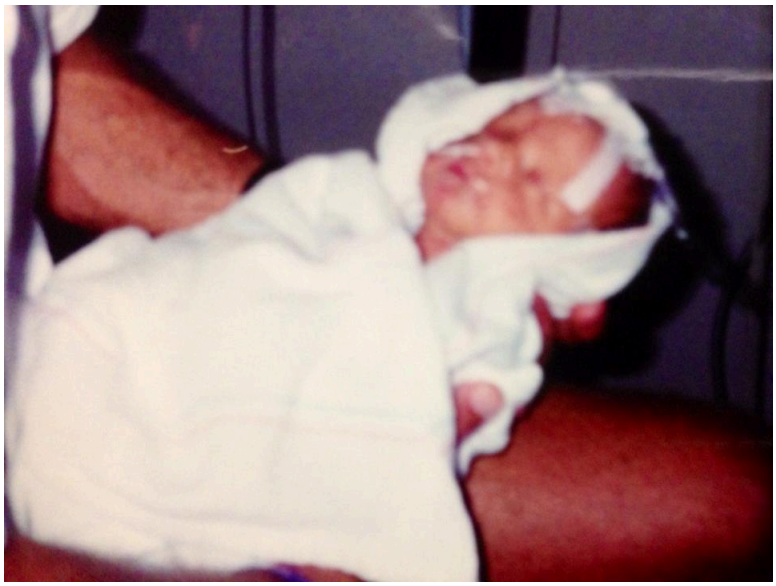

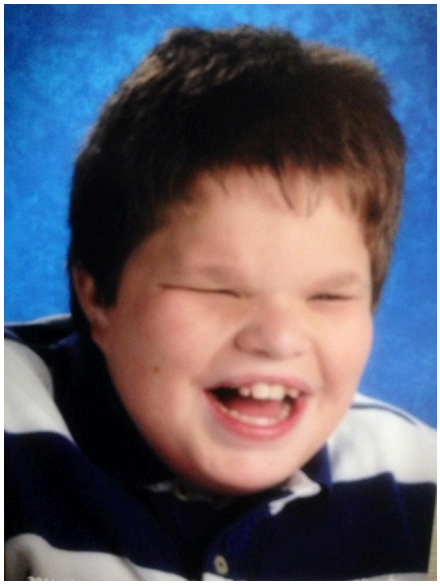

Samuel was born in 1998 and was diagnosed seven weeks after birth with mosaic trisomy 13. He lives in the United States. Participation in the TRIS project began in 2008 at the age of 158 months.

Mother was 34 and father was 36 years old at time of conception. Mother reported general knowledge of rare trisomy conditions due to training as a nurse. Mother also notes experiences preeclampsia, toxemia and gestational hypertension during pregnancy.

Child with rare trisomy was the third of three pregnancies (gravida, 3; para, 3). No siblings have been diagnosed with a genetic, terminal, medical, or neurological condition.

Birth Information

Male infant was delivered via emergency caesarian section at 34 weeks gestation. Infant was born weighing 2238 grams and measured 46 centimeters in length in November 1998. No APGAR scores were provided.

At birth, infant presented with low-set ears, small jaw, several hemangiomas and small genitatlia. Infant also presented with the following medical issues: ventricular septal defect (VSD), feeding difficulties, jaundice and hypoglycemia. Infant was diagnosed with mosaic trisomy 13 mosaic seven weeks after birth.

Infant received the majority of his post-birth care in a specialized hospital's neonatal intensive care unit (NICU). Infant was on the unit for 10 days. A heart monitor and pulse oxymeter were used prior to discharge. A Haberman feeder was also necessary for feeding. Discharge weight was 1871 grams.

Mother notes that an NICU nurse told her genetic testing was ordered. Medical staff did not inform parents of suspicion of a genetic disorder. Discharge occurred before the results were received. Infant's pediatrician referred parents to a geneticist. Infant was admitted at six weeks of age to a regional children's hospital. Results came back a week later for mosaic trisomy 13.

