



Kayden was born in 2001 without a prenatal diagnosis.

He was diagnosed with an unbalanced translocation from the 18th to the 13th chromosome (t18q;13). He lives in the United States. Participation in the TRIS project began in 2008.

Mother was 24 and father was 27 years old at time of conception. Trisomy condition was not suspected prenatally. Child with rare trisomy was second of four pregnancies (gravida, 5; para, 3 – one molar pregnancy and currently pregnant). No siblings have been diagnosed with a genetic, terminal, medical, or neurological condition.

Birth Information

Condition was not diagnosed prenatally but at 30 weeks, it was noted that the fetus exhibited cardiac arrhythmia. Mother also experienced polyhydramnios during her pregnancy. Male was delivered vaginally on June 19, 2001 after 45 minutes of labor at 40 weeks gestation weighing 2126 grams and 44.5 centimeters in length.

Infant was tested at three days of age. Diagnosis of t18q;13 was confirmed at the age of two weeks. At birth, the infant presented with a small jaw, and cleft palate. Infant presented with the following medical issues: respiratory difficulties, heart murmur, ventricular septal defect (VSD), feeding difficulties, jaundice, abdominal hernia, and bowel and intestinal malrotation.

Post-birth care was received at a neonatal intensive care unit (NICU) in a specialized hospital. Infant was intubated soon after birth. Other respiratory equipment used was an apnea monitor, tracheostomy with a ventilator, and a pulse oximeter. Tracheostomy was placed at three days of age. Feeding methods included Haberman feeder, nasogastric tube and via gastrostomy tube. Gastrostomy tube placement occurred at two weeks of age.

An audiology examination, echocardiogram, intubation, ultrasound, and x-ray series were performed while in the NICU. Infant was discharged after 9 to 12 weeks on the NICU. Discharge weight was 2268 grams.

Surgeries

Initial surgery was at three days of age to place a tracheostomy. At two weeks, infant underwent surgery for malrotation of bowel repair and gastrostomy tube (g-tube) placement. At two months of age, abdominal hernia repair was performed. Jaw distraction was performed at six months.

There were two surgeries to repair child's cleft palate: first was at 10 months old and second shortly after child's second birthday. Child underwent an adenoidectomy, and uvulectomy when he was two and a half years old. Child has undergone to ear surgeries for cholesteatoma repair: first at 72 months and second at 149 months, which also included repair of the ear canal. Both surgeries were for the left ear.

Child also received Botox injections for his jaw and hip abductors at approximately six years of age. Injections were every three months for one year. Injections were discontinued due to seizure activity, which caused child to stop breathing.

Cardiac

Child was diagnosed at birth with a heart murmur and ventricular septal defect (VSD). The VSD did not require a surgical repair. Child was also diagnosed with upper pulmonary stenosis at one year of age. The condition resolved without intervention around the child's sixth birthday.

Respiratory

At the age of three days, a tracheostomy was placed. It was removed when the child was 21 months old. In the past, the child has used a pulse oximeter, supplemental oxygen as needed, bi-level positive airway pressure (Bi-PAP) machine, ventilator, suction machine, and a nebulizer for respiratory support. The child was diagnosed with obstructive apnea at 30 months of age and an adenoidectomy, and uvulectomy were performed. Child uses a nebulizer when necessary which is approximately once a year due to illness. Albuterol and Pulmicort are used via the nebulizer to maintain an open airway.

Currently, the child continues to use a pulse oximeter to monitor saturation level, supplemental oxygen as needed, suction machine, and a nebulizer for respiratory support. Child has not needed supplemental oxygen since approximately six years of age.

Neurological

At 72 months, child was suspected to have seizures associated with stoppage of breathing. Keppra was prescribed and he was bagged as needed during these periods. Medication was effective with no reported side effects. At 84 months, child was diagnosed with seizures. Currently, they are continuing to be controlled with medication.

Renal

Child has not been diagnosed with any renal disorders.

Gastrointestinal

Infant was diagnosed soon after birth with reflux and prescribed Prevacid. Child experienced long periods of crying and fussiness due to stomach discomfort/excessive gas and constipation. Miralax and enemas were also prescribed. Child still experienced stomach discomfort/excessive gas. Miralax is prescribed for constipation but was not reported to be effective. As a result, Miralax was discontinued soon after it was prescribed.

Nutrition

At two weeks of age, a gastrostomy tube (g-tube) was placed. This continues to be the primary method for feeding.

Orthopedic

Beginning at 12-18 months of age, child used a manual wheelchair, supine stander, gait trainer, and orthopedic braces. Child was diagnosed with minor scoliosis at six years of age. Condition remains but does not require use of a brace or surgical intervention.

TRIS Follow-up Survey data indicates that at 117 months child used a supine stander, a manual wheelchair, and a specialized bed (SleepSafe™). At 131 months, the child continued to use a supine stander, a manual wheelchair, a specialized bed, and a crawl trainer (Creepster Crawler™).

Oral health

Cleft palate was repaired via two surgeries: first was at 10 months old and second shortly after child's second birthday. Child receives routine dental care from a dentist specializing in working with children. Until several years ago, child required sedation and antibiotics before

routine dental care. Child no longer needs this intervention. At 117 months, the child was treated for excessive plaque with additional cleanings.

Vision

At five years of age, child was diagnosed with astigmatism.

Auditory

At 72 months, a tumor was removed from child's left ear. Within the past 12 months, a second surgery was performed to remove a second tumor from the left ear and repair the canal.

Child currently receives treatments for wax buildup and/or blockage for the ear(s). It is also noted that the child has small ear canals. Otitis media has been experienced multiple times. Each occurrence has been resolved with antibiotics.

Immunizations

Immunizations were started late due to child's low birth weight. By 84 months, child had received the following immunizations: Diphtheria, Tetanus and Pertussis (DTP), Haemophilus influenzae type B (HIB), Hepatitis A (HepA), Hepatitis B series, Inactive polio virus, Measles, Mumps and Rubella (MMR), Pneumococcal pneumonia, Respiratory syncytial virus (RSV), Varicella ("Chicken pox"). No adverse reactions were reported for any vaccination.

Current Education and Therapy Services

Child receives 60 minutes of occupational therapy, physical therapy, and speech and language therapy in the home each week. Child has consistently received four hours a week homebound school services and three hours out bound services. Assistive technology services are received at a clinic. Audiology services are provided at a center. He receives cranial sacral therapy as well.

Developmental milestones

Presently, child can sign "mom" and "dad" and will say "momma". He shakes his head for "no" and also will push things away to indicate he is done. He enjoys pictures and will use pictures to indicate what he wants. For example, if he has two pictures of preferred toys, he will point to the one he wants. Mother is also working on object recognition using pictures (e.g., given picture of a kitten and a puppy, asked to point to the kitty, child points to the kitty). Mother notes that loud music calms him.

Child produces a variety of facial expressions to communicate including happy, sad, mad and uncomfortable. Mother explains that he pretend cries to gain attention. When an adult turns

toward him while he is “crying”, he laughs. He enjoys playing peek-a-boo. He will look at an adult and quickly turn away. He, then, begins to turn back. When he sees the individual is still looking at him, he turns away again.

Child uses both hands to activate switches. He will also reach and grasp items. He is able to place items into containers such as a jar when asked. He also shakes or smacks hand-held items. Child activates cause and effect toys such as pushing buttons for lights or noise. Child also enjoys participating in craft projects. Child is given hand over hand assistance for these activities. If an item is out of reach, he will roll or scoot on his bottom to reach it. He can transfer from the sofa to the ground independently. He can also transfer from supine (on his back) to a sitting position.

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>