RARE CONDITIONS AND DISEASES

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Issue Topic: Rare Conditions and Diseases

Welcome to the 74th issue of our Pediatric e-Journal. In this issue, we are focusing on rare conditions and diseases that are likely to be confronted in pediatric palliative and hospice care. As we delved into this subject area, we learned there is a very broad category of challenges that can confront children, families, volunteers, and professionals who work in pediatric palliative and hospice care. We do not, therefore, expect a single issue of our e-Journal will cover every possible type of rare conditions and diseases. However, it is our hope to guide readers in many important ways of addressing these issues.

This e-Journal is produced by the Pediatric e-Journal Workgroup and is a program of the National Hospice and Palliative Care Organization. The Pediatric e-Journal Workgroup is co-chaired by Christy Torkildson and Melissa Hunt. Chuck Corr is our Senior Editor. Archived issues of this publication are available at www.nhpco.org/palliativecare/pediatrics/.

Comments about the activities of NHPCO's Pediatric Advisory Council, its e-Journal Workgroup, or this issue are welcomed. We also encourage readers to suggest topics, contributors, and specific ideas for future issues. We are open to suggestions for our 75th issue in May 2024 and for the other two issues to follow later this year. If you have any thoughts about potential topics for these future issues and/or potential contributors (including yourself?), please contact Christy Torkildson at Christy.Torkildson@gcu.edu or Melissa Hunt at Melissa.Hunt@optum.com.

Views expressed in this and other issues of the Pediatric e-Journal are exclusively those of the authors and do not necessarily reflect the views of the Pediatric e-Journal Workgroup, the NHPCO Pediatric Council, or the National Hospice and Palliative Care Organization.

Produced by the Pediatric e-Journal Workgroup

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What I Learned from My Girl

Melonie Stothers,
Mom to Izzy (and Phineas)

I was asked to share my experiences and lessons arising from living with a child with a rare condition and experiencing her death. I don't know if the lessons came from Izzy dying from a rare condition or simply from caring for a very sick child for a very long time, helping her come to terms with her terminal condition, all while trying to balance taking care of our other child, maintaining a marriage, working full-time, and staying sane. Either way, I learned so many things about myself, my amazing family, and about dying that I could write an entire book. Instead, today I share these four points that seem to cover it all:

- I learned that the truth hurts, but acknowledging it, helps you heal.
- I learned that true strength comes from having every reason to be angry at the world yet choosing instead to smile and see the wonder and beauty all around.
- I learned that even when you know what's coming, you can't be prepared for the ache of losing your child.
- I learned that you can't put the rest of your life on hold while searching for answers and caring for your sick child because your life might not be there when you come back and you'll need something to come back to when faced with the void left by your child's death.

I am a lawyer by trade. I excel in research and finding needles in haystacks. It's what I do. When Izzy was diagnosed in utero with heart defects and a chromosomal deletion and when she was later diagnosed with pulmonary hypertension, I researched as much as I could on the conditions. With each nugget of information we learned about Izzy, I researched. I spent years learning that her specific combination was extraordinarily rare. I read medical journals, scoured the internet–blogs, carepages, facebook groups searching for clues about other babies and kids who may have the same conditions. I wanted to know they were okay. When we were told Izzy had reached the edge of science, I went into manic research mode. I contacted every specialty center in the country. I requested opinions from any medical team that would listen. We traveled across the country where we were told heart failure just happens in some kids like Izzy at some point and the medical community did not know why.

Admitting Izzy to hospice meant giving up my deep research in finding answers and cures for her heart. I still trolled the medical journals and I spent my spare time raising money to fund medical research, but I had to let go of the control freak inside of me yearning for answers because there were none for me to find. Izzy's specific set of conditions were too rare for the research she had needed for a "cure" to be funded.

It was one thing to know that my daughter would not be healed—that she was going to die. It was quite a different thing to acknowledge that truth, but once I did, I was able to grieve for the loss of what I had wanted for her life. It helped me shift gears to concentrate on what Izzy could do in her condition. And Izzy did pack as much as she could into her short 15 years. She had more best day evers than anyone anywhere. We tried to help her enjoy every moment that she could and we were able to do that by acknowledging the truth about her health.

As Izzy matured and grappled with her own mortality, she got angry often at the limitations of her body, at medical science unable to fix her, and at all of us who took care of her for not doing more. I cannot even begin to estimate the number of times I had to explain to Izzy that we tried, we searched, we scoured the globe for answers, and there was nothing more to be done. Even though all of these facts about her life made her angry and sad, Izzy did not let those limitations define her. She still smiled. She still danced even if she was stuck in bed. She would hold the anger and grief long enough to learn from it for herself, but then would inevitably and always decide she wanted her time here to be pleasant. Nobody would have blamed her for being angry and sullen all of the time, but she chose kindness, caring, love, and laughter even when she was in constant pain. My Izzy taught me true strength.

As we shared Izzy's final days, living out the scenarios we had prepared for and planned with Izzy, I was grateful for the planning. We had promised Izzy we would read to her at the end even if she was not awake and we did. We fulfilled all of Izzy's requests for her funeral and it was so beautiful. I'm beyond grateful that we were able to have it planned and that Izzy helped in so many ways. But despite years of planning, innumerable conversations, and lots of support from family, I was not prepared for the ache of not having my girl with me.

The only solace I have these days, now that Izzy is gone, is that we put the work into our marriage, our family, and our careers while Izzy was with us. My husband and I spent years in counseling, helping us come to terms with Izzy's terminal illness and how that affected us individually and in our marriage. We have that now as a foundation for helping us move forward together while we grapple with the screaming void left by the loss of Izzy.

My husband and I both worked while Izzy was ill. We did hybrid work from home and the office when she was here so that we each got time with her and each had time to continue our work. It helped us continue to pay the bills, maintain health insurance, and not force either of us to give up time with Izzy. It was extraordinarily hard to do. But with the loss of Izzy, we still have work to return to. Going to the office is weird, but also comforting because there, I know what I'm doing and what is expected of me. So much of my life these days is spent trying to figure out who I am without Izzy here. But at work, I know who I am. Izzy is at work with me (as she always was in my thoughts and heart), but work is one corner of my life that was not totally defined by Izzy's life so I do not have to redefine that part of me because of her death. That is a solace.

