Some information about epilepsy caused by mutations in the DNM1 gene:

DNM1 is a gene that contains the instructions to make the Dynamin 1 protein. Dynamin 1 is an enzyme that is important for synaptic vesicle function in the brain, especially during postnatal development. This protein is essential for communication between nerve cells.

What medical conditions are associated with mutations in this gene?

- Early Infantile Epileptic Encephalopathy Type 31 (EIEE31)

What are the some of the features of DNM1 epilepsy (EIEE31)?

- Epileptic encephalopathy
- Delayed development (in some patients)
- Developmental regression
- Seizures, often not response to treatment
- Intellectual disability, severe to profound
- Lack of speech
- Inability to walk or difficulty walking
- Hypsarrhythmia seen on EEG as well as multifocal discharges, spike-wave discharges and background slowing
- Cerebral atrophy (in some patients)
- Poor or absent visual fixation
- Hypotonia
- Often inherited from one parent in an autosomal dominant manner or may occur de novo (not seen in either parent)
- Onset of symptoms in the first months of life

Who can I contact for more information about this type of epilepsy?

Please contact these groups for more information about DNM1 epilepsy:

- Mutations in Gene DNM1 Facebook Group

*If you know any other groups that can be added to this list, please email EGI@CUREepilepsy.org.*
References


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