



Emerson was born in 2011 and was prenatally diagnosed with partial trisomy 18q. He lives in the United States. Participation in the TRIS project began in 2012 at the age of 13 months.

Mother was 29 and father was 32 years old at the time of conception. Child with rare trisomy was fifth of six pregnancies (gravida, 6; para, 5). One male sibling was also diagnosed with partial trisomy 18q and lived for nine hours. Additionally, mother had a miscarriage at seven weeks four days during a subsequent pregnancy due to an unknown cause. Parents suspected a monosomy condition. A sibling was born this summer.

Mother has been diagnosed with a balanced translocation between the 13<sup>th</sup> and 18<sup>th</sup> chromosome.

### **Birth Information**

Infant was diagnosed with partial trisomy 18q at 17 weeks gestation after a routine ultrasound and amniocentesis. Prior to birth, infant was identified with clenched hands, club foot, an abnormal ventriculus tertius (middle brain ventricle), and decreased cardiac output.

Infant was delivered via planned vaginal birth after 6.5 hours of labor at 40 weeks gestation at 2693 grams and 47.6 centimeters on June 23, 2011. APGAR scores at one and five minutes after birth were 6 and 8 respectively. Infant was identified with low-set ears, small jaw, microcephaly, club foot (right foot), and low, wide-spaced nipples.

Infant was cared for in his mother's room at birth hospital. He remained in the hospital for two days. A pulse oximeter was used for monitoring. Infant was fed via nasogastric tube beginning at one day of age until seven months. Infant was discharged weighing 2523 grams.

### **Surgeries**

Initial surgeries at seven months of age included right-sided diaphragmatic hernia repair, stage 1 of mandibular distraction, ear tube placement and gastrostomy tube placement (button). At 10 months, child underwent stage 2 of mandibular distraction (distractors removed), and an adenoidectomy. At 16 months, child received a port placement. Two weeks later, a port removal and port reinsertion was necessary. Six days later, the port was removed.

again and a Broviac catheter was inserted. Finally, at 33 months of age, surgery was performed on child's right eye to correct ptosis (droopy eyelid) with a frontalis sling.

### **Cardiac**

Infant was diagnosed with coarctation of the aorta and double superior vena cava. Coarctation of the aorta was treated with Propranolol from three to 22 months of age when he "outgrew" the coarctation. Child did not need treatment for double superior vena cava because one emptied into the right atrium and other into the right coronary sinus.

### **Respiratory**

At 13 months, infant used a nebulizer and Dexamethasone drops for the treatment of congestion and upper airway obstruction. In addition, infant was prescribed Augmentin for a sinus infection from four to 10 months. Morgagni diaphragmatic hernia was discovered at two months and right side repaired at seven months of age (same time as mandibular distraction, gastrostomy tube surgeries and placement of bilateral ear tubes).

Infant was diagnosed with obstructive apnea, which was treated with an adenoidectomy at 10 months and a mandibular distraction at seven and 10 months. At 25 months, child was still experiencing episodes of obstructive apnea and was treated with Xopenoz, Pulmicort, and an increase in bilevel positive airway pressure (Bi-PAP) settings.

At 37 months, child's apnea was treated with a Bi-PAP, and oral Prednisone (given for 10 days with limited benefit). Child was also diagnosed with pneumonia at 30 and 32 months and was treated with Omnicef and Rocephin.

### **Neurological**

Infant was diagnosed prior to birth with microcephaly. Infant has not been reported to experience seizures.

Child started experiencing insomnia at 10 months. Cranial sacral therapy was initiated and met with some success. This type of therapy is continuing.

### **Renal**

At 15 months, child was diagnosed with nephroblastomatosis, which required chemotherapy by central line for five months. Condition will be monitored by a pediatric oncologist for five years.

### **Gastrointestinal**

Infant was prescribed Prevacid for acid reflux starting at 2.5 months until 15 months. At 25 months, child's reflux and constipation was treated with homeopathic digestive enzymes and a probiotic. In addition, child received acupuncture and massage therapy for the treatment of nausea with chemotherapy. At 37 months, child was treated for reflux and constipation with GTL herbal supplement and a probiotic supplement. Mother notes that the supplements are effective in treating these conditions.

### **Nutrition**

Infant was fed breast milk until 22 months of age and then transitioned to blended table foods by 27 months. Child was identified with the following food sensitivities: dairy, soy, wheat, corn, eggs, peanuts, hemp, oats, and buckwheat during testing at 15, 20 and 33 months of age. Blended diet must be monitored for these ingredients.

A nasogastric tube was used after discharge until gastrostomy tube placement at seven months. At 13 months, infant received all nutrition via bolus feedings. At 25 and 37 months, child was receiving nutrition primarily by gastrostomy tube. Oral intake is limited to a few bites one to two times a day.

### **Oral health**

At the present time, child has not experienced any dental issues. Child first saw a dentist at 34 months and follow up visits every six months.

### **Vision**

Child was diagnosed with hyperopia (farsightedness) at six months and is not currently receiving treatment for this condition. Child was also diagnosed with strabismus (cross-eye) at five months and was treated with patching until 19 months. Child wears glasses.

