Natalia was born in 2000 and was not prenatally diagnosed (trisomy 13). She lives in the United States. Participation in the TRIS project began in 2007.

Mother was 39 and father was 49 years old at the time of conception. Mother declined AFP test at 20 weeks due to a previous false positive. Child with rare trisomy was seventh of seven pregnancies (gravida, 7; para, 4). No siblings have been diagnosed with a genetic, terminal, medical, or neurological condition.

Birth Information

Female infant was delivered via planned caesarian section at 38 weeks gestation**.** Trisomy 13 was suspected after birth due to congenital malformations and confirmed with genetic testing a few weeks later. Child was also retested at the age of 18 months when family changed pediatricians.

Infant was born weighing 3401 grams and 52 centimeters on August 25, 2000. Records show APGAR score was 7 and 9 at one and five minutes, respectively. At birth, the infant presented with ocular hypotelorism noted, a two-vessel umbilical cord, low-set ears, left cleft lip and bilateral cleft palate, postaxial polydactyly of both hands, rocker bottom feet, and aplasia cutis congenita (pea sized scalp lesion) on the head. Infant also presented with the following medical issues: hydronephrosis, ventricular septal defect (VSD), atrial septal defect (ASD), feeding difficulties, and jaundice.

Infant was transferred to a neonatal intensive care unit (NICU.) She remained on unit for 11 days. A nasal cannula provided oxygen for two days and a pulse oximeter was used prior to

discharge. Infant was fed via multiple methods prior to discharge including a Haberman feeder due to clefts, oral gavage/mouth tube feeding, and nasogastric tube (n-g tube). An audiology exam, echocardiogram, ophthalmology exam, ultrasound, and x-ray series were performed in the post-birth care unit. Infant also received occupational therapy, physical therapy, developmental therapy and feeding therapy consultations. At time of discharge, infant's weight was 3380 grams.

Surgeries

At six weeks, infant had umbilical hernia repaired, an appendectomy, and Ladd's procedure for complete bowel malrotation. At nine weeks, cataract removal surgery on left eye was completed. At 11 weeks, cataract was removed from right eye and ankyloglossia ("tongue tie") was clipped. At seven months, cleft lip was repaired, duplicate postaxial digits on both hands were removed and tympanostomy tubes were inserted. At 18 months, cleft palate was repaired and separating of cleft lip suture scar was corrected.

Cardiac

At birth, infant was diagnosed with an ASD and VSD. Both resolved without medical intervention before 30 months of age. Mother notes a doctor agreed to repair both by the time child reached three years of age if needed.

Respiratory

At baseline survey completion at 79 months, the child was noted to use a suction machine and a nebulizer for congestion as needed. After diagnosis of combined immune deficiency and treatment with Sub-Q gamma globulin shots in 2012, there have been no long-term upper respiratory issues. Identification of this condition (combined immune deficiency) came after work up for pustules in child's buttocks when child was 11 years old.

Neurological

Child experienced myoclonic jerks between 4-18 months. A neurologist monitored this condition. Phenobarital was initially prescribed but discontinued after it created lethargy. Topomax was tried next and produced clogs in child's g-tube. Lamictal was then prescribed and effective in controlling the myoclonic jerks. Additionally, she was diagnosed with cerebral palsy due to trisomy 13 with epileptic retardation at one year. Seizure activity ceased at three years of age. Medication was discontinued until child reached 10 years of age when seizures began again (tonic-clonic type).

Between 79 and 110 months, child was taking melatonin and experienced myoclonic seizures and two grand-mal seizures. Melatonin and essential fatty acids (EFA; over the counter fish oil) was discontinued after grand-mal seizures. Child was additionally diagnosed with tonic-clonic seizures and partial complex seizures. Klonopin and Keppra were used at age 10 for approximately a year. Keppra was ineffective due to agitation. Lamictal was added at age 11. At approximately age 12, child began to experience partial complex seizures with status epilepticus due to Lamictal**.** Klonopin was continued, and Onfi was prescribed. Lamictal was tapered to Onfi with positive results. Oxygen and a pulse oximeter are kept at home in case of seizures. In addition, child was diagnosed with microcephaly at approximately seven years of age.

At present, seizure activity has resumed. Medications are in the process of being adjusted to manage this recurrence. In addition, Diastat is used in controlling seizures when needed, as are oxygen and a pulse oxymeter.

