

# SACRA FAMILY

## MYRTLE BEACH, SC, USA



### GFPD WARRIOR: TJ

- TJ is currently 24 years old.
- He has a peroxisomal biogenesis disorder in the Zellweger spectrum (PBD-ZSD).
- Skeletal/Femur length abnormalities diagnosed in utero. Jaundice, feeding issues and orthopedic issues present at birth.
- First eye surgery at 6 months and first seizure at 8 months.
- Failure to thrive, pneumonia, and liver disease led to testing and elevated VLCFA's led to diagnosis of peroxisomal disorder.
- Diagnosed unknown type of PD at 18 months and confirmed ZSD at age 5 by Dr. Hugo Moser and Steven Steinberg at age 5 years.
- PEX 1 Variants Nt2098insT and c.2528G>A



### OUR FAMILY INCLUDES

- Katie, Mom
- Ted, Dad
- TJ, GFPD Warrior
- Donna, TJ's Grandma
- Many "Chosen Family" too!

Originally from Maryland, we are a family dedicated to serving others through our careers and volunteer work. We "work hard, play hard!" and have a passion for all things tropical! We love time by the pool or at the beach.

### FAMILY VALUES

Honesty, Hard Work, Empathy and Open-Mindedness



### ALL ABOUT TJ

- Determined!
- Has the best laugh!
- Loves brushes, spatulas, mama snuggles, playing ball, car rides, wheels on the bus, baths, and communication.
- He is a mama's boy!

Most positive impacts in TJ's life have been bone surgeries, cochlear implants, total communication approach, and deafblind intervention.

### CARE TEAM HAS INCLUDED

- |                              |  |
|------------------------------|--|
| • Audiologist                | • Neurologist  |
| • Dentist/Oral Surgeon       | • Ophthalmologist  |
| • Pediatrician               | • Orthopedic Surgeon                                       |
| • Developmental Pediatrician | • Otolaryngologist   |
| • Dietician                  | • Pulmonologist  |
| • Endocrinologist            | • Urologist  |
| • Gastroenterologist         | • Physical Therapist                                       |
| • Hepatologist               | • PT, OT, SLP, O&M, TVI, TOD and Special Education Teacher |
| • Geneticist                 | • DeafBlind Intervener                                     |
| • Hematologist               |  |

The GFPD has helped our family by providing a foundation to connect families and professionals.



### SYMPTOMS EXPERIENCED

- |                           |                            |
|---------------------------|----------------------------|
| • Seizures                | • Dental symptoms          |
| • Balance problems        | • Adrenal insufficiency    |
| • Feeding difficulties    | • Hearing and vision loss  |
| • Reflux                  | • Cardiac symptoms         |
| • Abnormal liver function | • Kidney symptoms          |
| • Low muscle tone         | • Respiratory difficulties |
| • Bone loss               | • Neurogenic bladder       |
| • Bone abnormalities      | • Vasovagal syncope        |

Advice to New Families: Feeling a full range of emotions, including anger, is okay. There's no need to maintain a constant smile. This is hard. Remember, you're not in this alone.