Life is Too Short

Rachel Henderson,
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Yesterday I started my day with a one-year milestone session in the studio. Mama asks me to please keep his scar in his photos—the one from his chemo port, she explains, from the chemo treatments he has been through for exactly half of his life. Do you remember the lump on his neck at his newborn session, she asks. And I do. I remember how soft it was, at the base of his neck, and how delicately I handled him. It doesn't hurt him, she says. But I handle him so gingerly...just in case. It was nothing, the doctors had said. We can remove it when he's old enough to be more safely under sedation, at six months old, they said. And at six months old he started chemo for the cancer that had grown there even before his birth. In the midst of my shock, she quickly announced his cancer is now officially in remission, just declared this week! We celebrate together, marvel at how big he is, how happy he is, how strong he is. His hands grasp firmly onto my fingers as he breaks out into an assisted run, laughter pealing throughout the studio, as we head from spot to spot. I tell his mama how strong he is, but how strong SHE is. How I see her, truly see her, and all she has been through. With my first baby at that, she whispers.

Life can be so short. I cling onto the hope that his will be ever so long.

I start to clean up smeared cupcakes and icing from the floor. I pull out a gown—a gown I've been eyeing for a while, but have shied away from the cost. Until I had the most compelling reason to take the plunge. A mama, with her baby, who was taken far too soon. I prepare the set for this next session, one to honor a mother, her partner, and her womb. Empty as it should not be. We honor it as it should be.

She carries in the tiniest urn I have ever seen in person. Her son. I pick him up gently, so slowly. I call him by his name. I have held many babies in my hands. Many babies who have departed before I arrive for photos. Babies at 39 weeks, babies at 20 weeks. But he is my youngest, at just 15. We take the photographs that honor mama as she should be. In a beautiful gown, cradling his first home in her womb. Cradling him against her. We take all of the photos that should've had him cradled in her rounded stomach. Cradled in the warmth of her arms.

Life is too short.

I clean up and head home to prepare for my third session of the day. A mama I was connected with because of my many years of experience working with families like hers. A mama whose little girl has a .005% chance of survival. She arrives to our location and steps out of her car, the biggest belly I've seen in a while. I wonder how many people stop her and ask questions about how excited she is. How far along she is. What she's having. She is having a little girl, her second, and what do you say after that? She is quiet and doesn't quite know what to say. These are my first professional photos, she tells me. I tell her not to worry, that I'll walk her through every step of the way.

Halfway through she laughs and tells me her little girl—Maddie, she tells me—is pushing her bottom up high. She pats her stomach and says right here, and she's moving a lot, too. I claim that she must be excited to have her photos taken. We share a laugh. And I watch mama's eyes swell with tears. We dance with her little girl, 2.5 years old, and tell her to give her sister kisses. I keep saying her name. Maddie. Maddie. Maddie. The same as one of my own.

After photos are completed we walk and talk together. We discuss the many different outcomes I may be photographing next. They realize how far my experience goes and ask me a dozen questions. They want to know about my other families. They want to know how I can do what I do. It's simple, I say. I hold all of my babies as though they are any baby in my arms. I dress them. Brush their hair. Talk to them. There is no difference if breath is

in their lungs. That is their baby, and I am honored to hold them and love them. The hard part, I say, is what the families endure. The grief in the room, that is what I struggle with. They want to know the truth of how I feel, and I describe it. I see mama's face and ask if I can give her a hug. She clings onto me, hard. She presses her face into me and I feel her tears. I cry myself, and quickly apologize—they tell me it brings them comfort that I feel it all so deeply. They ask me every question they can think of. I answer them all. Mama looks at me and says she just keeps thinking...what if she makes it? I say mama, we are going to hold on to that .005%. We prepare for it all, but we can still hold onto hope. She asks what photos I will take if she makes it. I tell her I'll be there for her at her birth. I'll be there in the NICU if she beats the odds. And I'll happily be there to photograph her homecoming, too. The doctors have said if she even makes it to a live birth, she may only be there for mere seconds. But .005% is not 0%.

I ask mama and dad if they would like me to share their photos with the world. Maddie's dad looks at me with tear filled eyes and says he wants everyone to know her, and to know her story.

I'm rambling, because it's the only way I know to process things sometimes. I'm sharing, because these children deserve to be known. Talked about. Loved. These families deserve to be known. The trauma, the pain, the grief...but also the love they have for their sweet children. I have often heard parents say that one painful thing about losing your child is that people shy away from talking about them, when talking about them is what keeps them alive in a sense. I'll never forget that. I'll never forget them.

Integrating Principles of Pediatric Palliative Care in Children with Rare Conditions

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This excellent article by Bridget McCrate Protus, PharmD, listing the range of relatively rare severe pediatric conditions, describes the setting in which pediatric palliative care principles are often impactful and appropriate. Why would the principles of pediatric palliative care (PPC) be appropriately followed soon after determination of a severe or chronic or potentially life-limiting condition? Why not wait until more information about diagnosis and prognosis is available? The answer is that PPC principles, including decision-making support, psychosocial, emotional, and spiritual support of the child and family, management of pain and symptoms, a focus on quality life, and logistical support for the family, are all appropriate regardless of diagnosis or prognosis. When the diagnosis and/or prognosis, are uncertain, decision-making needs to be "fluid" and be adaptable when more data are available. In the situation of uncertainty, a time-limited trial of treatment, possibly with a back-up plan, would likely be appropriate. Even when the diagnosis is more certain, there is still often a range of prognoses. It is better to provide **time-limited** supportive treatment with defined success and failure outcome measures, rather than withhold potentially life-saving treatments in the setting of uncertain prognosis.

As a neonatologist, I provided many prenatal and post-birth consultations. I informed families about how our joint decision-making would likely proceed. I emphasized the need to support the newborn as we gathered the information to determine the **range** of treatment options that were medically and ethically appropriate. As more diagnostic and prognostic data were available, the treatment options often changed.

This is a stressful time for the child, the family, and the medical and nursing teams. Understanding and integrating the principles of PPC early in the course of the condition when diagnosis and prognosis may be uncertain are important to the care team and the family.

