

Amelia, NS-RIT1

with Noonan syndrome - RIT1
21 Months Old
As Reported by Samantha, Mother



Newborn

Recent



Amelia - with Noonan Syndrome - RIT1



HOBBIES:

Loves puzzles and matching games

Determined.
Smart.
Miracle.
Inspiring.
Sassy.

8 weeks

Amelia - with Noonan Syndrome - RIT1



Most challenging cardio issue:
Hypertrophic cardiomyopathy (HCM), mitral valve regurgitation. Diagnosis of pulmonary valve stenosis (PVS) is now cured.

Most unique challenge:
Managing our eczema, a common side effect from the Trametinib. She is at the age to where she will constantly scratch it. We have recently found a new OTC cream that has really helped.

3 months

5 months



Amelia - with Noonan Syndrome - RIT1

Medical success story:

- Anatomy scan at 20 weeks' gestation lymphatic found a small build up on both sides of her neck.
- Led me to 4 different maternal fetal medicine specialists for the rest of my pregnancy.

- Within 1 week of the build-up findings, they resolved!
- I was still followed by the 4 physicians in the clinic for the remainder of my pregnancy, no other concerning factors found.



5 months

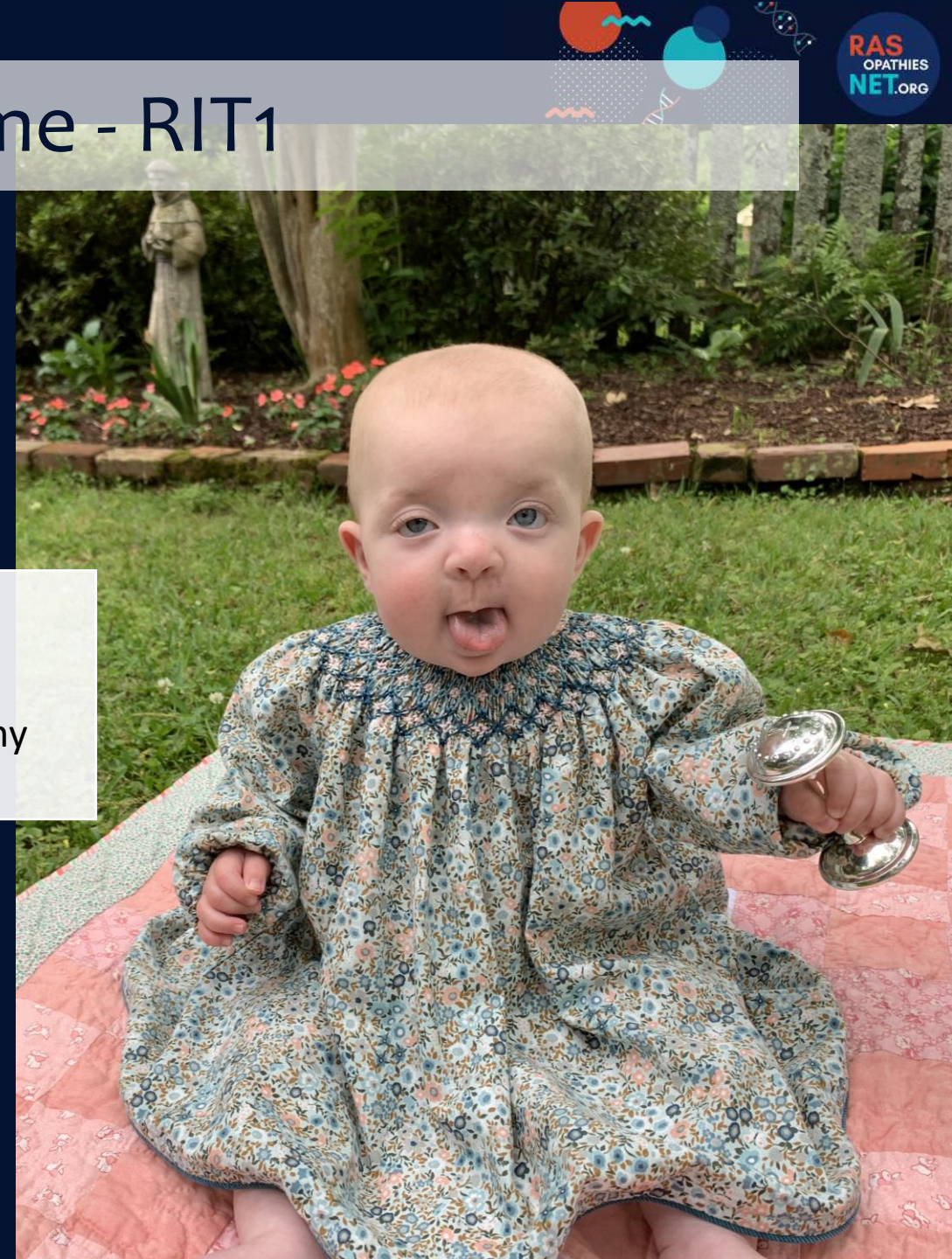


7 months

Amelia - with Noonan Syndrome - RIT1

- At ~30 weeks gestation I was diagnosed with polyhydramnios, thought to be from borderline gestational diabetes.
- Born at 37 weeks, 5 days, normal birth weight 7 lbs 1 oz. healthy except for a slight, innocent murmur, referred to cardiology.

8 months



