Paloma Juarez and Brian Way Testimony Infantile Onset Pompe's Disease

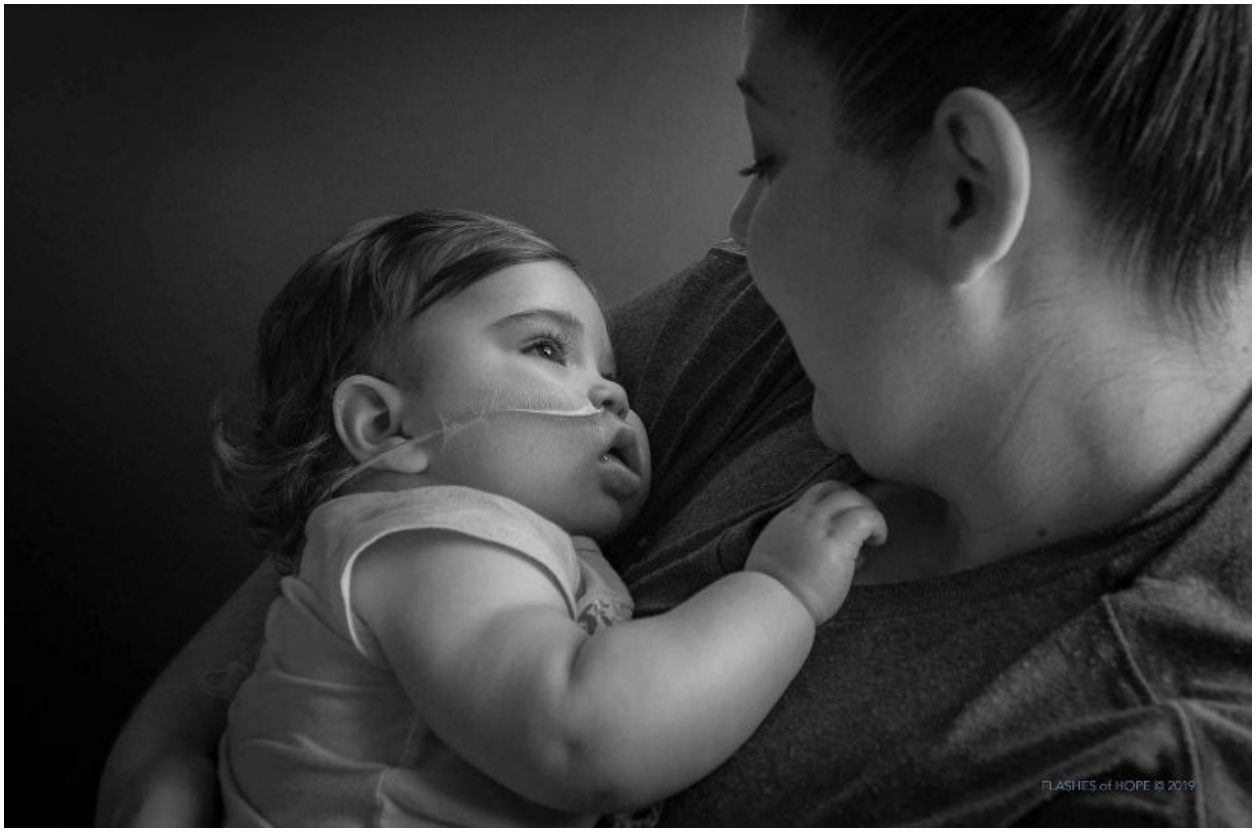
If you Google Pompe's disease you'll find this: "Pompe disease is a genetic disorder in which complex sugar called glycogen builds up in the body's cells. The disease results from the deficiency of an enzyme called acid alfa glucosidase (GAA), which breaks downs complex sugars in the body. This buildup occurs in organs and tissues, especially in muscles, causing them to break down." Next you'll learn there are two types- infantile onset and late onset.

If you decide to read the next line or paragraph, you learn some devastating information. "Without treatment, infants with Pompe disease will die. Many of the people with Pompe disease have respiratory (breathing) problems, heart problems, and almost all are plagued with muscle weakness. Most people will have to use oxygen and wheelchairs at some point."

These were the words Brian and I were met with when researching Pompe's disease just seconds after a nurse called about the results of our son, Vaun's, Missouri newborn screening. Almost 5 years later we have learned so much about IOPD. Many call us and parents like us "experts" in the disease. The thing that we have learned is MOST important and crucial in these children and families lives will be influenced by the decision you in this room make today.

Early diagnosis is imperative. It will allow these children the opportunity to LIVE. It will also (and more importantly) determine the quality of life that they will have.

