

# Caring for Simon: A Case Study in Care for an Infant with Trisomy 18

## Background

Trisomy 18 is a genetic disorder in which the presence of extra material from chromosome 18 causes a variety of medical conditions. The literature cites live birth prevalence ranging between 1/3,500 and 1/8,000.

Simon was born on September 7, 2010 at 38 weeks gestation. He weighed 1900 grams and presented with patent ductus arteriosus (PDA), ventricular septal defect (VSD), coarctation and a bilateral cleft lip. He was diagnosed at three days with full trisomy 18 via fluorescence in situ hybridization testing.

Simon lived for 88.5 days in a Level III neonatal intensive care unit of a mid-sized hospital in the Midwest.

Simon's team of care providers included neonatologists, a pediatric cardiologist, a pharmacist, nurses and respiratory therapists. This group worked closely with Simon's parents to make treatment decisions and manage daily care. When disagreements occurred, often due to his trisomy 18 diagnosis, the majority of Simon's nurses honored his parents' preferences. Nurses also provided support and information to Simon's parents and older brothers.

## Class Design



## Decision making

- **Cardiac surgery:** Simon's cardiologist advocated for surgery once he stabilized and gained weight (no goal weight established). Additionally, there was collaboration between Simon's neonatologist, cardiac surgeon and cardiologist across two hospitals. Cardiac surgery is not typically recommended for infants with trisomy 18. Simon's PDA closed on its own.

- **Breastfeeding:** Initially, Simon received total parenteral nutrition (TPN) feeds. Breast milk was then offered via syringe and, later, bottle. He was also given opportunities to nurse. Feeds were also introduced via nasogastric (NG) tube with formula thickened with rice cereal. Change to TPN for approximately 48 hours after Simon aspirated on Day 60. Return to breast milk via NG-tube.

- **Medications:** Simon was placed on prostaglandins on Day 2; diagnosis on Day 3. Beth, one of Simon's nurses, was surprised, "they don't give trisomy 18 babies prostaglandins". Parents were pressured to withdraw prostaglandins. Same nurse advised parents to discuss this with hospital's Ethics Committee which parents did.

"...stopping the prostaglandins is our decision...I heard that one doctor was going to just stop it on his own. When I asked later he said he would have checked with us first. Well how do I know that? We keep being pressured to lower the dose or turn it off."

(Sheryl, Simon's mom)

- **Immunizations:** Debate among Simon's medical team and his parents regarding immunization schedule.

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## Overview

Literature describes the prognosis for newborns with full trisomy 18 as poor citing only approximately 10% of newborns surviving to their first birthday. Cause of death is usually related to complications due to central apnea and/or cardiac anomalies such as ventricular septal defect (VSD). Several recent studies describe medical interventions for surviving newborns including provision of supplemental oxygen, continuous positive airway pressure and related mechanical ventilation. There needs to be further examination of the care received by newborns with full t18 during the prenatal, neonatal and immediate perinatal periods.

## Simon and his family



## Conclusion

Simon's life offers guidance for nursing practice including blending medical treatments and family-centered care in the face of a bleak diagnosis. In addition, there is a need for care that is professional and compassionate regardless of diagnosis.

"We will make sure that Simon is treated as an individual and give him all the same options as any other baby." (Sheryl, Simon's mom)

## Basic and specialized care

"I connected with Simon through my heart and not as a medical case...[For example] if he didn't like certain music, he would let you know. At night he stayed awake and interacted with me...Sometimes, if he was angry, he wouldn't look at me, but usually he looked at me clearly. He had a hooked finger and he hooked everyone's heart."

(Mary, one of Simon's primary nurses)

Simon received basic care including bathing, monitoring his weight and other activities of daily living. His parents, brothers and extended family were given opportunities to participate in.

Simon also received specialized care including oxygen via canula, total parenteral nutrition (TPN) feeds and some medications through a peripherally inserted central catheter (PICC) line.



## Implications

"On my first day caring for Simon I noticed how many diverse opinions Scott and Sheryl were getting from caregivers," she recalled. "They've not gone through this situation before, their family is just beginning to deal with devastating news and they're being bombarded with opinions from people they don't know. It's overwhelming. My heart went out to the family. How were they processing this? Recommendations about heart and plastic surgery and feeding and respiration options. Pharmacy issues with the prostaglandins. They needed a consistent approach with all their doctors and nurses. [They needed] a care plan. When a baby is born with Trisomy 18 the plan lays out how the medical team should respond. We need to all be coming from the same direction, in agreement."

(Lola, one of Simon's primary nurses)

- Care encompasses the provision of basic and specialized care along with ongoing interactions with parents and family members. Joint decision making is a necessity.

- Need to address needs and make decisions based on individual infant *not* diagnosis: "Treatment of children with Trisomy 18 is planned on a case-by-case basis. Which treatments are used depends on the patient's individual condition."

(Downloaded from

<http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002626/>)

- Efforts must continue to increase awareness of trisomy 18 and other rare trisomy conditions. Training and related efforts must be undertaken and/or continued with professionals across a range of medical fields. Training should also extend to university level coursework and specialized post-graduate programs.

## TRIS project

Additional information about The Tracking Rare Incidence Syndromes (TRIS) project is located at <http://web.coehs.siu.edu/Grants/TRIS/> and contact by email at [tris@siu.edu](mailto:tris@siu.edu)