

A Pompe Story

My name is Trae. I am a 36 year old and was diagnosed with Pompe disease in 1991. I have been married to my husband Paul for thirteen years and we have a twelve year old daughter named Haley.

As I said I was diagnosed in 1991 during my third year of college. I must say I already knew I had the disease prior to diagnosis as my sister was diagnosed years earlier with Pompes. She has always been weak and is now trached and unable to move. I was weak as a child but nothing like my sister. While in college I began to exhibit tell tale signs of the disease. I was devastated as I knew what was in store for me. I had watched it slowly rob my sister of all mobility and dignity. I knew we would neither grow to be elderly. My sister would never get to be a mother.



I however became pregnant in 1993. I was elated. My goal in life was to raise a child. In my sixth month it became hard to walk and in my seventh month I was put on a bi-pap machine to breathe. Due to my critical state my daughter was taken six weeks early to save both of our lives. Haley McKay was born on September 9th, 1993. A healthy 4lb baby girl. Truly my reason to live.

After Haley's birth I began to deteriorate. I went from being independent to relying on many people. I was a mother who needed others to help raise her daughter. I am still a mother who needs others to raise her daughter, just on a much larger scale.

My current condition? Where do I start? I can still walk at times. If I do walk I use a walker and my breathing only allows me to walk short distances without needing my ventilator. I currently have only 17% lung function. I must use my vent most of the time. I can be off of it for short periods of time however I build up CO2 and become quite sick. If I fall down I can't get up without someone picking me completely up. If I fall on my back I must pray someone is close as I can't right myself and I have NO ability to breathe when on my back. If my head falls backwards my daughter or husband must push it back up. I have to have my husband turn me over in bed or sit me up should I need to get up. My daughter helps me shower and must pick me up off of the toilet when I go to the restroom. I can't brush my hair as I can't lift my arms to my head. I can't even lift my arm to scratch my nose or face. I can't do any of my Adel's without help. Finally, I have a feeding tube as my swallowing has become dangerous and choking is a daily occurrence.

This disease has devastated my life as well as my families. My daughter sees me getting worse and lives in fear that I will stop breathing at anytime. She has become 'the mother' and takes care of me in everyway. She cooks, cleans and helps me with my daily needs. She has to watch her aunt slowly die before her eyes and know that her mother is on the same path.

Without Myozyme being approved soon I face a certain future. A future where I can't move or breathe. I do not have two years to wait. I was in the LOPOS (observational study) and was told I was too severe to continue in the LOTS (late onset treatment study). TOO SEVERE is how they put it. My only hope is that the FDA approves Myozyme on a broad based label for infants AND ADULTS. There are many adults who are mild and have time to wait. However there are many adults that are dying before their families eyes. I pray the FDA will have the understanding and knowledge that Pompes is one disease and it affects us all, infants, children and adults. We all have beating hearts and are in need of Myozyme. Please help us, please help us ALL.

Trae



Trae with two of her cousins