The Ethical Implications of Bias in Counseling Parents of Children with Trisomy 13 and 18

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ABSTRACT

Many physicians continue to see serious chromosomal abnormalities as lethal or "incompatible with life." A diagnosis of trisomy 13 or 18 is associated with a high risk of perinatal death, but children who survive the neonatal period may continue to live for several years. In sharing diagnoses with family members, physicians may convey their bias with family members. Shared decision making, using a model of patient-centered care, may help physicians to avoid conveying such bias.

INTRODUCTION

A generation ago there was implicit agreement among physicians that life-sustaining measures were not indicated for a diagnosis of trisomy 13 or 18 due to the severity of the anomalies.[1] Recent studies show this viewpoint persists among physicians, despite several societal changes that have influenced the approach to counseling families of children with serious illness.[2] These changes include a shift from paternalism to an emphasis on shared decision making,[3] changes in our ideas about treatment of people with disabilities,[4] and an increase in social media that allows parents to connect with each other outside of the healthcare system.[5] Despite these changes, most healthcare providers who encounter these serious chromosomal abnormalities are still likely to use terms such as "incompatible with life" or "lethal" as they think about how to describe the prognosis to the family. A diagnosis of trisomy 13 or 18 is associated with a high risk of perinatal death, with a median survival rate that is typically less than two weeks.[6] However, children who survive the neonatal period may go on to live for several years. In a retrospective cohort study carried out over 21 years (1991-2012) in Canada, the one-year survival rate for children with trisomy 13 was 19.8 percent and 12.6 percent for children with trisomy 18.[7] The 10-year survival rate for children with trisomy 13 was 12.9 percent and 9.8 percent for those with trisomy 18.[8] A significant number of the children in this study underwent surgeries ranging from myringotomy (surgery in which an incision in the eardrum to reduce pressure behind it) to complex cardiac

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repair, and had one-year survival rates of around 70 percent.[9] In a multi-state study of survival in children with trisomy 13 or 18, preterm birth and major organ birth defects, such as omphalocele (a rare defect in which the intestine, liver, and other organs are located outside the abdominal wall) were found to be most predictive of death in the neonatal period.[10] Finally, in a population-based analysis of mortality in children with trisomy 13 and 18, girls and African-Americans had a higher rate of survival.[11] Surviving children with these aneuploidies (chromosomal abnormalities) uniformly have cognitive and physical disabilities, although with significant heterogeneity throughout the population in terms of specific organ anomalies. One consistent feature is the severity of neurocognitive deficits. These studies report that treatment for children with trisomy 13 and 18 is not unequivocally futile in terms of survival.

Children with severe impairments can still be perceived by their families and careproviders as having an excellent quality of life.

Healthcare professionals need to consider future quality of life when they counsel a family about what they feel is in the best interest of the child. In a recent editorial, Lantos described treatment decisions for children with trisomy 13 and 18 as "in a stable gray zone" in bioethics.[12] The factors that contribute to this "gray zone" include a high mortality rate, severe neurocognitive deficits, and potential for high burden of treatment.[13] For this reason, he suggests that parental values should drive decisions, with tolerance of variability in choices and outcomes.[14] Lantos also highlights a traditional bias among healthcare professionals that children with trisomy 13 or 18 have a very poor quality of life.[15] He reminds us that quality of life should not be equated with physical or neurological impairment.[16] Parents report that their children with an aneuploidy are typically comfortable and experience the good things in life; they smile and laugh, and they bring joy and meaning to their families.[17] Children with severe impairments can still be perceived by their families and careproviders as having an excellent quality of life.

It is important to note that families may also have bias. Many families now have access to information in addition to what they are given by the medical team, especially via the internet and social media. Depending on what families choose to read and how they interpret the information, they could easily be exposed to material that is biased toward a more positive outlook. Families may be drawn toward stories of children who survived against the odds, and they may develop an understandable hope that their child will also outlive medical expectations. Janvier and colleagues surveyed 332 parents who were members of support groups for their children diagnosed with trisomy 13 or 18, with the purpose of describing the parents' lived realities and points of view.[18] Janvier and colleagues concluded that parents in support groups were often able to find positive accounts of children living with trisomy 13 and 18, which could then lead to differences of opinion between parents and healthcare providers about the appropriateness of medical interventions.[19] Thus, families may tend to be biased toward thinking about the best case scenario and how the intervention will benefit their child. Healthcare professionals, on the other hand, tend to take a more utilitarian view toward decision making when the situation falls into the "stable gray zone," balancing the benefits and risks of an intervention for the individual patient against the burden on the healthcare system in terms of the utilization of resources. This may bias healthcare professionals toward thinking about the most likely medical outcome, rather than the best case scenario.

