



Aniella was born in 2005 and was prenatally diagnosed with full trisomy 18. She lives in the United States. Participation in the TRIS project began in 2011.

Mother was 36 and father was 32 years old at the time of conception. Child with a rare trisomy condition was fifth of six pregnancies (gravida, 6; para, 6). No siblings have been diagnosed with a genetic, terminal medical or neurological condition. Neither parent was aware of rare trisomy conditions prior to their child's diagnosis.

Birth information

Ultrasound at 19 weeks indicated choroid plexus cysts on fetus's brain, ventricular septal defect (VSD) and clenched hands. An amniocentesis was performed at 19 weeks. At 22 weeks, results confirmed full trisomy 18 with no growth restriction or other anomalies.

Mother had preterm labor at 36 weeks gestation and decided for a spontaneous vaginal birth at that time. Decision was also made to withhold treatment or heroic measures.

Female child was born after 13 hours of labor at 36 weeks gestation weighing 1769 grams and 41 centimeters in length on May 24, 2005. APGAR scores at 1, 5, and 10 minutes were 2, 2, and 5 respectively. Infant presented with the following physical features: low set ears, small jaw, microcephaly, webbed toes, rocker bottom feet, 1st and 4th digits of both hands overlapping 2nd and 3rd digits, and left eye shut by an acrochordon (skin tag). Infant presented with respiratory issues. Infant was released after two days at 1644 grams.

Infant was taken to a different hospital due to dehydration upon physical examination of pediatrician on the day after discharge from birth hospital. As a result, she was admitted to another hospital for five days to stabilize her. Infant was treated for jaundice and dehydration only. Echocardiogram showed atrial septal defect (ASD), patent ductus arteriosus (PDA), ventricular septal defect (VSD) and pulmonary stenosis. Infant was not offered supplemental

oxygen or provided with an apnea monitor. Weight at discharge was 1814 grams. After release, cardiopulmonary resuscitation was required twice during her first month of life due to aspiration pneumonia.

Infant received hospice services until four months of age. At that time, child was taken and admitted to a Children's Hospital in a neighboring state for a full work-up. During that time, a nasal cannula, apnea monitor, continuous positive airway pressure (CPAP), heart monitor, pulse oximeter, and gavage feeding were provided. An audiology exam (bone conduction audiometry), echocardiogram, and ophthalmology exam were also performed. An acrochordon (skin tag) on left eyelid was also removed during her 17-day stay.

The following sections describe medical concerns related to specific organs or body systems.

Cardiac

Child was diagnosed with an ASD, a VSD, and pulmonary stenosis. To date, no surgeries have been performed to correct these conditions. Mother reported recent discussions with a pediatric cardiologist who is willing to consider surgery for child's VSD and pulmonary stenosis. Mother emphasized the doctor's agreement to a preventative approach to address these issues prior to any future complications.

From the age of four to 36 months, child was prescribed Lasix for management of VSD. From four months to time of survey completion in late 2011, Digoxin was also prescribed.

At current time, child has a cardiologist visit every two years. Child's last appointment was in January of 2018. At present, child does not take any cardiac medications. Child's VSD and pulmonary stenosis are still unrepaired, however, and both are in stable condition.

Respiratory

Child's mother listed her daughter's most critical need as resolving airway difficulties. Child was diagnosed soon after birth with obstructive apnea and had an adenoidectomy at 23 months and tonsillectomy at 48 months. An emergency tracheostomy was placed at 48 months (May 2008). Child has needed supplemental oxygen in September 2013 due to pneumonia. Child also needed supplemental oxygen in 2014 when her lingual tonsils and adenoids were removed.

Successful decannulation occurred on November 17, 2018 at the Children's Hospital of Philadelphia. Child was in the hospital for one day post-procedure.

Renal

Child was diagnosed with crossed fused renal ectopia (kidneys are fused and located on the same side of midline) in the post birth care unit. Both kidneys are on the left side. Child's left kidney is anatomically correct, while the right kidney is placed in the left pelvic region. The condition is monitored on an annual basis with ultrasounds.

Gastrointestinal

From the age of 10 to 13 months, child was reported to experience long periods of crying. It was discovered this was due to a malrotation of her lower intestine. At 13 months, the malrotation was corrected (lower bowel was untwisted and most moved to left side to address above renal issue). No further problems were noted post-repair. An appendectomy was also performed during this surgery for preventative purposes.

Nutrition

Child received gavage feeds until 18 months of age. At the age of 21 months, a gastrostomy tube was placed. Starting at the age of 12 months, child was prescribed Prevacid for reflux. At 14 months, a gastrostomy tube was placed. Since the age of 12 months, Lactulose has been prescribed for constipation relief. At the age of 48 months, Miralax was added to child's medication regimen.

Since February of 2018, child receives 30mg of Nexium daily for her reflux. Child also receives 4 teaspoons Miralax and 5ml Senna twice daily.

Child is currently primarily fed by gastrostomy tube and supplemented with table food as tolerated. Currently, the only tolerated table food is yogurt. Child also is cup fed Boost Breeze® (Honey Thick consistency).