Brief Summary of Some Life-Limiting Conditions

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Anencephaly is a neural tube defect (NTD) in which a baby is born without parts of the brain and skull. NTDs happen within the first 4 weeks of the pregnancy as the neural tube forms but fails to close properly. When the upper neural tube has incomplete closure the brain, skull, and scalp do not develop. The forebrain and cerebrum are absent and remaining brain tissue often remains exposed. Infants born with anencephaly are usually blind, deaf, unconscious, and unable to feel pain. The baby may exhibit reflexive actions such as breathing or response to touch. The cause of anencephaly is unknown and there is no standard treatment or cure. If the infant is born alive, the prognosis is very poor with a life expectancy of hours to days.

Batten disease is a fatal, inherited disorder of the central nervous system, also known as juvenile neuronal ceroid lipofuscinosis (JNCL). Children with Batten disease may appear to develop normally but begin to display early symptoms between the ages of 5 and 10 years. Common presentation may include vision changes, seizures, personality or behavior changes, clumsiness, or stumbling. These symptoms become increasingly severe with progressive cognitive impairment, loss of sight, and loss of motor function. Batten disease is usually fatal by late adolescence. Because vision loss is an early sign, the optometrist or ophthalmologist may be the first clinician to suspect Batten disease, as the loss of cells in the eye are noted on exam. Neurologists usually diagnose JNCL based on additional testing of blood, urine, skin samples, electroencephalogram (EEG), and computed tomography (CT). There is no cure for Batten disease but a recently FDA-approved therapy, cerliponase alfa (Brineura®) may delay loss of ambulation in patients age 3 years and older with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2). Anti-epileptic medications are used to control seizures; other symptoms are managed as they arise. Physical therapy can help maintain the child's functional level for as long as possible.

Cortical dysplasia, or focal cortical dysplasia (FCD), is a malformation of cortical development and the most common cause of refractory epilepsy in pediatrics. Epilepsy is the main symptom of FCD and early, severe seizure onset is associated with intellectual disability. FCD also leads to cognitive impairment and other neurological problems. FCD can refer to a wide range of cortical malformations which are classified by severity and timing of development in embryo. FCD can be grouped into 3 main types: Type I has milder, later onset, changes seen in the brain's temporal lobe, and is more often seen in adults; Type II is more severe, presents in childhood and changes occur primarily in the frontal lobes or may be hemispheric; Type III involves a type I or type II dysplasia associated with another principal lesion in the hippocampus, vascular malformations, or an acquired childhood pathology. Magnetic resonance imaging (M□I) may assist in differentiating between FCD types and in determining if surgical resection of the lesions is possible and□or beneficial for patients with drug-resistant epilepsy. If the epileptic seizures are intractable, surgery is the next line of treatment with resection of the lesion, the lobe, or hemispherectomy. If the entire lesion can be resected, up to 80% of patients may remain seizure-free□ subtotal resection may result in seizure relapse within 6 months.

Fabry disease is an inherited inborn error of metabolism (IEM) caused by the deficiency of alpha-galactosidase-A enzyme. This gene mutation causes insufficient breakdown of lipids, which accumulate to toxic levels in the eyes, kidneys, autonomic nervous system, and cardiovascular system. Fabry disease is one of several lipid storage disorders, but the only X-linked lipid storage disease. Since the gene that is altered is carried on a mother's X chromosome, male children have a 50% chance of inheriting Fabry and female children have a 50% chance of being a carrier. A milder form is common in females, and occasionally some affected females may have severe manifestations similar to males with the disorder. Symptoms usually begin during childhood or adolescence and include burning sensations in the hands that gets worse with exercise and hot weather and small, non-cancerous, raised reddish-purple blemishes on the skin. Some males will also have eye manifestations, especially corneal

cloudiness. Lipid storage may lead to impaired arterial circulation and increased risk of heart attack or stroke. Cardiomegaly and progressive renal dysfunction also occur. Other signs include decreased sweating (hypohidrosis or anhidrosis), fever, and gastrointestinal difficulties. Enzyme replacement therapy with agalsidase alfa (Replagal®), agalsidase beta (Fabrazyme®), or pegunigalsidase alfa (Elfabrio®) is FDA-approved for treatment of Fabry disease. Enzyme replacement therapy can reduce lipid storage, ease pain, and improve organ function. MigALAstat (Galafold®) is an oral treatment for adult patients with a specific galactosidase alpha gene (GLA) variant. Acroparesthesias, a type of neuropathic pain, may be treated with antiepileptic medications such as gabapentin, phenytoin, or carbamazepine. Gastrointestinal hyperactivity resulting in diarrhea, nausea, vomiting, cramping, and intestinal malabsorption may be treated supportively as symptoms arise. Some individuals may require dialysis or kidney transplantation.

Fragile X syndrome (FXS) is the most commonly inherited disorder causing intellectual disability and behavior disorders. FXS is caused by a mutation in the FMR1 gene. Diagnosis of FXS is usually made between 12 and 36 months. Males tend to be diagnosed earlier as they typically have more severe disability. Distinctive physical features (e.g., prominent forehead and chin, long face, and protruding ears) are present in males, becoming most apparent during puberty. Motor and language delays and behavioral problems develop over time. Individuals with FXS may have frequent ear infections, dental and vision problems, low muscle tone, seizures, and cardiac problems including mitral valve prolapse. Treatment of FXS includes special education programming, occupational therapy, and speech therapy. Other treatment is symptomatic and supportive. Additional related disorders also associated with the FMR1 gene mutation are Fragile X-associated tremor/ataxia syndrome (FXTAS), more common in men, and primary ovarian insufficiency (POI) in women.

Galactosemia is an inherited metabolic disorder occurring when the galactase-1-phosphate uridyl transferase (GALT) enzyme is missing or malfunctioning. Without the GALT enzyme, a milk-based sugar, galactose, cannot be broken down and accumulates in the blood. Infants develop feeding difficulties, diarrhea, vomiting, failure to thrive, jaundice, and liver damage. Children with galactosemia are at increased risk of developmental delay, cataracts, and intellectual disability. Because life-threatening complications occur within a few days of birth if galactosemia is not promptly identified and treated with a low-galactose diet, screening for GALT is routinely performed on all newborns in the United States. Affected individuals must avoid all milk, milk-containing products, and other foods that contain galactose to lessen exposure. Endogenous galactose production cannot be prevented; symptoms cannot be completely avoided by dietary restriction. Ataxia, growth delays, female hypogonadism and ovarian failure, and speech difficulties such as verbal dyspraxia may present as the child develops. Calcium and vitamin D3 supplements are necessary to prevent bone demineralization. Medications containing lactose or lactulose must be avoided. Nutritional supplements with casein hydrolysates are not recommended because they contain trace amounts of bioavailable lactose. Liver dysfunction can develop leading to coagulopathy, jaundice, hypoglycemia, and failure to thrive.

