

# Leigh's Story, Sarah's Mom

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## My daughter's complicated journey

At the age of 19, my daughter was diagnosed with Lambert-Eaton Myasthenic Syndrome (LEMS) and another rare condition. Her diagnostic journey was long and complicated. At the age of 15, she began experiencing unexplained weight loss. She dropped more than 30 pounds in a few short months. Other symptoms began to surface like difficulty standing from a sitting position, falling, difficulty walking, tripping, passing out, sensory loss in her hands and feet. Her eyes quit moving, she had GI issues and eventually stroke-like episodes which left her unable to walk for long periods of time.

She had a very comprehensive work up by many specialists. More than 26 doctors have seen her since this all started. Her medical work up consists of over 400 blood tests, 50 diagnostic procedures including 2 muscle biopsies, 5 lumbar punctures, EMGs, nerve conduction studies and over 25 ER visits.

## Searching for treatment

After her diagnosis we were told she needed a medication that was not FDA-approved, but we did not know how or where to get it. I tried to get her into a clinical trial, but the timing of her diagnosis made her ineligible for trial admission. I began to call compounding pharmacies to have the medication compounded and found one locally. Sadly, the dose was too high, which made her ill, and we didn't realize we had to refrigerate the medicine. She discontinued that after a week.

## A turning point

Then in the spring of 2015 she read about a LEMS conference in Orlando. We could not wait to attend. There we connected with a doctor that understood LEMS and learned about a new, investigational medication available through expanded access programs. An unexpected highlight of this conference was meeting and talking with other people and families living with LEMS. This was a pivotal point of transformation in her journey. She had a spirit of hope breathed into her that I had not seen before.

