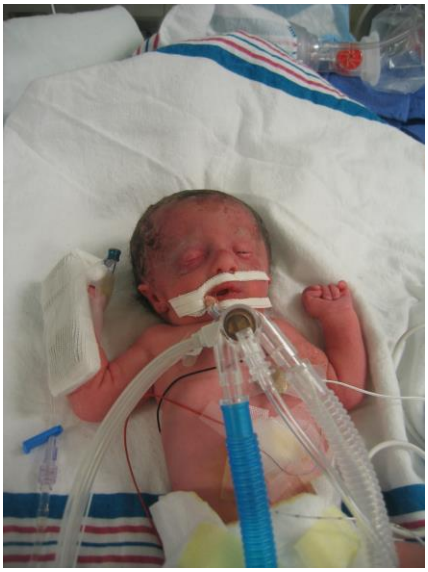

TRIS project
2014

Maristella was born in 2009. Trisomy 18 was suspected but not confirmed before birth. Child lives in the United States. Participation in the TRIS project began in 2010.

Mother was 39 and father was 56 years old at the time of conception. Child with a rare trisomy condition was third of three pregnancies (gravida 3, para 2; ab 1). Parents were aware of rare trisomy conditions from a previous pregnancy.

Birth Information

Trisomy 18 was suspected prenatally after alpha-fetoprotein (AFP) screening. Further testing was refused due to associated risks. Prior to birth, infant was identified with a possible ventricular septal defect (VSD), microcephaly, and small for gestational age. Mother also experienced preeclampsia and gestational diabetes during pregnancy.

Female infant was born by emergency caesarian section at 35 weeks weighing 1800 grams and 45 cm in length on December 31, 2009. APGAR scores at 1 and 5 minutes were 4 and 5, respectively. Infant was diagnosed with trisomy 18 by results of fluorescence in situ hybridization (FISH) test at two days of age and skin biopsy at 11 days of age. Karyotype report was received approximately 20 days after FISH test.

Infant presented with phenotypic features of low-set ears, small jaw, microcephaly, and clenched hands. The following medical conditions were present: heart murmur, VSD, coarctation of the aorta, feeding difficulties, and jaundice.

Infant was born in a regional hospital with a specialty care nursery (Level II). Eight hours post birth she was transferred and received care at a neonatal intensive care unit (NICU) in a

specialized hospital and was discharged at the age of six months and six days. While in the post birth care unit, a nasal cannula, apnea monitor, ventilator, continuous positive airway pressure, heart monitor, pulse oxymeter, and gavage tube, gastrostomy tube, and IV feeding were used. An audiology exam, blood transfusion, echocardiogram, intubation, ultrasound, and x-ray series were also performed. During this time, open-heart surgery to repair child's VSD and coarctation of the aorta and a tracheostomy were performed.

Surgeries

Repair of VSD and coarctation of the aorta were performed at three months of age. At approximately four months of age, a tracheostomy and gastrostomy tube was placed. At 46 months, an uvulopalatopharyngoplasty was performed. Child underwent several bronchoscopies from age 4 months to present.

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

Cardiac

Child was diagnosed with heart murmur, VSD, and coarctation of the aorta soon after birth. Child's VSD and coarctation of the aorta were repaired at three months of age.

Respiratory

A tracheostomy was placed and child was put on a ventilator for the treatment of central apnea. Child was on a ventilator 24/7 from the age of four months to one year. After this time, child only required the ventilator when asleep until the age of 27 months.

At 21 months, child was diagnosed with obstructive apnea. Mother notes a formal diagnosis of mild central apnea was not made until March 2012 when child was 26 months old. This was after child's initial sleep study (none were ordered during infancy). A subsequent sleep study in March 2013 showed no sign of central apnea, only obstructive apnea. A bronchoscopy was performed at four months for further information about child's obstructive apnea.

At 29 months, child was experiencing episodes of central apnea and receiving supplemental oxygen, Budesonide (inhalation suspension) for airway maintenance and Albuterol Sulfate (inhalation solution) every 4 to 6 hours as needed for cough, shortness of breath or wheezing when ill. These medications were also noted at 41 months.

Since child reached 36 months of age, central apnea has been considered resolved. This was confirmed with an additional sleep study at 44 months of age. Obstructive apnea was still noted to be present. At 45 months of age, child underwent an uvulopalatopharyngoplasty (surgery to remove excess tissue in the throat to widen the airway) to address obstructive apnea. An additional post-surgery follow-up is scheduled. Child was successfully decannulated March 2014. Since decannulation, child uses an airway clearance device twice a day for maintenance (The Vest®) and also during sickness such as congestion. The device uses high-frequency chest wall oscillation (HFCWO) to dislodge mucus from the bronchial walls and also thins thick secretions.

Renal

No renal conditions are present. Conditions were ruled out after a complete diagnostic retroperitoneal ultrasound (provides images of kidneys, abdominal aorta, common iliac artery origins and inferior vena cava) when child was 27 months old and, again, at 39 months of age.

Gastrointestinal

A gastronomy tube was placed at four months to address feeding needs. Around this time, child was diagnosed with stomach discomfort/excessive gas. Child was on Reglan for first 18 months. Prevacid was also provided at the same time. Medications were discontinued once mother began blenderized diet (rather than infant or specialized formula) through the gastrostomy tube. Mother also provides essential oils including fennel tea to aid with constipation, bloating and excessive gas as well as to boost child's immune system.

Child continues to experience stomach excessive gas intermittently. Mother uses fennel teas and oil to relieve excessive gas.