Meet Analeigh. Born in Alabama, she had a low O2 level at 5 months old. The ER doctor assumed she had a virus. In only 48 hours, the severity of her symptoms became known, and she was put on life support. After a month, Analeigh was diagnosed with IOPD. She had severe hypertrophic cardiomyopathy. With enzyme replacement therapy, currently the only treatment for Pompe's disease, baby Salmeron is now 2.5 years old. She doesn't walk and can only sit for 20 minutes unassisted. With severely weakened oral muscles, she does not talk and is fed from her gtube. Unable to potty train, her parents current challenge is what preschool opportunities will be available for their silly, animal loving daughter.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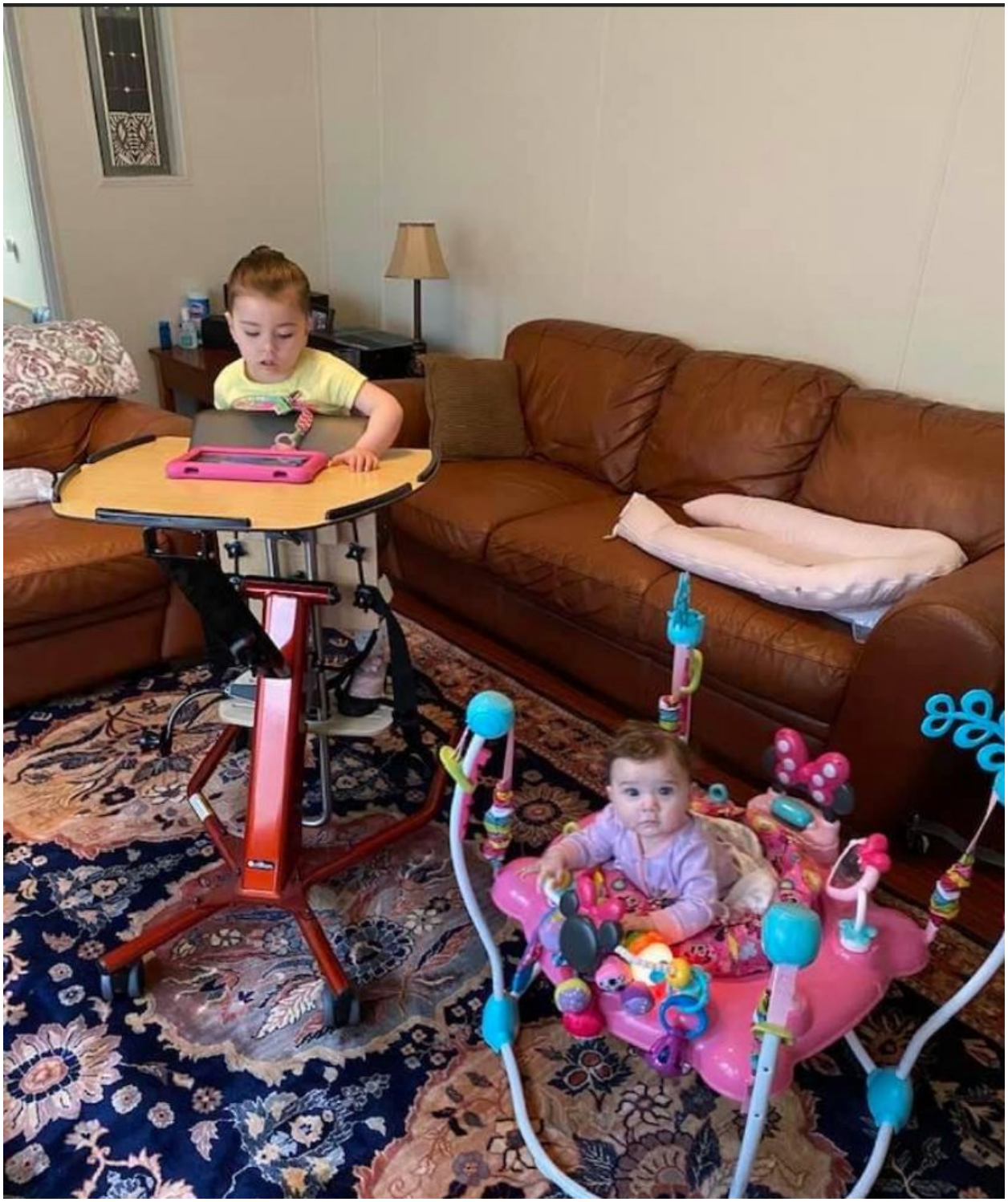
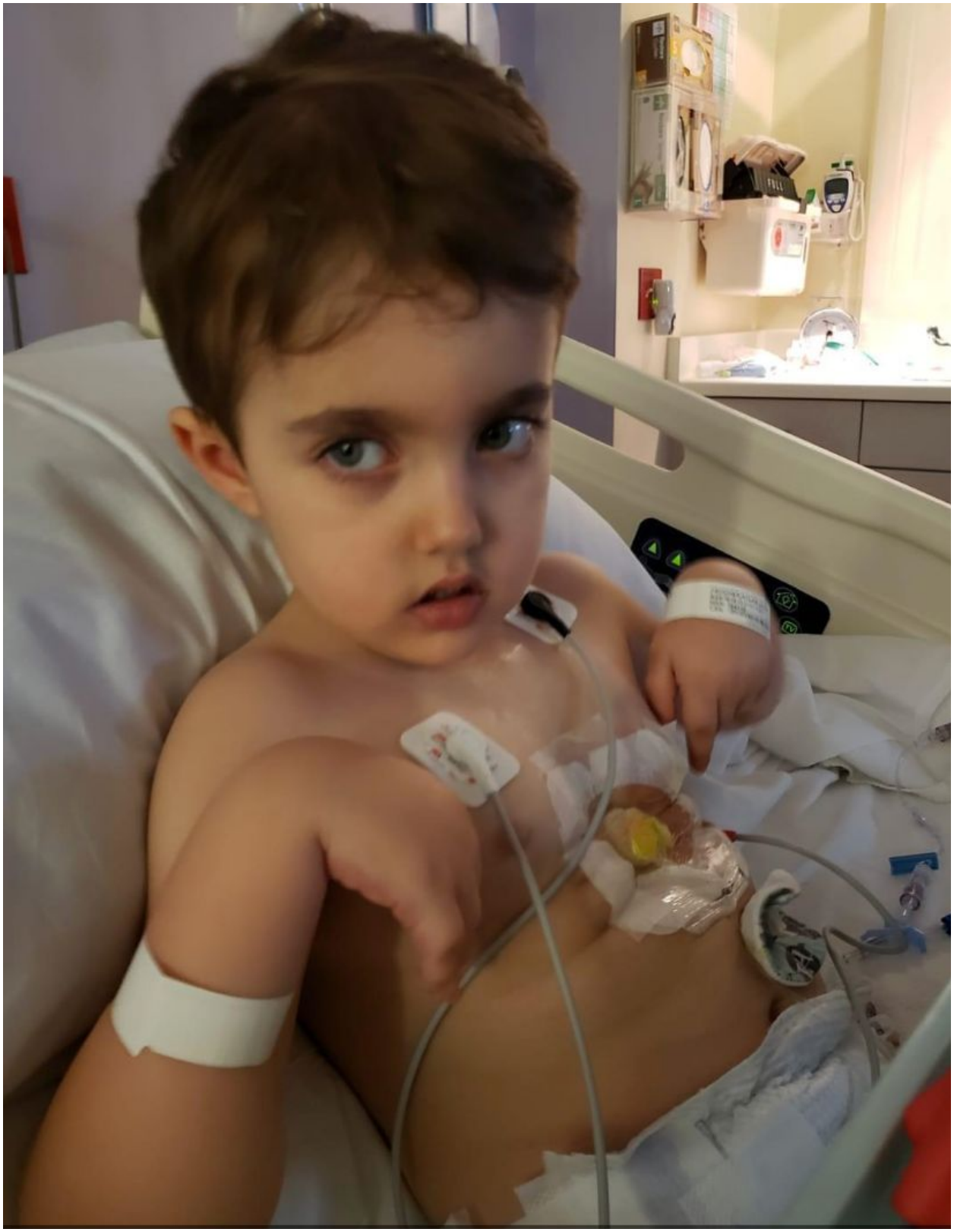
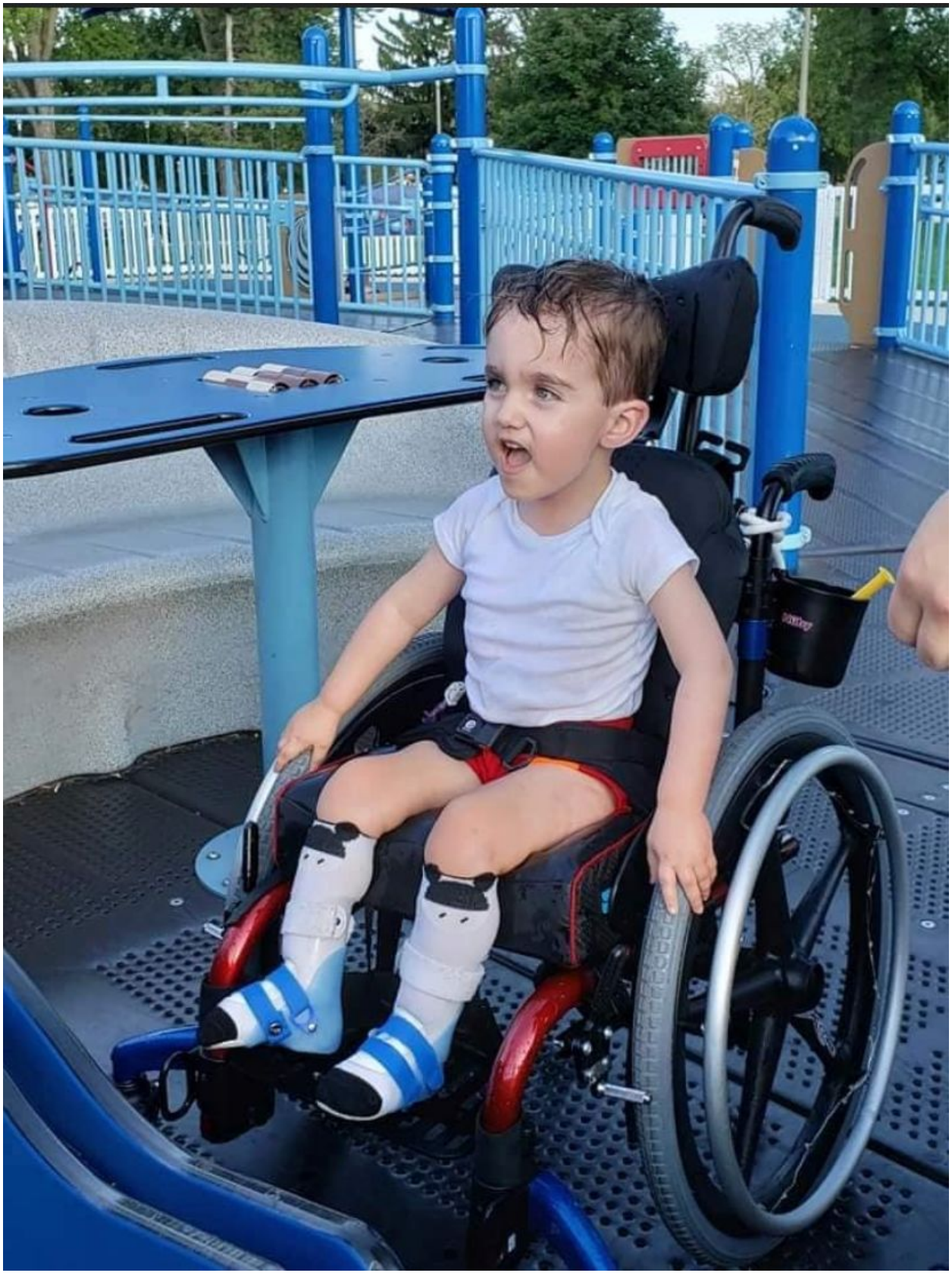
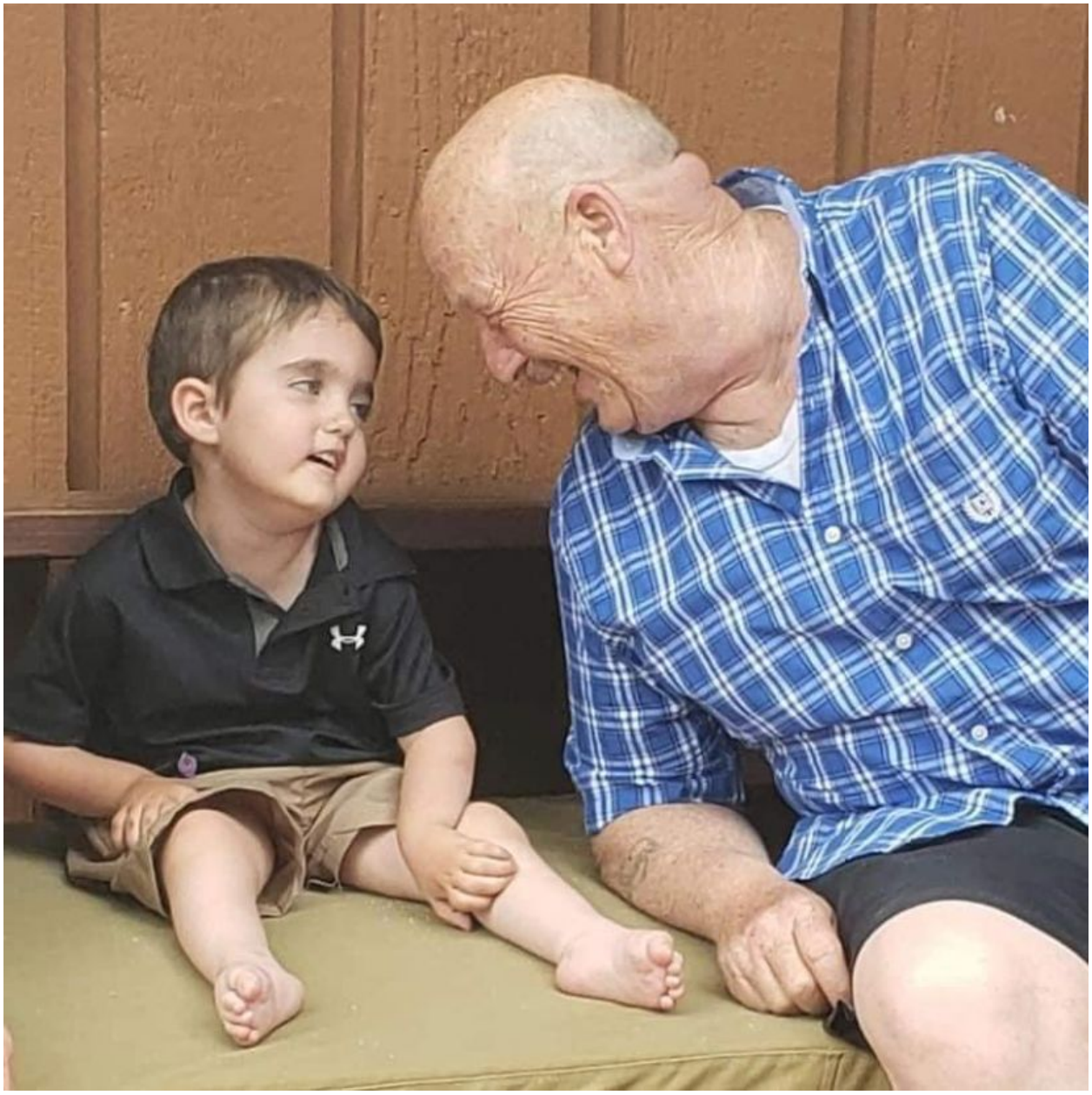


