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Ethical considerations in pediatric intensive care palliative care: a case study of a patient with Edwards syndrome

Abstract

Introduction: This study addresses Edwards syndrome, a chromosomal condition affecting approximately 1 in 6,000 live births. The syndrome exhibits diverse phenotypic features, diagnosable prenatally or postnatally, with reserved life expectancy. Families face complex psychosocial challenges due to the syndrome's implications, impacting emotional adjustment, treatment decisions, and coping with socioeconomic barriers.

Case presentation: A patient with Edwards syndrome is described, emphasizing the severity of malformations and a multidisciplinary approach. The patient, with severe cardiac malformations, was diagnosed shortly after birth. Due to dependency on hospital equipment, the multidisciplinary team opted, in agreement with the family, for palliative care until death. The patient passed away 20 days after the initiation of palliative care, seemingly free of pain or visible discomfort.

Discussion: The ethical approach in palliative care, especially in Edwards syndrome, involves orthothanasia, aiming to respect the overall well-being and dignity of the terminally ill patient. Effective and empathetic communication, preparation for hospital discharge to allow for death at home, and post-decision follow-up are crucial aspects. The correct practice of orthothanasia involves technical competence and ethical sensitivity, emphasizing a multidisciplinary and transdisciplinary approach.

Conclusions: This case study highlights the importance of palliative care in Edwards syndrome, emphasizing the need for a holistic and compassionate approach considering the physical, emotional, and social needs of the patient and their family. Collaboration strategies, open communication, health professional training, and the implementation of family-centered care are essential to providing effective and compassionate palliative care in these complex cases.

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Keywords: Edwards syndrome, palliative care, orthothanasia, holistic approach, effective communication, professional training

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Introduction

Chromosomal anomalies, categorized as numerical or structural, include trisomies, characterized by an additional chromosome [1]. Trisomy 18, or Edwards syndrome, is one such condition named after its discovery by Edwards in 1960 [1]. It occurs in approximately 1 in 6,000 live births, ranking as the second most prevalent trisomy [2]. This syndrome manifests diverse phenotypic features, impacting various organs and systems [3]. Diagnosis can occur prenatally through ultrasound and invasive tests [2] or postnatally [4]. Survival is reserved, with 30% succumbing within the first month and 90% not surpassing the first year, varying with the severity of complications [5]. Discharge home for these children also varies, with some remaining in the hospital throughout their lives, while others, with multi-professional support and family assistance, manage hospital discharge and maintain care at home.

The psychosocial impact on families of children with severe congenital conditions, such as trisomy 18, is extensive, influencing various aspects of family and social life. Parental emotional adjustment is a significant challenge, involving a range of emotions like distress, worry, loneliness, and hopelessness when confronting their children's diagnoses, medical and surgical procedures, and prognoses [6, 7]. Making crucial decisions about treatment and education, including choices regarding neonatal surgeries and pediatric intensive care unit (ICU) admissions, presents another complex challenge for these parents [8]. Additionally, socioeconomic issues such as low income, lack of access to health and education services, and social barriers are common challenges for families of these children [7, 9]. The complexity of congenital conditions also affects attachment and family relationships, hindering social relationships due to constant attention to the well-being of the children [10]. Comprehensive knowledge about the syndrome and ongoing support are essential for parents and families to manage their children's physical, cognitive, and emotional development and facilitate their social and school integration. Seeking medical attention and psychological support is a constant, often challenging need, especially in regions with limited access to healthcare services [6–10].

Including trisomy 18 patients in palliative care is crucial to ensure a holistic and compassionate approach that considers the physical, emotional, and social needs of the patient and their family. In severe cases requiring hospitalization from birth to death, the palliative care multidisciplinary team needs to work together to provide emotional and psychological support to parents and families, in addition to adequate medical care [10]. A 2016 study emphasized the importance of open communication and continuous support for families of children with trisomy 18, highlighting that family-centered care can significantly improve the palliative care experience [8]. Furthermore, health professional training and education can play a crucial role in the early identification of the need for such care [6]. Therefore, implementing collaboration and communication strategies, along with proper training, can contribute to the provision of more effective and compassionate palliative care for trisomy 18 patients and their families. This case report aims to describe the process used by the pediatric ICU team to address palliative care in a patient with Edwards syndrome from birth and diagnosis to death. The patient's family authorized the acquisition of medical record information and photographic records for the preparation of this article.

Case presentation

The patient, born at term (38 weeks and 5 days) *via* cesarean section, had the umbilical cord clamped after 40 seconds. Positive pressure ventilation was required due to a heart rate below 100 bpm, using a manual T-piece ventilator with a blender. Subsequently, the patient was transferred to the ICU due to continuous positive airway pressure support. At birth, Apgar scores were 5 at the first minute and 8 at the fifth minute, classified as small for gestational age (SGA) and low birth weight (2,420 g). The patient exhibited a male phenotype and morphological characteristics suggestive of trisomy 18, including a head circumference of 30 cm, chest circumference of 27.5 cm, and abdominal circumference of 26 cm. Maternal obstetric history revealed complications due to the use of psychoactive substances and alcohol, along with recurrent suicide attempts and self-mutilation, with support from the municipal Social Assistance Reference Center. This birth represented the third gestation, with the second resulting in a miscarriage and the first in a newborn with similar clinical signs but without a chromosomal diagnosis, dying at 18 days.