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- 6 weeks later, official diagnosis: hypertrophic cardiomyopathy (HCM) and pulmonary valve stenosis (PVS).
- **We were immediately sent 5 hours from home for a valvuloplasty, which was unsuccessful.**
- It was after the heart cath/valvuloplasty procedure that the specialists highly suspected Noonan syndrome.

9 months

Amelia - with Noonan Syndrome - RIT1



- Dr. Kleinmahon mentioned a treatable drug he read about in Canada, but with only a clinical diagnosis of Noonan syndrome, we had a 95% likelihood of not having the RIT1 gene that showed benefit from the drug intervention.
- At 7 weeks old, Dr. Jake Kleinmahon thought that **heart transplantation would more than likely be inevitable.**

11 months

Amelia - with Noonan Syndrome - RIT1



- We were kept inpatient for one week for monitoring and to complete the transplantation workup, including labs, echoes, and ultrasounds of other various organs.
- **Amelia was deemed healthy enough and a candidate for transplant until the time was thought to list for transplant.** We were discharged to stay locally on a Friday.
- The following Monday we received our **RIT1 diagnosis.**

13 months

Amelia - with Noonan Syndrome - RIT1

- Dr. Kleinmahon had us readmitted to the hospital 3 days later to start **trametinib**.
- It took *only one week* for the drug to stabilize her HCM.
- We were seen every week then every 2 weeks, then 4 weeks, then 6 weeks, 8 and now we go every 12 weeks. That is the longest we are expected to space our appointments out.

