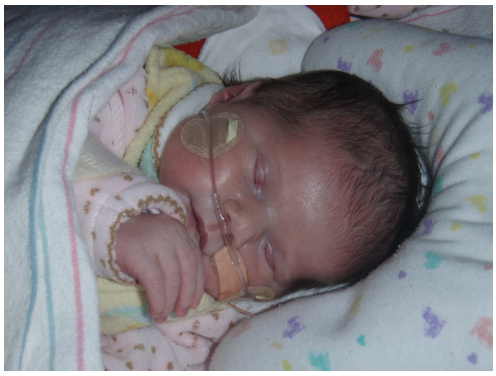

Mackenzie was born in 2009 and was diagnosed three days after birth (trisomy 9 mosaic). She lives in the United States. Participation in the TRIS project began in 2013 at the age of 44 months.

Mother was 40 and father was 37 years old at time of conception. After conception, parents reported general knowledge of rare trisomy conditions. Child with rare trisomy was the second of two pregnancies (gravida, 2; para, 2). Older sibling has been diagnosed with an autism spectrum disorder.

Birth information

Female infant was delivered via emergency caesarian section at 39+5 weeks gestation. Infant was born weighing 3020 grams and 46 centimeters in length in November 2009. APGAR scores at one and five minutes were 2 and 2, respectively and 8 at ten minutes. Infant was identified with larger than typical sized ventricles in the brain prior to delivery.

At birth, infant presented with low ears, small jaw, small & sunken appearing eyes, short palpebral fissures small anteverted nostrils, widely arched palate, thumbs with tightness to abduction, small chest, deep sacral dimple, dorsiflextion feet with limited plantarflexion, torticollis, increased generalized tone. Infant also presented with the following medical issues: respiratory difficulties, heart murmur, patent ductus arteriosis (PDA), ventricular septal defect (VSD), aberrant left subclavian artery, hypocalcemia, thrombocytopenia, congenital hypothyroidism (continues to be treated with Levothyroxine), feeding difficulties, and jaundice. Infant was diagnosed with trisomy 9 mosaic three days after birth.

Infant received the majority of her post-birth care in a specialized hospital's neonatal intensive care unit (NICU). Infant was on the unit for 3.5 weeks. Infant was also on a respirator and a ventilator for first 24 hours after birth. Oxygen was delivered via nasal cannula after first 24 hours. A heart monitor and pulse oxymeter were also used prior to discharge. Infant was fed via IV feedings, nasogastric tube (n-g tube) and, later, after surgery, gastrostomy tube (g-tube).

An audiology exam, echocardiogram, ophthalmology exam, ultrasound, x-ray series and blood transfusion were also performed in the post-birth care unit. Infant's feeding difficulties where repaired through placement of a gastrostomy feeding tube 18 days after birth. Discharge weight was 3165 grams.

Infant received palliative care after discharge from the post-birth unit. Palliative care was discontinued at age three after feeding was stabilized.

Surgeries

Infant's feeding difficulties (oral motor dysfunction and aspiration) were addressed 18 days after birth with placement of a percutaneous endoscopic gastrostomy tube (PEG g-tube). It was surgically replaced with a Mic-Key button at the age of six months.

Also, at six months of age, child received ear tubes for otitis media (recurrent middle ear infections). At seven months after birth, infant had surgery to correct dysplasia of right hip and adductory longus for left hip contracture. A spica cast was needed for eight weeks and a brace for the following six weeks. In addition, to address obstructive apnea, infant was treated with two surgical procedures: adenoidectomy and tonsillectomy at 26 months. Child's strabismus was corrected at 29 months.

Child used a thoracic lumbar sacral orthosis (TLSO) for approximately three years and had a tethered spine release procedure done by a neurology team in April 2018. Eight pins were placed for a Halo procedure in September 2018. A spinal fusion procedure followed four weeks later (October 2018). Procedures were conducted at Gillette Hospital in St Paul, MN. The tethered spine surgery was to release the spinal column to promote growth. The pin placements and fusion were a combined procedure to stop child's kyphosis and scoliosis from progressing and impeding breathing and overall growth.

