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Surviving with Trisomy 13: Provider and Parent Perspectives and the Role of the Pediatric Palliative Care Program

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Trisomy 13 typically denotes an overall poor prognosis in the setting of multisystem anomalies. Through a provider and parent perspective, this case illustrates the benefit of hope, communication, and teamwork through the integration of a palliative care team in the care of a medically complex child with trisomy 13, resulting in enhance survival and perceived quality of life for patient and family. © 2016 Wiley Periodicals, Inc.

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INTRODUCTION

Trisomy 13 is a rare chromosomal anomaly that results from aberrations during cell division, or meiosis, culminating in an extra copy of chromosome thirteen. This abnormality is generally associated to multi-organ anomalies, including holoprosencephaly, cleft lip, palate, seizures, in addition to cardiac, gastrointestinal, and genitourinary malformations [Saikia et al., 2014; Suda et al., 2015]. The median survival has been reported to be 8.5 days, with over ninety percent 1 year mortality. The most common causes of death include cardiopulmonary arrest secondary to congenital heart defects and pneumonia [Brewer et al., 2002; Boghossian et al., 2014]. A study by Janvier et al. [2012] notes that parents of trisomy neonates are most often times advised that the condition is incompatible with life and would result in diminished overall quality of life for patients and families. Springett and Morris [2014] also established increased detection of trisomy 13 prior to 15 week gestation with antenatal cytogenic testing commonly used for Down Syndrome.

Recent studies, however, noted that many with trisomy 13 are outliving their expected survival. A multi-state study by Meyer et al. [2016] noted a 5-year survival of 9.7% and this increased survival could be attributed to aggressive medical interventions. Along with enhanced survival, many of these children must undergo a multitude of surgeries throughout their lifetimes. Baty et al. [1994] notes that these children usually have on average of 2.2 surgical interventions in their first year of life. Bruns and Campbell [2014] describes the varying types of surgical interventions and Janvier et al. [2012] noted increased survival to 1 year in children who receive such interventions [Janvier et al., 2012; Bruns and Campbell, 2014].

The following case demonstrates that despite apparent odds of survival, strong interdisciplinary team involvement as well as an unrelenting sense of hope and communication amongst providers and families are the foundation of palliation for children with trisomy 13.

CLINICAL REPORT

Provider Perspective

Sofie[1] was born to a 46-year-old mother, G5P5005, at 38 weeks gestation. Ultrasound and amniocentesis at 30 weeks confirmed trisomy 13. Upon birth, Sofie was monitored in the NICU for 3 days and required oxygen via tent for only a few hours. She received wound care and bandaging for cutis aplasia, a scalp anomaly. Because of compromised suck due to cleft palate, Sofie received an NG tube for assistance with feeds. She was transferred to another

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NICU on day three for higher level of care with regards to cutis aplasia but no surgical intervention was pursued. Sofie was also found to have bilateral cataracts, which were eventually repaired. An echocardiogram detected atrial septal defect and patent ductus arteriosus, which closed spontaneously without need for surgical intervention. Sofie has obstructive sleep apnea but required no oxygen or intervention. Sofie also needed bilateral Eustachian tube replacements. Her medical course was also complicated by frequent hospitalizations for surgeries and sepsis and currently at 2 years of age, Sofie also underwent repeated surgeries for small bowel obstruction, for which she became TPN dependent.

Since birth, Sofie had a total of ten hospital readmissions for surgical interventions related to anomalies associated with trisomy 13. Despite numerous surgeries, Sofie thrived with support from her parents and four older siblings. They described her as center of their world and caring for Sofie brought warmth and unity to the family. Whether it be providing for Sofie's needs, holding her, or singing her favorite grungy Nickelback songs, everyone in the household had a special sense and purpose.

The journey, however, was not without its obstacles. Her mother, Kristina,[1] noted that on initial diagnosis, she was advised by her geneticist to terminate the pregnancy. Her family's subsequent hospital visits were also met with hesitancy and doubt cast by the medical community given overall poor prognosis. During Sofie's initial hospitalization for management of cutis aplasia, the family overheard a training physician inquire his supervising attending about the futility of additional medical interventions. He expressed an unrelenting desire for no-treatment and to encourage the family to pursue comfort measures.

Amidst these insurmountable doubts, Kristina and her husband still fervently believed in the livelihood of their child. The family is spiritually grounded and their Christian faith factors eminently in their unwavering sense of hope. They also received tremendous support from SOFT (Support Organization for Trisomy 18 and 13) and became tireless advocates for the organization. Through SOFT, the family was referred to physicians in the community who were attentive to Sofie's needs. Sofie was followed early on by a knowledgeable and compassionate primary care physician who provided supportive listening and valuable family education. Along with an interdisciplinary team of child-life specialist, chaplain, psychologist, social worker, nutritionist, chiropractic, physical and occupational therapists, the physician was able to help Sofie continue to thrive at home. She would later be enrolled in Calfornia's Pediatric Palliative Concurrent Care program for patients with medically complex illnesses. On a follow up visit to our palliative care clinic, Sofie smiled incessantly and engaged tactilely with providers and continued to be her playful self.

Sofie's case illustrates the interwovenness of science and the art of palliative medicine. There is inherent hope on multiple dimensions: hope within the family, community, and amongst providers who were attune to the family's needs. While medical interventions may certainly have a role in prolonging survival, hope, communication, and teamwork are equally as important. It is important to note that while palliative care intervention is important, the above elements should be incorporated by all providers taking care of medically fragile children.