Gaucher disease is the most common of the inherited metabolic disorders known as lipid storage diseases and is caused by a deficiency of the enzyme glucocerebrosidase. Lipids accumulate in the spleen, liver, lungs, bone marrow, and brain. Symptoms may include skeletal disorders, enlarged spleen and liver, liver malfunction, anemia, and yellow spots in the eyes. There are 3 clinical subtypes of Gaucher disease. Type 1 (non-neuropathic) is the most common. Symptoms can appear at any age. Individuals with Gaucher type 1 may bruise easily due to low blood platelets and experience fatigue due to anemia. There are no signs of cognitive impairment or seizures, but hepatosplenomegaly, lung, and kidney impairment may occur. Individuals with a very mild form of the disorder may show no symptoms. In Type 2 (acute infantile neuropathic), liver and spleen enlargement are apparent by 3 months of age. Individuals usually die before 2 years of age. In Type 3 (chronic neuropathic), hepatosplenomegaly is variable, and signs of brain involvement, such as seizures, gradually become apparent. Major symptoms also include skeletal irregularities, eye movement disorders, respiratory problems, and blood disorders. Enzyme replacement therapy (ERT) is available for most people with types 1 and 3 Gaucher disease and includes taliglucerase alfa (Elelyso®), velaglucerase alfa (Vpriv®), or imiglucerase (Cerezym®) infusion. Oral therapy with migLUstat (Zavesca®) or eliglustat (Cerdelga®) is available for adult patients unable to use ERT. ERT may decrease liver and spleen size, reduces skeletal anomalies, and may reverse other symptoms of the disorder, including abnormal blood counts. Bone marrow transplantation can reverse the non-neurological effects of type 1 Gaucher disease but is rarely performed due to high risk. Splenectomy may be required, and blood transfusions may benefit symptomatic anemia. Joint replacement surgery can improve mobility

and quality of life. There is no effective treatment for severe brain damage that may occur in persons with types 2 and 3 Gaucher disease.

Hunter syndrome, or mucopolysaccharidoses type II (MPS II), is an inborn error of metabolism and occurs almost exclusively in males. MPS II is caused by a mutation in the iduronate sulfatase (IDS) gene which is needed to metabolize carbohydrates in the body. Abnormal metabolism leads to accumulation of glycosaminoglycans in body tissues including the skeleton, joints, brain, heart, lungs, and liver. Symptoms associated with Hunter syndrome appear between 2 to 4 years of age, with growth delays, joint stiffness, restricted movement, hearing loss, hepatosplenomegaly, and macrocephaly. Two distinct forms of Hunter syndrome are clinically recognized. Individuals with late-onset, mild form (MPS IIB) may present with only slight intellectual ability impairment and slower disease progression; while those with early-onset, severe form (MPS IIA) present with profound mental retardation and progressive cardiac and lung disease leading to death between 10 and 20 years of age. Treatment of symptoms include developmental, physical, and occupational therapy, interventions to improve breathing (e.g., tonsillectomy, adenoidectomy, positive pressure ventilation), and procedures such as cardiac valve replacements or hernia repair as needed. Special care is required during any surgical interventions requiring anesthetic because of poor pulmonary function. Enzyme replacement therapy (ERT) with idursulfase (Elaprase®) is approved for treatment of the enzyme deficiency but has only been studied in milder disease with no benefit for severe pulmonary or CNS disease. See also paragraph on Mucopolysaccharidoses (MPS).

Hurler disease, or mucopolysaccharidoses type I (MPS I), is an inborn error of metabolism caused by a deficiency in the alpha-L-iduronase enzyme. Milder forms of MPS I are known as Scheie syndrome, while intermediate disease may be referred to as Hurler-Scheie syndrome. There are no biochemical differences in the disease, the names indicate only a level of severity. Individuals are better described as having either attenuated or severe MPS I to avoid confusion and guide therapeutic options. Infants with severe MPS I appear normal at birth with early indications being non-specific umbilical hernia or frequent upper respiratory infections. Between ages of 1 and 3 years, facial features coarsen, spine deformity may occur, and skeletal dysplasia progresses. Progressive intellectual disability, lack of growth, and hearing loss follows; death from cardiopulmonary failure is common before 10 years of age. For individuals with attenuated MPS I, severity of complications may limit life expectancy to less than 30 years, with milder cases having a normal life span but progressive joint malformations similar to non-inflammatory arthritis. Hearing loss and heart valve disease are common for all levels of disease severity. As with MPS II, treatment includes developmental, physical, and occupational therapy, interventions to improve breathing (e.g., tonsillectomy, adenoidectomy, positive pressure ventilation), and procedures such as cardiac valve replacements or hernia repair as needed. Enzyme replacement therapy (ERT) with laronidase (Aldurazyme®) can benefit non-CNS symptoms by improving liver size, joint mobility, linear growth, breathing, and sleep apnea. Hematopoietic stem cell transplant (HSCT) may slow the course of cognitive decline in children with severe MPS I and may increase survival when provided before age 2 years. Special care is required during any surgical interventions requiring anesthetic because of poor pulmonary function. Bacterial endocarditis prophylaxis with antibiotics should be provided for all individuals with cardiac involvement. See also paragraph on Mucopolysaccharidoses (MPS).

Hydranencephaly is a congenital disorder in which the brain's cerebral hemispheres are absent, replaced with sacs of cerebral spinal fluid (CSF). Infants born with hydranencephaly may appear normal at birth with expected reflexes for sucking, swallowing, crying, and limb movement. Within the first few weeks to months, irritability, hypertonia, hydrocephalus, and seizures can develop. Other symptoms are visual impairment, hearing loss, paralysis, and intellectual disability. Prognosis is poor with life expectancy of 1 year, though some children may survive several years. Hydrocephalus is treated with a shunt; other symptoms are managed with supportive care.