Renal

Infant was diagnosed with hydronephrosis (swelling of the kidney due to reflux) of the left kidney shortly after birth. Condition resolved at five years but then returned shortly after and affected child's right kidney. Currently, right kidney has double collecting system with history of mild bilateral reflux on both sides. Prophylactics antibiotics have been and are successfully used to ward off urinary tract infections (UTI). When child is diagnosed with a UTI, Keflex (Cephalexin) and Bactrim are used and are effective. The condition is further monitored with annual renal ultrasounds and prophylactic dose of Nitrofurantoin to avoid urinary tract infections. Sulfratrim was used initially. A small cyst is present in one kidney. A specialist monitors the condition.

Child began menstruating at 12 years. At present, there are no issues with monthly menses. At 13 years, child was diagnosed with wandering spleen for which a laparoscopic mesh splenopexy ("sandwich" technique) was performed. Treatment was **s**uccessful.

Genital

At 139 months, child was treated for pustules. An enflamed cyst was surgically removed in the labial area. Treatment was effective and issue is ongoing. Remaining cysts are monitored.

Gastrointestinal

Ladd's procedure for complete bowel malrotation was successfully performed at six weeks of age. A nissen fundoplication was performed at 12 months.

Nutrition

Immediately after birth, the infant was fed primarily by nasogastric tube (n-g tube). Infant left hospital using a Haberman bottle-feeding system and no feeding tube. At 6 weeks, infant was hospitalized for "failure to thrive." A gastronomy tube (g-tube) was placed after testing confirmed reflux.

At 12 months, a nissen fundoplication was performed and g-tube was reinserted. At 18 months of age due to continued motility issues, formula was changed from elemental predigested formula to Pediasure. Prevacid (for stomach discomfort, gas and reflux), Miralax (for constipation), Erythromycin (for motility) and Simethicone were also prescribed. All four are still used currently, however, Eryped is now used in place of Erythromycin along with Prevacid and Miralax. This combination has proved effective. Positioning was also adjusted to assist tolerance of bolus feedings. At baseline survey completion at 79 months, the child continued to receive bolus feedings through a g-tube with by push hourly feeds at smaller volume and pump feeds at night.

Currently, child is fed by g-tube hourly (90 to 120cc + as tolerated) and by pump at night (80cc per hour). Child uses a Bard foley catheter. Phazyme treats stomach discomfort and excessive gas. At 160 months, Prevacid, Miralax, Eryped and Phazyme, are taken daily and are effective.

Orthopedic

Child began to walk with assistance at age three. A prone stander was used to build strength. Ankle foot orthotics (AFO) were used for short period of time. At age five, child used a reverse K-walker with adult guidance, began crawling and equine therapy. At age six, child began to use a pummel swing to assist with trunk control and balance. A year later, at age 7, she was able to walk hand in hand with an adult. By age nine, child's balance improved and she was able to walk on her own in the home but was easily fatigued. Sinemet (a medication used to treat Parkinson's disease and dystonia) was added to her regimen to assist with muscle tone. She was also able to get on and off a pummel swing independently. At 139 months, child walks unaided during daily activities at home and with a one on one aid at school and outside the home such as from house to the car, car to classroom and parking lot to doctor's office within close walking distance.

Currently, child uses a Meywalk Gait Trainer at school and is able to transfer independently. Child uses a transport wheelchair (Zippie IRIS Pediatric Tilt-in Space Wheelchair with shoulder strap, support at hip, and abductor between legs and five-point harness for trunk alignment) for longer distances.

Child was diagnosed with scoliosis at one year of age. Mother reported that increasing child's wheelchair support decreased curvature from 15 to 7% by age three. TLSO back brace was used for a short period of time. It was discontinued due to poor balance when walking. Stretching exercises to help manage scoliosis and kyphosis are provided by school personnel, physical therapist and mother. At age seven, osteomyelitis bone infection in the spine was diagnosed and treated. Increased kyphosis was also noted. X-rays showed destruction of T12 and L1 due to presumed osteomyelitis. Infectious Disease specialist prescribed Rocephin via port (catheter placed in the upper chest area) for a period of six weeks.

Oral health

Child began dental treatments at 24 months and was diagnosed with excessive plaque. At 110 months, out-patient surgery was performed to remove six baby teeth. At 139 months, child was diagnosed with an ectopic molar eruption into the maxillary sinus, which was removed by a maxillofacial surgeon at 156 months.

