



Decision-Making in a Newborn with Trisomy 18 and Esophageal Atresia: The Role of Clinical Ethics Consultation

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Case Report

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Abstract

Esophageal atresia associated with tracheoesophageal fistula is one of the congenital anomalies that may occur in Edwards syndrome. The birth of an infant with these malformations confronted the treating team with the complex decision of whether surgical repair should be undertaken. Given the infant's overall clinical condition, the team also had to determine whether intensive care interventions should be initiated in the event of clinical deterioration. A clinical ethics consultation, conducted according to the standards of the American Society for Bioethics and Humanities, enabled all parties involved to reach a decision that respected the moral values of each participant while focusing on the infant's best interests.

Keywords: Esophageal Atresia; Clinical Ethics; Clinical Ethics Consultation; Informed Consent in Minors; Medical Futility; Proportionality of Treatment; Edwards Syndrome

Introduction

The American Society for Bioethics and Humanities (ASBH) states that: «The general goal of clinical ethics consultation is to improve the quality of healthcare through the identification, analysis, and resolution of ethical questions or concerns regarding a specific patient. Key to clinical ethics consultation is identifying and analysing the nature of the value uncertainty or conflict that underlies the consultation as well as facilitating resolution of conflicts in a respectful atmosphere with attention to the interests, rights, and responsibilities of all those involved. It involves being attentive and responsive to dimensions of the clinical encounter, such that the care delivered to each patient fits within the standards of the healthcare professions and ethical norms while accounting for the goals, values, rights, and responsibilities of involved parties [1]. As stated in the document approved in Trento on October 10, 2013, by the

Italian National Working Group on Clinical Ethics and Ethics Consultation in Healthcare: «All those involved in the process of care experience the ethical dilemma: what is the right thing to do in this situation? No one is excluded or exempt: from the patient to the healthcare team, from the family to the healthcare institution. Ethics is therefore an integral and not merely ancillary component of clinical practice» [2]. An editorial published in The Lancet in 1997 further observed that: «The ethics industry needs to be rooted in clinical practice and not in armchair moral philosophy. Debate on ethical matters is as much an integral part of everyday doctoring as choosing the best treatment for patients» [3]. Against this background, the validity of ethical reasoning in addressing medical dilemmas can only be assessed by comparing the values adopted and the methods applied with concrete clinical situations. Because of the complexity of the ethical issues involved, the case presented here provides an opportunity to move beyond abstract theoretical discussion



and to evaluate the contribution of clinical ethics consultation to decision-making in medicine.

Case Presentation

At 37 weeks of gestation, a young woman delivered a female infant who had received a prenatal diagnosis of trisomy 18 (Edwards syndrome). Birth weight was 1,500g, and the infant was breathing spontaneously. She was admitted to the neonatal intensive care unit (NICU) of an Italian university hospital. The infant showed signs of intrauterine growth restriction and several dysmorphic features consistent with Edwards syndrome, including cardiac and neurological abnormalities. On the second day of life, diagnostic investigations revealed esophageal atresia with tracheoesophageal fistula. The esophageal atresia was severe and prevented even the passage of a nasogastric tube. The infant was therefore managed with parenteral nutrition and intravenous hydration. Because oral feeding was impossible and there was a potential risk of aspiration, a pediatric surgeon was consulted to assess the indication for surgical repair. The surgeon considered the procedure technically feasible and proposed two possible approaches: closure of the fistula with primary esophageal anastomosis or closure of the fistula with placement of a percutaneous endoscopic gastrostomy (PEG). The treating team believed that, if surgery were to be performed, it should be undertaken within the following hours. At the same time, an urgent decision was required regarding whether invasive life-sustaining treatment should be initiated in the event of clinical deterioration. Trisomy 18, also known as Edwards syndrome, is a rare chromosomal disorder caused by the presence of an additional chromosome 18. The reported prevalence among live births ranges from approximately 1 in 3,600 to 1 in 10,000. Over the past two decades, the overall prevalence of trisomy 18 has increased, mainly due to the rising average maternal age [4]. In the United States, the estimated overall prevalence is approximately 1 in 2,500 pregnancies, whereas the prevalence among live births is approximately 1 in 8,600 [5]. Approximately 94% of infants with Edwards syndrome have complete trisomy 18, meaning that the additional chromosome 18 is present in every cell of the body. Partial trisomy 18, by contrast, is characterized by the presence of additional chromosome 18 material in only a portion of the genome. The clinical phenotype depends on the amount of additional genetic material involved and its distribution. Mosaic trisomy 18, in which the additional chromosome is present only in a proportion of cells, may result in a different clinical presentation depending on the percentage and localization of affected cells. Infants with trisomy 18 frequently present with intrauterine growth restriction and low birth weight. Congenital heart defects and abnormalities involving other organs may also be identified before birth. Other characteristic features