16 months

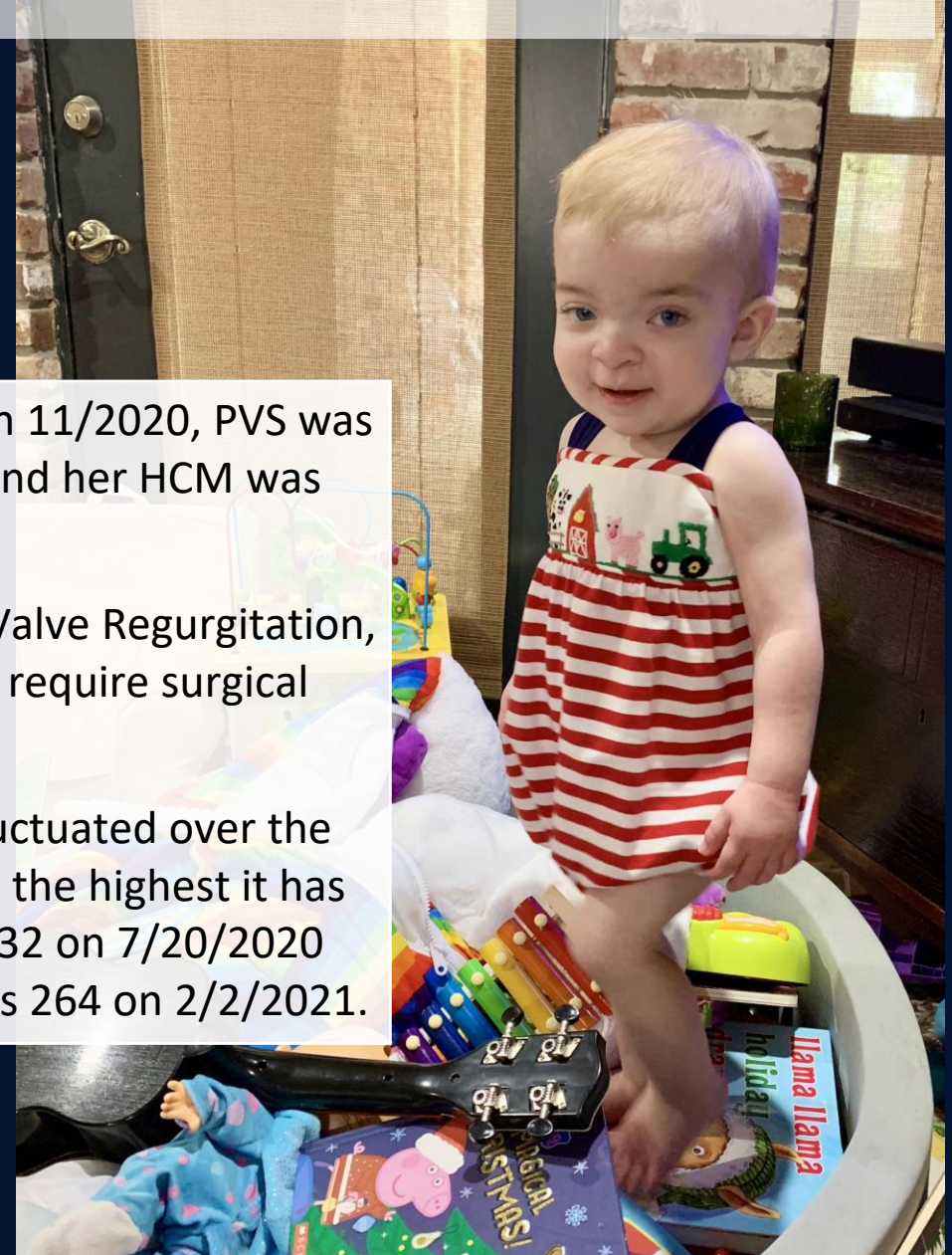


Amelia - with Noonan Syndrome - RIT1



- At latest heart cath 11/2020, PVS was completely gone and her HCM was stable.
- Does have Mitral Valve Regurgitation, but not enough to require surgical intervention.
- NT-Pro BNP has fluctuated over the last two years, but the highest it has ever been was 2,832 on 7/20/2020 and the lowest was 264 on 2/2/2021.

20 months



Amelia - with Noonan Syndrome - RIT1

- **The only side effect from the drug was acute dermatitis (eczema).** In January 2020, it was at its worst, causing impetigo. Dr. Kleinmahon had us go to the hospital 5 hours away and be admitted for monitoring and IV antibiotics.

- Since then, we use OTC hydrocortisone and Aveeno eczema creams that have it under control.

