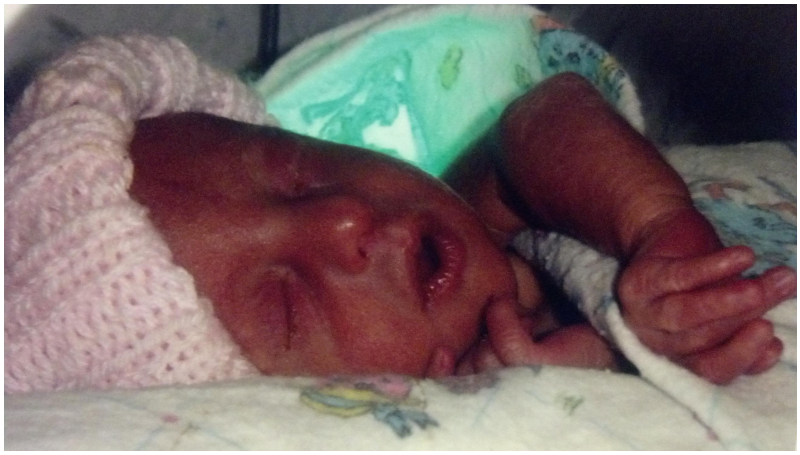

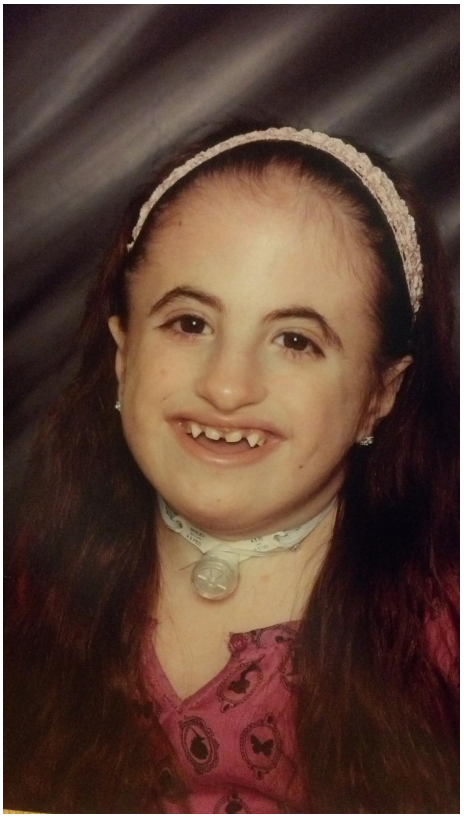

Aniella was born in 2005 and was prenatally diagnosed with full trisomy 18. She lives in the United States. Participation in the TRIS project began in 2011.

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 41centimeters 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 her 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. Acrochordon 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 wiling 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 the current time, child does not receive any cardiac medications and has been stable.

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) and is still necessary for optimal respiration. No plans are in place for decannulation. Child has only needed supplemental oxygen in September 2013 due to pneumonia.

Renal

Child was diagnosed with crossed fused renal ectopia (kidneys are fused and located on the same side of the midline) in the post birth care unit. The condition is monitored on an annual basis with ultrasounds. Both kidneys are on the left side.

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 gastronomy tube was placed. Child is currently primarily fed by gastronomy tube and supplemented with table food. Since the age of 12 months, Lactulose has been prescribed for constipation relief. At the age of 48 months, Miralax was added to the regimen.

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.

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.

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.

Auditory

Tubes were placed at age three. One tube fell out and the other remains intact. At the time of survey completion, 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.

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.

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.

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 does not produce verbal or gestural communication skills beyond the three-month level (coos or social smiles) but is able to

communicate needs to caregivers and other adults such as demonstrating preferences for favored toys. 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. Notably, child is now able to independently ambulate for short distances (≤15 feet).

Child also attempts to assist with dressing activities. In addition, mother reports child is 90% potty trained. She uses the toilet daily for both voiding and bowel movements. She is able to remain dry during the day and, sometimes, overnight.

For more information on the Tracking Rare Incidence Syndromes (TRIS) project:

Homepage: http://web.coehs.siu.edu/grants/tris/

Case studies page: http://web.coehs.siu.edu/Grants/TRIS/casestudies.html

Facebook page: https://www.facebook.com/TRIS.Trisomy.project