively pursuing new treatment options



Overview and Methodology

The data for this analysis was collected using an online survey of 964 patients and caregivers living in the United States with a reported diagnosis of tuberous sclerosis complex (TSC).

Key Areas of Research

- ✓ Symptom history
- ✓ Impact of the disease on daily life
- ✓ Experiences obtaining a confirmed diagnosis
- ✓ Familial history of seizures
- ✓ Seizure “triggers”
- ✓ Challenges, frustrations, and support needs
- ✓ Valued sources of information
- ✓ Experience with various treatments
- ✓ Seizures and how well they are controlled



Common TSC Symptoms

Most survey respondents experienced the following initial symptoms. More information about experiences with the diagnosis of TSC can be found on page 7 of this report.



Seizures



Skin Abnormalities



Development Delays



Tumors /Tubers



Behavioral Issues



Cognitive Issues



Infantile Spasms



Epileptic Spasms



Other

Treatments Used to Manage Seizures

Survey respondents reported having experience with a broad range of treatment options. More information about treatments can be found on page 12 of this report.



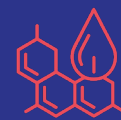
Prescription Medications:
Anti-tumor/
Anti-seizure



Surgery
(Including VNS
or Implants)



Diet



Non-FDA-
Approved CBD
Available Online, in
Stores, or in
Dispensaries



Other

Top Social Media Platforms

Survey respondents reported using the following social media platforms to get and share information about TSC. More information can be found on page 14.



Facebook



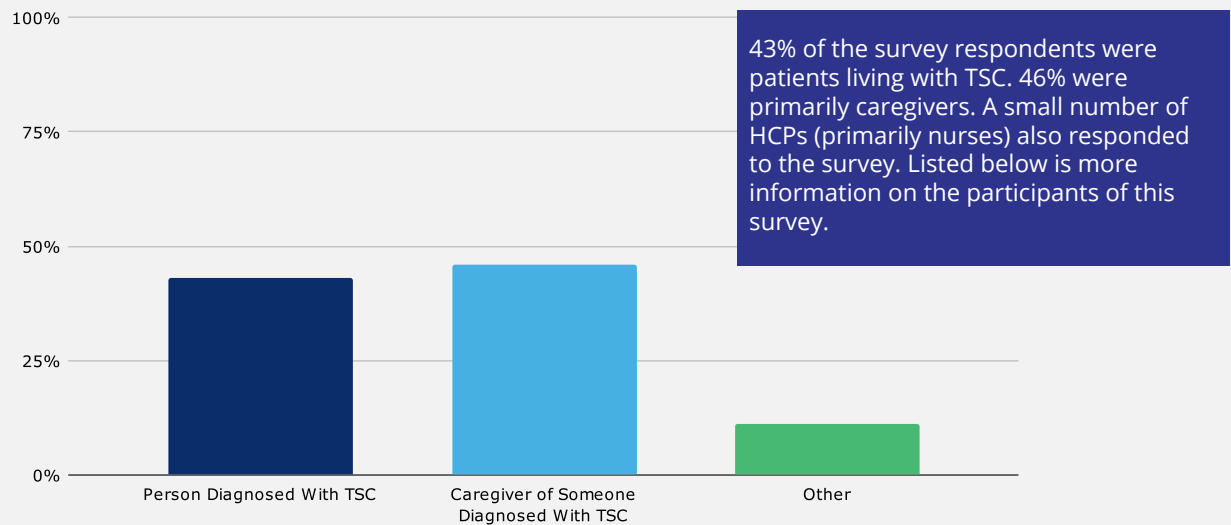
YouTube



Twitter

A Closer Look at Who Participated in This Study

The TSC survey respondents were a blend of patients, caregivers, and other healthcare professionals.

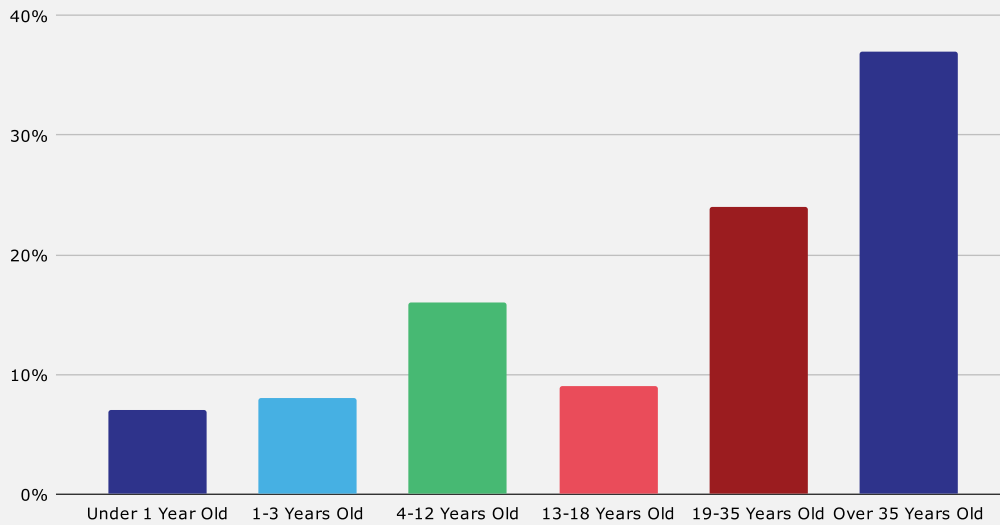


Input From a Broad Range of People Whose Lives Have Been Impacted by TSC

- ✓ TSC patients
- ✓ Parents
- ✓ Grandparents
- ✓ Children
- ✓ Siblings
- ✓ Aunts/Uncles
- ✓ Legal guardians
- ✓ Teachers/Coworkers
- ✓ Other paid caregivers
- ✓ Healthcare professionals (nurses)