Inborn errors of metabolism (IEM)) are single gene mutations that alter primary protein structures or the amount of a protein that is synthesized. The function of the protein, whether it is an enzyme, receptor, transport vehicle, membrane, or other structural element, is altered or compromised. Depending on the mutation, relatively mild to potentially severe metabolic abnormalities will occur. Two primary categories of IEM are disorders that result in toxic accumulations (disorders of protein metabolism, disorders of carbohydrate intolerance, lysosomal storage disorders) and disorders of energy production and utilization (fatty acid oxidation defects, disorders of carbohydrate utilization and production, mitochondrial disorders, peroxisomal disorders). Severe forms of IEM usually are apparent at birth or within a few weeks of birth. Common characteristics of IEM include: an infant appearing normal at birth and

becoming symptomatic later in life; the nature of the mutation varies from family to family with variation in phenotype; the earlier the appearance of clinical symptoms the more severe the disease; majority of conditions are autosomal recessive traits; successful control of symptoms with early diagnosis is often possible especially if diagnosis and treatment begins before damage to the brain occurs. Screening of all newborns for a range of IEM mutations is routine in the United States, although the screening disorder list varies by state. State screening lists are available from the Health Resources & Services Administration Newborn Screening Information Center (https://newbornscreening.hrsa.gov/your-state).

Lissencephaly is a gene-linked brain malformation characterized by the absence of the normal convolutions of the cerebral cortex and may be paired with microcephaly. Lissencephaly translates from Greek origin to "smooth brain." Defective neuronal migration during embryonic development prevents the proper formation of cerebral cortex grey matter. Symptoms of lissencephaly include unusual facial appearance, failure to thrive, muscle spasm, seizures, deformed fingers and toes, and impaired motor ability. Feeding and nursing difficulties may be managed with gastrostomy tube placement. Anti-epileptic medications can help manage seizures. As with hydranencephaly, shunting may be required if hydrocephalus develops. Individual development and prognosis depend on the degree of malformation. Most children will die before 10 years of age, though some may have near-normal development and intelligence.

Menkes disease is an x-linked, genetic disorder of copper metabolism caused by mutations in the ATPase gene (ATP7A). Because Menkes is x-linked recessive, primarily male newborns are affected. Abnormally low levels of copper accumulate in the brain and liver, while high levels accumulate in the intestinal lining and kidneys. Infants with classic Menkes disease appear healthy until age two to three months, when loss of developmental milestones, hypotonia, seizures, and failure to thrive occur. Menkes disease may be suspected when infants exhibit neurologic changes and characteristic changes in hair (short, sparse, coarse, twisted, and often lightly pigmented). Temperature instability and hypoglycemia may be present in neonates. Daily copper injections may improve outcomes, but only when initiated within days of birth. There is no newborn screening test for Menkes, but a genetic assay for ATP7A is in development. Families with increased risk of ATP7A mutation from prior pregnancy or family history may be offered prenatal screening. Survival past 3 years of age is uncommon.

Metachromatic leukodystrophy is an autosomal-recessive inherited disorder characterized by the accumulation of fats (sulfatides) in cells. Sulfatide accumulation in myelin-producing cells causes progressive destruction of white matter (leukodystrophy) throughout the central (CNS) and peripheral nervous systems (PNS). White matter damage causes progressive deterioration of intellectual functions and motor skills, peripheral neuropathy, incontinence, seizures, paralysis, inability to speak, blindness, and hearing loss. The most common form of metachromatic leukodystrophy, late infantile form, usually appears in the second year of life. Affected children lose speech, become weak, and develop gait disturbance. Muscle tone at first decreases (hypotonia) then increases to spasticity and rigidity. In 20-30% of individuals with metachromatic leukodystrophy, onset occurs between the age of 4 and adolescence (juvenile form). The first signs of the disorder may be behavioral problems and increasing difficulty with schoolwork. Progression of the disorder is slower than in the late infantile form and affected individuals may survive for about 20 years after diagnosis. The adult form affects about 15-20% of individuals with metachromatic leukodystrophy.

Mitochondrial diseases result from failures of the mitochondria, a cell component that creates energy for cellular processes, is involved with metabolic pathways, and enzyme synthesis. Mitochondrial disease may be inherited or result from spontaneous DNA mutations. Mitochondria are present in every cell of the body except red blood cells. When mitochondria fail, insufficient energy is generated within the cell, resulting in cell injury and death. Accumulation of damage from mitochondrial diseases primarily causes destruction to cells of the brain, heart, liver, skeletal muscles, kidney, and the endocrine and respiratory systems. Depending on which cells are affected and how severely, symptoms may include loss of motor control, muscle weakness and pain, gastro-intestinal disorders and swallowing difficulties, poor growth, cardiac disease, liver disease, diabetes, respiratory complications, seizures, visual and hearing problems, lactic acidosis, developmental delays, and susceptibility to infection. Because there are so many different functions of mitochondria within each cell, hundreds of mitochondrial diseases exist. The same DNA mutation (genocopies of mitochondrial disease) causing mitochondrial failure may not produce the same disease or spectrum of symptoms in different individuals likewise, differing mutations can lead to the same disease expression through a

different mutation pathway (phenocopies of mitochondrial disease). Additional information on the extensive list of mitochondrial diseases is available from the United Mitochondrial Disease Foundation (https://www.umdf.org).

Mucopolysaccharidoses (MPS) are lysosomal storage diseases caused by inborn errors of metabolism. MPS results from the absence or malfunctioning of enzymes needed to break down long carbohydrate molecules (glycosaminoglycans). Glycosaminoglycans are present in the fluid that lubricates joints and are components of bone, cartilage, tendons, corneas, skin, and connective tissue. Individuals with mucopolysaccharidosis either produce insufficient amounts enzymes required to metabolize glycosaminoglycans into proteins and simpler molecules. MPS may not be apparent at birth, but signs and symptoms develop as damaged cells accumulate. Glycosaminoglycans can accumulate in all body cells, including blood and connective tissues, resulting in permanent, progressive cellular damage that affects the individual's appearance, physical abilities, organ and system functioning, and cognition. All MPS diseases are autosomal recessive (both parents are carriers), except for MPS II in which a male child inherits the x-linked disorder from the mother only. Additional information on MPS diseases is available from the National MPS Society (https://mpssociety.org). See also paragraphs on Hurler disease (MPS I), Hunter syndrome (MPS II), and Sanfilippo syndrome (MPS III).

