Junctional Epidermolysis Bullosa-Herlitz Type: two cases involving a palliative care team

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Summary

Junctional Epidermolysis Bullosa-Herlitz Type (JEB-H) is a devastating mechanobullous disease characterized by internal and external blister formation from minor trauma that can ultimately lead to early mortality. Most patients die before the age of 3. Minimal literature exists on pain management and even less on Palliative Care (PC) for these patients. Goldsneider et al. have recommended involving PC to co-treat these patients as a means to improve their quality of life (3). A qualitative study looking at the needs of parents who have lost their child to JEB-H suggested involving a structured network of PC providers (4). However, no literature exists describing the participation of a PC team. We present two cases of JEB-H that demonstrate the involvement of a PC team.

KEY WORDS: palliative care; palliative medicine; pediatrics; epidermolysis bullosa.

Introduction

Epidermolysis Bullosa (EB) is a mechanobullous disease characterized by blister formation from minor trauma. It is classified into three major categories: simplex, junctional, and dystrophic. Junctional EB can be further subtyped into Herlitz or non-Herlitz. Herlitz subtype (JEB-H) is transmitted in an autosomal recessive pattern and characterized by extensive external and internal mucocutaneous blistering that can present at birth and lead to an early mortality. The mean age of death is 5 months, and most die before 3 years (1). Death is often secondary to respiratory compromise due to airway sloughing, failure to thrive, or sepsis (2). Minimal literature exists on pain management and even less on Palliative Care (PC) for these patients. Goldsneider et al. have recommended involving PC to co-treat these patients as a means to improve their quality of life (3). A qualitative study looking at the needs of parents who have lost their child to JEB-H suggested involving a structured network of PC providers (4). However, no literature exists describing the participation of a PC team. We present two cases of JEB-H that demonstrate the involvement of a PC team.

Case Reports

Patient 1

A 6-day-old female born at 36 weeks gestation with presumed epidermolysis bullosa was admitted for worsening blisters. Dermatology made wound care recommendations and performed a punch biopsy. Nutrition was consulted for caloric intake recommendations, given her increased metabolic requirements as a result of her extensive skin lesions. She was discharged home with instructions to follow up with her Primary Care Physician (PCP) and Dermatology. Biopsy results were pending upon discharge. She had one routine examination following discharge at 12 days of life, which demonstrated adequate weight gain. Parents followed up with Dermatology but did not continue follow-up with PCP, due to financial constraints. This patient had multiple outpatient visits with Dermatology for wound care management. Biopsy confirmed junctional EB at 29 days of life. At 43 days of life, Dermatology noted disease progression as evidenced by weight loss and hoarseness. At 47 days of life, Dermatology consulted PC to address pain control and limitations of care. At 50 days of life, the PC team coordinated an outpatient visit with Dermatology and Pediatrics, resulting in Pediatric Intensive Care Unit (PICU) admission for aggressive pain management, severe dehydration, and failure to thrive. Upon admission, the family opted for full, aggressive care. Her course involved multiple procedures and interventions including a fiberoptic laryngoscopy, central line placement, total parenteral nutrition, and high-flow nasal cannula. The PC team assisted with pain management utilizing an opioid (morphine) and engaged the family in advance care planning. By hospital day 7, the family “did not want to put her through anything that would hurt her and may not help her”. Limitations of care were subsequently established at 57 days of life, and thus a do-not-resuscitate/do-not-intubate order was placed in the chart.
She died 5 days later in the PICU after her parents chose to withdraw high flow nasal cannula. Of note, genetic studies confirming JEB-H did not come back until after the patient died.

**Patient 2**

A 3-day-old male was transferred to our NICU with extensive mucocutaneous blistering concerning for EB. The NICU consulted the PC team at 17 days of life for family support while awaiting biopsy results. Given the severe phenotype, the PC team made early recommendations for pain management including the initiation of an opioid (morphine) and gabapentin (for neuropathic pain and pruritis) (5). The PC team also coordinated multiple interdisciplinary team and family meetings to discuss treatment options, including experimental therapies (bone marrow transplant), and to address goals of care. At 17 days of life, biopsy confirmed junctional EB. At 34 days of life, his family opted for comfort care only and limitations of care were defined. The family did not want their child to endure any procedures or interventions, thus no tubes or lines were placed. Given the family’s goals of care, the PC team explored options including discharge home on hospice or to a facility. Parents chose to keep him in the hospital because of their limited ability to provide the level of skin care in the home that he required. It was not until 134 days of life that genetic studies confirmed Herlitz subtype. Throughout his course, the PC team provided recommendations regarding medication titrations to ensure comfort and reduce pain associated with dressing changes. Medications administered included opioids (morphine, hydromorphone, methadone), neuropathic agents (clonidine, ketamine, gabapentin), and benzodiazepines. During the last few days of life, palliative sedation was initiated (dephenhydramine, phenobarbital, pentobarbital prn, fentanyl). Of note, all medications were given via oral route, including palliative sedation, in accordance with the family wishes to avoid needle sticks and other painful procedures. At 162 days of life, he died comfortably in the hospital on the General Pediatric floor with family members at the bedside. Given the rarity of EB, most staff had not seen this devastating disease previously. Staff struggled with dressing changes, visible wasting, and had a constant fear of the patient enduring an acute airway obstruction. The PC team met regularly with the staff to address their concerns as well as provide anticipatory guidance and support, given his prolonged hospital course.

**Discussion**

Junctional EB-Herlitz type is a life-threatening diagnosis that causes significant pain and suffering. It may take several weeks to confirm the diagnosis of junctional EB and several months to confirm Herlitz subtype. Early diagnosis of JEB-H is crucial in helping families engage in discussions regarding goals of care to facilitate decision making and potentially shift their focus from life-prolonging measures to comfort care (6). These cases demonstrate the value of involving a PC team as a way to co-manage these patients in conjunction with the primary team while awaiting biopsy results. Because there is such variability in phenotype for patients with JEB-H, there is also a wide range in pain severity. Therefore, pain management needs to be individualized (3, 7). A PC team can offer expertise in the evaluation and management of pain. Patient 2 had a more severe phenotype upon presentation. For this patient, the PC team helped the primary team early on by providing recommendations on the titration of opioids and the addition of neuropathic agents and benzodiazepines.

A PC team can also help facilitate communication amongst providers and with family members, whether the patient is in the hospital or at home. As soon as PC was consulted for patient 1, the team bridged communication between the PCP and Dermatology, ultimately expediting her hospital admission. PC clinicians can lead discussions focusing on goals of care, which aim to identify parental preferences based on the patient’s medical condition, treatment options, and prognosis. Having these discussions can help the medical team more easily partner with the family in making decisions for the patient. Given the strong clinical suspicion of JEB-H in patient 2, the PC team was consulted earlier in his disease trajectory, thereby leading to earlier discussions regarding prognosis, treatment options and parental preferences. Having these conversations early on enabled the family to make important decisions and ultimately avoid of unwanted interventions. These cases offer concrete examples of patients with JEB-H being co-managed by a PC team. PC teams improve overall care for patients with JEB-H by providing expertise in effective pain and symptom management and facilitating communication between families and providers. Communication needs may include identifying goals of care, engaging in advance care planning, providing decision-making support, and facilitating discussions between providers and families or amongst providers alone. Palliative care teams help providers and families focus on optimizing the very limited time with a child affected by JEB-H. We believe offering these interventions early will maximize quality of life and reduce patient and family suffering.

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