The three procedures were successful. However, the tethered spine release had a side effect of a 20% change in the curvature of the kyphosis six weeks immediately post-procedures due to not using a prescribed TLSO, which caused the Halo procedure (with fusion) to occur before planned. Child's family had hoped to push off the procedure to give child more time for thoracic growth.

Recovery time for the tethered cord release surgery was two weeks. Her Halo procedure was an in-patient process due to being in traction for four weeks. After the fusion procedure, child was under sedation for 36 hours. Child was out of bed within 24 hours and walking after 48

hours. Child was released from the hospital a week post-op. At two weeks post-surgery (early November 2018), child returned for half days then to full days a short time later.

Overall, child has shown significant improvement from the recent surgeries and has not needed to wear new TLSO since 3 months post-surgery. However, since the fusion ended at the T1, kyphosis has led to a need for child to wear a cervical collar to prevent additional kyphosis from starting at the top of the fusion. The TLSO is worn overnight as well.

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

Cardiac

Infant presented with a heart murmur, PDA, VSD, and aberrant left subclavian artery. No surgical interventions were necessary to correct these conditions. All heart conditions are resolved.

Respiratory

Prior to discharge from the NICU, infant required oxygen, respirator, and ventilator assistance but did not need these items after discharge. Child is also noted to have micrognathia.

At nine months, Pulmicort and Albuterol were prescribed to prevent pneumonia. Child still uses these prescriptions as needed application with a nebulizer. Airborne allergies were reported in the follow-up survey at 56 months and treated with diphenhydramine HCI oral solution.

At 26 months of age, child required removal of adenoids and tonsils to prevent obstructive apnea. Mother reports loud snoring was the only symptom of this condition. Sleep apnea was never diagnosed.

Neurological

Mother reports seizures due to obstructive apnea shortly after child was born lasting for approximately 24 hours. No medications were necessary to treat this condition. Child was moved from supine to prone. Seizures stopped with the change in positioning allowing chin to move forward to counteract infant's jaw moving backward and cutting off her airway.

A neurologist did the child's tethered spine release. Post-surgery, the neurologist is no longer following the child. A new spine specialist has joined the child's medical team.

Renal

No renal conditions reported in baseline data. Follow-up data indicates constipation as a concern, but no treatment is indicated. More free water needed in diet.

Currently, child takes polyethylene glycol when needed for constipation. She is given free water at the same time she is given Levothyroxine (two 25mg tabs on Monday and Fridays; 1 ½ tabs the other 5 days). Child sees an endocrinologist and is now receiving growth hormone injections. Child receives one 5mg/1.5ml SOLN injection a night subcutaneously.

Gastrointestinal

Child was diagnosed with gastroesophageal reflux (GER) and aspiration while still in the NICU. Condition continues to be effectively treated with Zantac.

As of early 2019, child still experiences GER with no episodes of aspiration. Child still uses a g-tube (Mini button) as primary delivery system for nutrition since does not eat enough orally to meet daily caloric needs.

Nutrition

Immediately after birth, infant was fed primarily by nasogastric tube (n-g tube). Infant was not permitted to latch on to be fed by bottle or breast due to aspiration issues. A gastronomy tube was surgically placed 18 days after birth.

At time of baseline survey, child received g-tube feedings primarily. At Year 1 Follow-up, child continued to receive g-tube feedings as well as pureed foods orally. Child has also passed a swallow study allowing for additional oral feeding to start. A gastroenterologist is involved in the child's care.

Child's recent data states she eats a variety of solid foods especially pizza, crackers, and craisins and larger quantities (sufficient for one meal a day). Child also takes liquids such as orange juice and pink lemonade orally as well as soda, which is child's favorite. Mother reports child is often found raiding the refrigerator or food left on the counter or any family member's plate at home and in the community.

Oral health

Child began dental visits at the age of 27 months with a dentist specializing in working with children. Child requires sedation for routine dental care. Mother reports child has excessive plaque, due to lack of oral feeding and GER. Mother notes that plaque is diminishing as more oral stimulation is provided and oral intake increases.