Muscular dystrophy (MD) refers to a group of more than 30 genetic diseases characterized by progressive weakness and degeneration of the skeletal muscles that control movement. Most forms of congenital MD are autosomal recessive disorders. Some forms of MD are seen in infancy or childhood, while others may not appear until middle age or later. The disorders differ in terms of the distribution and extent of muscle weakness (some forms of MD also affect cardiac muscle), age of onset, rate of progression, and pattern of inheritance. The prognosis for people with MD varies according to the type and progression of the disorder. There is no specific treatment to stop or reverse any form of MD. Treatment is supportive and may include physical, respiratory, and speech therapies. Orthopedic appliances are used for support and corrective orthopedic surgery may be offered. Medications for symptoms include corticosteroids, antiepileptic medications, immunosuppressants, and antibiotics. Some individuals may need assisted ventilation to treat respiratory muscle weakness or a pacemaker for cardiac abnormalities. Duchenne muscular dystrophy is the most common form of MD among children, while Becker muscular dystrophy is a similar, but milder form of MD; together these are known as Duchenne Becker muscular dystrophy (DBMD). New therapies include antisense oligonucleotide infusions [casimersen (Amondys 45), eteplirsen (Exondys 51), golodirsen (Vyondys 53), viltolarsen (Viltepso)], a corticosteroid-like drug vamorolone (Agamree®), and gene therapy (Elevidys®).

Niemann-Pick disease refers to a group of inherited metabolic disorders, or IEM, known as lipid storage diseases. Toxic accumulations of lipids occur in the spleen, liver, lungs, bone marrow, and brain. Symptoms may include lack of muscle coordination, brain degeneration, eye paralysis, learning problems, loss of muscle tone, increased sensitivity to touch, spasticity, feeding and swallowing difficulties, slurred speech, and an enlarged liver and spleen. Corneas may cloud and a characteristic cherry-red halo develops around the center of the retina. The disease has 4 related types. Type A, the most severe form, occurs in early infancy and is characterized by an enlarged liver and spleen, swollen lymph nodes, and profound brain damage by six months. Children with this type rarely live beyond 18 months. Type B involves an enlarged liver and spleen, usually developing in the pre-teen years; the brain is not affected. Bone marrow transplantation may improve symptoms in Type B. In types A and B, insufficient activity of the sphingomyelinase enzyme causes accumulation of sphingomyelin, a lipid present in every cell of the body. Types C and D may appear early in life or develop in the teen or adult years. Types C and D are caused by a lack of the NPC1 or NPC2 proteins. Neonates can present with ascites and severe liver disease from infiltration of the liver and/or respiratory failure from infiltration of the lungs. Other infants, without liver or pulmonary disease, have hypotonia and developmental delay. The classic presentation occurs in mid-to-late childhood with onset of ataxia, vertical supranuclear gaze palsy (VSGP), and dementia. Dystonia and seizures are common. Dysarthria and dysphagia eventually become disabling, making oral feeding impossible; death usually occurs in the late second or third decade from aspiration pneumonia. Adults are more likely to present with dementia or psychiatric symptoms. Affected individuals have only moderate enlargement of the spleen and liver, but brain damage may be extensive, causing difficulty walking and swallowing and progressive loss of vision and hearing. Types C and D are characterized by a defect that disrupts the transport of cholesterol between brain cells. Type D is limited to people with an ancestral background in Nova Scotia. Individuals with Type C or D may be placed on cholesterol-lowering medications and a low cholesterol diet, but these interventions do not seem to be of much benefit. Avoid medications that cause excess salivation, exacerbate seizures, or interact with

antiepileptic drugs. A medication used for Gaucher disease, migLUstat (Zavesca®) may be used off-label in Niemann-Pick type C disease.

Osteogenesis imperfecta (OI) is a group of genetic disorders primarily affecting bone development. Most forms of OI have an autosomal dominant inheritance pattern. Bones break easily, often from mild trauma or with no apparent cause. Genetic factors are used to define the different forms, for example the most common 4 types are related to defects in the COL1A1/2 genes. Severity varies from person to person with severe cases including fetal bone fractures. Additional features include blue sclerae, short stature, hearing loss, respiratory problems, and tooth development problems. Infants may have abnormally small, fragile rib cage and underdeveloped lungs breathing problems can be fatal. Milder forms of OI are characterized by bone fractures during childhood and adolescence that often result from minor trauma. Fractures occur less frequently in adulthood. People with mild forms of the condition may have a blue or grey scleral coloring, develop hearing loss in adulthood but reach normal or near normal adult height. Treatment may focus on physical therapy, including teaching safe handling techniques to parents and caregivers, limb bracing and orthotics to promote stability of deformed limbs. Chronic daily bone pain may be associated with fractures and myofascial pain from connective tissue disorder. Therapy may include bisphosphonates, denosumab (Prolia®), or teriparatide (Forteo®) to help increase bone mass and bone strength.

Potter syndrome also known as Potter syndrome, refers to a sequence of events that impacts the physical condition and appearance of a newborn when insufficient amniotic fluid (oligohydramnios/anhydramnios) is present in utero. Features may include "Potter facies" with a flattened nose, recessed chin, epicanthal folds, and low-set abnormal ears. Additionally, eye malformations, heart defects, and features of Eagle-Barrett syndrome, also known as "prune belly", are caused by lack of abdominal muscle development causing the skin to wrinkle. Potter syndrome can also result from other conditions including polycystic kidney disease, malformed (dysplastic) or underdeveloped (hypoplastic) kidneys, and obstructive uropathy, in which urine cannot be voided from the bladder and builds up within the kidneys. Potter syndrome is often fatal at birth, mainly due to the pulmonary hypoplasia. The underlying cause of the condition is often undetermined, but may be genetic, with the inheritance pattern dependent on the specific genetic cause. Treatment, when possible, depends on the severity and nature of the abnormalities present. Treatment may be palliative focused only for neonates with bilateral renal agenesis, severe neonatal respiratory distress due to associated pulmonary hypoplasia, and spontaneous pneumothorax. Management of pulmonary hypoplasia with mechanical ventilation and chest tube placement for pneumothorax may be necessary. Renal dysfunction management includes correcting electrolyte abnormalities (hypocalcemia and hyperphosphatemia), treating anemia with iron and erythropoietin stimulating agents, controlling hypertension with diuretics, ACE inhibitors, beta blockers, or calcium channel blockers, and potentially use of human growth hormone (HGH) to stimulate growth and development.

