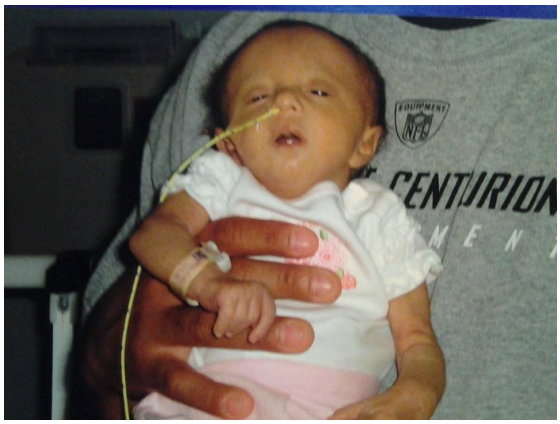

TRIS project
2014

Madison was born in 2008 and was prenatally diagnosed trisomy 18. She lives in the United States. Participation in the TRIS project began in 2009.

Mother was 30 and father was 31 years old at the time of conception. Child with rare trisomy was the first of two children (gravida 2, para 2).

**Birth Information**

Infant was diagnosed with full trisomy 18 at 18 weeks gestation by amniocentesis and alpha-fetoprotein screening. Infant was identified with atrial septal defect (ASD), ventricular septal defect (VSD), clenched hands, omphalaocele, horseshoe shaped kidney, rocker bottom feet, and intrauterine fetal growth restriction at time of diagnosis.

Female infant was delivered via planned caesarian section at 38 weeks gestation. Infant was born weighing 1984 grams and 44 centimeters in April 2008. APGAR scores at one and five minutes were 2 and 7, respectively. At birth, the infant presented with a single veined, herniated umbilical cord, low-set ears, small jaw, microcephaly, webbed toes, rocker bottom feet, right preauricular tag, and a small opening on the right cheek. Infant also presented with the following medical issues: respiratory distress, kidney problems, ASD, patent ductus arteriosis (PDA), VSD, feeding difficulties, and jaundice.

Infant was born at a hospital with a well-equipped neonatal intensive care unit (NICU). Oxygen was delivered via nasal cannula. Infant was also on a ventilator for three days after surgery to repair omphalocle. A pulse oximeter, apnea monitor and bilirubin lights were also used prior to discharge. Infant was fed via nasogastric tube (n-g tube) and IV feeding. An audiology exam, echocardiogram hepatobiliary (HIDA) scan, renal ultrasound, and x-ray series were performed in the post-birth care unit. Infant's omphalocele was repaired and appendix removed in surgery on first day of life. She remained on unit for 31 days. Discharge weight was 2179 grams. Discharge was intended for family to have time at home with their infant, not due to resolved health concerns. Frequent follow-ups with specialists initially to closely monitor health status.

**Surgeries**

Infant's omphalocele was repaired at one day after birth and an appendectomy were performed. At three months, infant had a liver biopsy and a cholecystectomy (removal of gall

bladder). A gastrostomy tube was also placed during the same surgery. At 4 months, ASD, PDA, and VSD were repaired. At 34 months, a partial right nephrectomy was performed and ear tubes were placed.

**Cardiac**

At birth, infant was diagnosed with ASD, PDA, and VSD. All three defects were repaired at four months. Cardiac issues resolved after surgery. Prior to repairs, infant was prescribed Lasix. Currently, child is on a two year follow-up schedule with cardiologist.

**Respiratory**

Prior to discharge from the NICU, infant was required oxygen and ventilator assistance. Amoxicillin and nebulizer treatments were prescribed for reoccurring sinus infections and colds. Child still uses antibiotics when she has a sinus infection but does not use a nebulizer currently. Child has been prescribed Singulair and Zyrtec for the management of airborne allergies. Management of allergies also included removal of carpeting, pets, and smoke in the home.

At 34 months, child contracted RSV that required hospitalization and ventilation. Child was then weaned from ventilator to a nasal cannula and back to room air. During this hospitalization, it was discovered that narcotics including morphine, and Dilaudid caused respiratory distress and arrest. RSV was contracted while in the hospital after kidney surgery.

