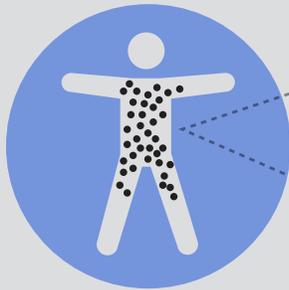
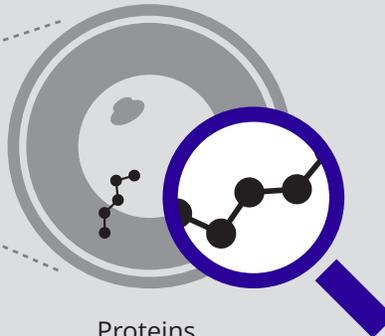


# SLC13A5 Deficiency

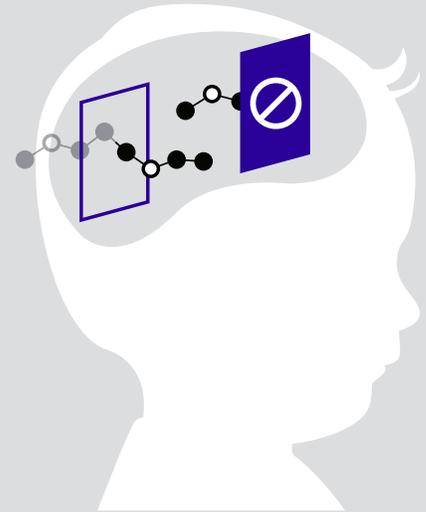
A genetic form of epilepsy and developmental delay with seizures beginning within the first days of infancy



Cells  
(= building blocks)



Proteins  
(= transporters)



SLC13A5 Deficiency  
(= less citrate to brain)

## ABOUT SLC13A5 DEFICIENCY

SLC13A5 Deficiency is a newly diagnosed form of genetic epilepsy and developmental delay with seizures beginning within the first days of life.

The body is made up of building blocks, called cells. Cells need proteins, called transporters, that act as “doors” through which nutrients and other chemicals enter and leave the cell.

Diagnosed patients have a deficiency of one such transporter, called SLC13A5. This transporter brings in a chemical called citrate into brain cells. When this transporter does not work, brain cells likely have less citrate and this is believed to cause seizures.

## DIAGNOSIS

A simple genetic blood test or saliva test is now available through Baylor, GeneDx, Blueprint Genetics, Courtagen and several other laboratories to test for SLC13A5 Deficiency.

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**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.**

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## SLC13A5 DEFICIENCY SYMPTOMS

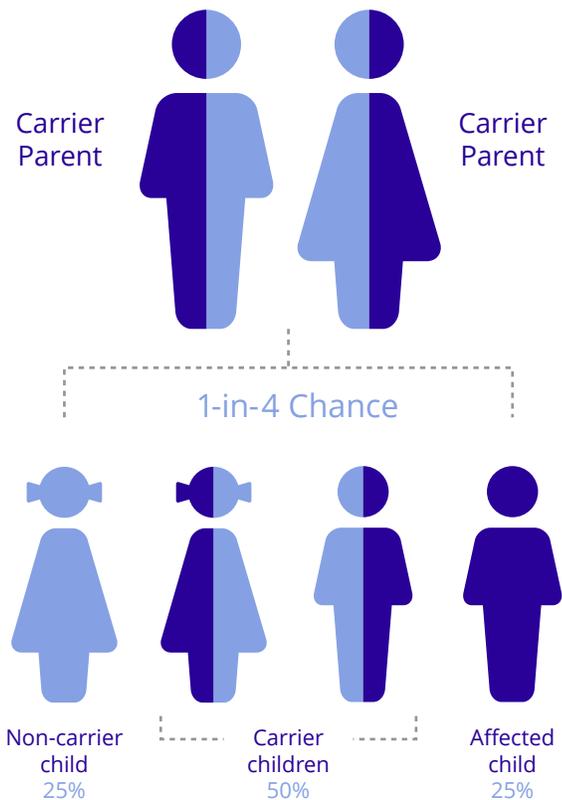
- Seizures beginning soon after birth
- Persistent seizures of multiple types
- Severe difficulty with speech production, with better ability to understand language
- Movement and coordination problems (low tone and unsteady)
- Episodes of body stiffening or weakness lasting a few minutes to a few hours
- Brain MRI that appears normal or has subtle changes in the white matter
- Tooth enamel abnormalities
- Mildly elevated Citrate levels in blood samples

## TREATMENT OPTIONS

At this time, there are no specific treatments that cure the disease. Treating seizures in children with SLC13A5 Deficiency has proven difficult in some patients. Children can succumb to complications of the seizures. However, the oldest known child with this disease is now 26 years old, fairly stable, but dependent on caregivers.

Please contact TESS Research Foundation to inquire about best practices and any currently available clinical trials. Please speak to your neurologist for specific treatment options for your child.

## AUTOSOMAL RECESSIVE INHERITANCE – HOW THE ABNORMAL GENE GETS PASSED DOWN THROUGH FAMILIES



Genes are a blueprint for making proteins including SLC13A5. With few exceptions, all people have two copies of every gene. In many cases, the extra copy can act as a “back up” if one of the copies is defective. So far, it appears that you need to have a change in the DNA sequence of both copies of your SLC13A5 gene in order to have SLC13A5 Deficiency. This means that you must inherit a copy with an abnormal sequence from both your mom and your dad (autosomal recessive inheritance). Because mom and dad have just a single defective copy of the gene, and they appear healthy, it appears that you only get the disease when you have two abnormal copies.

## SLC13A5 Deficiency

A genetic form of epilepsy and developmental delay with seizures beginning within the first days of infancy

### RESOURCES

If you or someone you know possibly has SLC13A5 Deficiency, we encourage you to reach out to the following resources for more information:

**Kim Lodato Nye, Founder and President,**  
TESS Research Foundation and mother  
of two children with SLC13A5 Deficiency  
[kim@tessfoundation.org](mailto:kim@tessfoundation.org)

**Brenda Porter, MD, PhD,**  
Associate Professor of Neurology,  
Stanford University  
[brenda2@stanford.edu](mailto:brenda2@stanford.edu)

**Matthew Bainbridge, PhD**  
Associate Director of Clinical Genomics Research,  
Rady Children's Hospital  
[mbainbridge@rchsd.org](mailto:mbainbridge@rchsd.org)