Benefits of Early Referral to Pediatric Palliative Care for a Child With a Rare Disease

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Myhre syndrome is a rare connective tissue disorder. Signs and symptoms include fibrosis of the skin and internal organs (heart, lungs, gastrointestinal system), intellectual disability, distinctive facial features, and skeletal abnormalities.[1,2] Myhre syndrome is caused by a mutation in the *SMAD4* gene. It typically occurs for the first time in an affected person.[1] A clinical case is described in a child whose family received a diagnosis of Myhre syndrome after he died. Although for some families the specific diagnosis is important, this mother did not feel that the information would have changed the course of her child's life. We outline the benefits of palliative care supporting the child and family with attention to individualized symptom management, improved communication, and support making difficult decisions.

PARENT NARRATIVE

My son Connor had an undiagnosed condition affecting several body systems including his airway, his heart, and his development. He also had subtle features including small hands and feet and skin that appeared thicker or fluid-filled. Although he was managed by an excellent pediatrician, the silos of medical care in our community at the time meant that crises were addressed by multiple different people on different teams, each of whom had an incomplete knowledge of Connor's specific medical issues.

The research program (Care4Rare)[3] at the Children's Hospital of Eastern Ontario in Ottawa, Canada, performed whole exome sequencing of Connor's DNA. They identified a mutation in the gene *SMAD4*. This specific change (c. 1499>C, p.Ile500Thr) has been seen in other children with a multisystem condition known as Myhre syndrome; from a genetic standpoint, this was molecularly confirmed as the cause for Connor's differences.

These results, however, came exactly 5 years after Connor's death.

When you have a child born with differences, you are thrust into a journey of "fixing" what can be surgically repaired and focusing on the differences to identify ways to improve development. Your child is managed by subspecialists for each system of the body in which differences occur, more than 15 in Connor's case.

He didn't have a unifying diagnosis to provide some explanation or anyone interested in finding that answer or investigating. I felt alone in the management of his care and quickly became the medical expert on all things Connor. Connor was medically fragile, aptly described by his first cardiologist as "a boy who didn't read the textbook," and from whom we all learned to expect the unexpected.

PHYSICIAN NARRATIVE

When I think of Connor, the image that comes to mind is that of standing near the island in my kitchen, talking on the phone with his mother, Mindy. It was

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often in the evening, after dinner but before bedtime. Mindy was always apologetic about calling ("Sorry, Dr Chris!"), but the worry in her voice was obvious. I mostly provided reassurance that the things she was doing were the right things. We spoke when Connor wasn't tolerating feeds, or seemed uncomfortable, or wasn't settling for sleep, or had changes in his breathing.

PARENT NARRATIVE

Connor had episodes in which he would stop breathing, turn pale or blue, have a gray protruding tongue, and would go cold as ice in his extremities. Doctors wondered if these were seizures or cardiac spells but offered no form of treatment or plan to prevent the episodes. Our working assumption for Connor was that these were pulmonary hypertensive crises that he for some reason was surviving.

At age 5, after a lengthy stay in the PICU, we were referred to the pediatric palliative care (PPC) team and Roger's House (now Roger Neilson House).[4] We were told by the acute care team that all that could be done for Connor was being done and that whether he pulled through this latest crisis was up to him.

We had a new team and the hope that they could make a difference for Connor. We were still reeling from the disappointment of nothing having been done to help him up to this point, however, and couldn't shake the feeling of being "passed on."

PHYSICIAN NARRATIVE

The primary team caring for Connor was reluctant to refer him to the PPC team. They weren't sure what was wrong, but didn't think he was going to die, at least not yet. When the role of palliative care in children was explained (support any time of the day or night, symptom management, help with decision-making, respite for the child and family with high medical care needs), a referral was made, but there was an element of surprise that the team would manage him. From our team point of view, Connor was fragile, but it was initially unclear whether he would get better or worse. Our approach is to accept the child and family to be managed but review the case periodically. If the child improves, we will sometimes "sign off" with the agreement that if the medical situation deteriorates, we would reassess the need for our involvement. More often the child remains fragile or begins to deteriorate, and we continue to support the child and family (and primary team). The family has become the expert medical caregivers and the advocates for their child but benefit from support when they are not sure that what they are doing is the right thing.

PARENT NARRATIVE

Connor was so much more than his medical chart. He was a little boy who lived each day to the fullest. The PPC team recognized this in him through their observations as well as extensive discussions with us as to who Connor was and what we wanted for him. At the time, our focus was on finding a way to help him with the episodes we feared would take his life. The only clarity was the toll the episodes were taking on our family mentally. Having no idea of their physical impact on Connor, I wondered each time if this was the way he would die.

Everything would change for Connor and our family after a call to the palliative care doctor on call. Connor had begun breathing again albeit shallowly. When she arrived, the doctor would inject what we referred to at the time as a "rescue med" (morphine) intended to help relax Connor's breathing, and we waited to see the effect. I thought, finally someone is trying something. I was grateful. The decision to try that medication led to its use prophylactically and the management of these episodes in Connor.