Orthopedic

At the time of baseline survey completion, child used a manual wheelchair, a walker, and leg braces. Currently, she is able to walk independently for short distances (≤ 15 feet). She is also able to transfer from couch to the floor, crawl to a stable surface and pull to stand in preparation for walking.

At current time, child is able to climb on and off of the couch. She can stand up by using the couch to gain balance and walk short distance. However, she is still unsteady and at risk for falling. Child uses a walker or pushed in a wheelchair or adapted stroller for longer distances.

In June 2018, child had a hamstring and tendon release surgery on both legs due to contractures. The surgery was successful. The recovery process is ongoing including physical therapy five times a week with increased mobility in lower extremities. However, although child stands straighter, assistance is necessary when walking.

Oral health

Child is seen by a pediatric dentist bi-annually and is given antibiotics before and after each routine dental procedure. At the time of survey, child had all but two deciduous teeth.

Currently, child has had no oral procedures. Child brushes her teeth four times daily.

Vision

Child was diagnosed with myopia and strabismus at the age of four months and was given prescription eyeglasses at that time. All vision needs are met with prescription eyeglasses that are updated annually by an ophthalmologist to check for vision abnormalities.

Auditory

Tubes were placed at age three. One tube fell out and the other remains intact. Mother reported excessive earwax that blocks the ear canal and diminishes hearing capabilities in both ears. Child's family doctor monitors this condition to prevent hearing loss.

Child still has excessive ear wax. She had an auditory brainstem response (ABR) hearing test in November 2018. However, no hearing loss was reported.

Immunizations

Currently, all immunizations are up to date but were not started at the recommended ages. Initial doses were given at four months while admitted to the children's hospital. Child has received 15 doses of Respiratory syncytial virus (RSV), six doses of influenza, four doses of Diphtheria, Tetanus, and Pertussis (DPT), Haemophilus influenza type B (HIB), and Pneumococcal pneumonia. She also had three doses of Inactive poliovirus. She received two doses of Measles, Mumps, and Rubella (MMR). She received one dose of Hepatitis A (HepA), Hepatitis B series, and Varicella. No adverse reactions were reported after any immunization.

In October of 2018, child received an HPV vaccination. Child also annually receives a preventative flu shot. No adverse reactions noted.

Current education and therapy services

Child attends her neighborhood school. She is placed in a general education classroom for homeroom and "specials" (Physical education, music etc.). She also receives special

education services focusing on functional life skills including activities of daily living. At present, child is learning to sort by color and uses a Big Mac switch to activate application on an iPad. Mother reports a large receptive language repertoire. Mother has also purchased a Go Talk 4 system for child's use. An augmentative communication evaluation is scheduled.

Child received physical therapy two hours a week at home and two hours a week at school. She received one hour of speech therapy at school and two hours a week at home. She also received 30 minutes of feeding therapy per week at a specialized clinic.

At current time, child receives educational services at a special school for children with disabilities. Child's favorite part of the school day is recess.

Parent disagrees with placement recommendation on child's Individualized Education Plan. Parent is requesting for child to be placed into an inclusive high school, where she can be integrated with peers her age. Parent is currently working on this placement for June 2019, since child graduated from middle school in June 2019 and will begin attending high school in August 2019.

As requested by parent, child receives one-on-one therapies. Child receives physical therapy three times a week at school, each a 30 minute session. Child receives occupational therapy twice a week at school, each a 30 minute session. Also, child receives speech therapy five times a week at school, each a 30 minute session. The only therapy provided in the home is additional physical therapy twice a week, each a 45 minute session.

Developmental milestones

Child has attained most developmental milestones to the age of 12 months with some skills extending to the 18-24 month range. Child communicates needs to caregivers and other adults such as demonstrating preferences for favored objects. Child also participates in social play and explores objects with hands. Child also demonstrates affection to preferred adults and siblings. She engages with eye contact, smiling and laughing.

Child also attempts to assist with dressing activities. In addition, mother reports child uses the toilet daily for both voiding and bowel movements. She is able to remain dry during the day and, sometimes, overnight. Notably, child is now able to independently ambulate for short distances (≤ 15 feet).

At current time, child has reached new developmental milestones. The orthopedic section outlines the child's current gross motor skills. For fine motor milestones, child is able to pick up

objects in her reach. Child will either throw or kick the object. Child uses problem solving skills, such as when a desired object is not in arms reach, she will move closer to it.

Child recognizes her name, colors, and desired TV shows. Child is fully dependent with her daily living skills. However, when dressing she will help by taking her arms in or out of shirts.

Child is non-verbal, and produces a few American Sign Language signs such as, *yes*, *thank you*, and *book*. Child demonstrates receptive language through responses to one step instructions such as, *'take the book out of your mouth'*, *'go get it yourself'*, *'stand up'*, *'sit down'*, *'do you want juice'*, *'do you want yogurt'*, *'do you want to watch sports'*, and *'do you want to watch a game show'*. She also responds when her name is spoken. She will nod her head to indicate "Yes" or sign "*Thank you*".

For social and emotional milestones, child enjoys going to school and being in public. In June 2019, child walked down the aisle at her 8th grade graduation and will attend the community high school where she will have inclusion with her peers. Her new program in high school will continue to include a focus on life skills.

Child participates in a variety of community activities with her family including going to the beach, and apple picking in the fall.