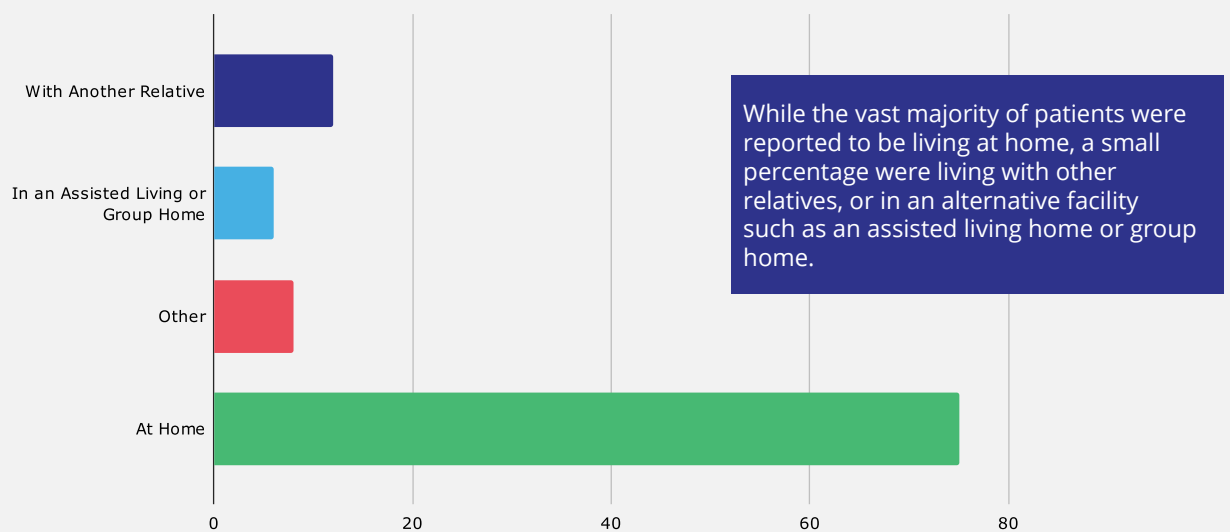
Patient Age at the Time of This Study

The patients represented in this survey are distributed across all age groups from under 1 to over 35 years old. Currently, most (61%) are adults, 18 years of age or older.



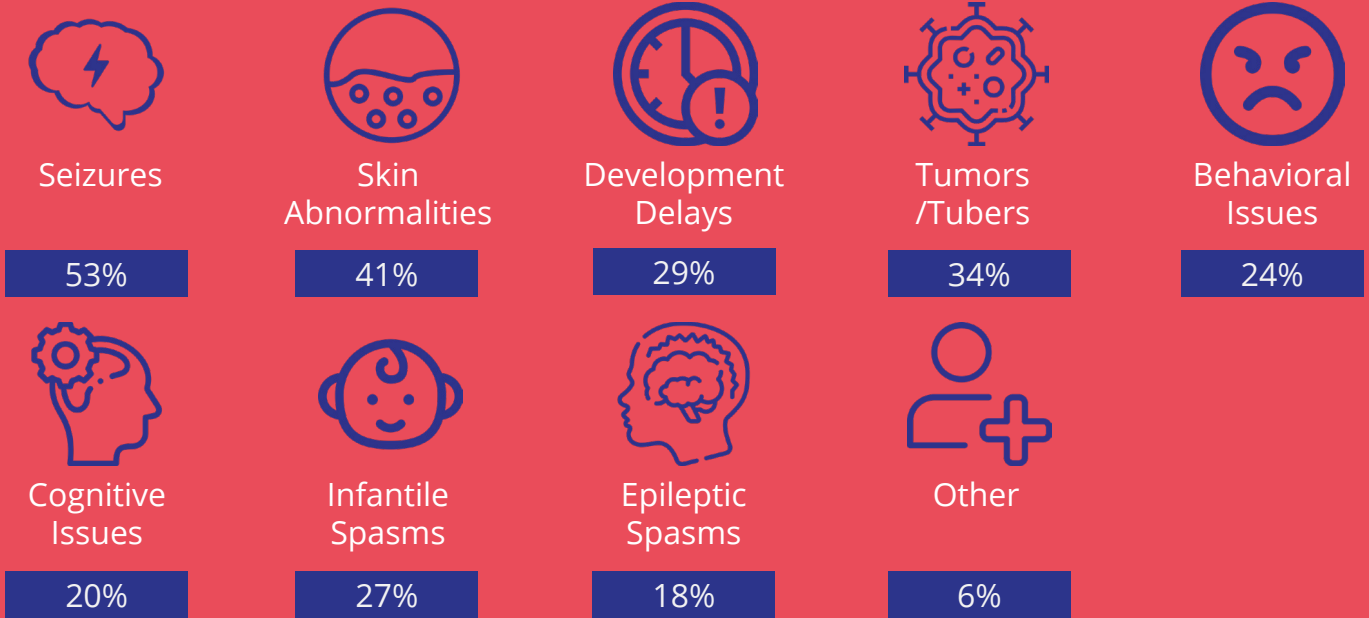
Living Situation

Most TSC patients referred to in this study were living at home at the time the survey was taken.



