

# What is Lafora Disease?

Lafora Disease is the most severe form of human epilepsy. It is an inherited myoclonus epilepsy syndrome. Most cases of Lafora Disease are caused by mutations in one of two known genes: EMP2A and EMP2B. Both genes are in chromosome 6. The gene EMP2A makes the protein called Laforin and the gene EMP2B makes the protein called Malin. A few cases of Lafora Disease are caused by a yet unidentified gene(s). The disease most commonly starts as epileptic seizures in adolescence. Rarely, it begins in 5- to 6-year-old children as a learning disorder. There is a higher incidence of the disease in children of Middle Eastern, Southern European (Spain, France, and Italy), South Asian (India and Pakistan) and North African descent. The disease appears to affect males and females equally.

## THE SYMPTOMS

- Recurrent, Increasingly Intractable Seizures
- Cognitive Decline
- Ataxia (difficulty controlling muscles)
- Myoclonus
- Difficulty Walking
- Difficulty Eating
- Speech Difficulty
- Dementia



## WHAT DOES LAFORA DISEASE LOOK LIKE?

The extent of the disease is devastating. Although the child is born with Lafora, the disease typically does not manifest itself until adolescence. Children grow up normally, with no perceivable problems until they begin to suffer with seizures. That is one of the cruelest things about Lafora. A normal, beautiful, and apparently healthy child who has everything to live for is faced with nothing more than a “death sentence.” Most patients are misdiagnosed with JME, Juvenile Myoclonic Epilepsy until seizure medications become ineffective. Genetic testing is used to determine a Lafora diagnosis. Therapy is primarily palliative and aimed at reducing seizures. From manifestation, which is normally the first seizure, a Lafora victim will die typically within 10 years. There is no cure.

## OUR MISSION

**To improve the lives of those affected by Lafora Disease and help accelerate the development of treatments.**

This requires raising awareness about Lafora Disease and the children it afflicts. Chelsea's Hope engages key stakeholders and supports the Lafora community with connections, assistance for families in the form of education, emotional support, and general assistance.