We met Atlas and his parents at Duke's Pediatric Pompe's conference. Atlas and Vaun are just one month apart in age. At 4 months old Atlas went in to test for pneumonia. The xray came back and found out that his heart was enlarged. After more tests, they found he had cardiomyopathy heart failure. Five days later Atlas was diagnosed with

IOPD. Due to complications from IOPD, Atlas began a more aggressive treatment plan in his first year of life. Today, he receives ERT twice a week. Now 4 and a half, Atlas continues to make progress against the many challenges presented from his diagnosis. What is considered a common cold means a 5 day stay in the hospital for him and his parents. He uses the assistance of a wheeler chair and recently met the milestone of pushing up on his arms from laying on his belly. Hecontinues to practice eating and swallowing while still using the assistance of his Gtube. Atlas' parents are very active in the Pompe's community and used their family's story to help get Pompe's added to Wisconsin's newborn screening.



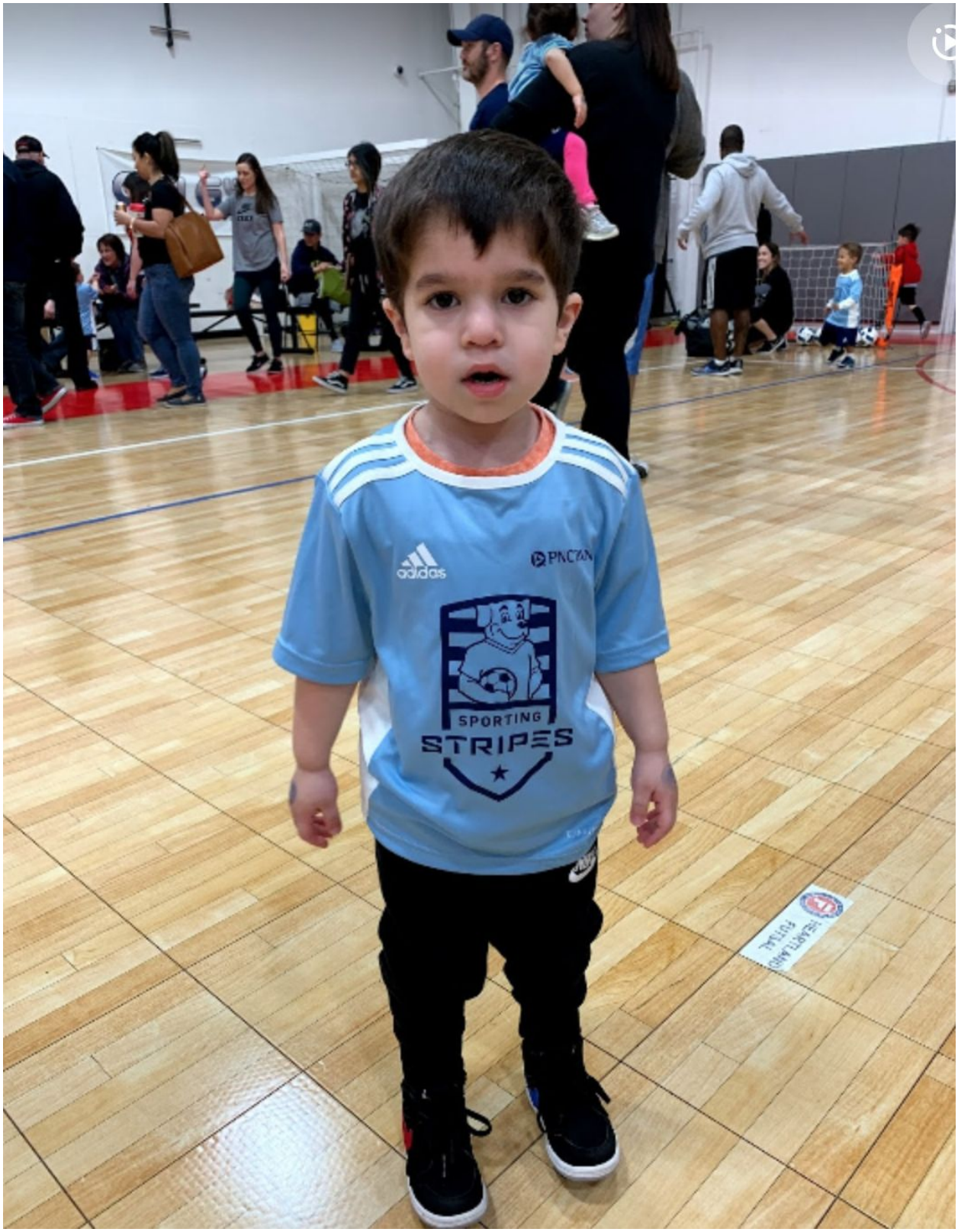




These stories are unfortunately not uncommon. ERT is most successful for IOPD kiddos when begun in under 30 days of life. Even a clinical diagnosis at 5 weeks old leads to hearing loss and developmental delays in speech. Clinical diagnosis at 3 months old has lead to life threatening complications from cardiomyopathy, wheelchair