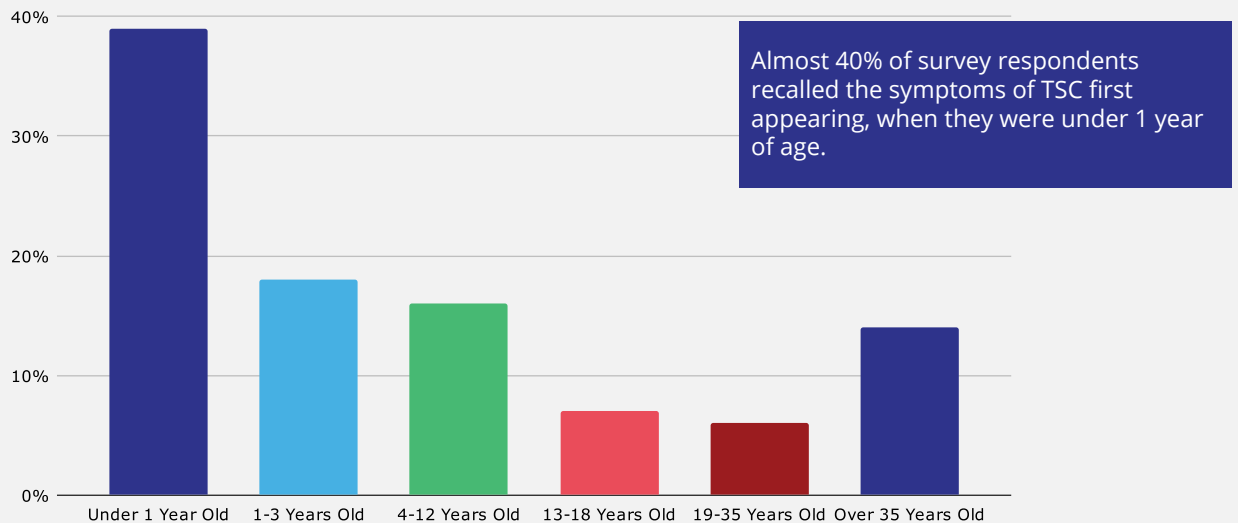
Early Symptoms

Most survey respondents recalled noticing the following initial symptoms of TSC. More information about the group's experiences with diagnosis can be found on page 7.



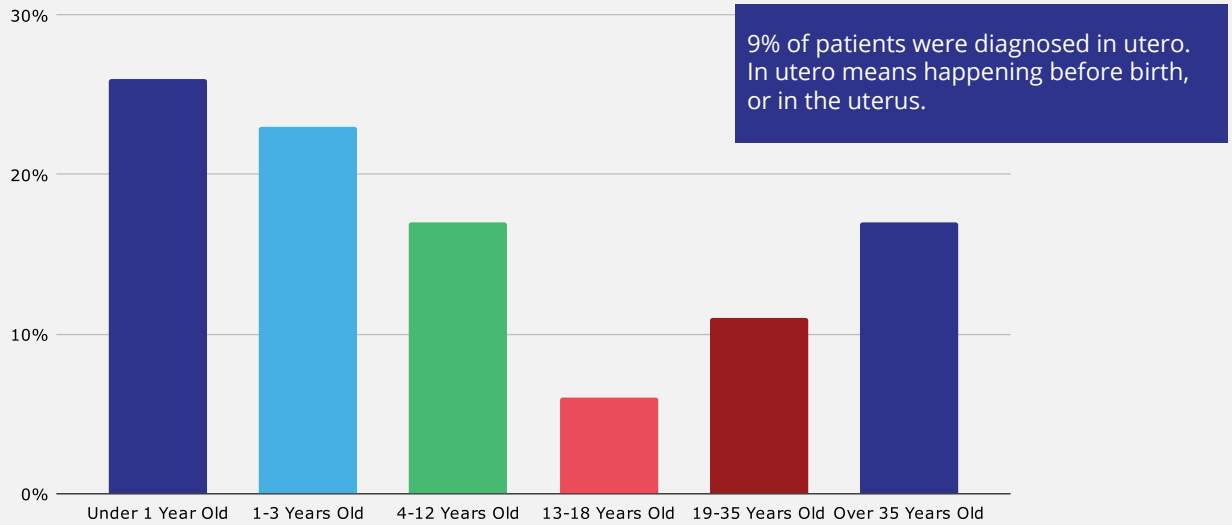
Percentage of survey respondents who recalled these symptoms occurring.

