An 8-week-old infant with trisomy 13: dilemmas for medical decision making

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Abstract: Patau syndrome, trisomy 13, is a rare genetic condition with notable physical and mental characteristics and an average reported lifespan of 9 months. For years, trisomy 13 was regarded as a lethal condition; however, a few infants survive for many years, creating a dilemma for both the medical practitioner and the family in terms of the appropriateness and timeliness of specific medical interventions. Even in the face of severe mental and physical limitations, most families of children with trisomy 13 report their experiences as enriching. Appropriate and timely surgical interventions and medical treatments have been shown to increase survival for some infants. Early palliative care helps to limit physical and emotional suffering, and allow the family to create a legacy that their baby's life had meaning, regardless of how long they survive. We present a case of an 8-week-old infant with trisomy 13 to illustrate some of the medical decision making dilemmas faced by both medical practitioner and the family.

Keywords: Trisomy 13; Patau syndrome; ethics; survival; aneuploidy; palliative care

Received: 08 December 2017; Accepted: 15 December 2017; Published: 26 January 2018.
doi: 10.21037/acr.2018.01.03
View this article at: http://dx.doi.org/10.21037/acr.2018.01.03

Introduction

Patau syndrome, trisomy 13, has live birth prevalence between 1:7,000 and 1:29,000 depending on the source of the data, and is the third most common autosomal trisomy (1-5). The average length of survival is approximately 9 months with 90% of patients dying before the age of 1 year (6). The standard of medical care has mainly focused on education of the family, and palliative care measures, and withholding of invasive interventions. The combination of short life span and severe physical and mental anomalies poses a challenge for medical practitioners and family when deciding which, if any, treatments would be appropriate. Recent studies have shown increased survival of some children with trisomy 13 into adolescence with earlier recognition and better treatments; though these children still have significant mental and physical disabilities. Many more mosaics are identified with new genetic testing modalities, which present with more mental limitations than physical anomalies (7). These children may live longer and will require a multifaceted approach to care. The causes of death in infants with trisomy 13 include severe brain malformations, cardiopulmonary arrest, congenital heart defects, sepsis, pneumonia, and withdrawal of life support (2).

Case presentation

An infant confirmed to be of 38-week gestational age was delivered in the prehospital setting (in the ambulance on way to the hospital) via spontaneous vaginal delivery to a 33-year-old mother who did not receive prenatal care. She had meconium stained amniotic fluid, and her membranes ruptured at delivery. Apgar scores were 3 and 6 at 1 and 5 minutes, respectively. Mother was unaware of her current pregnancy and had a history of preeclampsia, and 2 prior spontaneous abortions. There was no history of maternal medication use during pregnancy; however, a urine drug screen was positive for amphetamines, methamphetamines and benzodiazepines. Mother's history was also significant

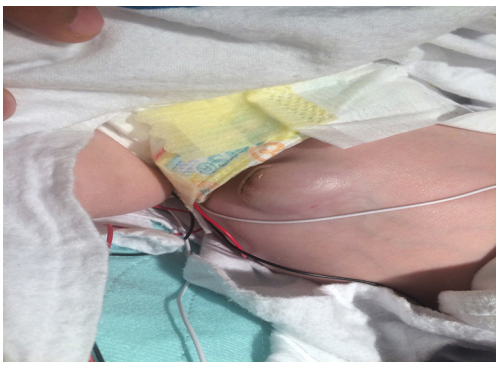

Figure 1 Unruptured omphalocele.

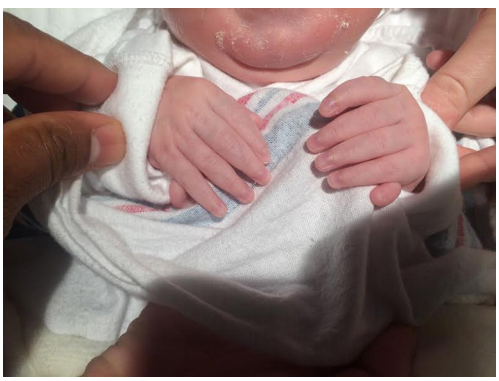

Figure 2 Hyper convex nails and postaxial polydactyly of both hands.

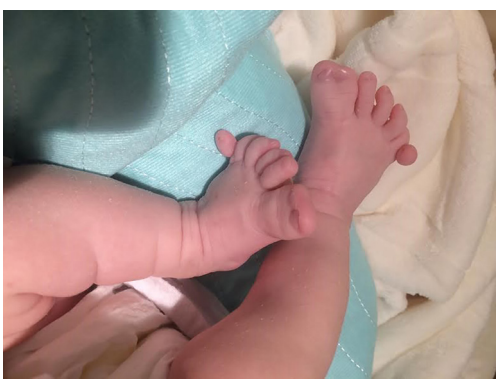

Figure 3 Postaxial polydactyly and syndactyly of both feet.

for a prior term twin delivery.

The infant was found to have an un-ruptured omphalocele (*Figure 1*), postaxial polydactyly of both hands (*Figure 2*) and toes (*Figure 3*), partial syndactyly of bilateral halluxes (*Figure 3*), and dysmorphic facial features. A chest X-ray showed meso or dextrocardia. A loud, systolic murmur was noted and echocardiogram showed a small secundum atrial septal defect (ASD), large malaligned ventricular septal defect (VSD) or possible double outlet right ventricle (DORV) and moderate sized patent ductus arteriosus (PDA) with mild biventricular hypertrophy.

