



IOPD... THIS PARENT'S VIEW

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*You don't have to be a nurse to be
a caregiver! The journey is YOURS!



Two of a kind...
literally!
Bruce, 10
Myra, almost 3!

If our genetics are a
cookbook,

Mom has a typo (mutation) in
Chapter (gene) 5,

Dad is missing (deletion) in
Chapter 18...

This combination creates the
unique recipe for low GAA
activity, the natural occurring
enzyme that breaks down
glycogen



Our first diagnostic story -

- Bruce failed kick counts and stress tests; born by c-section and *"looked normal"*!
- Leg swelling began at two days old...transferred to a NICU in Kansas City at 3 days and diagnosed with a thickened heart (HCM) and electrical cardiac issue (WPW)
- First SVT episode (heart rate EXTREMELY fast) at day 3 and **that** got docs' attention
- He was diagnosed clinically (by visualization and diagnostic tests) in 2011
 - NBS started in Missouri in 2013 after using his blood for testing calibration

So, you have a diagnosis too -

- Enzyme Replacement Therapy (ERT)
 - Treatment vs. Cure
- Early and consistent ERT is critical and individual!
 - Missing one dose here and there is going to happen and that's 'ok' in relative terms

For reference, Bruce started ERT at 13 days old & Myra at 5 days old

*Mom-RN tip: Never be afraid to watch nurses like a hawk and ask question when accessing, deaccessing, and cleaning the lines used to give medication! If you see something new or different, take a moment to learn or question

And then Little Sister's story..

- Rule #1 of nursing school...don't get pregnant!
- Myra was diagnosed in utero by amniocentesis at 20 weeks old
- Had 2 fetal ultrasounds to detect heart involvement that started later in the pregnancy
- Mom took boards at 36 weeks pregnant...started Master's same week...
- Born by c-section at 38 weeks and started ERT at 5 days old in the NICU

What the first decade brought us -

- Finding home nursing can be hard - advocate for safe delivery of medications and completely INVOLVED nursing...ERT is a long and tricky process, start to finish!
- Two kids with ports is exhausting....what is THIS fever now?!
- Find a medical team you LOVE
- Know your resources - Patient Advocacy and Palliative Care are there for your child and YOU as their caregiver
- Rare is the new normal

That decade brought us two pretty dang, healthy kids!

What we continually learn -

- Nothing is the same
- Nothing stays the same
- Power in advocacy and self-education
- The research and advocacy world is constantly working for Pompe
- Find tools to support home life - pulse oximeters, thermometers, nursing!

Use your resources...

Hospital, State, Insurance, Lawyer

Fellow Pompe Warriors!



BE A VOICE!
(but at your speed)

There is a balance, and
you will find it. Just know
it will evolve!

These kids are warriors!
We have good days,
we have GREAT days, and
we have days we even
forget about Pompe!

Educate yourself!

We have a
Day 1 Guide for that!

PompeInformation@gmail.com



THANK YOU, POMPE ALLIANCE!

John, Sarah, Bruce & Myra