include microcephaly with abnormal cranial morphology, micrognathia, a small mouth, clenched fists with overlapping fingers, and, in many cases, severe intellectual disability. Approximately 40% of affected fetuses die during labour, and around one-third of those who survive are born prematurely. Median survival has been reported to range from 3 to 14.5 days. Survival rates are estimated at 60–75% during the first week of life, 20–40% at one month, and approximately 10% at one year. Overall, only 5–10% of patients with Edwards syndrome survive beyond the first year of life [6]. Other studies have reported a median survival of 14 days among live-born infants with complete trisomy 18, with a 3-month survival rate of approximately 20%. One-year survival is estimated at 8%, compared with approximately 70% among live-born infants with mosaic trisomy 18 [7]. Major gastrointestinal abnormalities have been described in approximately 16% of patients with Edwards syndrome, including esophageal atresia (4%) and tracheoesophageal fistula (6%), which may occasionally occur together. Although esophageal atresia with tracheoesophageal fistula is a potentially life-threatening condition that can only be effectively treated through corrective surgery, no previous studies have specifically evaluated the effectiveness of esophageal atresia repair in patients with trisomy 18. One study analysed the clinical outcomes of 24 patients with trisomy 18 and esophageal atresia who were admitted between 1982 and 2009 to two neonatal intensive care units at a Japanese hospital and received intensive treatment, including surgery [8]. Nine patients underwent palliative surgery only, whereas the remaining 15 underwent definitive repair. No intraoperative deaths or anaesthesia-related complications were reported. Enteral feeding was established in 17 patients, including three who achieved oral feeding. Three patients were discharged home. The one-year survival rate was 17% overall: 27% among patients who underwent definitive repair and 0% among those who underwent palliative surgery. Most deaths were related to cardiac complications. The authors concluded that, among patients with trisomy 18 receiving definitive repair of esophageal atresia combined with intensive cardiac management, esophageal atresia itself had limited prognostic significance.

Discussion

The complexity of the case led the treating team to promptly request a clinical ethics consultation, which was provided by the hospital's clinical ethics consultant. The first issue considered concerned the legal responsibilities of the different parties involved. Since the infant was unable to express her own wishes because of her age, the parents' preferences were sought as her legal representatives. After several meetings aimed at providing comprehensive information, the parents declined to consent to the proposed surgical intervention. The treating team therefore

considered whether to apply to the competent guardianship judge for authorization to proceed with surgery despite the parents' refusal. Under Italian law, parental decision-making authority may be temporarily overridden when necessary to authorize medical interventions that, if withheld, could result in serious harm to the health of a minor. The discussions with the parents revealed a clear opposition to treatments they considered medically futile. The treating physicians also expressed their reluctance to pursue overly burdensome interventions, in accordance with the ethical obligation not to initiate or continue clinically inappropriate and ethically disproportionate treatments (Article 16 of the Italian Code of Medical Ethics). The proportionality of the proposed intervention was therefore assessed in relation to the realistic goals of care that could be achieved. The available scientific evidence did not demonstrate any improvement in survival or quality of life following surgical repair of esophageal atresia in patients with trisomy 18. Although procedure-related suffering was considered unlikely, it could not be completely excluded. Moreover, the proposed intervention was technically complex and associated with a significant risk of complications.