Given the potential for bias and the bioethical gray zone of making treatment decisions, Haug and colleagues advocate for and describe a patient-centered care approach to counseling families, based on the recommendations made in 2001 by the Institute of Medicine (IOM).[20] These patient-centered concepts were established to define quality in healthcare delivery and include dignity and respect, information sharing, participation, and collaboration.[21] The IOM recommendations list specific approaches that can help careproviders to facilitate decision making in an ethically sound manner that considers the well-being of all involved. Haug and

colleagues take the recommendations and specifically focus on counseling after a prenatal diagnosis, but the concepts included in these recommendations are applicable to treatment decisions throughout a child's life. We will discuss the application of these concepts in the setting of Cora and her family's experience, as told by her mother, Joy Salls, one of the authors.

DISCUSSION

The Salls Family's Experience

Cora was born at a small, rural hospital. My husband and I did not have a prenatal diagnosis and received her diagnosis of trisomy 18 when she was three days old. We have been fortunate to have support from pediatric palliative care as well as many other specialties to help us navigate our journey. Cora has had many struggles. She has had surgeries to open her eyelids, place a gastronomy tube, and stabilize her gut; an appendectomy; cardiac repair; sutures on her eyelids; and surgery to release her tethered spinal cord. She had the most pain from teething, but is otherwise generally happy and interactive, which vouches for her quality of life, even through many difficult trials.

Cora is the light of our world and has an amazing bond with her little sister. Cora turned three in September 2016.

PATIENT-CENTERED CONCEPT NUMBER 1: DIGNITY AND RESPECT

The majority (98 percent) of parents of children with trisomy 13 or 18 who responded to a questionnaire about their experience felt that their family was strengthened and enriched by the birth of their child, regardless of their child's longevity.[22] The same group of parents reported four reasons for continuing the pregnancy after receiving a prenatal diagnosis of trisomy 13 or 18: moral/religious beliefs or personal values (77 percent); child-centered reasons, such as the baby was already a loved member of the family (64 percent); patient-centered reasons such as wanting to get to know the baby even if time was limited (28 percent); and practical reasons, such as being unable to terminate the pregnancy (6 percent).[23] Thus, the majority of parents who participated in the study based their approach to decision making on moral or religious beliefs and their child's identity as their son or daughter, despite being given this life-changing diagnosis. This differs from the perspectives of many healthcare professionals, in which there is a tendency to focus on the diagnosis and prognosis, rather than on individual dignity and the family's values.

Among other ways to support dignity and respect, Haug and colleagues advocate for assessing values, beliefs, and preferences throughout the continuum of care; supporting parents in making decisions that fit with their values; and creating opportunities for careproviders to listen to the healthcare experiences of patients.[24]

The Salls Family's Experience

When Cora was six months old, she and I were invited to speak at Grand Rounds at Fletcher Allen Health Care in Vermont. It was an honor, as many of her doctors and specialists were in attendance. There were also many there whose only knowledge of trisomy 18 was that it meant an infant would not live very long. I know there are medical professionals who do not believe in providing a range of treatment options to parents of children with trisomy 18 because of this perception. When I spoke to this room of doctors, I told them, "Cora is perfect. She was perfect from the first breath she took, and will be perfect until the last breath she takes." I believe Cora had an impact on many that day, and hopefully, sharing her story changed their perception of how to work with a family who has a child with a trisomy diagnosis.