assistance, inability to feed independently, loss of speech. (And these are just the people we have met in our journey)

Newborn screening not only saved Vaun's life, but has given him the opportunity to live that life. We welcomed our second IOPD child, Koen, this past August. Due to early diagnosis, Koen is developing and enjoying life the way his twin brother, Zavier does.

Vaun walks independently and eats unassisted. He loves to SING LOUD and soccer is his favorite activity. Early diagnosis will continue to change what we know about this disease and the lives that children who were once just fortunate enough to be alive can now lead. Thank you.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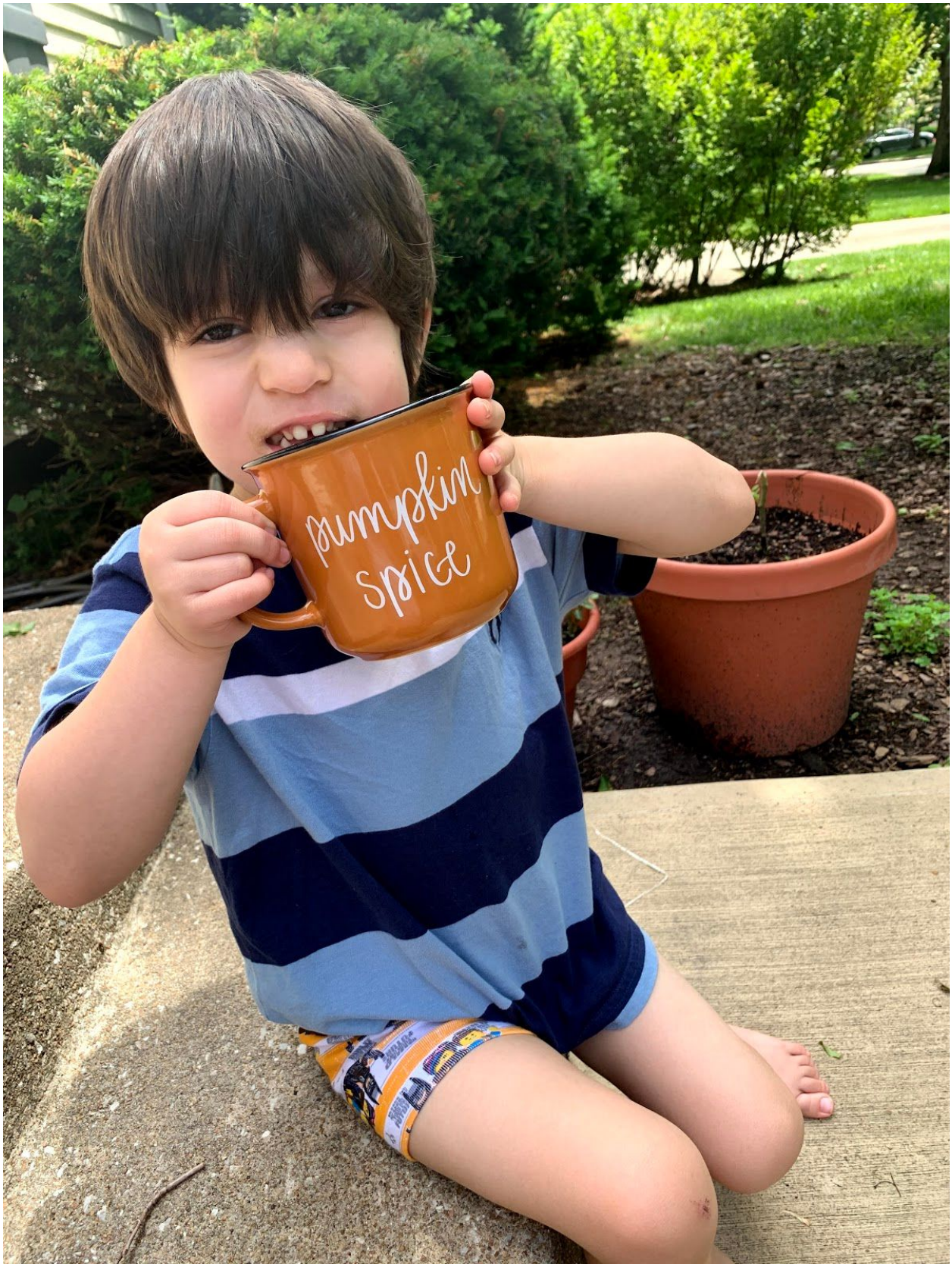
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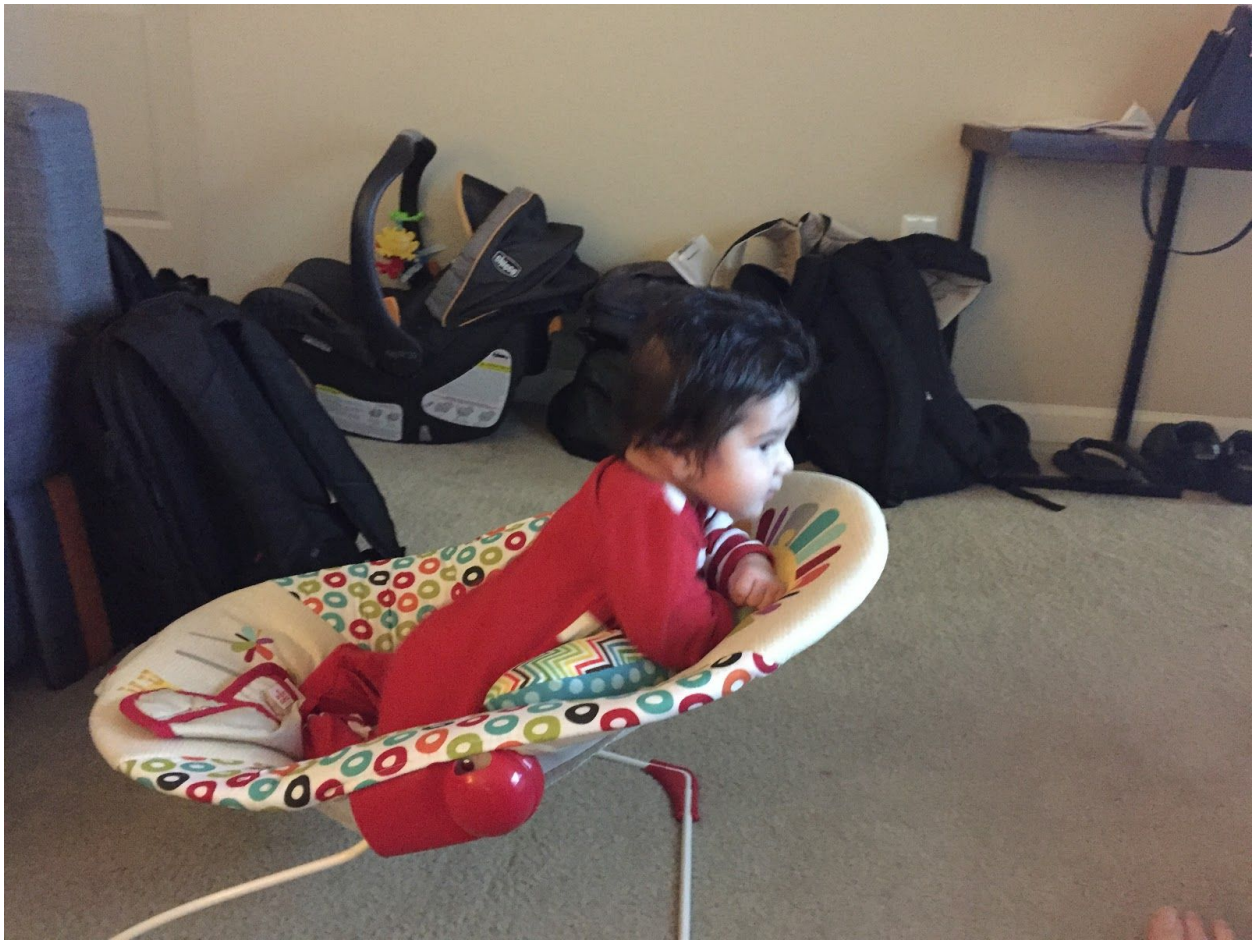

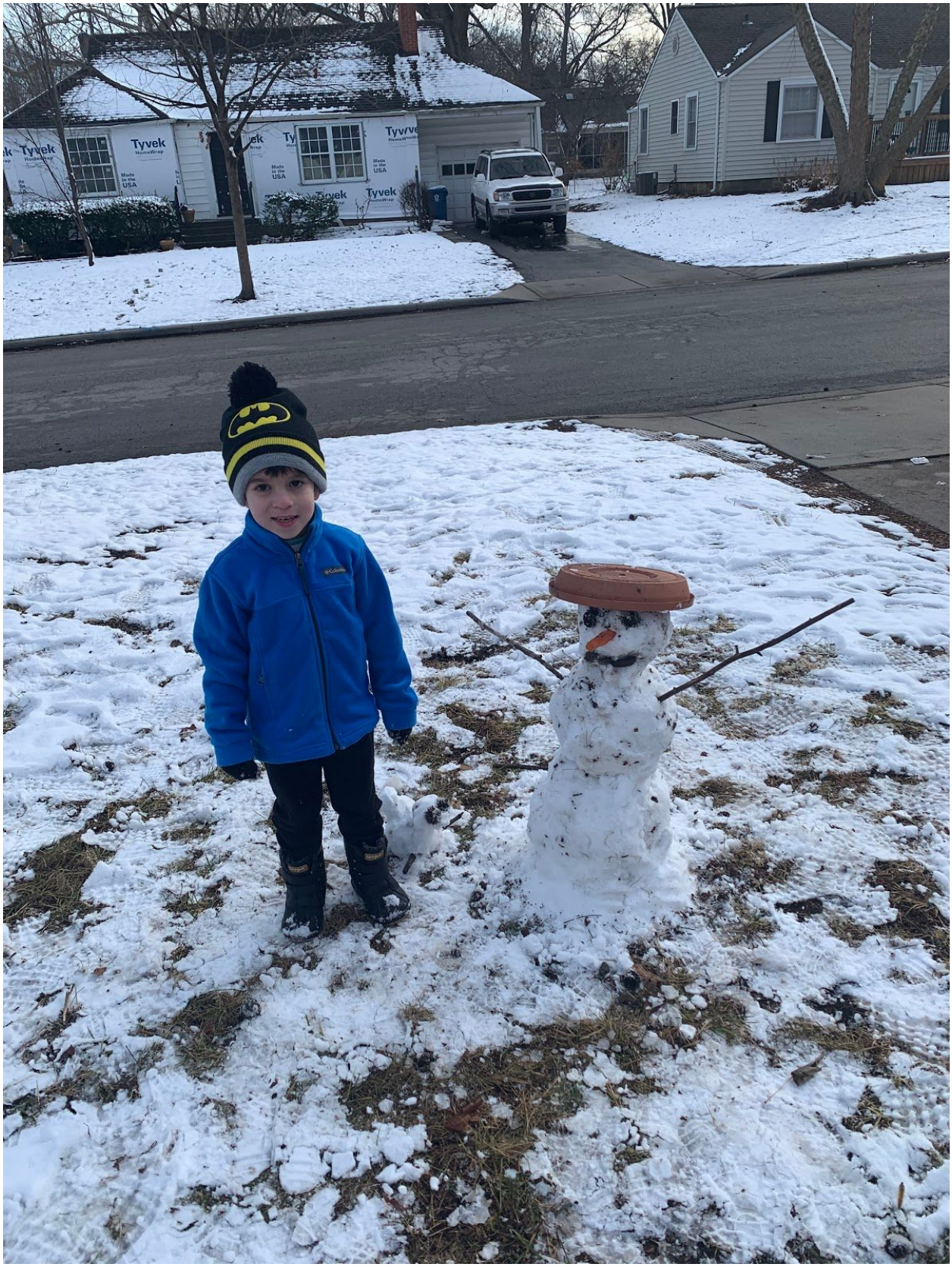
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