A further decision concerned the therapeutic approach to be adopted should the infant's currently stable clinical condition deteriorate. In particular, the team had to determine whether endotracheal intubation and invasive ventilatory support should be provided in the event of respiratory failure, considering the parents' opposition to interventions aimed primarily at prolonging life for a limited period without a realistic expectation of meaningful benefit. The proportionality of these interventions was also evaluated considering the limited available evidence, which continues to question the appropriateness of invasive life-sustaining treatments in similar situations. The possible palliative role of available interventions was also considered. The multidisciplinary team concluded that, in the absence of convincing evidence supporting a meaningful benefit from surgical repair, overriding the parents' refusal would not have been ethically justified. The prognosis would have been substantially different in the presence of mosaic trisomy 18, a possibility that could not initially be completely excluded because the diagnosis had been established prenatally. However, further genetic investigations performed a few days later ruled out this possibility. The only potential justification for surgical treatment would have been to enable oral feeding. However, discharge home would not have provided a meaningful benefit, as the parents did not consider themselves able to provide the complex care required. Similarly, the use of invasive life-sustaining treatments was considered disproportionate.

The decision was therefore made to continue standard supportive care, including parenteral nutrition and

hydration. At the conclusion of the decision-making process, neonatologists, pediatric surgeons, and the infant's parents reached a shared decision not to proceed with surgical repair and not to initiate invasive life-sustaining treatments. The infant died a few weeks later from causes unrelated to the oesophageal condition, without ever leaving the neonatal intensive care unit. As stated by Pellegrino and Thomasma [9], every therapeutic decision requires, above all, a careful definition of the good to be pursued for the patient. In defining the biomedical good, it is essential to understand how a proposed treatment may modify short- and long-term prognosis as well as the natural history of the disease. In this case, all available evidence suggested that the proposed surgical intervention would not have altered either the prognosis or the natural course of the disease. Likewise, resuscitative interventions in the event of hemodynamic instability would most likely have resulted only in a short-term prolongation of life. Regarding the expression of the patient's preferences and the acquisition of informed consent, decision-making for minors in Italy is regulated by Article 3 of Law No. 219 of 22 December 2017, which states that informed consent for medical treatment of a minor may be given or refused by those exercising parental responsibility or by the legal guardian. The same provision further states (paragraph 5) that, when the legal representative of a minor refuses a proposed treatment but the physician considers that treatment appropriate and necessary, the matter may be referred to the competent guardianship judge by the legal representative, by the individuals identified under Articles 406 et seq. of the Italian Civil Code, by the physician, or by the legal representative of the healthcare institution.

The central question was therefore whether the proposed interventions could be considered appropriate and necessary, thereby creating the conditions for judicial review. In this case, Article 1, paragraph 7, of Law No. 219/2017 was not applicable because there was no emergency or urgent situation requiring immediate action. Consequently, careful assessment was required of the consequences of the parents' refusal of the proposed treatment. The physicians had to determine whether the parents' decision could reasonably be considered a serious threat to the infant's health, sufficient to justify referral to the Juvenile Court through the guardianship judge. In this regard, the lack of proportionality of the proposed interventions represented a crucial element, making it difficult to classify refusal of treatment as a decision likely to cause serious harm. The main challenge concerned the absence of a universally accepted definition of treatment proportionality, since the concept does not have a single, unambiguous meaning [10]. The principle of proportionality is recognized in Italian Law No. 219/2017, which states in Article 2, paragraph 2, that unnecessary or disproportionate treatments should not be provided in patients with an unfavourable short-term prognosis or imminent death.

Similarly, Article 16 of the Italian Code of Medical Ethics requires physicians not to undertake or continue diagnostic procedures or therapeutic interventions that are clinically inappropriate and ethically disproportionate, when no reasonable expectation exists of a meaningful benefit for health or quality of life.