Rett syndrome is a neurodevelopmental disorder and primarily affects females. Rett syndrome is caused by a mutation in the methyl CpG binding protein 2 (MECP2) gene but not everyone with the MECP2 mutation develops ett syndrome. Early infant and childhood growth and development appears normal but is followed by a slowing of development, loss of purposeful use of the hands, distinctive hand movements, slowed brain and head growth, progressing to problems with walking, seizures, and intellectual disability. Rett syndrome course, including the age of onset and the severity of symptoms, varies from child to child. Before the symptoms begin, the child generally appears to grow and develop normally, although there may be subtle abnormalities in infancy, such as loss of muscle tone (hypotonia), difficulty feeding, and jerkiness in limb movements and difficulty with eye contact. The loss of functional use of the hands is followed by compulsive hand movements such as wringing and washing. The onset of this period of regression is sometimes sudden. Apraxia (inability to perform motor functions) interferes with every body movement, including eye gaze and speech. Children with Rett syndrome often exhibit autistic-like behaviors in the early stages. Other symptoms may include walking on the toes, sleep problems, a wide-based gait, teeth grinding, difficulty chewing, slowed growth, seizures, cognitive disabilities, and breathing difficulties while awake such as hyperventilation, apnea (breath holding), and air swallowing. Supportive care may include antiepileptic drugs to control seizures, low-dose antipsychotics or antidepressants to help with agitation, and medications to help with sleep, GI upset, and constipation. The only FDA-approved treatment, trofinetide (Daybue®), was approved in 2023. Trofinetide is an oral solution developed to reduce inflammation in the brain, leading to potential improvement in communication, fine motor skills, and breathing. Potentially severe GI side effects including nausea, vomiting, and diarrhea are reported frequently and may require therapy discontinuation.

Sandhoff disease is an autosomal recessive, lysosomal storage disorder resulting in the progressive neurodegeneration. Sandhoff disease is caused a mutation in the HexB gene and subsequent deficiency of hexosaminidase-B enzyme, resulting in the accumulation of lipids in the brain and other organs. Sandhoff disease is clinically similar to Tay-Sachs disease. Onset of the disorder usually occurs within the first 6 months of life (acute infantile Sandhoff disease) but timing of onset may vary with onset at 2-5 years (subacute juvenile Sandhoff disease) and older teenage-young adult years (late-onset Sandhoff disease). Neurological symptoms may include motor weakness, startle reaction to sound, early blindness, progressive mental and motor deterioration, macrocephaly, red spots on macula noted on eye exam, seizures, and myoclonus. Other symptoms may include frequent respiratory infections, unique facial appearance, and an enlarged liver and spleen. Supportive treatment includes proper nutrition and hydration and keeping the airway open. Antiepileptic drugs may control seizures. Avoid antipsychotic medications like risperidone, haloperidol, and chlorpromazine which may worsen movement disorders with limited psychiatric benefit. Prognosis is poor for children with acute infantile onset with mortality by 3 years of age.

Sanfilippo syndrome is an inherited, autosomal recessive mucopolysaccharidosis disorder (MPS III) with progressive CNS neurodegeneration leading to severe intellectual disability, developmental regression and may also result in autism-spectrum disorder. MPS III has 4 main subtypes depending on the specific gene and enzyme affected. Type A is the most severe form caused by missing or altered form of heparan N-sulfatase. Type B (alpha-N-acetylglucosaminidase), type C (acetyl-CoA:alpha-glucosaminide acetyltranferase), and type D (N-acetylglucosamine 6-sulfatase) is missing or insufficiently produced. Unlike other forms of MPS, symptoms appear after the first year of life and decline in learning ability typically occurs between ages 2 and 6. The child may have normal growth during the first few years, but final height is below average. Delayed development is followed by deteriorating cognitive ability. Other symptoms and complications include behavioral problems, coarse facial features, diarrhea, sleeping difficulties, blindness, hearing loss, seizures, stiff joints, and difficulty walking. Urine tests show large amounts of heparan sulfate (a glycosaminoglycan) in the urine. Enzyme replacement therapies, gene therapy, gene modified stem cell therapy are in clinical trials. Supportive care includes antiepileptic drugs to manage seizures, antipsychotics to managing agitation, hyperactivity, and aggressive behavior, and vitamin D to support bone mineral density. See also paragraph on Mucopolysaccharidoses (MPS).

Short bowel syndrome (SBS) is a malabsorptive state resulting from either surgical resection or congenital disease of the small or large intestine. The amount of resection and remaining bowel dictates the degree of malabsorption and the need for specialized enteral nutrition or parenteral nutrition (PN). Children may be dependent on PN to maintain minimal energy and fluid requirement for growth. Common causes of SBS in infants and children include necrotizing enterocolitis (NEC), mid-gut volvulus (twisted intestinal loop), intestinal atresia, and gastroschisis (incomplete formation of abdominal wall causing protrusion of intestines outside the body). Close monitoring of nutritional status, early introduction of enteral nutrition when possible, and prevention and treatment of infections of the parenteral nutrition intravenous catheter and control of bacterial overgrowth in the bowel can improve prognosis. Diarrhea is common and if severe or prolonged, can cause life-threatening dehydration. Deficiencies in vitamin and mineral absorption may create additional symptoms: vitamin A (vision and skin problems), vitamin B (stomatitis, macrocytic-megaloblastic anemia, peripheral neuropathies, tachycardia), vitamin D (rickets, tetany, paresthesias), vitamin E (ataxia, edema, vision problems), vitamin K (petechiae, ecchymoses, purpura), iron (glossitis, weakness, fatigue, dyspnea), and zinc (alopecia, stomatitis, poor wound healing). In patients with severe short bowel syndrome, the absorption of medications may be an important issue. Higher doses of medications may be required to offset the lower percentage of absorption. In general, extended-release medications should be avoided. Non-enteral delivery systems (e.g., transdermal patches, nasal administration, or parenteral routes) should be considered for medication administration. Medications to reduce stomach acid production, histamine-2 receptor antagonists (H2RA) and proton pump inhibitors (PPI), as well as pancreatic enzyme replacement therapy and bile-acid resins may help with digestion of enteral nutrition. Teduglutide (Gattex®) is FDA-approved in patients ages ≥ 1 year and adults for treatment of short bowel syndrome dependent on parenteral nutrition support. Teduglutide is a glucagon-like peptide-2 (GLP-2) analog that increases intestinal and portal blood flow and inhibits gastric acid secretion to reduce intestinal losses and improve intestinal absorption.