Child still experiences recurring sinus infections and colds that required suctioning and Amoxicillin or similar antibiotic. At 48 months, Nasonex was added for the management of airborne allergy symptoms. Per mother's report, medication seems to help with nasal drainage from child's small nasal passages.

**Neurological**

Infant was diagnosed with microcephaly at birth. By 16 months, head circumference kept pace with body growth, but some fluid had been reported to collect at the back of the brain. No spasms or seizures were reported at 16 months. In addition, no seizure activity noted since 16 months to present to date. At 41 months, child was also diagnosed with Dandy-Walker syndrome.

At 53 months, child was reported to take Melatonin for the treatment of sleeplessness. At 65 months, medication was changed to Benedryl after it was determined that the Melatonin was ineffective. Mother reports medication seems to help but going to sleep at night is an ongoing issue. Mother explains, "child bangs her head repeatedly when tired. When she was younger, it was only her head from neck up while still laying down. As she has gotten stronger and more active, she now gets on her knees in a crawling position and has her hands under her head while she bangs. It will continue until a person intervenes to hold or lay beside of her to make her stop. It has continued for hours and she will cause a hematoma on her head. Often, repetitive vocal noise goes along with the banging. Once she goes to sleep, she usually sleeps well but getting to sleep is a difficult process. I usually sleep with her at night time due to the headbanging".

**Renal**

During the perinatal period, infant was diagnosed with hydronephrosis, duplicated upper right pole, cysts, and double ureters (right kidney). Actigall was prescribed at birth to treat liver dysfunction. It was discontinued at three months of age after a liver biopsy was preformed to find the cause of continuing liver dysfunction. Infant was also noted to have an abnormal stalk on gallbladder. Once gallbladder was removed, infant no longer had increased bilirubin levels.

At 16 months, child was diagnosed with persistent urinary tract infections (UTI) and was catheterized intermittently to ensure complete emptying of bladder from approximately 51-63 months. Child's fourth summer was when it became more persistent. Mother questioned if child's time in a pool had any impact on increased UTIs. UTIs have been unsuccessfully treated with Furadantin, which caused vomiting and loose stools. Child was then prescribed Trimethoprim and was effective. At the current time, catheterization is no longer necessary; discontinued at 63 months.

At 34 months, partial nephrectomy of nonfunctioning right kidney was performed. At approximately 50 months of age, child's prescription was changed to Bactrum for the prevention of UTIs. Mother also reports that when child's constipation issues were treated, child experienced less UTIs. Child receives daily liquid glycerin suppository to treat constipation.

Since birth, child receives an abdominal ultrasound every six months to monitor these conditions and monitor for Wilms tumors.

**Gastrointestinal**

At 3 months, a cholecystectomy (removal of gall bladder) was performed. Between the ages of 4 and 7 months infant was prescribed Prevacid (Lansoprazole) for the treatment of vomiting caused by reflux. At 12 months, infant started to experience excessive gas and stomach discomfort that was treated with elevated positioning. At 40 months, child was again prescribed Prevacid for reflux. Several additional reflux medications were used before child reached four years of age including Carafate and Zantac.

Beginning at 18 months, child was prescribed Miralax to use as needed for the treatment of constipation but it was not effective. At 24 months, medication was changed to Senna laxative but it caused gas with no bowel movements. Since the age of 62 months, liquid glycerin suppositories have been used and were immediately effective. UTIs have decreased as well.

At 62 months, child was changed to a blended diet four times daily by bolus feeds through gastrostomy tube. This change has reduced vomiting, reflux, and constipation. An example follows: breakfast blend is: 3 Tbsp coconut oil, 1 cup coconut milk (could use almond milk or regular milk), 1 cup baby multigrain rice cereal, 8 oz. Greek yogurt, 2 pack baby food prunes, 2 pack baby food bananas, 3 large jars mixed fruit baby food (makes approximately 7 one cup servings). Breakdown: 261 calories, 24 carbs, 14 fat, 4 protein, 26 sodium, 18 sugar. Example dinner blend: 1 cup cooked quinoa, 2 tbsp milled flaxseed, 7 oz baby food carrots, 2 jars butternut squash pear baby food, 2 jars mixed veg. and turkey dinner baby food, 3 jars beef

and broth baby food, 6 Tbsp extra virgin olive oil, 8 oz baby food squash, 5 oz applesauce, 1.5 cups unsweetened almond milk, 2 pack prunes baby food (makes approximately 7 one cup servings). Breakdown: 281 calories, 27 carbs, 16 fat, 7 protein, 90 sodium, 13 sugar.