Even within legal and professional frameworks, however, the meaning of proportionality requires interpretation. It would be impractical to expect legislation to provide a precise definition of proportionality, a concept whose evaluation should remain within the sphere of clinical judgment and professional responsibility. Pope Francis has also recognized the intrinsic complexity of assessing treatment proportionality, stating that therapeutic decisions require “an additional measure of wisdom.” Addressing participants at the European Regional Meeting of the World Medical Association, he noted that: «In the face of critical situations and in clinical practice, the factors that come into play are often difficult to evaluate. To determine whether a clinically appropriate medical intervention is proportionate, the mechanical application of a general rule is not sufficient. There needs to be a careful discernment of the moral object, the attending circumstances, and the intentions of those involved» [11].

To define the therapeutic strategy, two formal meetings with the clinical ethics consultant were organized. Participants included physicians from the neonatal intensive care unit, pediatric surgeons, the clinical geneticist, a representative of the hospital administration, and the infant's parents. The methodology used to reach the final decision deserves particular emphasis. Clinical ethics consultation, conducted according to the standards proposed by the American Society for Bioethics and Humanities, played a decisive role in the decision-making process. The consultant acted as a facilitator during meetings with all parties involved, helping them identify, analyze, and address the ethical dilemmas that emerged. The proportionality of the proposed interventions was assessed by considering not only their technical feasibility and associated risks, but also the goals of care they were intended to achieve and their ethical significance. Medico-legal aspects were also considered.

Conclusions

This case confirms the importance of shared decision-making between the healthcare team and the parents, as well as the need to evaluate the most appropriate therapeutic strategies on an individual basis, considering the patient's clinical condition, prognosis, and available resources [12]. It also highlights the ongoing efforts in Italy to promote the implementation of clinical ethics consultation within healthcare settings, with the aim of supporting informed

decisions that are consistent with the moral values of patients, families, and healthcare professionals [13]. Furthermore, this experience confirms the effectiveness of a facilitative approach to clinical ethics consultation, in contrast to an authoritarian or directive model. In this case, the ethics consultation provided valuable support to the healthcare professionals involved, preventing them from having to bear sole responsibility for difficult and ethically complex decisions. By shifting attention from individual interventions to the overall goals of care, the consultation process broadened the range of possible alternatives to be considered: interventions aimed at prolonging survival versus palliative care; hospital-based care versus home-based care. Finally, it enabled all parties involved to identify a rational ethical justification for the decisions taken, thereby reducing the likelihood that these decisions would later be questioned or reconsidered. However, this case also revealed several critical issues requiring appropriate interventions, involving both organizational structures and professional culture. In particular, the lack of adequately developed and accessible home-based pediatric palliative care services limited the possibility of offering alternatives to continued hospital management.

Similarly, a professional culture that is not sufficiently oriented toward the identification, analysis, and discussion of ethical issues may hinder the recognition of problems requiring further reflection. Wider implementation of clinical ethics consultation services within healthcare systems would therefore be desirable to promote ethically informed decision-making and contribute to improving the overall quality of medical care. The consultation process also enabled all participants to understand the ethical justification underlying the final decision, thereby reducing the likelihood of future uncertainty or moral distress regarding the course of action that had been taken. At the same time, this case revealed important organizational and cultural challenges that deserve further attention. In particular, the limited availability of structured pediatric palliative care services capable of supporting children at home restricted the range of realistic alternatives to prolonged hospitalization. Likewise, a professional culture that is not routinely oriented toward the identification and analysis of ethical issues may prevent clinically relevant ethical questions from emerging in a timely manner. For these reasons, wider implementation of clinical ethics consultation services within healthcare institutions should be encouraged. Integrating ethics consultation into routine clinical practice has the potential to improve not only the quality of ethical decision-making but also the overall quality of patient care, especially in complex cases involving uncertainty, conflicting values, and high-stakes treatment decisions.

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