



Luka was born in 2009 and was prenatally diagnosed (partial trisomy 9). Child lives in the United States. Participation in the TRIS project began in 2009 at the age of 2 months.

Mother was 34 and father was 39 years old at the time of conception. Trisomy condition was suspected prenatally. Mother reported irregular findings at 22-week ultrasound with a follow-up with a specialist including bulbous nose, splayed cerebellum, enlarged kidney, and enlarged ventricles (not present in later ultrasounds). An amniocentesis was performed as well as a more extensive genetic testing (chromosome microarray analysis with fluorescence in situ hybridization) which found partial trisomy of chromosome 9 from bands p22-p24. Child with rare trisomy was second of three pregnancies (gravida, 3; para, 2). 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 diagnosed with partial trisomy 9 at 26 weeks gestation.

Infant was born weighing 1673 grams and 43 centimeters on February 2, 2009. APGAR scores were 2 at one minute and 3 at five minutes. Prior to birth, infant was identified with hydronephrosis and Dandy-Walker malformation. A computerized tomography (CT) scan and magnetic resonance imaging (MRI) after birth ruled out the latter condition. At birth, infant presented with low-set ears, small jaw, feeding difficulties, jaundice, and hydronephrosis.

Infant was cared for in the neonatal intensive care unit (NICU) at his birth hospital. He remained on unit for eight weeks. The extended NICU stay was due to recurring bradycardias and aspiration during most feedings. Several swallow studies were completed including videofluoroscopies. Infant also received nasogastric tube (n-g tube) feedings. These procedures were completed in order to establish a treatment plan for discharge, which was dependent on passing a final swallow study.

An abdominal ultrasound for hydronephrosis was also conducted. Infant was found to have an enlarged left kidney. The condition was treated with antibiotics to reduce infection was thought to be related to infant's prematurity. Ultrasounds and antibiotics continued until the condition resolved at 12 months of age.

Infant was discharged weighing 2948 grams. Infant was also sent home with a heart monitor and pulse oximeter to monitor for bradycardia episodes while feeding. Mother reports infant did not experience any episodes after discharge from the NICU.

Cardiac

Infant was prescribed a home heart monitor and pulse oximeter that was used for two weeks after discharge from the NICU. Infant was not diagnosed with any heart defects.

Respiratory

Child has not experienced any chronic or acute respiratory issues.

Neurological

Child has not experienced infantile spasms or seizures.

Renal

Infant was diagnosed with hydronephrosis prior to birth at 30 weeks gestation. Infant was scheduled to receive abdominal ultrasounds every two months to monitor this condition. Omoxicillin was used starting at birth to treat this condition. Hydronephrosis was resolved at approximately 12 months of age. No additional renal conditions have occurred.

An attempt was made to descend the child's testicles at 15 months. This was successful. Doctor noted that one testicle is quite small and probably not functional. Hormone levels were found to be in normal range. Currently, mother reports that one testicle has retracted. This is being monitored annually to determine if an additional surgery is needed.

Gastrointestinal

Infant was prescribed Prevacid for reflux while on the NICU. Infant continued on Prevacid until approximately 10 months of age when reflux issues resolved.

Child has not experienced any acute or chronic digestive issues since discharge.

Nutrition

Infant was bottle fed breast milk when discharged from NICU. At 16 months, infant accepted all food and liquids orally. At 23 months, child was able to self-feed with hands. At 43 months, child was able to self-feed with a spoon and uses a sippy cup. At present, child eats all types of table foods and continues to use a sippy cup.

Oral health

No dental issues have been reported.

Vision

No vision issues have been reported.

Auditory

At 23 months, child experienced an ear canal blockage that required ear tubes. Treatment was successful.

Immunizations

Child's immunizations began at six months of age and are currently up to date. Child did not experience any adverse reactions.

Orthopedic

Child required hand splints from 12 to 15 months along with hand massage to keep palms open. Currently, child wears supra malleolar orthosis (SMO). Use of SMOs began at approximately 20 months of age. Mother reports the orthoses are being phased out at the present time due to child's progress and recommendation of physical therapist.

Developmental milestones

Infant was two months old at completion of baseline survey resulting in few developmental milestones.

Mother reports child crawled at 12 months and began walking at 24 months. Child's speech became recognizable at approximately three years of age and continues to work on articulation and phonology. Child could manipulate blocks, Legos and similar objects. Fine motor strengths also include using appropriate grasp on crayons when child was younger and writing his name, with some prompting, at the present time. Mother also notes child has always been social such as playing nursery games and interacting with peers. Child is initially shy but warms up quickly and engages in social activities.

Child has some difficulties with dressing and other activities of daily living but mother reports he can drink from a water fountain at school as well as use a straw. At home, he demonstrates difficulty with similar tasks. Mother notes this may be intentional as child is the youngest in the family. Child is also in the process of mastering bladder and bowel control.

Currently, child can say most letters of the alphabet as well as can write and spell his first name. Child can answer "wh" questions (who, what, where, when) using factual information in 3-5 word phrases (may require verbal prompts).

Education and Therapy Services

Beginning at two months of age, infant received early intervention services including developmental therapy, occupational therapy, physical therapy, and speech therapy in the home once a week for an hour. At 16 months child was receiving 60 minutes each of physical, speech, and occupational therapy in the home per week. He was also receiving feeding therapy and attending a music class. Child began receiving 60 minutes of developmental and speech therapy once per week in a center-based facility at mother's workplace at 18 months. Mother was able to attend and assist during therapy sessions. When facility was closed, services were provided at the local EI clinic. Child continued to attend a music class and began an Aqua Babies class.

Feeding therapy was provided during the first three years but was intermittent for up to three months at a time. Infant was also receiving vision services at a vision center.

At 43 months, child was receiving 60 minutes of occupational, physical, and speech therapy once per week at a center. He also attended a social therapy play group and physical

therapy boot camp. Between 36-40 months, child was receiving educational services in an early childhood special education classroom in a half-day program. When child returned to school at 43 months in August, he received services in a blended, inclusive half-day classroom. This placement continued until 51 months of age.

At 53 months, child switched schools and was again placed in a half-day inclusive, blended classroom led by a special education teacher. Child also received pull out occupational, physical and speech therapy services for 30-60 minutes a week for each area. Mother also notes private services for physical, occupational and speech therapy (60 minutes each per week). At 56 months, child was again receiving 60 minutes of feeding therapy once per week at a center. He also continued to attend social therapy play group.

Currently, at 66 months of age, child will attend a full day kindergarten class this coming school year. Child will continue to receive physical, occupational and speech therapy in the school setting along with private occupational and speech therapy. Child also began attending a dance class in June 2014.

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>