I wanted someone from the medical community to show an interest, to see our suffering and act. I needed someone who was hands on, intimately knowledgeable about my son, and interested in his long-term quality of life across all domains including school. I wasn't looking for a diagnosis or even prognosis, just a plan to help Connor to live his best life despite his differences.

PHYSICIAN NARRATIVE

In caring for children with life-threatening conditions, we get to know families of children with rare diseases. Their child may be 1 of only 100 children in the world, or 1 of 10, or the only living child with a particular genetic difference. For some families it is important to have a name, even if it is rare. Coming to an emergency department in crisis with a name of a syndrome somehow makes it easier to navigate the system, even if no one has ever heard of that syndrome. For other families, finding a name or a genetic difference or a cause is important for future family planning or for prognostication. There are also parents for whom finding a name or cause is nice, but it doesn't change their day-to-day (or minute-to-minute) life much at all. Symptom management is a key component of the care we add, and we have expertise in use of medications that many deem "too strong" for fragile children. Morphine is a mainstay of our symptom management, both for pain control and for relief of dyspnea. This is despite the many voices that tell parents that morphine is "too strong" or "too dangerous" or "will stop them from breathing."

PARENT NARRATIVE

The answer I was looking for was found in PPC, not in primary care or the acute care system that was wonderful at being reactive but failing to support, treat, or prevent these episodes. The answer wasn't found in the diagnosis; we had none.

The PPC team focused on what we needed: what Connor needed each and every day and especially when changes in his health status occurred. Because of their attentiveness and our trust and mutually respectful relationship, difficult conversations were possible. Discussions with the PPC team included helping us choose when to intervene and when we would not. We were dedicated to ensuring that Connor died on his terms, when his body no longer had the reserves to continue, and without machines that prolonged his suffering.

PPC does not always mean imminent end of life. It is reliable support for the child and family throughout the life of the child, focused on trying to live every moment to the fullest. It could be the end of suffering through pain and symptom management. It could mean the end of isolation by meeting other families who "get it." It could mean you won't have to do this alone and the 24-hour, on call supportive care you receive from the PPC team extends beyond the hospice to your home, community, hospital, or school. It could mean the end of worry and fear and the peace of the present moment. All of this was true for our family.

Diagnoses may be delayed for many reasons. In 1 sense it doesn't matter. Connor lived and died without knowing what "it" was called. Labels may allow access to funding or may not. At some point you have to move past the "what" to the present moment. When you live life 1 day at a time, the name of your overarching illness, especially if it is rare, doesn't offer the same comfort as an around the clock team supporting you with day-to-day struggles. ". . . Though pediatric palliative care is underpinned by the same philosophy as adult palliative care, children who have life-limiting conditions and their families have particular needs that distinguish them from users of adult palliative care. For example, at a physical level children are more likely than adults to have non-malignant conditions that follow trajectories in which children oscillate between feeling relatively well and acutely unwell . . ."[5] This was particularly true in Connor's case, making the most important factor our team that supported us continuously.

Connor's outcome would not have been much different if genetics had managed to diagnose him before his death. It may have prevented more surgeries, intubations, and anesthetics, but maybe not. Our palliative care team gave us the advice the studies I have read about Myhre would have provided. Observations made by the PPC team that he was difficult to ventilate and that we should think seriously before putting him under ultimately helped us to decide not to do any elective surgeries on Connor again.

CONCLUSIONS

This case is described to provide a voice for families who are caring for a child with an undiagnosed or life-limiting illness and especially for the families who won't consider PPC either out of fear or not knowing it exists. Policy and funding frameworks need to support the reality of PPC and our home care system needs to be flexible to the fluctuating needs of children or the reality will be that services are not there when required, and we could miss supporting a family at home when that had been their wish all along. Physicians owe it to their families to become educated about PPC. In the relationships physicians create with their patients and families, they should have a clear understanding of what the family needs in terms of support.[6] If that is something better suited to the PPC team, it is important to offer the option.

ABBREVIATION

PPC: pediatric palliative care

REFERENCES

1. Starr LJ, Lindor NM, Lin AE. Myhre syndrome. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews*. Seattle, WA: University of Washington, Seattle; 2017. Available at: https://www.ncbi.nlm.nih.gov/books/NBK425723/. Accessed August 30, 2017
2. National Organization for Rare Disorders. Myhre syndrome. 2015. Available at: https://rarediseases.org/rare-diseases/myhre-syndrome/. Accessed August 30, 2017
3. CHEO Research Institute. Care for rare. Available at: www.cheori.org/EN/care4rare. Accessed August 30, 2017
4. Vadeboncoeur CM, Splinter WM, Rattray M, Johnston DL, Coulombe L. A paediatric palliative care programme in development: trends in referral and location of death. *Arch Dis Child*. 2010;95(9):686–689
5. Kang TI, Munson D, Hwang J, Feudtner C. Integration of palliative care into the care of children with serious illness. *Pediatr Rev*. 2014;35(8):318–325; quiz 326
6. Ekberg S, Bradford N, Herbert A, Danby S, Yates P. Healthcare users' experiences of communicating with healthcare professionals about children who have life-limiting conditions: a qualitative systematic review protocol. *JBI Database Syst Rev Implement Reports*. 2015;13(11):33–42

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