As indicated from most recent follow-up survey, child sometimes has excessive plaque. Child sees a pediatric dentist for dental surgery and regular check-ups every 6 months. Deep cleaning needs to be done under anesthesia.

Vision

Child was diagnosed with strabismus at approximately nine months. This condition was surgically corrected at 29 months.

Child's initial vision-related diagnosis was intermittent alternating exotropia at 32 months. At 38 months of age, diagnoses of optic atrophy ("dull" optic nerve), hypermetropia and astigmatism were made when child were added. Hypertropia was identified at 44 months. An ophthalmologist follows the remaining conditions at six-month visits. Glasses are not needed at this time.

Currently, child sees an ophthalmologist every six months. Child still lacks depth perception and is being monitored at each check-up. Glasses or magnification of any type are not needed at this time.

Auditory

Child has a documented hearing loss (mild to moderate in left ear; wears a hearing aid) based on auditory brainstem response (ABR) results. In addition, otitis media was first diagnosed at one month of age. Child had tubes placed at five and 26 months. The latter was during tonsil and adenoid surgeries. Child has also used hearing aids since 18 months of age as well as American Sign Language.

Immunizations

Immunizations began and continued at the ages recommended by the American Association of Pediatrics for Diptheria, Tetanus and Pertussis (DPT), Haemophilus influenzae type B (HIB), Hepatitis A (HepA), Hepatitis B series, Inactive polio virus, Influenza, Measles, Mumps and Rubella (MMR), Pneumococcal pneumonia, Respiratory syncytial virus (RSV) and Varicella. No adverse reactions are noted.

Orthopedic

At birth child presented with torticollis, thumb in palm, hip dislocation, and tightness of dorsal structures bilaterally at ankles and feet. At seven months, infant had surgery to correct dysplasia of right hip and adductor longus for left hip contracture.

Child was also diagnosed with scoliosis and kyphosis at 3.5 years. An orthopedic surgeon monitored the child at six-month intervals for the first three years and, now, yearly. Currently, child is followed by two orthopedic surgeons (one focused on walking, the second for spine). Both are seen every six months.

Child has used McKie hand splints for approximately the first seven months after birth and ankle foot orthotics until age four. After child's fourth birthday, a change was made to a foot orthotic with flexible metal shoe insert to providing bracing to calf. In addition, child went from supra malleolar orthotic (SMO; bracing to the ankle) back to an ankle foot orthoses (AFOs) because was unable to maintain a frame stance (standing with her feet spread to remain stable) due to weak ligaments in knees, which cause pronation. Left foot can maintain position but has low tone, which contributes to pronation. When right foot and frame stance improves, SMO will be reintroduced.

Developmental milestones

At completion of baseline survey at 44 months of age, child demonstrated most 12-month milestones including standing, demonstrating preference for toys/objects, and using two hands to explore objects. In addition, child was able to scribble with a crayon, sort objects based on one characteristic (e.g., size), use signs and gestures to communicate needs, attempt to throw or kick a ball and play cooperatively with sibling (a 36 month skills).

At follow-up at 56 months, child was able to walk independently. Child is using American Sign Language to communicate wants and needs to her parents and teachers. Child was also better tolerating oral stimulation and accepting more food orally.

Developmentally, child uses sign language to communicate (approximately 50-60 signs) and vocalizes sounds during music activities at preschool (e.g., mama, baba). When presented with two songs or preferred items, child demonstrates choice making. Child enjoys playing with toys with lights and music as well as sensory play activities including painting. Fine motor skills such as approximations of age appropriate grasps and copying vertical and horizontal lines using a slant board are improving. Child is able to interact with apps on an iPad. Finally, child is showing increased interest in same age peers including observing them during small group activities.

Child participates in indoor and outdoor gross motor activities with adult support including playground equipment and an adapted tricycle. Mother notes child especially enjoys

climbing on various play structures. Child is also noted to be cooperative during dressing activities. Child can also complete some steps of hand washing with little to no adult assistance.