The infant received supportive care and no reparative interventions for heart defects. The infant was sent home in the care of a foster family. The infant continued hospice care for 8 weeks and then died while at home.

Discussion

Trisomy 13 was first described in 1657 by Bartholin; later, Patau delineated its chromosomal abnormality in 1960 (1,2). Risk factors include advanced maternal age, but a link between ethnicity, race, or geographic location has not been established (2). Advanced genetic testing shows that Patau syndrome is most commonly the result of nondisjunction, translocation, or mosaicism (2). The standard for diagnosis is a karyotype analysis to determine aneuploidy (2). Chorionic villus sampling (CVS) and amniocentesis allow prenatal diagnosis. An increased number of cases are being identified with the use of comparative genomic hybridization testing (6).

Based on genetic testing, it is now possible to provide more detailed information regarding specific chromosomal variations and more complete genetic counseling. The advances in medical care have given us the opportunity to extend the life of these children; however, it is a subject of much debate and discourse whether or not to provide a range of comprehensive treatment, including invasive modalities, to extend lives of infants with trisomy 13 (2-13). Nelson *et al.* showed a survival of 20% of trisomy 13 patients to 1 year and a 12% to 10 years of age (14). The median survival was 12.5 days; fewer deaths occurred after 3 months of age. Infants who survived to 6 months of age lived for 10 years or longer. Most of the children living longer than 1 year had multiple major procedures on organ systems including cardiac, genitourinary, respiratory, otolaryngology, or musculoskeletal; each with their own inherent risk and cost. This data did not include mosaics, which may skew results for complete trisomy. A study by Costello *et al.* advocates for surgical repair of congenital heart disease (12). The operative mortality was reported between 29% and 80% for patients alive after 116-day follow up (12). A 50% survival rate was reported with supportive care prior to hospital discharge with a median

survival to 32 days (7-9). Many children with trisomy 13 live beyond the first decade; early artificial ventilation and treatment for intractable epilepsy may be typical in most of these children (5).

Though most studies focus on survival, a few discuss the developmental potential of infants with trisomy 13. Many of the mental deficiencies are severe and the child may only attain minimal to no discernible skills (10). Bruns, in a sample of 8 trisomy 13 children, reported developmental range of 3–9 months in children aged 15–33 months (13). Children were able to explore objects and perform voluntary movements such as kicking legs, sitting without support, and rolling. Some children were able to imitate open vowel sounds and 1 child was able to imitate simple gestures, scribble, and step with assistance. None of the children were able to assist with dressing or self-feeding. Liang, *et al*. reported children with limited communicative function; only 2 patients in their study were able to use one word phrases (10).

Families are able to find support groups and social networks. Appropriate counseling and communication to families is important. Janvier *et al.* reported that families of a trisomy 13 child have been told their child's diagnosis was incompatible with life (87%), would live a life of suffering (57%), be a vegetable (50%), live a meaningless life (50%), ruin their marriage (23%), and ruin their family (23%) (8). More thoughtful and empathetic phrases were used much less frequently such as, their child may enrich their family (16%), might have a short and meaningful life (60%), or might survive for many years (43%) (12). Although most parents described their child having severe neurodevelopmental disabilities, 97% described their child as "happy" and had a positive view of family life and quality of life for their child (8,9). This reaffirms the importance of thoughtful dialogue between the medical practitioner and families when discussing goals of care and establishing rapport.

Regardless of the specific interventions and surgeries, the responsibility to provide a balance of hope and reality to the family, is often placed on the medical practitioner. Parents should be given the facts without making a judgement of their perception of quality of life for their child. Early palliative care may help limit physical and emotional suffering and offer nontraditional care opportunities to allow the family to create a legacy that their baby's life had meaning, regardless of the duration of survival. Palliative care including nursing consistency, private family time, and undisturbed sleep have been seen to improve family dynamics and clinical outcome (4,15).

Conclusions

It is not certain that all children with trisomy 13 will have an extended life, as shown by survivorship curves (10). Medical practitioners should neither mandate full resuscitation nor support all medical or surgical interventions chosen by the family. Medical practitioners should offer an evidence based understanding of the condition in a transparent and unbiased way to allow families to make informed decisions. Further research into the genomics of full trisomy versus mosaics may offer better understanding of the variable expression into phenotypes of trisomy 13. Employing early palliative care helps the family get resources and networking which will enrich the lives of the children with trisomy 13 and their families (12,14,15). According to Fenton, "*We perform ethical analysis assuming we truly understand how to apply beneficence or maleficence to a child with trisomy 18 or 13 or any other disorder in which there may be profound disability. As if the child can tell us what he or she is feeling. But we can do our best to assess and treat pain and discomfort. Smiles and laughter need no score pad. We know what they mean*." (16).

Acknowledgements

None.

Footnote

Conflicts of Interest: The authors have no conflicts of interest to declare.

Informed Consent: It was waived as it was not required in the writing of this article. Case data has been entirely de-identified from person, location, and date.

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doi: 10.21037/acr.2018.01.03
Cite this article as: Staso P, Paitl S, Patel DR. An 8-week-old infant with trisomy 13: dilemmas for medical decision making. AME Case Rep 2018;2:3.

