



Empowering Children. Supporting Families. Building Brighter Futures.

**Resource 071 | Genetic and chromosomal conditions**

# Tuberous sclerosis complex

## What it means

Tuberous sclerosis complex (TSC) is a genetic condition that can cause growths in several organs and affect brain development. Seizures, learning differences, and behavioral or communication needs may occur, but not every child has developmental impairment. Ongoing surveillance helps detect problems before they become obvious. [1]

## Causes and risk factors

Changes in TSC1 or TSC2 affect a pathway that regulates cell growth. A change may be inherited or arise for the first time in the child. Features vary widely, including within families. Genetic counseling supports understanding of the result and options for appropriate family evaluation. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



## Signs and everyday impact

Possible early signs include pale skin patches, seizures, infantile spasms, or heart growths found before birth. Later concerns can involve skin, kidneys, eyes, learning, attention, autism, or behavior. Some organ problems are initially silent. Repeated short spasms or loss of skills need prompt attention. [1,2]

## Assessment and diagnosis

Diagnosis uses established clinical criteria and genetic testing when appropriate. Specialists assess the brain, kidneys, heart, skin, eyes, and other relevant systems, alongside development and behavior. EEG and imaging are scheduled according to age and clinical findings. Surveillance remains important even when a child seems well. [1,2]

## Treatment and therapy

Treatment is directed at specific problems. Seizures may need syndrome-specific medicines, and selected children may benefit from surgery or targeted medicines affecting the growth pathway. Organ growths require specialist monitoring or treatment. Developmental therapies, AAC, educational support, and care for attention, sleep, and emotional needs complement medical treatment. [1,2,3]

## Support at home

Keep a shared surveillance calendar and written seizure plan. Learn what the child's usual events look like and report changes. Support communication and meaningful play, with goals based on the individual child. Ask about family support and how developmental, behavioral, and medical teams can coordinate appointments.

## When to seek help

Suspected infantile spasms require same-day urgent assessment. Use the seizure plan for prolonged events. Severe pain, blood in urine, breathing difficulty, or marked reduction in alertness also needs urgent medical review.

## Questions for your care team

- Which surveillance tests are due and why?
- How should we recognize and respond to spasms or seizures?
- Are targeted treatment and developmental supports appropriate?



## Podcast preparation

Use this outline to develop a later podcast script.

### Suggested opening

Welcome to Mariel Early Intervention. Today we discuss tuberous sclerosis complex. Tuberous sclerosis complex (TSC) is a genetic condition that can cause growths in several organs and affect brain development. This is general education; each child needs individualized care.

**Opening:** Define the topic and its developmental context.

**Understanding:** Explain causes and signs without inviting self-diagnosis.

**Assessment:** Describe evaluation and distinguish screening from diagnosis.

**Support:** Discuss treatment, therapy, and practical family supports.

**Closing:** Explain urgent signs and one clear next step.

Misconception to address: Myth: all children with TSC have severe learning difficulties. Developmental effects vary widely.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

### Terms in plain language

- TSC1 and TSC2: genes involved in regulation of cell growth.
- Surveillance: scheduled checks for possible health complications.
- mTOR pathway: a cellular signaling system involved in growth regulation.

### Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

**[1] Tuberous Sclerosis Complex**

<https://www.ninds.nih.gov/health-information/disorders/tuberous-sclerosis-complex>

**[2] INFANTILE EPILEPTIC SPASMS SYNDROME (IESS)**

<https://www.epilepsydiagnosis.org/syndrome/west-syndrome-overview>

**[3] 6 Treating childhood-onset epilepsies**

<https://www.nice.org.uk/guidance/ng217/chapter/treating-childhood-onset-epilepsies>