Nutrition

Child has never accepted food orally.

Initially, infant received gavage feedings. Gastrostomy tube was placed at four months of age. Infant was fed with fortified breastmilk until six months of age. Then, infant formula was provided until age one. From age one to 18 months, specialized formulas were used without

success (Pediasure, Peptamen Jr, Compleat). At 18 months child received a blenderized diet via gastronomy tube.

Child underwent a barium swallow study in 2011. No restrictions on oral intake were given but child continues to demonstrate a high level of oral aversion.
Child was diagnosed with a vitamin D deficiency and is given Vitamin D3. Values are checked annually and adjustments to the dosage are made as needed. Child also receives calcium carbonate daily. Currently, Vitamin D3 levels are in the normal range.

Mother notes use of MediHoney for relief of occasional skin problems around the site of the child's gastrostomy tube.

Neurological

Child was diagnosed with microcephaly at birth. No seizure activity has been reported.

Due to plagyocephaly (asymmetrical distortion/flattening of the skull), child developed torticollis (asymmetrical head or neck position). Child was denied a molding helmet during infancy. At 21 months, which mother notes is significantly past the typical recommended wearing period for effectiveness of molding helmets, child's new Physical and Rehabilitation doctor prescribed a molding helmet as child's skull was still soft enough for its use. Within one year, child wore two helmets. The initial helmet was replaced after six months due to growth. Resulting skull changes were significant and torticollis significantly improved as well. Child now has full range of motion but tends to keep head turned to the right. Mother also notes that child's hearing loss is more significant in her right ear and may be the cause of the preference to the right side.

Orthopedic

Child wore hand splints for the treatment of clenched fists. Splints were on one hour every four to five hours or when asleep. At approximately one year of age, child started wearing thumb-splints for a few hours each day. Child can now fully open her hands although still tends to keep them fisted.

A thoracolumbosacral orthosis (TLSO) was used in the treatment of scoliosis (20 degree C-shaped) from 30 to 36 months. Child's Physical and Rehabilitation doctor and mother agreed to discontinue its use since it restricted child's movements.

Child uses a Squiggles Stander (Leckey) for approximately 30 minutes a day. However, the current focus is on crawling. A Creepster Crawler is used to encourage gross motor skill development.

Oral health

After the age of two, child experienced excessive tooth decay, which was treated with additional cleanings. At initial dental visit at 30 months, child was diagnosed with bruxism. Follow-up with dentist is done every six to nine months.

Vision

Child was diagnosed with esotropia, a form of strabismus in which one or both eyes turn inward, at 30 months old. Condition is monitored every six months by an ophthalmologist as well as an annual visit.

Auditory

Child has extremely small ear canals. Between the ages of two and four, child experienced ear infections, which were successfully treated with antibiotics. The child also experienced excessive wax build up which was treated with occasional Debrox drops (no longer used) and cleanings when child was undergoing surgical procedures.

Child was diagnosed with a moderate conductive hearing loss and wears BAHA hearing aids. Audiology visits are scheduled every 6 months.

Immunizations

Child did not receive any immunizations prior to NICU discharge at six months and six days of age. First immunization shot was administered at approximately seven months. Child is now up to date with all immunizations and has not had any adverse reactions. Child has also not received RSV shots.

Current Education and Therapy Services

Up to the age of three, child was followed by Early On (early intervention program) and received physical and occupational therapy. Data from TRIS Follow-up Survey indicated at 28 months, child received physical therapy and occupational therapy for 120 minutes per week at home. She also received therapies including speech and language therapy 60 minutes per week at a clinic as well as audiology and vision services. In total, child received up to four hours a therapy a week depending on health and local weather conditions.

Between age 3 and 3.5 years, child received one hour a week homebound services from a teacher. At present, child is homeschooled and receives physical, occupational and speech therapy at a clinic for a total of three to six hours a week.

Initially, parents receive 360 hours of respite care per month. This was reduced to 240 hours per month but typically only 160 hours per month were covered. This level of respite care was terminated due to child's decannulation in March 2014. Starting in June 2014, the child's family will receive 180 hours a year of respite care.

Developmental milestones

Child turns head to her name and can distinguish familiar and unfamiliar faces. Child also demonstrates understanding of different environments such as brother's classroom, hospital for appointments and church through facial expressions and activity level. Child also indicates when completes or is no longer interested in a task or activity by putting an arm across her eyes to block contact with the situation and/or individual. In addition, mother notes how child actively attempts to stop her or home care nurse from changing the tracheostomy (child has purposefully decannulated herself several times). Child inconsistently follows one-step directions. She has recently learned to wave "Hi/Goodbye" following a verbal cue. Mother report that child smiles unless feeling ill. Overall, mother describes child as laid back and able to assert herself when necessary.

Child attends to a variety of visual stimulation including watching football with father. In the past six months, she has changed her preferences from children's shows such as Sesame Street to Dinosaur Train and Wild Kratts. Child will attend for up to 30 minutes.

Although child's fine motor skills and dexterity are limited, mother notes child consistently pulls her eyeglasses off her face and then swings them in the air like a trophy. Child can grasp toys and hand held items for a few seconds. Presently, the focus of gross motor development is crawling with use of a Creepster Crawler. Child can sit with support.

Child produces sounds. Although they are not consonant-vowel sounds, they differ depending on the situation such as a high-pitched sound for attention or a grunt to express disappointment. Child only cries when in pain such as a stomachache or toothache.
In the area of daily living skills, child can push her feet into shoes and help push her arms through a shirt.

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