Vision

Child has limited vision in right eye and no vision in left eye. In addition, cataracts removed at approximately three months of age. Contacts were used to assist with up close vision when child was 1-3 years of age. Glasses for close distance were prescribed and used between 3-5 years of age. Distance glasses helped child crawl at age 5. At 6 years of age, child was prescribed bifocals for up close and distance vision. Bifocals were effective until 10 years of age. Child's school staff suggested near and far distance glasses when she was 11 years old. Prescription was updated at 110 and 139 months. Mother reported child wears glasses mostly at school and is able to navigate without them at home.

Auditory

At age six, child required removal of a brachial cleft cyst behind left ear.

Immunizations

Currently, all immunizations are up to date. Initial vaccinations began at two months of age with DTaP for diphtheria, tetanus, and pertussis. Child received a total of five doses of DTaP

and one of TDaP. She is up-to-date on Measles, Mumps, and Rubella (MMR) (two doses), Haemophilus influenza type B (HIB) (three doses) as well as Hepatitis A (HepA, two doses), Hepatitis B series (three doses) and Varivax (varicella, one dose). She also had four doses of inactive poliovirus and two doses of Prevnar (streptococcus pneumonia). Finally, child has also received annual or biannual flu shots since the age of seven and the H1N1 vaccine in 2009 (two doses). No adverse reactions were reported after any immunization.

It is also important to note that nebulizer treatments are used for sinus infections and upper respiratory infections (Albuterol sulfate initially then added Ipratropium bromide). Treatments are only continued as needed in the past year since starting gamma gobulin shots to address child's immune system deficiencies.

Current Education and Therapy Services

Child receives educational services in a special education classroom. She also receives occupational, physical and speech and language services in the school setting as well as adaptive physical education twice a week for 30 minutes at a time. Physical therapy is provided daily for 30-60 minutes with one-on-one aide, language and speech group for 30 minutes a week and occupational therapy consult once a month. Child also receives orientation & mobility training monthly (15 minute consultation). Mother notes more one-on-one therapy sessions in the primary grades (2nd through 4th grade). It is important to note child has had the same one-on-one aide since second grade. Mother notes child has had vision, speech, OT, PT as well as orientation and mobility integrated within child's Individual Education Plan (IEP). In addition, having the same one-on-one aide has helped to integrate individual and group therapy sessions and what is provided during daily instructional activities.

A private physical therapist worked with the child to help her crawl and walk. Therapist also has consultations with school staff twice a year to discuss optimal stretching and movement activities at school. Currently, treatment focuses on stretching and movement to address kyphosis and scoliosis issues.

Developmental milestones

Child has attained most developmental milestones to the age of 9 months with some skills extending to the 12-18 month range.

Currently, child using a Big MAC button, pull switch, and 4-way switch for communication at home and school. She also uses facial expressions and a few signs/gestures to

communicate her wants and needs. Child demonstrates affection to preferred adults and her siblings. She engages with others using eye contact, smiling and giggling aloud. She also vocalizes and is able to hum a few recognizable tunes at appropriate times, such as the "I love you" song from Barney.

Cognitively, child can problem solve. For example, if an object is in her path, she will walk around it. She also demonstrates choice making to go to and from different areas of her home, such as the bathroom to play with water in the sink, clothing and videos. She is able to respond to one step requests such as stand up. Child also demonstrates an understanding of the concept of "one more time". For activities of daily living, child cooperates with dressing by lifting her arms and legs. Child is still in diapers but uses the toilet facilities at both school and home with assistance to remove diaper. She cooperates with necessary health care routines such as tooth brushing.

Additional developmental skills include: displays reaction to familiar person nearby, looks at face of person speaking to her, visually focuses on objects and screens, touches objects when placed nearby, turns towards sounds and responds to hearing name with head turn toward sound. In the fine motor area, child can grasp and release toys items, activate cause and effect toys and transfer hand-held objects. She also reaches for and leads adult to a desired object. In general, child expresses needs and wants by using vocalizations, gestures, and singing. She maintains attention for the duration of exchanges with familiar adults. She recognizes her image in the mirror. Child anticipates tickles and demonstrates excitement when going to or from school. She is also able to adjust to changes in schedule and environment without difficulty. Mother notes child's positive demeanor. Child laughs and smiles in response to daily activities and interactions with family members and familiar adults.

Mother notes that addition of EFAs assisted child's development especially gross motor skills (crawling and walking). Seizures were noted at age eight so the supplements were discontinued. Child was entering puberty during this time as well.

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