Diet began with mostly baby foods and foods already in puree form. Since purchase of a Vitamix blender (able to purchase at a discounted price with a medical prescription), mother reports use of more fresh and frozen foods. Mother further reports there was initial hesitancy to try the blended diet but "it has been the best change we have made!!! Life changing!! No more pump!!"

Mother reports no food allergies to date. Pears and prunes help with constipation as child is having some bowel movements on her own but still mostly relies on suppository for complete bowel emptying.

**Nutrition**

Immediately after birth, infant was fed primarily by nasogastric tube. Infant was not able to latch on to be fed by bottle or breast. A gastronomy tube was surgically placed at 3 months. At 28 months, infant still received all nutrition and fluids by gastronomy tube and formula was changed from Pregestimil to Elecare to reduce stomach discomfort.

At 41 months, child was reported to drink from a cup for supplementary fluid intake with remainder of nutrition given via gastrostomy tube. At 53 months, child was introduced to pureed foods with some success. Child attempts to feed herself with a spoon as well. At 68 months, child still received most of her nutrition form bolus feedings four times daily of a blended diet. Child also received a Duocal calorie supplement.

Child was offered opportunities for exploration with food but not enough taken by mouth to meet nutritional needs. Child also has to be monitored for gagging when accepting food orally. Child has become more interested in food as she has grows older. Mother reports that change to a blended diet is a factor as reflux has been reduced which assists food to taste better.

**Orthopedic**

At 16 months, child used a prone stander. At 28 months, child was also fitted for orthotics (Supra-Malleolar Orthosis) and used a Leckey™ Squiggles stander and Rifton™ Mini Pacer Gait Trainer. At 41 months, child still required Supra-Malleolar Orthosis and used a Wenzelite Rehab™ Seat2Go Positioning Seat and a Hi/Lo Rifton™Activity Chair. At 68 months, child was using Ankle-Foot Othosis, Snug Seat™ Rabbit Stander, Sleep Safe Bed, Rifton™ Mini Pacer Gait Trainer, and Snug Seat™ Stingray Stroller.

Child sits up on her own, pulls to stand on furniture, crawls, and sits up on her knees and bounces.

**Oral health**

Child's first dental visit was at the age of 40 months. Attempted cleaning but child was frightened of the suction sound on the cleaning machine. As such, the cleaning was not completed. There was a follow-up visit at 50 months after losing first tooth. Child has had

regular cleanings (toothbrush only, not machine) and fluoride treatments every six months since then. Initial x-rays were taken at 72 months. Referral to a specialized dentist is expected to happen in future. There are no significant dental needs to address at this time.

**Vision**

At 12 months, infant was diagnosed with hyperopia, myopia, and strabismus. Treatment included prescription eyeglasses. At 28 months, child was reported to be resistant to wearing eyeglasses and effectiveness of treatment was not able to be determined. Child was additionally diagnosed with ptosis (weakness of the muscle which raises the eyelid) of the right eye.

Glasses were initially prescribed at 12 months. Prescription changed frequently (approximately every 6 months) for the first two years. Currently, prescription is checked yearly and six months appointments for follow-up. Child often chews on her glasses instead of wearing them.

**Auditory**

Infant was identified with small ear canals. At 10 months, infant experienced otitis media and was treated with antibiotics (Amoxicillin). At 28 months, child continued to experience otitis media and was successfully treated with Amoxicillin.

Child received ear tubes at 34 months with no relief in symptoms and reoccurring ear infections. Ear tubes caused bloody drainage and ear canal irritation due to size of child's ear canals and had to be removed due to significant irritation. Minor hearing loss was diagnosed in the left ear due to a structural abnormality and reoccurring infections. This condition was identified through a sedated ABR test. The loss is not significant enough to require hearing devices at this time. Child is noted to respond to sounds in her environment. Child is monitored yearly.