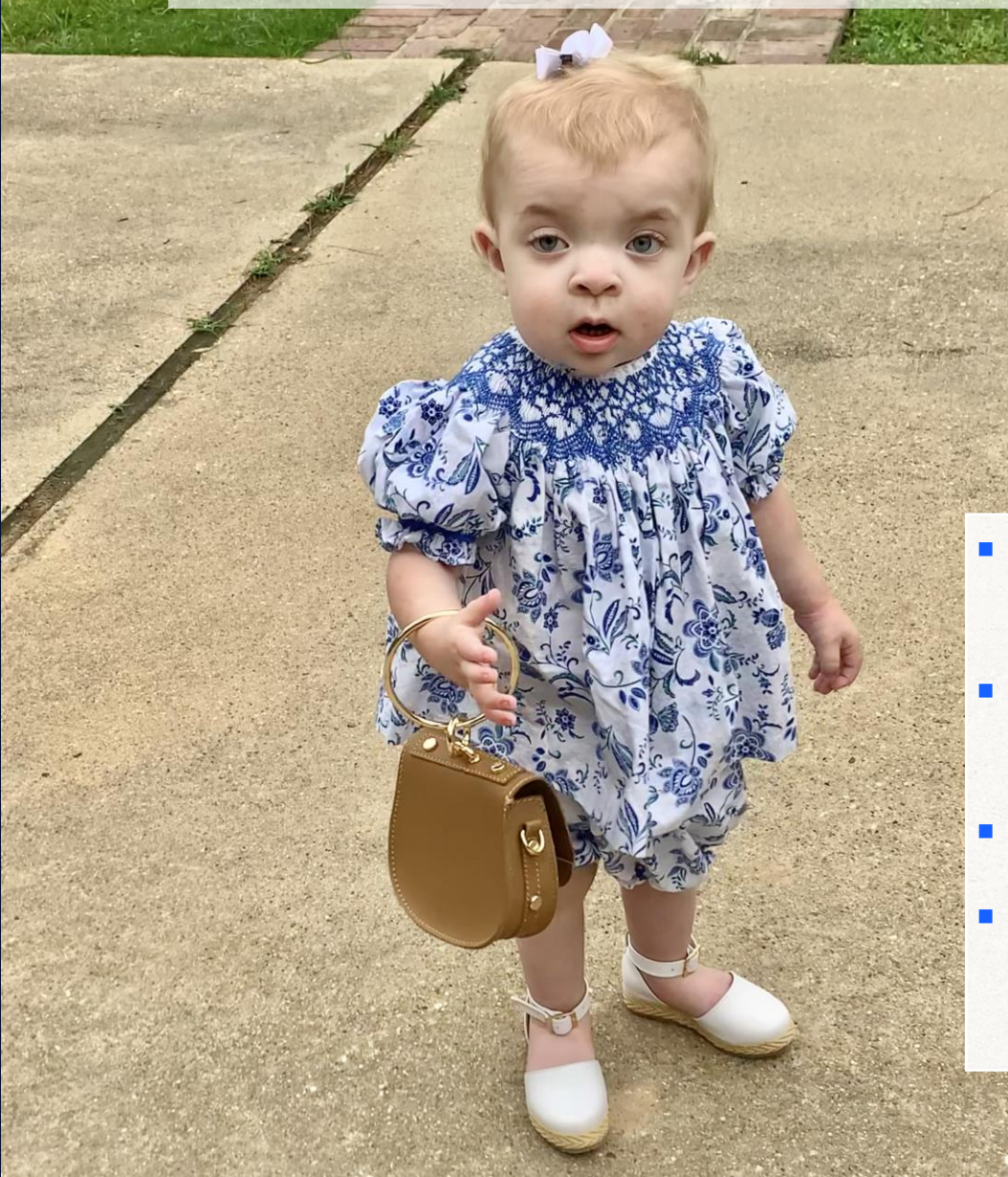
- Amelia is a thriving 21-month-old.

- She has met every one of her developmental milestones.