Surgeries

Child underwent gastrostomy tube placement and Nissen fundoplication at 22 months of age. A revision was necessary (noted in Year 1 Follow-up survey). In addition, adenoids and tonsils were removed when child was 34 months old. Child also underwent an Achilles tendon lengthening procedure at 47 months of age. A bilateral femoral osteotomy with left acetabulum shelf procedure was performed between 170 and 182 months as indicated in Year 2 TRIS Follow-up Survey.

A bilateral inguinal hernia repair was performed at 34 months and an ureteroneocystostomy at 43 months.

Cardiac

No cardiac defects are noted.

Respiratory

When child was younger, the use of supplemental oxygen, suctioning and a nebulizer were necessary. Child was also diagnosed with obstructive apnea. Adenoids and tonsils were removed when child was 34 months old.

At the time of completion of baseline survey and follow up surveys, child required suctioning and nebulizer treatments to treat colds, flu and similar conditions. Antibiotics are prescribed as needed. In addition, Biaxin, nebulized steroids, and oral Prednisone are used. Zyrtec is taken for allergies as well as monthly injections.

Neurological

Child experienced infantile spasms for an eight-month period between the ages of two to 10 months. Child's primary physician monitored the condition. Tonic-clonic seizures were noted at baseline survey. Depakote and Trileptal were prescribed. Trileptal was and continues to be effective in controlling seizure activity. No side effects were noted.

The left side of child's brain is smaller and includes a damaged area. It is believed the damaged area was present at birth. Child's neurologist explained that as the neuropathways developed and reached that part of child's brain, seizure activity began at seven years of age. An electroencephalography (EEG) showed continuous firing from the left side of the brain. An MRI showed the damaged portion on the left side. A neurologist is part of child's care team. Visits were yearly until age 14. Now, appointments are two years apart (next appointment will be spring 2015).

Renal

Constipation has been noted as well as urinary tract infections (UTIs). Septra was effective. The medication was taken between the ages of two months and five years. No side effects were noted. For the past two years, child has taken Septra and Macrodantin daily to prevent UTIs. One medication controls gram positive bacteria and the other gram negative bacteria. Child has not had a UTI since beginning the combination.

Child was diagnosed with hydronephrosis (swelling of a kidney due to a build-up of urine) at six weeks of age (same time as genetic testing). The condition was determined to be congenital. A ureteroneocystostomy was performed at 43 months to repair the blockage causing the hydronephrosis. Child is monitored yearly by abdominal ultrasound. Cysts are also noted to be present on one kidney. This condition has been monitored since child was approximately 11 years of age.

Child underwent bilateral inguinal hernia repair at 34 months. In addition, a urologist is involved in child's care.

Gastrointestinal

Between the ages of two weeks and 22 months, child experienced stomach discomfort, excessive gas and constipation. Gastroesophageal reflux (GER) was also diagnosed. Prevacid, Zantac and Reglan were prescribed. Initially, prior to nissen fundoplication, Reglan was most effective at treating GER. At time of baseline completion, child was taking Pepcid, which was noted to be effective in reducing acid in child's stomach and preventing discomfort. Miralax is taken to prevent constipation.

Child underwent gastrostomy tube placement and Nissen fundoplication at 22 months of age.

Nutrition

Immediately after birth, infant was fed with a Haberman feeder. Infant also received feedings via a nasogastric tube (n-g tube) until six months of age. Infant was able to accept liquids from a regular nipple at this time. Cereal was introduced at nine months. Stage 1 and 2 baby food was also introduced. Child had a small bowel follow through study at the age of 20 months and was found to be aspirating. Mother noted bottle feeding was allowed to continue until gastrostomy tube (g-tube) placement at 22 months of age. A g-tube revision was performed between 170-182 months of age.

