# MAPLE FAMILY

## TENNESSEE, USA

## GFPD WARRIOR: ARCHER

- Archer is currently 13 years old.
- He developed nystagmus at 6 months, which led to testing and a diagnosis of Leber's Congenital Amaurosis.
- After giving the geneticist information that LCA patients should be checked for PEX 1 mutations, he requested testing for Archer.
- He was diagnosed at 18 months with a peroxisomal biogenesis disorder in the Zellweger spectrum (PBD-ZSD).
- PEX 1 variants c.1777G>A (p.Gly593Arg), c.2916delA

## OUR FAMILY INCLUDES

- Parents
  - Ashley
  - Ryan
- Siblings
  - Sophie age 20
  - Lydia age 18
  - Madden age 16

### FAMILY VALUES

Determination, Hard Work, Service to Others.

### FAVORITE ACTIVITIES

Outdoor activities

## SYMPTOMS EXPERIENCED

- Balance Issues
- Feeding Difficulties
- Abnormal Liver Function
- Low Muscle Tone
- Bone Loss
- Dental Symptoms
- Adrenal Insufficiency
- Hearing Loss
- Vision Loss

## SUPPORT SYSTEM INCLUDES

- School
- Summer Camp
- State Program for Disabled Children
- The GFPD
- Extended Family Members

## ACCOMPLISHMENTS

- Archer began walking independently at 3.5 years old.
- Partially potty trained during the COVID quarantine.
- He is able to tell us when he needs to use the bathroom, too!

## ALL ABOUT ARCHER

Archer radiates joy every single day. He smiles from when he wakes up until he goes to sleep. His persevering spirit inspires others to keep on going through anything life gives you.

**The GFPD has helped our family by giving us a community of support through medical professionals and other families. The feeling of not being alone gives us such hope. We have a resource to always turn to for advice, which we've done countless times. The GFPD conference is the "vacation" we look forward to because we are surrounded by families that have challenges like ours, and they get it! We get to see that there are others out there fighting for a better future for GFPD warriors.**

GLOBAL FOUNDATION for PEROXISOMAL DISORDERS
GFPD
FAMILY • RESEARCH • HOPE

## PRIMARY HEALTHCARE SYSTEM

East Tennessee Children's Hospital
Vanderbilt Medical Center

## POSITIVE IMPACTS

- School/Summer Camps
- Archer LOVES being around others!

## ADVICE TO PROFESSIONALS

Go to the GFPD website and read the article, "Peroxisome biogenesis disorders in the Zellweger spectrum: An overview of current diagnosis, clinical manifestations, and treatment guidelines."

Advice to a New Families: Try to take it one day at a time, and not think too far into the future. I try to stay present to be the best mom I can be for Archer today. Reach out to other GFPD families, you are not alone!