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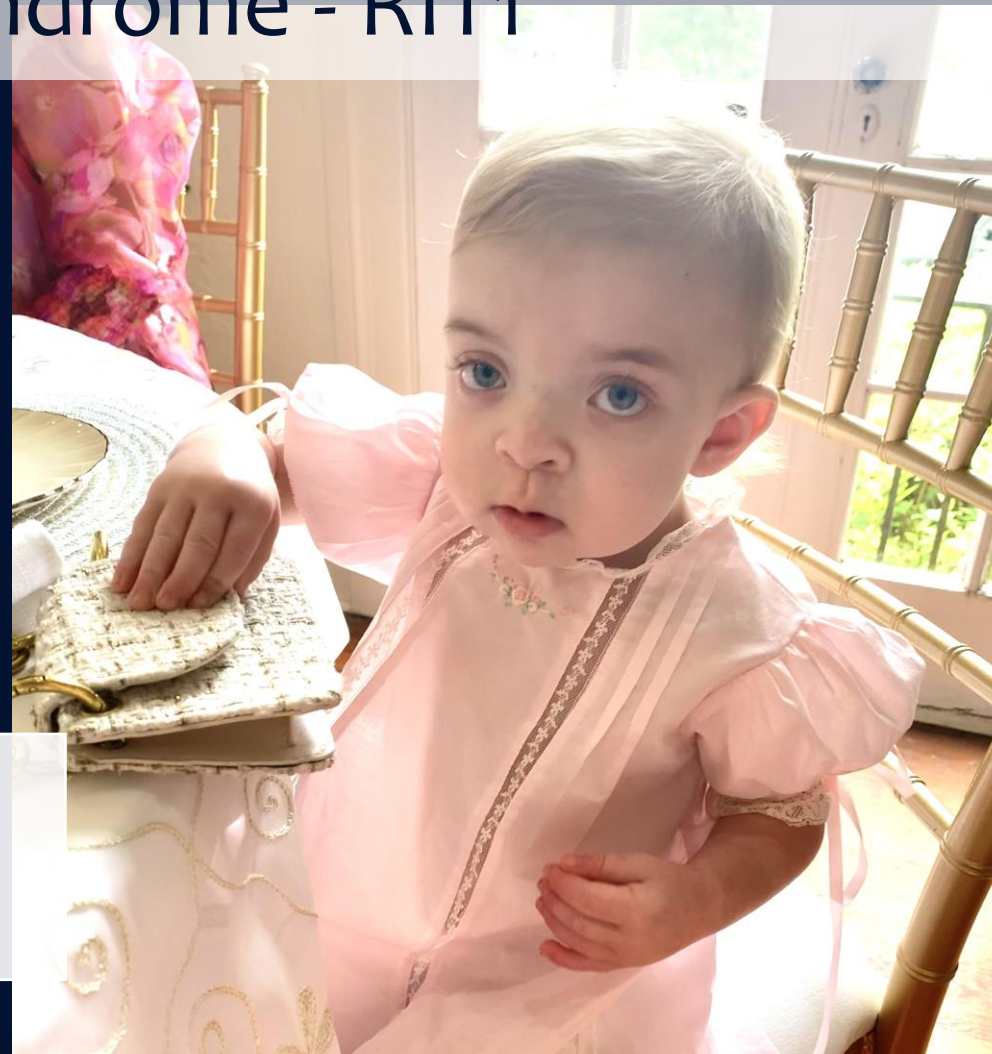
- Never had any signs of cardiac distress, even before starting trametinib.
- She walks, talks, makes her needs and wants known, and is very good at playing puzzles and matching games.
- Has the spunkiest personality that keeps us on our toes.
- We are so fortunate for Dr. Andelfinger's discovery and Dr. Kleinmahon's willingness and eagerness to try something new.

Recent

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Medical success story:

- It seemed like a Hail Mary treatment
- **The greatest decision and treatment I could have ever asked for my daughter**



Recent

I hope and pray that her data from the drug will be a pathway to help so many more children in the future.