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Sofie's family found hope through prayers, friendships built through their trisomy organization, as well as within each clinical improvement that Sofie seemed to make with medical interventions. This hope fueled their desire to do more to rally support for trisomy education. They are appreciative of the palliative care team who embarked on numerous life journeys with them, always making sure they felt heard. The team provided psycho-social-spiritual support through a comprehensive, interdisciplinary approach grounded in strong communication.

Although strong communication is especially important in the care of seriously ill children, a study by Davies et al. [2008] revealed many barriers to palliative care by pediatric providers. The most-commonly perceived factors center around uncertain prognoses, discrepancies in treatment goals between staff and family members, as well as language barriers. Furthermore, the study noted that many providers perceived they have little training in end-of-life care [Davies et al., 2008]. By contrast, in a cross-sectional survey of nurses, Feudtner et al. [2007] found that nurses with greater clinical experience, more palliative care education, and higher scores on the Adult Dispositional Hope Scale, demonstrated higher levels of comfort working with dying children and families. Valuable training in communication is a crucial step toward providing compassionate care and support to families of medically fragile children. It is important to provide current, accurate and comprehensive information about a child's disease process without imposing one's own values on the families.

Sofie's case also supports that early palliative care intervention is key. This palliative care intervention is not only by means of symptom management but also by establishing meaningful relationships earlier on during the diagnosis phase so that families have a team to turn to amidst medical doubts and uncertainties.

Recent studies, however, reported that palliative care medicine may be underutilized in the perinatal period. Marc-Aurele and Nelesen [2013] found that of the 66 women referred to a specialized home perinatal palliative care program between 2006 and 2012, only one third visited their palliative care team once or twice prior to delivery, indicating a need for earlier referral to provide more comprehensive palliative care [Leong and Nelesen, 2013]. Although Sofie's parents were unaware of palliative care until after delivery, the continuity of care provided was essential to the family's ongoing ability to cope with complex medical decisions.

While many would argue that having repeat surgeries and prolonged inpatient stays equate to having diminished quality of life, Sofie's family did not perceive it that way. Each medical decision was jointly made by the family in collaboration with their trusted providers. Each hospitalization and surgical intervention seemed to strengthen rather than weaken the family unit; they were bonded by their love for Sofie. Sofie was referred by the family as a teacher and moving life force given all that she has overcome. She has taught the family about the fragility of life, how to passionately care for a loved one, how important it is to live life with hope and faith. This sentiment was reflected in Janvier's work on trisomy parents' experiences in social networks. It was felt that despite being afflicted with severe disabilities, the vast majority of parents described their children as having a happy disposition. Moreover, parents reported these children enriched their family irrespective of the length of their lives [Janvier et al., 2012].

Sofie's perseverance is mirrored by her family's unconditional love and sacrifice. Her story resonates the resiliency of the human

spirit—that despite overbearing numbers and statistics, there is a human being with a seemingly fearless determination to beat all odds.

The following is a unique perspective from Sofie's parents.

Parent Perspectives

We had no idea what Trisomy 13 even was when we were given the diagnosis by the genetic counselor, who promptly informed us over the phone that it was a "fatal syndrome" and "not compatible with life," that our baby would likely not even live to birth and her recommendation was early termination at 30 weeks gestation. We did not make any haste decisions and quickly met with our regular doctor, who completely supported us and our position. We began to educate ourselves about this syndrome and the chances of long term survival. We immediately found SOFT, Support Organization for Trisomy 18 and 13, and began to read about hundreds of babies who survived years to decades after diagnosis. We fought to do everything possible to ensure full care and support of our unborn baby girl and her survival, as we would for any of our children. We recognized quickly, being frequently reminded by medical professionals that Trisomy 13 is a fatal syndrome that we all suffer from fatal syndromes. We are all going to die. Some will die young, some old, some expected and some unexpected. We have no real control of knowing when our lives will end so why would or should medical care be any different for Sofie, or any Trisomy 13 baby? We have fought long and hard and continue to do so, advocating for equal medical care and rights for Sofie and all Trisomy 13 babies.

Our life has changed immensely with the birth and blessing of our *Princess Warrior*, Sofie Marie. She has defied all the odds and proved many medical professionals wrong. She continues to grow and strive and fight for her life. We have had some rough patches, that would be questionable for even a "typically" abled child to overcome but our Sofie is a warrior and has battled through. Each day is a blessing and each milestone, a miracle. Sofie is a true joy and has brought so much love and happiness to our family. She has a way of touching the souls of people who met her and filling them with love. Sofie is a living miracle and cultivates change in others every day of her life. Her purpose on this earth is vast and we are beyond blessed and honored to be her parents. Experiencing life with Sofie has led us into new experiences and endeavors. We have a new found passion to change the way that Trisomy 13 is perceived. A passion to give all Trisomy 13 babies, and those with like syndromes, every chance for support and survival and to assist and support other Trisomy 13 families. The pediatric palliative care program has also been an integral part of our ability to cope with Sofie's condition as well as her ability to thrive. The team instilled in us a continued belief of our work and advocacy for trisomy children. We now serve on the Family Advisory Council of the children's hospital where Sofie receives care so that we can influence staff's understanding of the disease process to better support other families. In fact, we have also begun designing and developing assistive mobility devices to offer better solutions to what is currently available. All of this stemming from the influence of our precious Princess Warrior whose little life had no value to so many but will always remain priceless to us. The biggest and best gift we have been given by Sofie is the gift of complete and total, unbiased, unconditional love and joy. For this we are eternally grateful to have been blessed with this amazing baby with Trisomy 13 and we look forward with excitement and anticipation to the years of her life that are yet to come.

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