



Gabby was born in 2002 and was not prenatally diagnosed (karyotype revealed isochromosome 14; trisomy 14 mosaic was diagnosed several days after birth). She lives in the United States. Participation in the TRIS project began in 2008.

Mother was 25 and father was 25 years old at the time of conception. Child with a rare trisomy condition was second of three pregnancies (gravida, 3; para, 3). No siblings have been diagnosed with a genetic, terminal medical or neurological condition. Neither parent was aware of rare trisomy conditions prior to their child's diagnosis.

### **Birth Information**

Trisomy diagnosis was not suspected during pregnancy but infant was identified with a ventricular septal defect (VSD) and cleft lip prior to birth. Mother had preterm labor at 29 weeks gestation and was hospitalized. Mother elected to have a spontaneous vaginal birth at 33 weeks.

Female infant was born weighing 1865 grams and 17.75 centimeters after approximately 12 hours of labor. APGAR scores at 1, 5, and 10 minutes were 8, 8, and 9 respectively.

Infant presented with low-set ears, cleft lip, and arm and a leg abnormality (unequal leg length). Infant was also diagnosed with a VSD and jaundice soon after birth. Child's karyotype revealed isochromosome 14 (break on long q leading to duplication on chromosome 14). Infant's diagnosis was trisomy 14 mosaic which was also made several days after birth.

During the time spent in a neonatal intensive care unit (NICU) at a specialized hospital, a nasal cannula, apnea monitor, ventilator, heart monitor, pulse oximeter, and nasogastric tube were provided. An audiology exam, blood transfusion, echocardiogram, ophthalmology exam,

ultrasound of abdomen, and x-ray series were performed. Infant was released after three and a half weeks in the NICU at 2380 grams.

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

### **Surgeries**

A full VSD repair was completed at 18 months of age. A tympanostomy was performed to place tubes in both ears around 36 months of age and at 60 months. In 2009, an adenoidectomy and tonsillectomy were performed to address apnea. At 126 months, child had a mastoidectomy (surgery to remove cells in the hollow, air-filled spaces in the skull behind the ear).

### **Cardiac**

Infant was diagnosed with a VSD prior to birth. Child was prescribed Aldosterone until a full VSD repair was completed at 18 months of age.

### **Respiratory**

Child was diagnosed with obstructive apnea. In 2009, an adenoidectomy and tonsillectomy were performed during the same surgery. Child no longer experiences apnea episodes.

At 87 months, child used a nebulizer for administration of Pulmacort and Albuterol for asthma. At 100 months, the child continued to use a nebulizer for the administration of Pulmacort for the prevention of asthma symptoms. TRIS Follow-up Survey data indicates that at 100, 126, and 138 months, child was reported to experience airborne allergies to pet dander and dust mites and was prescribed Singulair and Clairtin. These medications successfully treated the allergies. Child continues to take both daily.

### **Renal**

Child has not been diagnosed with any renal conditions but has an ultrasound every six months for monitoring purposes.

### **Gastrointestinal**

Child was prescribed Lactulose for constipation for approximately two years after 36 months of age. Miralax has also been used. At the present time, child is not on any medication for this condition. No other gastrointestinal problems have been reported.

### **Nutrition**

Infant was bottle-fed breast milk after discharge. Child's sucking ability was weak so she was fed through an enlarged nipple to assist with swallowing and breathing. Child has eaten pureed

food since she was weaned. At 87 months, child still preferred purred foods due to aversion to textured foods. At 126 months, child was reported to eat mostly table food. At 138 months, child was reported to eat all table food.

### **Orthopedic**

Prior to 87 months, child used gait trainer/ walker to assist with mobility. Child was diagnosed at birth with one leg shorter than the other. A shoe lift has been used to equalize length. Surgery was performed at the beginning of April 2014. Child is recovering at this time.

Child has also been diagnosed with scoliosis (55% currently). Child used a brace from 2010-2013. Surgery is scheduled for summer 2014.

### **Neurological**

Child has not experienced seizure activity or been diagnosed with any brain anomaly.

### **Oral health**

Cleft lip was successfully repaired at 4 months. Child started seeing a dentist at 24 months and was diagnosed with too few teeth. Routine dental care is provided annually.

### **Vision**

Child was prescribed glasses for amblyopia at 72 months. Follow-up visits with an ophthalmologist are scheduled every six months.

### **Auditory**

Child was born with a malformed left ear from which she cannot hear. At 138 months, child was identified with profound hearing loss of the left ear.

Child has experienced otitis media since 24 months and wax build up since 24 months. An ENT monitors both conditions. A tympanostomy was performed to place tubes in both ears around 36 months of age and at 60 months. To present, a total of five sets of ear tubes have been placed. Child was also diagnosed with Meniere's disease and tinnitus at 24 months.

At 126 months, child had a mastoidectomy (surgery to remove cells in the hollow, air-filled spaces in the skull behind the ear). This procedure was found to be ineffective to treat conductive hearing loss.

### **Immunizations**

Child immunizations are up to date and were started at the recommended ages. No adverse reactions were reported.

### **Current Education and Therapy Services**

At 87 months, child attends her neighborhood school and participated in a life skills curriculum that included community-based instruction. She also received speech/language therapy at school.

At 100 months, the child was additionally diagnosed with Attention Deficit Hyperactivity Disorder (ADHD) after referral by the child's school. The child receives physical therapy, occupational therapy, speech and language therapy, audiology, and vision services at school. The child's time in school is shared between a general education and special education classroom. The child participates in tee ball through her local park district and in Special Olympics. Child competes in swimming, bowling, track and tee ball.

At 126 and 138 months, child was reported to be homeschooled. At the present time, she continues to be homeschooled.

### **Developmental milestones**

Child started crawling at 15 months. Child started pulling self onto furniture and taking steps with assistance at 24 months. Child was potty trained at 36 months. Child was able to engage in pretend play and play cooperatively with peers at 40 months. Child would sing nursery rhymes at 50 months, speech was difficult but was able to carry tune. Child could say alphabet and numbers 1-10 and dress self at 60 months. Identifies shapes and colors, says 6-8 word sentences, and accepts responsibility for personal belongings at 65 months. Child was able to catch and throw ball with two hands, attempts to tie shoes, retell previous events, and use scissors at 72 months. Child was completely potty trained by 126 months.