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A Parent's Journey: Incorporating Principles of Palliative Care into Practice for Children with Chronic Neurologic Diseases

Allyson Brown, MPA,[*,†] and Jonna D. Clark, MD, MA[†,‡]

Rather than in conflict or in competition with the curative model of care, pediatric palliative care is a complementary and transdisciplinary approach used to optimize medical care for children with complex medical conditions. It provides care to the whole child, including physical, mental, and spiritual dimensions, in addition to support for the family. Through the voice of a parent, the following case-based discussion demonstrates how the fundamentals of palliative care medicine, when instituted early in the course of disease, can assist parents and families with shared medical decision making, ultimately improving the quality of life for children with life-limiting illnesses. Pediatric neurologists, as subspecialists who provide medical care for children with chronic and complex conditions, should consider invoking the principles of palliative care early in the course of a disease process, either through applying general facets or, if available, through consultation with a specialty palliative care service.

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Introduction

Treating children with complex and incurable life-limiting conditions presents unique challenges in providing optimal medical care. Although advances in medicine have led to cures for historically fatal diseases, and supportive therapies have dramatically extended the duration of life for children with complex conditions, there remain many disease processes that have a high degree of prognostic uncertainty. Even when prognostication is less uncertain, curative therapies are not always available. During the search for a diagnosis and the "miracle" cure, the life of the patient becomes filled with frequent hospitalizations, consultations with a myriad of health care providers, innumerable diagnostic tests, and extensive trials of therapies. Although this extensive evaluation is necessary, equally important is a holistic approach to addressing the patient's and family's needs including physical, emotional, and spiritual well-being. As complementary to the curative model of care, the practice of pediatric palliative care provides tools to assist with medical decision making, reduce disease burden, and improve the quality of life, helping families to maintain hope, regardless of whether the child dies.[1-7] The following case-based discussion, through the voice of Allyson Brown, the mother of Hannah, a child with refractory epilepsy, demonstrates how the fundamental principles of palliative care work to improve the quality of life for children with life-limiting illnesses and their families.

Case Report

Hannah is a 9-month-old girl with intractable epilepsy, axial and appendicular hypotonia, intermittent choreathetoid-like movements of her hands and tongue, progressive microcephaly, and severe global developmental delay. She was delivered at 39 weeks' gestational age via normal spontaneous vaginal delivery with Apgar scores of 9 and 9 following an uncomplicated pregnancy. At 4 hours of age, she suffered from frequent generalized tonic-clonic seizures, requiring endotracheal intubation in the neonatal intensive care unit for respiratory failure. Workup included negative results on infectious evaluation, normal findings on brain magnetic resonance imaging (MRI), and normal electroencephalogram (EEG) results. She was treated with

From the *Department of Pediatrics, Division of Critical Care Medicine, University of Washington School of Medicine, Seattle, WA.
†Department of Pediatrics, Seattle Children's Hospital, Seattle, WA.
‡Treuman Katz Center for Pediatric Bioethics, University of Washington School of Medicine, Seattle, WA.
Address reprint requests to Jonna D. Clark, MD, MA, Seattle Children's Hospital, Critical Care Medicine FA.2.112, 4800 Sand Point Way NE, Seattle, WA. E-mail: jonna.clark@seattlechildrens.org

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phenobarbital and was discharged at approximately 3 weeks of life.

At 6 weeks of age, she was readmitted critically ill to the pediatric intensive care unit with a severe lactic acidosis, hyperglycemia, and acute respiratory failure. Infectious and metabolic evaluations revealed no etiology for her acute decompensation. Initially she responded well to supportive therapies; however, she developed worsening seizures, requiring the addition of levetiracetam to phenobarbital. A repeat MRI revealed potentially delayed myelination with an elevation in lactate level on spectroscopy. Additional EEGs showed normal findings. She was discharged at 2 months of age. Developmentally, she had a social smile, was able to bring her hands to midline, and appeared to be gaining appropriate head control.

Over the next 8 months, Hannah developed intractable epilepsy with progressive global developmental delay, requiring more than 80 hospital visits, including overnight stays and several intensive care admissions. She was placed on a multitude of additional antiepilepsy agents, in addition to a ketogenic diet, with minimal success. Her neurologic examination revealed intermittent horizontal nystagmus, dyskinesia with choreathetoid-type movements of her hands and tongue, and diffuse axial and appendicular hypotonia with hypokinesia. Developmentally, she had regression of her milestones, was unable to fix on or follow objects, lost her social smile, had minimal verbal communication with cooing, had poor head control with an inability to roll over, and lost her ability to bring both hands to midline. Her growth curve revealed worsening microcephaly.

Extensive neurologic, metabolic, and genetic evaluations were performed without revealing a unifying diagnosis. Repeat EEGs revealed worsening abnormalities, including findings of status epilepticus (Table 1). Repeat MRIs of the brain showed normal findings.

As incredible advocates, throughout the course of illness, Hannah's parents sought consultation with numerous subspecialists, retaining the glimmer of hope that a unifying and potentially treatable disease process would be identified. However, as each diagnosis was ruled out and each subspecialist no longer offered a curative mentality toward the disease process, Hannah's parents expressed concern regarding Hannah's quality of life. They recognized that her underlying disease process was progressive and remained hopeful that Hannah would be able to live her life to its fullest, cherishing each moment with her.

At 9 months of age, the Pediatric Advance Care Team (also known as palliative care team) convened an interdisciplinary group to discuss and prioritize the goals of care for Hannah. Her parents expressed that although Hannah was delayed, eating and breathing without artificial interventions gave her great pleasure. Out of extreme love for her, they determined that if Hannah lost the ability to eat or breathe independently, they wanted to prioritize her comfort and optimize her quality of life, acknowledging that the duration of her life would be limited. Following referral to an outpatient palliative care and hospice program, through consultation with the neurology team and her primary care provider, she continued to receive medical care both at the hospital, including the intensive care unit, and at home. As her disease progressed with the inability to pass stool, urinate, and feed without assistance, at the age of 16 months, she peacefully passed away in her mother's loving arms in the comfort of her home.

An autopsy revealed severe microcephaly (head circumference of 41 cm, smaller than fifth percentile); micrencephaly (weight 618 g, expected 1010 g); diffuse cortical, cerebellar, and spinal white matter gliosis and atrophy with intact myelin; patchy cortical gray matter and subpial gliosis; and patchy severe gliosis and atrophy of the caudate,

Table 1 Metabolic and Genetic Evaluation (all results negative unless indicated)

Plasma amino acids
Urine organic acids
Acylcarnitine profile
Urinary purine panel
Very long chain fatty acids (X-linked adrenoleukodystrophy)
Plasma alpha-aminoadipic semialdehyde (pyridoxine dependent seizure disorder)
Plasma piperideine-6-carboxylic acid (folinic acid responsive disorder)
Coenzyme Q10
CSF pyridoxal 5-phosphate
Ceruloplasmin (Wilson's disease)
Muscle biopsy- electron transport chain
Whole blood mitochondrial point mutation analysis
CGH micro-array analysis- *small 9p24.3 duplication of unclear significance*
POLG1
PCDH 19 (epilepsy and mental retardation limited to females)
PNPO (alternating hemiplegia)
SCN1A (sodium channel mutation associated with severe myoclonic epilepsy of infancy and generalized epilepsy with febrile seizure)
CACNA1A (calcium channel mutation associated with familial hemiplegic migraine and ataxia)
ATP1A2 (genetic mutation associated with familial hemiplegic migraine)
DHCR7 (7-dehydrocholesterol reductase) (Smith-Lemli-Opitz)
CDKL5