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One families journey with ALSP

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ALSP is a neurodegenerative disease caused by a mutation to the CSF1R gene. This gene mutation is autosomal dominant, which means that any child of a carrier has a 50% chance of inheriting the gene and the disease. ALSP makes up about 10 to 25% of all adult-onset leukodystrophies globally. There are approximately 10,000 people with this gene mutation in the United States alone. The average age of onset of symptoms is around 43 years old and the disease affects women and men equally. Each patient's symptoms are unique to them, but throughout their disease, most show symptoms of cognitive impairment, behavior and psychiatric issues, motor impairment, speech difficulties, and seizures. Once symptoms begin, they progress quickly. This is a devastating disease where a patient dies within an average of 5 to 7 years after initial onset of symptoms. Having seen 5 of my family members become symptomatic and die from ALSP, I understand that each patient has their own unique symptoms. My family has been affected by ALSP for over 20 years. My mom, aunt, and uncle died after being misdiagnosed with other diseases like Pick's Disease, Binswanger Dementia, Cortical basal Degeneration, and Lewy Body Dementia. Each had their own variety of symptoms, so the doctors weren't able to confirm they had the same disease based on their symptoms alone. However, after my uncle died, an autopsy was done and neurologists from the University of Pennsylvania who had been following my family confirmed the right diagnosis. My uncle, and then soon confirmed in my mom and aunt's tissues, had a mutation to the CSF1R gene, confirming the diagnosis of ALSP. Luckily for our family, my mom, aunt and uncle had donated brain tissue to the brain bank allowing researchers to confirm the gene mutation.

Biography

I am Heidi Edwards, President and founder of Sisters' Hope Foundation, a non-profit organization supporting families affected by adult-onset leukoencephalopathy with axonal spheroids and pigmented glia, or ALSP.

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