At the present time, child is demonstrating greater independence. Child is more willing to try new foods and has an easier time going to sleep at night. Child often asks for her favorite song using sign language.

In gross motor development, child continues to climb on a jungle gym, and pumps to swing (even if it is not in a straight trajectory). Child can go down a slide but is wary due to lack of depth perception. Child walks at a fast pace on a flat surface. Child goes up and down stairs holding a rail. Currently, PT is working on not holding the rail and going down a single step independently, not holding a rail or hand, in a familiar place. Child has limited depth perception so negotiating unfamiliar places cause anxiety. In addition, child can kick a ball and can catch, at times, with arms outstretched, but often misses. Child is working on getting into the family SUV independently but, because of her size, it is challenging.

In fine motor skills, turn pages of a picture book (frequently upside down), assemble chunky puzzles, open doors and can grasp and pick up small objects. Child also plays with chalk in the family's driveway.

In language development, child primarily uses American Sign Language to communicate. Frequently used signs include *Daddy, Mommy, brother, dog, juice, cracker, craisin, fish (for goldfish cracker), pump (to indicate hungry for a feeding), music, bike (for trike), swing, outside, sleep/bed, pants* and *shoes*. She is also learning to use an iPad with a communication app. Child follows one to two step instructions, demonstrates understanding of familiar stories, and points to requested objects.

In cognitive skills, child is working on identifying colors. Child also knows where items are in the house. When told to retrieve an item, child will successfully locate it. When child wants a feeding, child will bring her pump to a parent.

In daily living skills, child assists with dressing such as putting on pants, zipping and unzipping clothing items, and is currently working on putting on orthotics and shoes. When eating solid foods, child is working on using a spoon and fork. Child still wears a diaper during the day and night.

In social-emotional milestones, child displays a variety of emotions including happy, sad and upset as well as interacts with peers including older and younger neighbor children in age

appropriate activities such as kicking a ball. The latter has increased in the past year. Child has also started to take the initiative to do things she wants to do even if it is not allowed such as going outside to play without an adult.

Child tolerates long distance family road trips and had first international road trip to Canada (Quebec City) but didn't like Niagara Falls' Maid of the Mist experience (noisy and too much water). Child had a similar experience with air boats in Florida but enjoyed the ocean waves. Family hopes to take more international trips, possibly to other continents in the coming years.

Education and therapy services

Child received early intervention services up to 36 months of age including nursing and nutrition services. In addition, audiology and vision services were provided. Occupational, physical and therapy services were provided in-home. Developmental therapy was also provided. A developmental pediatrician was also involved in child's care.

After the age of three, child received services in both a general education and special education classroom in a public school.

At baseline survey completion, child was receiving 45 minutes of occupational therapy weekly at a clinic. Physical and speech therapy were each provided for 90 minutes per week also in a clinic setting. Child has used a gait trainer as well. Child's progression was sitting to using a stander for a brief time then a gait trainer, which was too heavy for child to successfully use, to a lighter weight gait trainer, which she could maneuver successfully, and, finally, walking independently.

At present, child attends school full time. On average, child will spend approximately 2.5 hours daily in the integrated third grade classroom 5 days a week for "specials" such as art, physical education, and reading activities with peers and balance of the time in a special needs classroom or therapies provided by the school district. Child continues to receive physical, occupational and speech therapy services at school and home. Child continues with a one-on-one aid during the school day. For lunch time, the school has an onsite nurse's aide to assist with g-tube feedings. When a nurse is not present, an RN is available on call. Finally, child attends Summer School for three weeks ½ days four days a week in July in a special needs classroom.

Child receives physical therapy twice a week both private and at school to address cervical kyphosis and work on balance/steadiness when walking. Child also receives

occupational therapy once a week both private and at school as well as speech and language therapy once a week both private and at school. Child is learning to work with an augmentative communication device to increase communication.

Child is involved in a local special needs T-ball league. Parents are also looking into hippotherapy.