DIAGNOSIS

Currently, the way to identify a citrate transporter disorder (SLC13A5 Deficiency) as the cause of a child having intractable seizures is to look at the DNA sequence of the SLC13A5 gene. Only if both the copies of the child's SLC13A5 genes are mutated, it is considered to be disease causing. For diagnosis, either whole exome sequencing (WES) can be performed OR targeted panel sequencing (SLC13A5 is included in many epilepsy panels) can be performed which is often less expensive and faster to get results.

Here are a few of the companies that can test for SLC13A5 mutations:

- Centogene AG-the rare Disease Company, Germany
- GeneDx, United States
- Prevention Genetics, United States
- Genetic Services Laboratory, University of Chicago
- Blueprint Genetics, Finland
- Medizinisch Genetisches Zentrum München MGZ München, Germany
- Asper Biotech ltd, Estonia
- Fulgent Genetics, United States
- Ambry Genetics, United States
- CGC Genetics, Portugal
- Greenwood Genetic Center Diagnostic Laboratories, United States
- VU University medical center metabolic Unite, Netherland
- Invitae, United States

So far it appears that you need to have a change in the DNA sequence of both copies of the SLC13A5 genes. This means that you might inherit a copy with an abnormal sequence from both your mother and your father. Because mom and dad have a single copy of the gene that is abnormal, and they seem well, it appears that you only get the disease when you have two abnormal copies.