PATIENT-CENTERED CONCEPT NUMBER 2: INFORMATION SHARING

Parents who receive a prenatal diagnosis of trisomy 13 or 18 often report that they were given very directive counseling, based only on the diagnostic category of trisomy 13 or 18, without consideration for the specifics of their child's unique clinical status.[25] Families said that they found it most helpful when healthcare professionals gave balanced and personalized information, when their choices were respected, and when they were provided with support. The plan of care (comfort care, limited interventions, or full interventions) that parents chose was significantly associated with the child's anomalies. For example, if the child had neither cardiac defects nor holoprosencephaly—when the embryo's forebrain does not develop into two hemispheres—the parents were much more likely to choose full interventions than if the child had both conditions. The patient-centered care model recommends presenting accurate figures for survival and outcome that take into account the unique clinical features present, avoiding unmodified use of the terms "lethal," "fatal," or "incompatible with life," and to give

information about educational resources, including support groups.[26]

The Salls Family's Experience

When Cora was born, despite not having a prenatal diagnosis, it was apparent something was wrong. Three days later we got her diagnosis of trisomy 18. The geneticist who gave us her diagnosis was amazing! She simply stated that Cora had trisomy 18. She gave us some information from one of the most reputable organizations, with the most up-to-date research. I asked what this meant for Cora, and she said, "She is going to have a difficult life." This was the truth, nothing more, nothing less. She didn't make assumptions, or tell us that Cora wouldn't live very long. My family is forever grateful for how she set us on the path for what was to be a very difficult, but rewarding, journey.

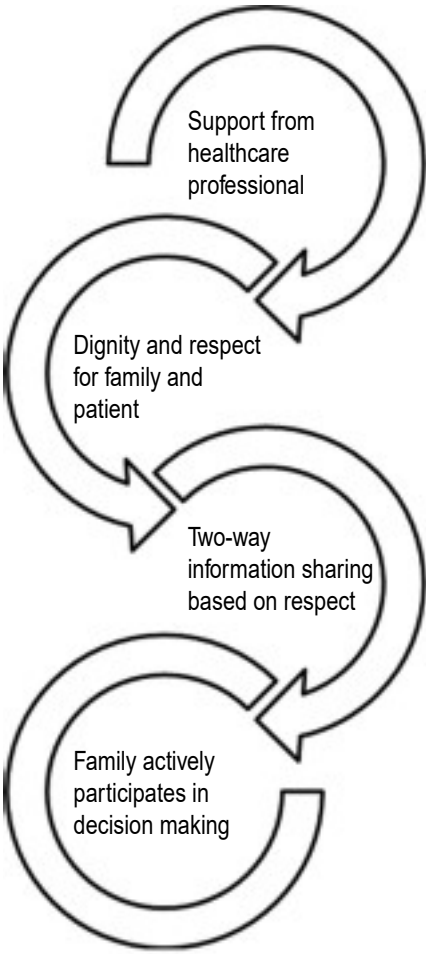
PATIENT-CENTERED CONCEPT NUMBERS 3 AND 4: PARTICIPATION AND COLLABORATION

Bias on the part of healthcare professionals can severely limit parents' ability to participate in medical decisions for their child with trisomy 13 or 18, as bias tends to limit collaboration. To increase parents' ability to participate and collaborate, it is essential that healthcare professionals discuss the range of options available in an objective way, even if a particular institution does not provide the type of care the family is seeking.[27] An ideal patient-centered care approach avoids coercion, attempts to keep lines of communication open, and establishes relationships with specialists and interdisciplinary teams that can provide coordinated care.[28] Even when families and healthcare professionals participate in a shared decision-making model and are able to look beyond their own biases, disagreements can still occur. This is when an ethics consultation may be beneficial. An ethics consultant creates a "moral space" to facilitate discussion between the family and medical team in order to elicit goals and values, ensure that interventions and outcomes are described more effectively, and weigh risks and benefits for a particular child.[29] This may be particularly helpful when parents ask for an intervention that the medical team feels is not indicated or is not in the best interest of the child.

The Salls Family's Experience

When Cora was seven months old, she went into heart failure due to her atrial and ventricular septal defects. Prior to this, she had been doing very well, with minimal interventions. She went into cardiopulmonary arrest, and was intubated. We knew the only way to save her life was to have her transferred to a medical center that could do the necessary cardiac repair. Her heart repair would normally be considered simple, as far as heart surgeries go, but due to her diagnosis of trisomy 18, we were not sure we would find a surgeon who would accept her case. We waited for days until we finally convinced a surgeon to operate on her little heart. That was two and a half years ago, and today Cora is doing great! She is learning to walk and is attending school. I still cannot fathom the thought of denying her a lifesaving surgery based just on the diagnosis of trisomy 18.