Patient Age When the Symptoms of TSC First Began



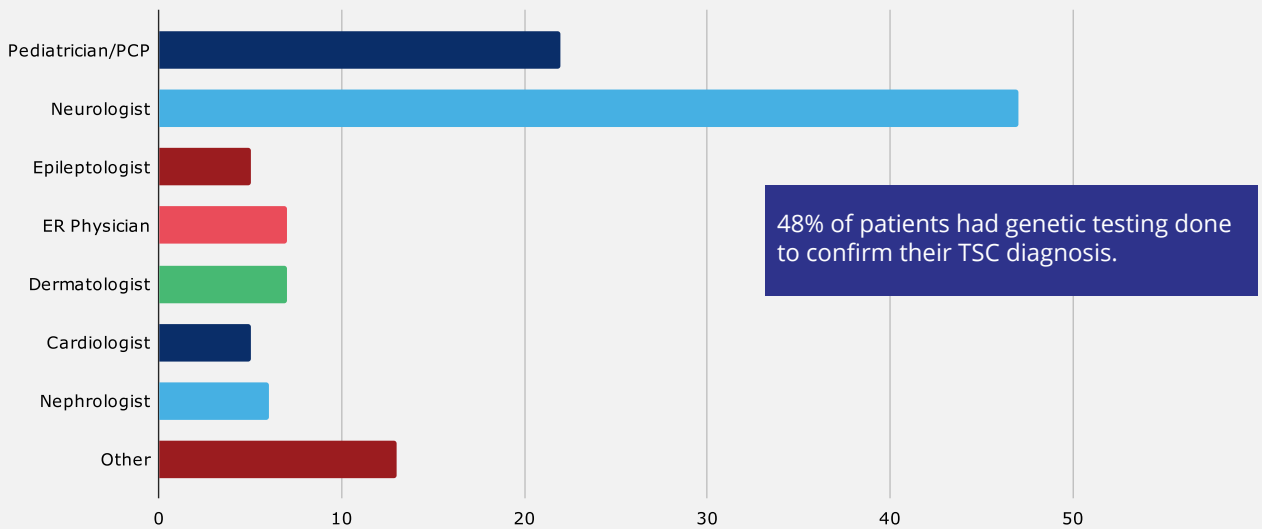
Patient Age at Diagnosis

Approximately 49% of the patients referred to in this survey were diagnosed relatively early in life, at 3 years of age and younger.



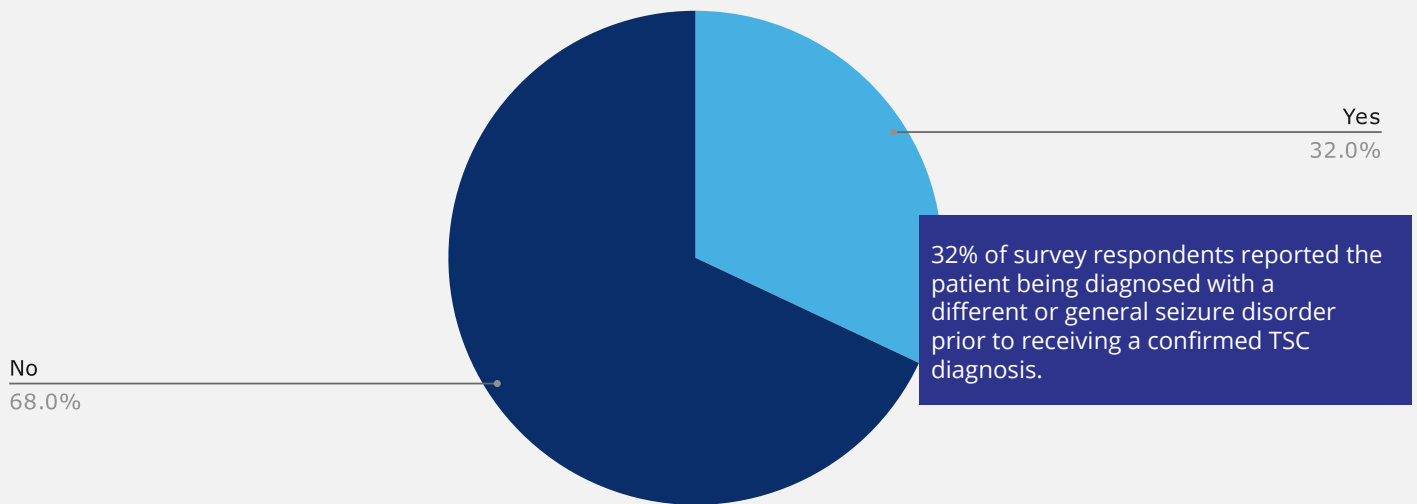
Diagnosing Physician

Over 20% of patients received a confirmed TSC diagnosis by their pediatrician. The remaining patients were diagnosed by a physician specialist. Of these, neurologists were responsible for diagnosing roughly half of patients.



Other Diagnoses

One-third of the patients referred to in this study were diagnosed with a different seizure disorder prior to being diagnosed with TSC.

