



Alana was born in 2009 and was diagnosed at 20 weeks gestation with mosaic trisomy 9. She lives in the United States. Participation in the TRIS project began in 2010 at the age of two months.

Mother was 38 and father was 38 years old at time of conception. Mother reported general knowledge of rare trisomy conditions due to a friend with a child with a genetic condition. Mother noted low amniotic fluid starting at 36 weeks gestation.

Child with rare trisomy was the third of three pregnancies (gravida, 3; para, 2). Sibling has not been diagnosed with a genetic, terminal, medical, or neurological condition.

### **Birth Information**

Female infant was delivered via emergency caesarian section at 37 weeks gestation. Infant was born weighing 2466 grams and measured 48 centimeters in length in December 2009. APGAR score was 9 at one minute.

At birth, infant presented with low-set ears and slightly recessed jaw. Infant also presented with the following medical issues: atrial septal defect (ASD) and patent ductus arteriosus (PDA).

Infant spent 12 hours in the birth hospital's neonatal intensive care unit (NICU) and was then transferred to the regular nursery. An audiology examination, echocardiogram and ultrasound were performed. Infant was able to take fluids from a bottle. Discharge weight was 2410 grams.

### **Surgeries**

Child has not require any surgeries.

### **Cardiac**

Infant presented with ASD and PDA. Both resolved by six-month cardiology check-up. Infant was discharged from cardiology at that time. Mother also noted a

structural abnormality with a major artery leading into the heart as located behind trachea. This condition does not and has not adversely affected child's functioning.

### **Respiratory**

Infant was hospitalized at four months of age due to suspected Respiratory syncytial virus (RSV). Infant was inpatient for four days with only oxygen support. More recently, general respiratory difficulties such as colds and strep throat and airborne allergies (grass and pollen) were noted in Year 2 Follow-up Survey. Treatment was with antibiotics and over the counter medication, respectively.

Child was noted to develop an allergy to amoxicillin at one year. Treatment consists of avoidance of the medication. Allergic reaction was hives all over the body. An allergist was seen in August 2013 for a complete allergy evaluation. None were found.

### **Neurological**

No neurological issues noted.

### **Renal**

At Year 1 Follow-up, a kidney issue was noted. Child was diagnosed with double ureter tubes leading from left kidney to bladder. This condition did not impact kidney or bladder functioning or require treatment. Child was recently discharged from urology follow-up. Mother noted that urologist told her to share this information if child ever requires surgery.

### **Gastrointestinal**

Stomach discomfort and excessive gas was noted at the time of the Year 3 Follow-up Survey. No treatment was noted. Year 1 and 3 Follow-up indicated constipation issues. Treatment included increased fruits and juices when child was younger and Miralax in the past year. No changes in condition but will be discussed at upcoming visit with gastroenterologist. Child also has ongoing constipation issues.

### **Nutrition**

Child used a bottle after discharge from hospital post-birth. On Follow-up surveys, beginning at age two, child was noted to use a cup, spoon and fork as well as accept solid foods.

### **Oral health**

Initial dental visit was at approximately 30 months of age. Check ups were initially twice a year. The need for additional dental cleanings due to tooth decay was noted in Year 3. In addition, two teeth were pulled in August 2014 (one for decay and one due to an unresolved trauma that loosened the tooth).

### **Vision**

Child began wearing glasses at 24 months of age to treat hyperopia (farsightedness). Risk for strabismus was noted in Year 2 Follow-up Survey but appears wearing glasses has addressed this. An ophthalmologist is involved in the child's care.

Mother notes that child will need to wear glasses throughout her life due to asymmetry of eyes (one eye is smaller and weaker than the other).

### **Auditory**

Otitis media is reported in Years 1 and 2 Follow-up Surveys. Treatment with antibiotics was noted to be successful but amoxicillin caused a body rash during time period covered in Year 1 Follow-up Survey. No tubes have been needed. Child's primary physician monitors this condition.

### **Immunizations**

At baseline survey at two months, infant's immunizations were up to date. Child received immunizations at the recommended times with no adverse reactions. Child also receives an annual flu shot and experiences no side effects.

### **Orthopedic**

Beginning with Year 1 Follow-up Survey at 21 months of age, child has used orthopedic inserts in shoes to assist walking. The first insert was a dynamic ankle foot orthotic (DAFO). An orthopedist follows the child's case.

A sedated magnetic resonance imaging (MRI) procedure was performed in August 2014 to check degree of scoliosis and presence of tethered spinal cord. Scoliosis is noted to be 16 degrees and no tethered spinal cord. Child was referred to an orthopedic surgeon to follow scoliosis. Surgery is a strong possibility if the curve reaches 50 degrees.

### **Developmental milestones**

Child was two months of age at completion of baseline survey. Skills included following moving objects with eyes, brings one or both hands to mouth, raises head and chest when lying on stomach and molds when held.

Child began to use sign language at approximately 12 months of age, walked independently at 18 months and spoke intelligible words at three years of age. At present, child knows the alphabet and counts to 20. At about 8 months sat without support. Has been able to scribble with markers since about 2, can now draw lines and circles, working on writing letters. She can undress as long as there are no buttons, just started to attach and pull zippers herself. Has been able to put mittens on since about 4. She interacts with peers to some degree, working on requesting peers to play what she wants to play and turn taking.

Infant's initial visit to a developmental pediatrician occurred at two months of age. Follow-up visits continued twice a year until age four. Visits are now once a year for treatment suggestions. Mother notes the developmental pediatrician recommended the special school child currently attends.

### **Education and therapy services**

Child began receiving early intervention (EI) services by two months of age. Initially, only physical therapy was provided. At each follow-up survey (21, 43 and 57 months), child received occupational therapy, physical therapy and speech and language therapy. At Year 2 Follow-up, a behavioral therapist provided therapy for 130 minutes per week. Remaining therapies ranged from 30-120 minutes a week. A special instructor was also part of EI services.

After the age of two, child began receiving services three days per week in a preschool setting with children with and without disabilities. At 42 months of age, child transferred to a full time placement at a Catholic special needs school's preschool program. Child continues to receive services at the Catholic special needs school. A special educator teacher provides services along with an occupational therapist, and a physical therapist. Teacher is also a speech therapist.

Child attended an infant-toddler music class. Child has also participated in recreational activities including Park District activities and attending sibling's school and sports events. Child has been involved in adaptive sports. Most recently, child has been involved with Variety Club Cheerleading, Challenger baseball and a summer camp program operated by child's school program. Child has also travelled with family to multiple cities, and loves amusement rides.

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>

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