Physical examination revealed a syndromic face, cleft lip, omphalocele, and bilateral congenital clubfoot. Additionally, severe cardiac malformations were identified, including extensive atrial and ventricular septal defects, patent ductus arteriosus with repercussion, tricuspid and aortic regurgitation, diffuse left ventricular hypokinesis, and mild systolic dysfunction, diagnosed through malformation screening *via* ultrasound (trans fontanelle and abdominal) and transthoracic echocardiogram. Genetic testing (karyotype) confirmed trisomy of gene 18 (47, XY, +18), solidifying the diagnosis of

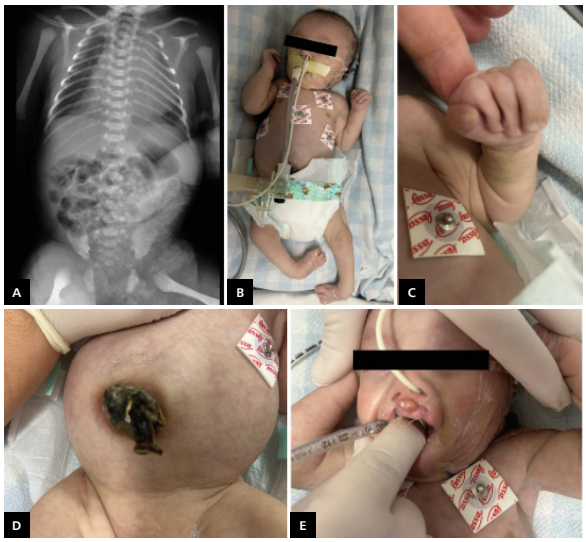

Figure 1. Patient with Edwards syndrome. Whole-body X-ray of the newborn (**A**); image of the newborn with Edwards syndrome showing morphological alterations: characteristic syndromic face and cleft lip, omphalocele, and bilateral congenital clubfoot (**B**); hands of the patient with Edwards syndrome (**C**); omphalocele of the patient with Edwards syndrome (**D**); cleft lip in the patient with Edwards syndrome (**E**)

Edwards syndrome. Given the severity of the condition, the need for orotracheal intubation, irreversible complications from malformations, and dependence on hospital equipment with no prospect of discharge, the multidisciplinary team, including intensive care pediatricians, surgeons, social workers, and other professionals, conducted a meeting with the mother, the patient's sole visitor, at this time the patient was 1 month old. In this meeting, it was mutually agreed that the approach would follow palliative care and end-of-life care guidelines, thus implementing comfort measures until the patient's death. After 20 days of initiating palliative care, the patient passed away, free from visible pain or discomfort. The clinical manifestations of the patient with Edwards syndrome are depicted in Figure 1.

Discussion

A multidisciplinary approach and effective, empathetic communication are crucial in managing patients with trisomy 18 and other complex genetic syndromes. The multidisciplinary team, including healthcare professionals such as intensive care pediatricians, surgeons, social workers, and nurses, is vital to guiding the mother and family comprehensively and emotionally supported, providing clear and honest information about the prognosis and available treatment options [11, 12]. As more than 150 anomalies may be present, Edwards syndrome can have various phenotypes, and the management of each differs due to the diverse needs of each patient [3]. In the case of patients with more severe comorbidities and a reserved prognosis, the presence of professionals skilled in palliative care becomes essential, as these patients may never be discharged home, remaining in hospital care from the first to the last day of life [13].

The ethical approach to palliative care involves orthothanasia, aiming for patients in the terminal phase to face death with some tranquility, respecting their overall well-being and dignity. Other terms used when addressing palliative care are misthanasia, euthanasia, and disthanasia. Misthanasia refers to errors in conduct; euthanasia is the abbreviation of life, a practice prohibited in Brazil, and disthanasia is the implementation of measures that prolong life without benefiting the patient [4]. Correct orthothanasia practice integrates scientific knowledge, requiring technical competence and human and ethical sensitivity in a multidisciplinary and transdisciplinary approach. Ideally, all healthcare professionals should integrate into practice since everyone, dealing with patients in the dying process at some point in their professional careers, needs to have knowledge of management in these situations [4].

The palliative care team must be prepared to address the specific needs of each patient, including shared decision-making and open, empathetic communication with the family about the situation, preparation for hospital discharge to allow death at home, and follow-up after the decision not to proceed with interventions that would not benefit the patient [4, 11, 12]. Orthothanasia practice can begin during pregnancy, as the gestation process of a fetus with a reserved prognosis brings forth anxieties and discomfort beyond those intrinsic to pregnancy. These pregnant women and other family members may perform better biopsychosocially when prepared and supported in the grieving process of the idealized gestation, of the child who may die either during pregnancy or days or months after birth, with the awareness that there is no one to blame for such a situation. These guidelines and work that can be carried out by the palliative care team can bring more comfort and acceptance to both the family and the patient when born, reduce abandonment in hospitals or shelters of these children, and obtain necessary care support when the possibility of hospital discharge exists [4, 13].

Article information and declarations

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Author contributions

ESC — contributed to the conception and design of the study, acquisition, analysis, and interpretation of data, and drafting of the article; NKC — contributed to the conception and design of the study, acquisition, analysis, and interpretation of data, and drafting the article; RMG — participated in the acquisition, analysis, and interpretation of data, and critically revised the article for important intellectual content.

Conflict of interest

The authors declare that they have no competing interests.

Ethics statement

This study was reviewed and approved by the Ethics Committee of Dr. José de Carvalho Florence Municipal Hospital/68078823.0.0000.5451. Written informed consent was obtained from the patient's legal guardian for the publication of any potentially identifiable images or data.

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Supplementary material

None.

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