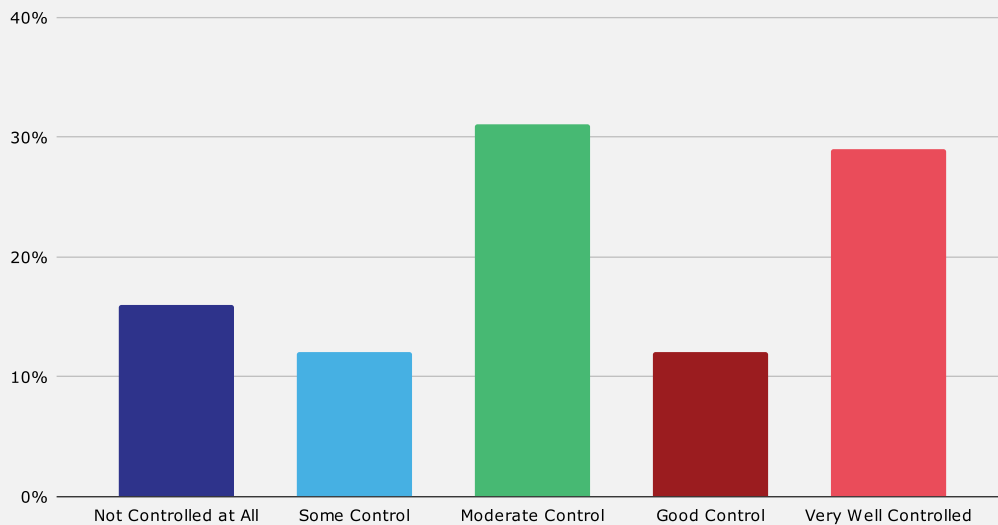


Prior Diagnoses Mentioned in This Study

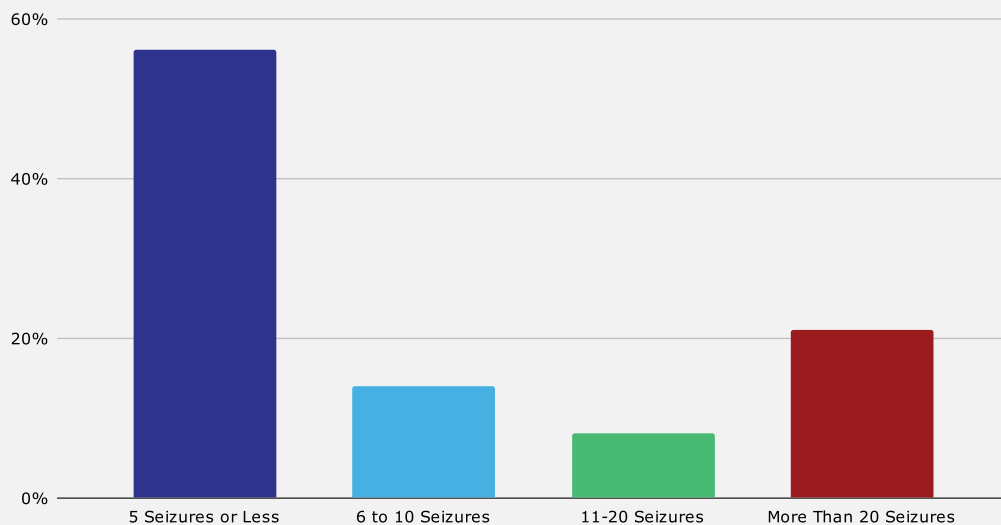
- ✓ Epilepsy
- ✓ Infantile spasms
- ✓ Inoperable tumors on the brain
- ✓ Mucopolysacridosis
- ✓ Schizencephaly
- ✓ Complex partial seizure disorder
- ✓ Multifocal seizures
- ✓ Subcortical band heterotopia
- ✓ Absence/focal seizures

Current Level of Seizure Control

While many survey respondents reported their seizures were very well controlled, almost 60% reported low to moderate degrees of seizure control, with 16% saying the seizures were “not controlled at all.”



Number of Seizures Experienced in an Average Month



How Seizures Relate to Other Health Concerns

Nearly 40% of survey respondents felt that the seizures associated with TSC were equally impactful versus other health issues resulting from the disease. 38% found the seizures to be the most impactful health issue they faced.