**Immunizations**

All immunizations are up to date and were started as scheduled. Mother reports spacing clusters of vaccinations instead of giving all on the same day. Child would receive several, wait a few weeks and receive the remainder so as to not overload her body. Infant was reported to have chicken pox 12 days after receiving the varicella virus immunization at 13 months.

**Education and Therapy Services**

At 16 months, child received 120 minutes of physical therapy (PT) per week and 60 minutes of speech and language therapy (ST) per week at home. Child also received early intervention services at home. At 28 months, child received 120 minutes of PT, 60 minutes of ST, and 60 minutes of occupational therapy (OT) per week. In addition, 80 hours of respite care were provided weekly by a community agency.

At 36 months, child received a diagnosis of developmentally delay and began to receive services in a special education classroom in child's local school district. Child began with 5 days per week for 2 hours per day the first year. Child experienced congestion and respiratory issues soon after she began attending Pre-kindergarten classroom. Child missed most of the Fall term

and, in December, contracted a respiratory virus requiring a week long hospital stay where child received additional oxygen support by nasal canula. Child was then changed to homebound services for the remainder of the school year.

At age 4, child attended 2 times per week for one and a half hours per day. In the Spring, this was increased to 3 days weekly for one and half hours per day. During this time, child's stamina and strength increased. Child's Individualized Education Plan (IEP) team decided since she missed the initial year of Pre-kindergarten, child would be offered an additional year to continue to build her stamina. During child's third year of Pre-kindergarten at age 5, she attended school 5 days weekly for 3 hours daily. Child has not had any major issues other than a sinus infection and colds. Child's strength and stamina have increased significantly.

When home-based early intervention services ended at 36 months, child received OT and PT through medical model home-based services. Speech services were not provided due to lack of providers. At 53 months, child received 90 minutes of physical therapy through medical model home-based services. Currently, child receives, 60 minutes of ST, OT and PT weekly through the school system.

Recently, at 68 months of age, child's diagnosis was changed to intellectually disabled/multiple disabilities at her triennial evaluation.

**Developmental milestones**

At 16 months, child had accomplished a variety of seven month developmental milestones such as rolls from stomach to back, explores objects with hands, indicates needs by crying and some skills extending in to the 9 month level such as showing preference for familiar caregivers.

At 28 months, child used a Big Mac communication button. At 41 months, child had success with use of a choice board when given two choices. At 68 months, child was exposed to an iPad to indicate between two choices of items and activities. Child is not always consistent with making choices but offered to develop visual attention to develop this skill.

Currently, child sits on her own, pulls to stand on furniture, and is crawling up to 10 feet at a time. Child can walk with assistance at her hips or holding her hands. Her favorite gross motor activity is bouncing. Child will jump on her knees independently, jumps when an adult holds her hands, and jumps in an infant jumper that can be attached in a doorway.

In the area of fine motor skills, child can reach and hold items with one or both hands such as puzzle pieces but cannot manipulate them to put in correct place without assistance. Child is not tolerant of hand over hand assistance for writing or coloring. Recently, child has shown interest in Lego blocks. However mother reports child holds them and spins them in her hands but doesn't attempt to put them together. Yet, child will seek these blocks out of a basket of toys. Child also enjoys dumping toys from toy baskets. Child will push her arm up when putting her shirt on but this skill is just developing. Child also needs full assistance for dressing, feeding and other self-care activities.

Child enjoys social games. Social interaction is her greatest strength. Child also effectively communicates without words with varied cries and noises when unhappy or not

feeling well. Child giggles, and belly laughs when happy. Child is very interactive and playful with her sister and will "wrestle" with her. Child enjoys playing with musical toys, pretend kitchen set and baby dolls. Child also enjoys having books read to her and listening to music. Mother also reports that child seeks a great deal of oral input and chews "everything in sight". Child must be closely monitored regarding what she places in her mouth. Lastly, child also has a very high pain threshold. Mother notes that by the time child get fussy (cries and/or whines) it usually indicates a sinus or urinary tract infection.

Mother describes child as having a "very patient, laidback demeanor and loves life".

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