YARBOROUGH FAMILY
Pompe Disease

These are my two beautiful boys. In 2014, when I was 20 weeks pregnant with my first son, Wesley, my husband had a seizure. We discovered he had brain cancer. It's been a long journey, but he is stable. We were excited to start life anew with our baby. He was born in January 2015 and simply perfect. We decided to have another child, Evander, and he was born September 2018. At 5 days old, we received a call from the pediatrician stating his **Newborn Screening** results came back with an abnormality. Through a long, terrifying, inconsolable period, we learned BOTH of our perfect boys had **Pompe Disease.**

Pompe disease is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body's cells. The accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally. Most individuals with late-onset **Pompe disease** experience progressive muscle weakness, especially in the legs and the trunk, including the muscles that control breathing. As the disorder progresses, breathing problems can lead to respiratory failure. However, in 2006, the first FDA drug was approved to treat Pompe disease, Lumizyme, which significantly slows the progression of the disease.

If my second son hadn't been born with the disease, we never would have known about my oldest as **Pompe disease** was NOT included in **Newborn Screening** in January of 2015. Many individuals spend over 10 years attempting diagnoses of this terrible disease. Because of NBS, we can get both of my children treatment immediately, before deterioration of muscles begin. Many have not been as fortunate.

Thank you – Jenna Yarborough (mother of Wesley and Evander)

Aurora, IL