28%

Other Health Issues Are Far More Impactful Than Seizures



34%

Seizures Are on Par With Other Health Issues

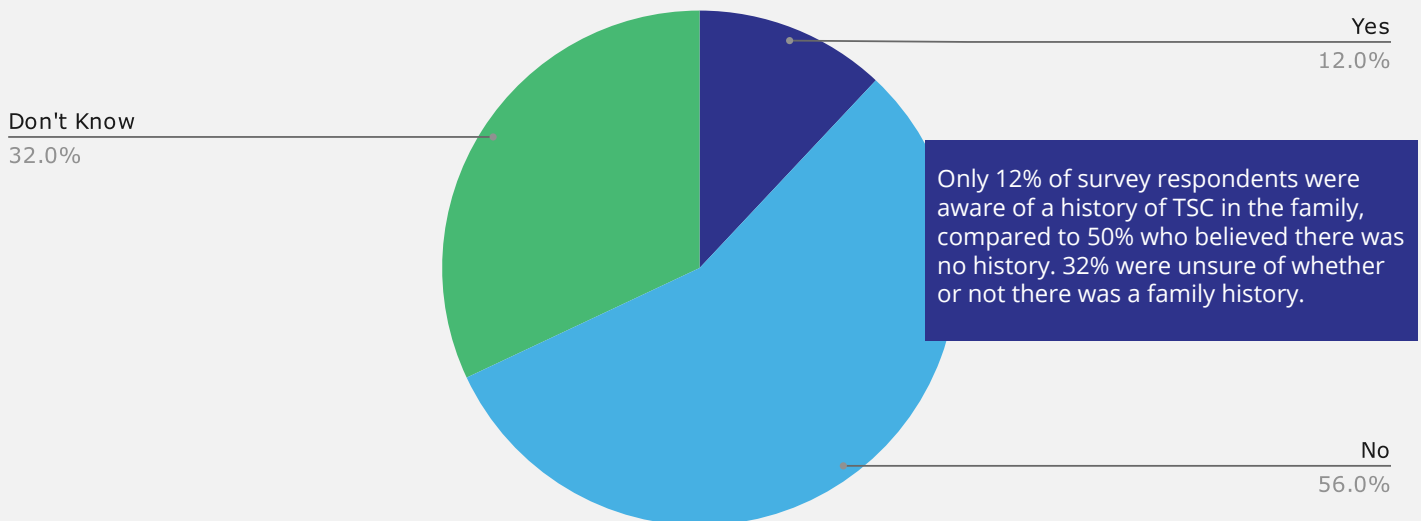


38%

Seizures Are by Far the Most Impactful Health Issue Faced

Percentage of survey respondents who reported the impact of TSC.

Family History of TSC



Seizure Triggers

Survey respondents reported the following activities and environmental stimulus contributing to the onset of seizures.



Strong Emotions

29%



Illness or Fever

21%



Overexertion

19%



Heat

18%



Upon Waking

16%



Flashing Lights

6%



Falling Asleep

13%



Bathing

5%



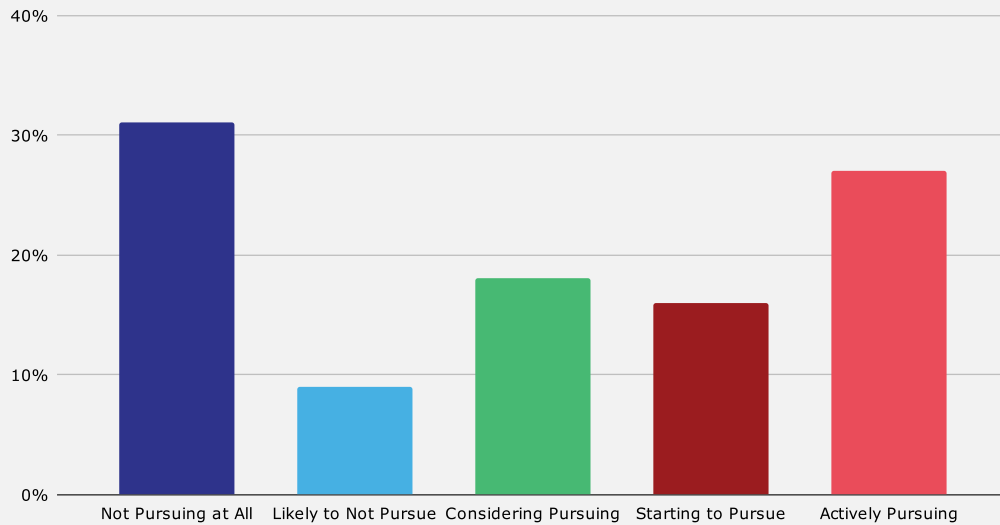
No Known Triggers

29%

Percentage of survey respondents who recalled these triggers occurring.

Looking for New/Different Seizure Treatments

Survey responses about pursuing new seizure treatments were mixed. 31% said they were not pursuing new options at this time, while 43% said they were starting to pursue or actively pursuing new options.



Treatments Used to Manage Seizures



Prescription Medications:
Anti-tumor/
Anti-seizure

24%/81%



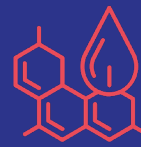
Surgery
(Including VNS
or Implants)

