

FARS2 DEFICIENCY

What is it?

FARS2 deficiency is a rare, autosomal recessive genetic disorder. The disease severity ranges between two phenotypes: infantile-onset epileptic mitochondrial encephalopathy and later-onset spastic paraplegia. The infantile-onset phenotype is characterized by epileptic encephalopathy with lactic acidosis and a poor prognosis. This phenotype accounts for 70% of affected individuals. The later onset phenotype is characterized by spastic paraplegia, less severe neurological symptoms, and a longer survival rate. This phenotype accounts for 30% of affected individuals.

Infantile-onset phenotype

(Typically diagnosed birth to six months of age)

- Seizures that may be difficult to control
- Truncal hypotonia
- Myoclonus
- Failure to thrive (FTT)
- Lactic acidosis
- Motor, speech and cognitive delays
- Feeding difficulties
- Visual impairment
- Sleep apnea

Later-onset phenotype

(Typically diagnosed 6 months of age and older)

- Seizures, brief and resolve over time
- Spastic paraplegia characterized by weakness, spasticity, and exaggerated reflexes of the lower extremities
- Walking difficulties
- Motor, speech, and cognitive delays
- Bladder symptoms (i.e. incontinence)

What is the treatment?

There is no cure for FARS2 disorder. Treatment is focused on the specific health issues and the related symptoms. A team of medical and developmental specialists are key to the best possible outcome. Those diagnosed with FARS2 deficiency will often be followed by numerous specialists which might include neurologists, neurodevelopmental pediatricians, and physiatrists. The pediatrician will often manage the multidisciplinary approach. Physical, occupational and speech therapy are also part of the treatment team for children with FARS2. Medications can be prescribed to reduce and control seizures. Treatments aimed at reducing spasticity include: anti-spasticity medications, Botox injections, and possible surgical interventions.



Kennedy Krieger Institute

The **Specialized Health Needs Interagency Collaboration (SHNIC)** program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Supporting students with FARS2 deficiency in the school setting requires the educators and parents/guardians to work as a team. A well coordinated plan promotes success for the student in the educational setting. Some accommodations to consider for an 504/IEP might include:

- Early intervention assessment for young children
- PT/OT/SLP/Vision specialist consults
- Emotional support
- Education about seizure management for staff
- Monitor poor motor skills
- Monitor visual, hearing, speech impairment
- Communicate behavior signs for when a break is needed
- Offer designated rest area
- Copies of notes, whiteboard, presentations in proper font size
- Consider assistive technology
- Adaptive PE
- Extended time for testing
- Provide support during transitions
- Accommodations if student is having surgery
- Revisiting work if student has seizures
- Extra time for assignments as needed
- Offer emotional support
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis including all affected systems and symptoms
- Communicate with school staff, parents, and provider any changes or concerns about the disease
- Current medication list for home and school
- Documentation of associated medical problems including seizures, feeding issues, bladder, etc.
- Documentation/log of seizures including type of seizure, description of, typical length, characteristics, triggers, warning signs, how often seizures occur, and student's behavior following a seizure
- Orders for emergency medications, when to administer, dose, route
- Feeding protocol including supervision, safety, positioning
- Nutrition orders, use of feeding tube and/or tube replacement policy
- Orders for ambulation, orthotics, or adaptive equipment
- Orders for medical devices (Baclofen pump, feeding pump)
- PT/OT/SLP/Vision services and assessment
- Rest period following seizures and return to class protocol
- Emergency Care Plan (ECP) related to seizures and other medical needs in the school setting and staff training as appropriate for each

Resources & Manuals

NCBI National Center for Biotechnology Information

<https://www.ncbi.nlm.nih.gov/books/NBK538658/>

NIH US National Library of Medicine-Genetics Home Reference

<https://ghr.nlm.nih.gov/gene/FARS2>