At time of baseline survey (158 months), child received g-tube feedings and supplemental table foods and liquids. At Year 1 Follow-up at 170 months and Year 2 at 182 months, child continued to use a cup for drinking and ate table foods. G-tube is used to deliver medications and when child is too ill to accept fluids by mouth. His primary nutrition is Ensure. Child is offered soft table foods but only accepts pudding and ice cream.

Oral health

Child began dental visits at the age of four years with a dentist specializing in working with children for routine care. Child requires sedation for specialized care such as filling cavities. Child was noted to have an extra baby tooth, which was replaced by an adult tooth. Child's dentist extracted the extra adult tooth.

No additional dental issues have been identified.

Vision

Child was diagnosed with coloboma (hole in an eye structure such as the iris, lens, retina, or optic nerve) in the right eye at four weeks of age. Child has very limited vision in that eye. An ophthalmologist monitors the condition.

Child was diagnosed with hyperopia (farsightedness) at the age of two years and was prescribed glasses but has never tolerated wearing corrective lenses or sunglasses. Child will continually pull glasses off his face. No additional vision issues have been diagnosed.

Auditory

No documented audiology issues.

Immunizations

Child's immunizations are up-to-date. Immunizations began at four months 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, and Varicella. No adverse reactions were noted.

Orthopedic

Child underwent an Achilles tendon lengthening procedure at 47 months of age. At Year 2 follow-up at 182 months, a bilateral femoral osteotomy with left acetabulum shelf procedure was noted.

No spinal malformations or defects have been identified. Child has used hand splints and orthotics. Specifically, child used a wrist splint with thumb support from the age of six months to six years.

Child underwent a right calcaneal osteotomy (controlled break of the heel bone to correct deformity of the foot and ankle) at 11 years of age. Child is also diagnosed with kyphosis. At present, it is a 65-degree curve. No treatment is recommended until the curve reaches 80 degrees. An orthopedist follows the child's case.

Developmental milestones

At completion of baseline survey at 158 months of age, child demonstrated a range of skills at the 24-36 month level including following two step instructions, expressing "no" to parent and caregivers, attempting to throw or kick a ball, and using the toilet when taken during daytime hours. In addition, child was also able to make preferences known vocalizing and gesturing. The abilities to walk independently for short distances and climb stairs with assistance were also noted. In addition, child assisted with dressing activities.

Currently, mother reports child is able to open the refrigerator and find his cup (and sometimes closes the refrigerator door). He has learned to take the lid off the cup and drink without spilling. Child can also use the remote control to turn channels on his

television. It is noted that child laughs at appropriate times. In social-emotional development, child will give hugs when asked.

At school, child is noted to select the correct color bean bag when given two choices. His mother also describes child's ability to recognize locations. For example, child knows the route to the park, school, church and physical therapy. Child also relearned how to walk after bilateral hip surgery (no weight bearing for three months post-surgery).

Child requires a sleep aid (Melatonin, 10 mg nightly). This medication has been the most effective of various sleep aids.

Education and therapy services

Child received early intervention services up to 36 months of age including developmental therapy and audiology services. In addition, occupational, physical and speech therapy services were provided.

After the age of three, child has received services included in a general education as well as in a special education classroom in a local public school. For kindergarten through second grade, child was in a general education classroom for 30 minutes daily and participated in Art, Music, Physical Education and Guidance classes with peers. After second grade, child only attended the related classes with an aide.

Beginning in seventh grade, child was placed in a self-contained classroom at the local high school. Child remains in that classroom and will continue until the age of 22. Peers come to the classroom as peer mentors. In addition, child attends school assemblies, pep rallies and football games.

At baseline survey completion, child was receiving 30 minutes of occupational, physical and speech therapy weekly in the school setting. Aquatic therapy was also provided to assist with stretching and balance. Child has qualified for 10 hours of respite services per month since early childhood.

At Year 1 and 2 Follow-up, child continued to receive occupational, physical and speech therapy. Occupational and speech therapy are 30 minutes a week. Physical therapy includes aquatics for a total of 60 minutes per week. Respite care is currently provided 10 hours a month. Child receives services in a special education classroom.