16%



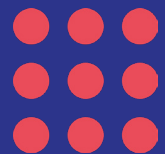
Diet

23%



Non-FDA-
Approved CBD
Available Online,
in Stores, or in
Dispensaries

18%



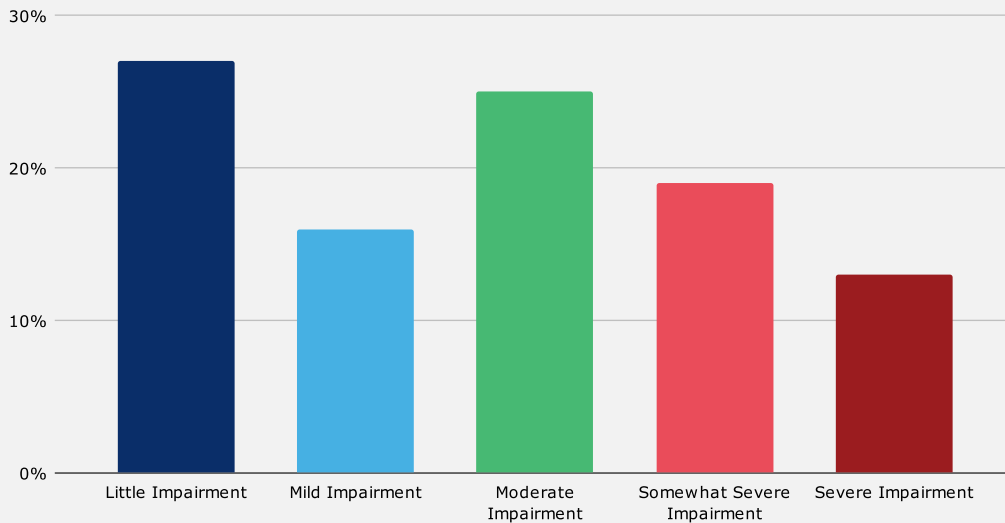
Other

9%

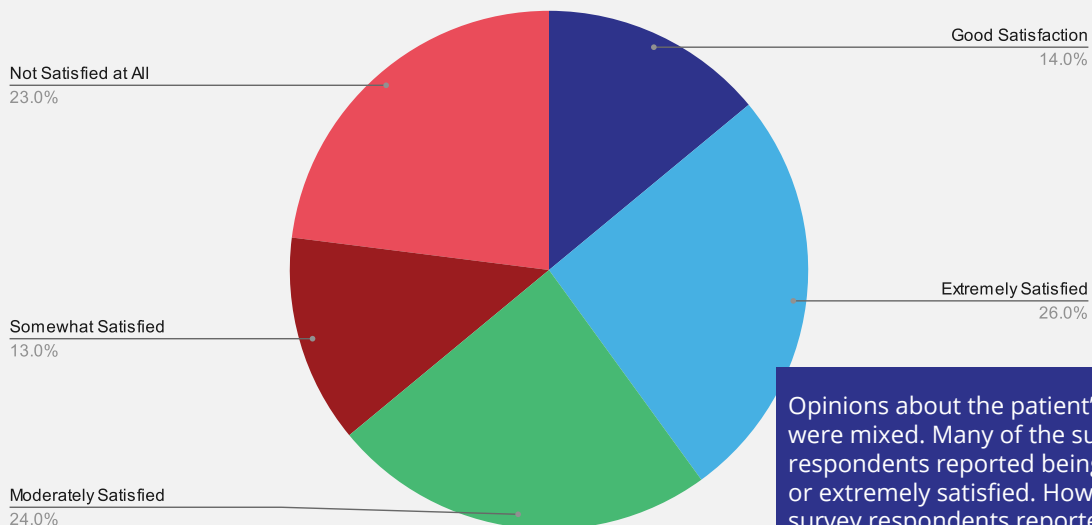
Percentage of survey respondents who used these treatments.

Impairment of the TSC Patient

Survey respondents provided their view of the impact TSC has had. Almost a quarter of the survey respondents felt the disease had little impairment on the patient's life, while an almost equal amount felt the disease resulted in moderate impairment. 13% of survey respondents felt the disease was severely impairing.

