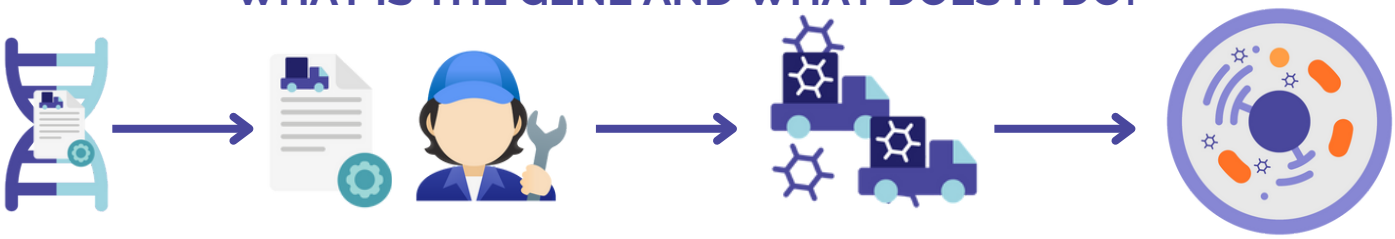
SLC13A5 EPILEPSY

A genetic epilepsy with global developmental delays and seizures beginning within the first days of birth

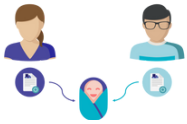
WHAT IS THE GENE AND WHAT DOES IT DO?



SLC13A5 is a gene in your DNA that tells your body how to build a citrate transporter protein. Citrate is a small molecule that is found throughout your cells and plays an important role in helping cells make energy. The protein carries citrate from across the cell membrane into your cells.

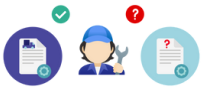
Researchers are still trying to understand whether the symptoms of this disorder are the result of not enough citrate in the cell, too much citrate outside the cells, or both.

HOW IS THIS DISORDER INHERITED?



Like all genes, you inherit one copy from each of your parents.

If both sets of instructions you inherit are functional, your body can effectively produce the citrate transporter.



Even if only one of the sets of instructions you inherit from either parent is functional, you can still produce the transporter. This is called being a carrier of the gene.

If neither copy is functional, the transporter your body creates is extremely ineffective at transporting citrate into the cell.

SLC13A5 Epilepsy is an autosomal recessive disorder. If both parents are carriers of an abnormal SLC13A5 gene, there is a 1 in 4 chance that their child will have this severe form of epilepsy.

SYMPTOMS

Severity of symptoms is on a spectrum for individuals with SLC13A5 Epilepsy and include:

- Seizures beginning shortly after birth
- Movement problems
- Behavioral and developmental delays
- Difficulty speaking and limited speech
- Sleeping and eating issues
- Poor teeth development / weak enamel

TREATMENTS

Currently, there are no treatments or cures. Individual symptoms can be managed with:

- Anti-seizure medications (often multiple)
- Physical therapy
- Occupational therapy
- Speech therapy
- Feeding therapy
- Assistive devices

JOIN OUR PATIENT REGISTRY: tessresearch.org/join-registry

TESS RESEARCH FOUNDATION

WHO IS TESS?

A patient-centered non-profit organization driving research for treatments and a cure for SLC13A5 Epilepsy

A worldwide community of doctors and scientists

An advocate and resource for families

We do this **across countries** because SLC13A5 Epilepsy has no geographic boundaries. We do this **collaboratively** because it takes a team to improve treatments and find a cure. We do this with **urgency** because our loved ones are sick now.

We will have succeeded when all children with SLC13A5 Epilepsy can live healthy, independent lives

12 new families added to our contact registry

Launched 3 in-person Natural History Study Sites

Attended 5 conferences

2022 At-a-Glance

12 US/Canada-based family support meetings

8 community electronic newsletters

Hosted an International Research Conference in Europe

4 family print newsletters

2 Scientific Advisory Board meetings

Over $2M in research funded since 2015

$250,000 in new SLC13A5 research awards

Organized and hosted Clinical Research Conference

10 Science Simplified articles

SCAN A QR CODE TO LEARN MORE

VIDEO: What is TESS Research Foundation?

ARTICLE: Stanford Medicine - Breaking the Code

VIDEO: TESS Research Foundation's Achievements

TESS RESEARCH FOUNDATION for SLC13A5 Epilepsy

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