FIGURE 1.
Patient-Centered Care for Children with Trisomy 13 or 18



This graphic is based on the authors' interpretation of M.J. Barry and S. Edgman-Levitan, "Shared decision making—pinnacle of patient-centered care," *New England Journal of Medicine* 3, no. 9 (2012): 780-1.

CONCLUSIONS

Not all cases turn out as well as Cora's. However, Cora's case provides a good example of shared decision making using the model of patient-centered care. Shared decision making using the model of patient-centered care can help healthcare professionals overcome biases about diagnoses like trisomy 13 and 18. Healthcare providers should encourage a discussion of values and preferences that enhance dignity and respect for the child and parents, provide information in a personalized and objective way, and invite participation and collaboration from the family and the interdisciplinary healthcare teams (see figure 1). Families who feel supported in this way may be more likely to make difficult decisions about treatment plans in a way that is consistent with their core values.

NOTES

1. J.D. Lantos, "Trisomy 13 and 18—Treatment Decisions in a Stable Gray Zone," *Journal of the American Medical Association* 316, no. 4 (2016): 396-8.

2. A.P. Jacobs et al., "Trisomy 18: A survey of opinions, attitudes, and practices of neonatologists," *American Journal of Medical Genetics* 170, no. 10 (2016): 2638-43.

3. J.P. Brosco and C. Feudtner, "Shared Decision Making for Children with Trisomy 13 and 18," *JAMA Pediatrics* 171, no. 4 (2017): 324-5.

4. Ibid.

5. Lantos, "Trisomy 13 and 18," see note 1 above.

6. R.E. Meyer et al., "Survival of children with trisomy 13 and trisomy 18: A multi-state population-based study," *American Journal of Medical Genetics* 170A, no. 4 (2016): 825-37; K.E. Nelson, L.C. Rosella, S. Mahant, and A. Guttmann, "Survival and Surgical Interventions for Children With Trisomy 13 and 18," *Journal of the American Medical Association* 316, no. 4 (2016): 420-8; S.A. Rasmussen et al., "Population-based analyses of mortality in trisomy 13 and trisomy 18," *Pediatrics* 111, no. 4, pt. 1 (2003): 777-84.

7. Nelson, Rosella, Mahant, and Guttman, "Survival and Surgical Interventions," see note 6 above.

8. Ibid.

9. Ibid.

10. Meyer et al, "Survival of children with trisomy 14 and trisomy 18," see note 6 above.

11. Rasmussen et al, "Population-based analyses," see note 6 above.

12. Lantos, "Trisomy 13 and 18," see note 1 above.

13. Brosco and Feudtner, "Shared Decision Making," see note 3 above.

14. Lantos, "Trisomy 13 and 18," see note 1 above.

15. Ibid.

16. Ibid.

17. Ibid.

18. A. Janvier, B. Farlow, and B.S. Wilfond, "The experience of families with children with trisomy 13 and 18 in social networks," *Pediatrics* 130, no. 2 (2012): 293-8.

19. Ibid.

20. S. Haug et al., "Using Patient-Centered Care After a Prenatal Diagnosis of Trisomy 18 or Trisomy 13: A Review," *JAMA Pediatrics* 171, no. 4 (2017): 382-7.

21. M.J. Barry and S. Edgman-Levitan, "Shared decision making—pinnacle of patient-centered care," *New England Journal of Medicine* 3, no. 9 (2012): 780-1.

22. Janvier, Farlow, and Wilfond, "The experience of families," see note 18 above.

23. J. Guon et al., "Our children are not a diagnosis: The experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18," *American Journal of Medical Genetics* 164A, no. 2 (2014): 308-18.

24. Huag et al., "Using Patient-Centered Care," see note 20 above.

25. Janvier, Farlow, and Wilfond, "The experience of families," see note 18 above; Guon et al., "Our children are not a diagnosis," see note 23 above.

26. Huag et al., "Using Patient-Centered Care," see note 20 above.

27. Brosco and Feudtner, "Shared Decision Making," see note 3 above.

28. Huag et al., "Using Patient-Centered Care," see note 20 above.

29. M.U. Walker," Keeping moral space open: New images of ethics consulting," *Hastings Center Report* 23, no. 2 (1993): 33-40.