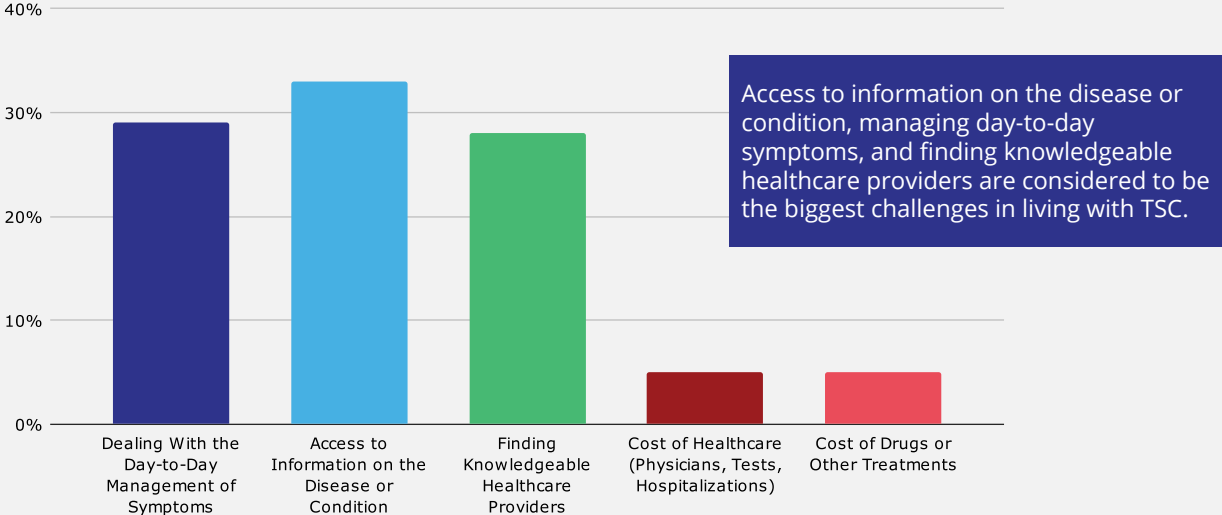


Quality of Life for the TSC Patient



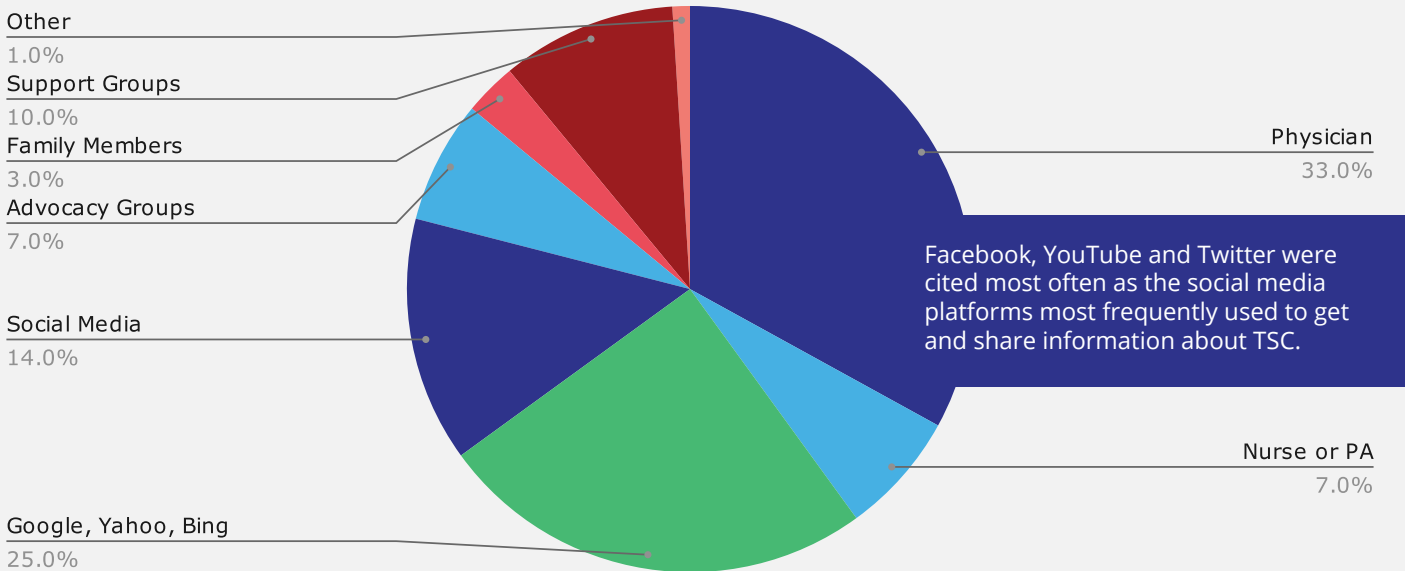
Opinions about the patient's quality of life were mixed. Many of the survey respondents reported being satisfied or extremely satisfied. However 23% of survey respondents reported not being satisfied at all with the current quality of life.

Challenges Caring for Someone With TSC



Valued Sources of Information

Not surprisingly, physicians were mentioned most frequently as a valued source of information. Online search engines and patient advocacy groups were mentioned next most frequently.



QUOTES

We'd like to thank many of our survey respondents for sharing their feelings and emotions with us. It is our hope that through these quotes, others will find comfort in knowing their experiences with TSC are shared by others, and that they are not alone.

"Not sure where to start?"

"He had brain surgery last year and we are on 5 meds and have tried keto."

"Meds work mostly, and seizures are currently controlled."

"She's been on a lot of meds. Her significant seizures now are non-neurological."

"I need help!"

"I'm looking for the right neurologist and I find neurologists that don't know much about tuberous sclerosis... I'm like why don't they know much ugh :("

"I've found what works for me and can't afford to risk having more seizures, at this time. My seizures have been under control since 2008."

"Always looking for better treatments."

"I feel like I've exhausted all options available right now."

"We are at the beginning of the journey."

"My son is extremely depressed. He has seizures daily, 2 brain surgeries, VNS implant, more than 25 different medications, and nothing works!!"

"Seizures have been controlled for over 11 years."

SUMMARY

Tuberous sclerosis complex (TSC), like many rare diseases, is clearly a challenging and difficult disorder to live with. Although each individual's experience is different, life after diagnosis has many challenges for the patients and their family and friends. This study, which includes experiences from over 950 patients and caregivers, captures the group's experiences across the diagnostic and disease management process.

TSC has clearly impacted our survey respondents early in life. Approximately 49% of the patients in this study were diagnosed before they were 4 years old, with almost 40% first experiencing symptoms before they were 1 year old. Seizures, skin abnormalities, tumors/tubers, and developmental delays were three of the most common symptoms which led to seeking medical care and ultimately to a confirmed TSC diagnosis. Further, many patients were diagnosed with a different seizure disorder prior to being diagnosed with TSC.

Clearly, a significant challenge faced by the survey respondents is managing the seizures associated with TSC. 38% felt that seizures were by far the most impactful health issue faced. Some of the most commonly reported triggers to seizures include strong emotions, illness or fever, and overexertion, (29% reported no known triggers). 27% of survey respondents reported actively looking for new and different treatments. Commonly used treatments used to manage seizures included prescription anti-seizure medications, CBD, surgery, and diet modification.

Finally, survey respondents voiced a high degree of dissatisfaction with the quality of life of the TSC patient. Approximately 36% of survey respondents reported not being satisfied at all, or only somewhat satisfied with the quality of life for the TSC patient. These findings underscore the need for new and different approaches to managing this disease.

ABOUT PATIENT JOURNEYS

At Patient Journeys, our mission is to support rare disease patients and their caregivers by helping them access the best care possible. We believe, that by sharing information and experiences with each other, we can accelerate the diagnostic process and help patients receive the right treatment sooner.

Patient Journeys was started by the parents of children living with rare disorders who have experienced first-hand the challenges that exist with getting accurate and relevant information, and the critical role patients and caregivers can play to help support others in need.

If you have any questions or comments, please email us at info@patientjourneys.org.