At 19 months of age, child received his first pair of prescription eyeglasses. At 33 months, child had an eyelid lift and brow sling surgery for treatment of ptosis.

### **Auditory**

At 36 months, child received a bone-anchored (BAHA) hearing aid for the treatment of conductive hearing loss due to delayed nerve development from ear to brain. The device is worn on a headband. Child also experiences recurring otitis media and wax build up due to small ear canals. Otitis media began at 16 months and occurs during illnesses. Otitis media is treated with Ciprodex antibiotic drops and an ENT treats wax build-up during office visits.

Child is also noted to have large perforations in both ears. This condition was diagnosed at 27 months of age.

### **Immunizations**

Immunizations began at 10 weeks with vaccines for Polio, Diphtheria, Tetanus, and Pertussis (DTP), meningitis and pneumonia. Three weeks later child was given immunizations for rotavirus and Hepatitis B. If child was ill when due for one or more vaccines, they were held off until he was healthy. Child's immunizations are currently up to date. No adverse reactions have been reported.

### **Orthopedic**

Casting was provided at 2-3 months of age for a total of five weeks for infant's club foot (right foot). At 13 months, child use – was prescribed Poinsetti orthopedic brace, which were not effective. The brace caused knee and hip pain due to the angle at which the bar connected

to child's feet and adversely affected child's sleep. A change was made to Supramalleolar Orthosis (SMO) braces at 25 months. The SMOs were worn while child was awake. Since then, child demonstrates equal use of right and left legs and has improved the quality of his sleep.

At 25 and 37 months, child was using a Lecky Squiggles® Stander. From 15 months, child received chiropractic adjustments for kyphosis, scoliosis, digestive health, and immune system response.

From 24 to 29 months, child received the Anat Baniel method of Feldenkrais therapy to improve physical mobility, which was noted to be effective. The cost was prohibitive to continue beyond the five-month period.

### **Developmental milestones**

Mother notes the following developmental milestones: smiling at 10 weeks, laughing at three months, shaking head "no" at approximately 12 months, signing "more", "all done" and "go" at approximately 27 months and babbling "da da da" at 34 months. Mother noted child said "ba bo" while playing with bubbles at 35 months. Child also began clapping at 22 months and demonstrated actions to songs such as Itsy Bitsy Spider (22 months) Pat a Cake (22 months) and Hokey Pokey (30 months with trunk support). Mother notes he began interacting with his older brothers at 12 months of age.

Child began rolling from prone to supine initially at three months, but the Poinsetti braces were then applied and he was unable to roll. Child relearned this skill at 33 months. Child rolled from supine to prone at approximately 22 months of age. Child began tripod sitting at 13 months and sitting independently for up to 20 seconds at 37 months. In addition, he began grasping objects at 32 months.

At 37 months, child was using a Big Mack® and a Texture switch for communication.

### **Education and Therapy Services**

Services began at two months of age with speech therapy (ST) for swallowing issues 60 minutes a week and occupational therapy (OT) for 60 minutes a week. Early intervention began at six months with 60 minutes per month of OT, physical therapy (PT) and developmental therapy. This was in addition to previously mentioned OT and ST from an outside clinic.

At 11 months old, additional PT began for 60 minutes a week, and ST increased to 120 minutes per week. At the time of completion of baseline survey (13 months), infant received 60 minutes of occupational therapy, 150 minutes of physical therapy, and 120 minutes of speech and language therapy per week in a clinic.

At baseline TRIS Survey completion at 13 months old, child was enrolled in a PT research study, and received 90 additional minutes of PT a week for three months. When the study finished, parents discussed child's measurable gains with early intervention providers and

fought for 60 minutes of OT and 60 minutes of PT weekly while continuing with outpatient OT, PT, and ST.

Beginning at three months of age, infant also received cranial sacral therapy for two months for the treatment of feeding difficulties. This therapy was not effective. Later, from 25-28 months, the therapy was reinstated weekly and also at present. Mother notes cranial sacral therapy is effective for child's gastrointestinal and sleep needs.

At 27 months, his outpatient OT and ST ended (providers felt their knowledge no longer met child's needs). Services were transferred to another outpatient clinic with 60 minutes of ST and 120 minutes of OT per week. The OT took over feeding therapy since child began having additional sensory issues with oral eating rather than swallowing.

At 30 months old, outpatient PT was transferred to the same clinic as OT and PT. Child received PT for 120 minutes per week with same therapy schedule with early intervention as well. Early intervention stayed at 60 minutes OT and 60 minutes PT per week and 60 minutes teacher per month. Aquatic therapy began to take the place of either an OT or PT session beginning at 32 months. Aquatic therapy continues at the present time for 45 minutes a week.

Currently, child will receive homebound services through the local school district as follows: PT 60 minutes twice a month, OT 60 minutes twice a month, and an Early Childhood Special Education teacher 60 minutes weekly. The public school-based team is hesitant to provide an SLP services. Mother suspects it is a financial issue rather than meeting therapy needs.

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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