Sickle cell disease (SCD) encompasses a group of symptomatic disorders associated with mutations in the HBB gene, defined by the presence of hemoglobin S (Hb S). Mandatory screening of all newborns born in the US establishes a diagnosis of SCD prior to the onset of any symptoms. SCD is characterized by intermittent vaso-occlusive events and chronic hemolytic anemia. Vaso-occlusive events cause tissue ischemia leading to acute and chronic pain as well as organ damage to the bones, lungs, liver, kidneys, brain, eyes, and joints. Dactylitis (pain and/or swelling of the hands or feet) in infants and young children is often the earliest manifestation. In children, the spleen can become engorged with blood cells in a "splenic sequestration crisis." The spleen is also particularly subject to infarction, with many individuals with SCD functionally asplenic by early childhood, increasing their risk for certain types of bacterial infections. Chronic hemolysis can result in varying degrees of anemia, jaundice, cholelithiasis, and delayed growth and sexual maturation. Individuals with the highest rates of hemolysis are predisposed to pulmonary artery hypertension (PAH), priapism, and leg ulcers. Prevention of these primary manifestations by maintaining adequate hydration, avoiding climate extremes, fatigue, and activities leading to inflammation is a therapeutic goal. Regular testing of lung, liver, and renal function, as well as iron studies, eye exams, complete blood counts (CBC), and arterial blood flow studies can help with earlier diagnosis of secondary complications. Severe pain during vaso-occlusive crises must be managed quickly and is most effectively treated with opioid analgesics. Because of splenic dysfunction, individuals with SCD are at higher risk of bacterial and viral infections requiring prophylactic antibiotics and immunizations. Reduced exercise tolerance from PAH may be managed with phosphodiesterase-5 enzyme inhibitors (PDE5-I), sildenafil (Revatio®) or tadalafil (Adcirca®). Hydroxyurea (Droxia®, Syclos®) may be started within the first 12 months and can help decrease pain, improve anemia, decrease hospitalization risk. L-glutamine (Endari®) is FDA-approved for patients 5 years and older to help prevent severe complications and may be use with hydroxyurea or as an alternative if patients do not tolerate hydroxyurea. Voxelotor (Oxbryta®) and crizanlizumab-tmca (Adakveo®) are FDA-approved to reduce frequency of vaso-occlusive crises in older children and adults. Voxelotor inhibits red blood cell sickling and deformation and reduces blood viscosity. Crizanlizumab inhibits interactions between endothelial cells, platelets, red blood cells, and leukocytes, to decrease platelet aggregation, maintain blood flow, and minimize sickle cell-related pain crises.

Spinal muscular atrophy (SMA) is an autosomal recessive genetic disorder causing motor neuron degeneration leading to progressive muscle weakness and atrophy. SMA subtypes 0-4 are based on severity and age of onset with lower numbers indicating younger onset and more severe disease. Type 0 is prenatal onset which may result in respiratory failure within the first month of life. Type 1 (Werdnig-Hoffmann disease) is usually diagnosed in infancy. Infants with type 1 may appear normal at birth but do not meet developmental motor milestones and are unable to sit or stand without assistance. Early childhood mortality is high due to progressive muscle weakness and respiratory failure. Type 2 (Dubowitz disease) intermediate form is diagnosed by two years of age. Children may be able to maintain a seated position without support but are unable to walk. Individuals with type 2 may survive into adolescence or young adulthood. Type 3 (Kugelberg-Welander disease) juvenile form onset may be in early childhood but may present as late as the teenage years. Children may be able to stand and walk independently but with disease progression may become wheelchair dependent; respiratory muscle function remains relatively normal. Type 4 of SMA has adult/late onset. People with type 3 or 4 may have a normal life expectancy. Disease modifying therapies including nusinersin (Spinraza®) is FDA-approved to treat children and adults with SMA targeting SMN2 mRNA. Onasemnogene abeparvovec-xioi (Zolgensma®) gene therapy targeting SMN1 is FDA-approved for children younger than 2 years. Risdiplam (Evrysdi®) is an oral drug for infants 2 months of age and older also targeting SMN2 mRNA. Gene therapy may halt motor neuron destruction and slow disease progression in individuals with SMA.

Tay-Sachs disease results from an enzyme deficiency (Hexosaminidase A, HexA) causing a group of neurodegenerative disorders. Tay-Sachs disease, or acute infantile variant HexA, is characterized by progressive weakness, loss of motor skills, decreased attentiveness, and increased startle response beginning between ages three and six months. Progressive evidence of neurodegeneration includes seizures, blindness, spasticity, and death, usually before four years. The juvenile (subacute), chronic, and adult-onset variants of HexA deficiency have later onsets, slower progression, and more variable neurologic difficulties including progressive dystonia, spinocerebellar degeneration, motor neuron disease, and, in some individuals with adult-onset disease, a bipolar form of psychosis. There are no approved treatments for Tay-Sachs. Supportive care includes nutrition and hydration support, managing infectious disease, protecting the airway, and controlling seizures. Antiepileptic drugs are used but

seizures are progressive and can change in type and severity. For individuals with adult-onset HexA deficiency who have psychiatric manifestations, antipsychotic or antidepressant therapy may be required. Tay-Sachs disease is prevalent primarily in people of Eastern European (Ashkenazi) Jewish descent and some French Canadians.

Trisomy 13 (Patau syndrome) is a chromosomal disorder caused by an extra copy of chromosome 13 in some (mosaic Trisomy 13), or all, of the body's cells. Trisomy 13 interferes with normal development, leading to severe intellectual disability, congenital heart defects, brain and spine abnormalities, cleft palate, microphthalmia, and poor muscle tone. Most Trisomy 13 is not inherited but results from random mutations during reproductive cell formation. Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within days to weeks of birth. Median survival is only about 2 years with aggressive management – surgeries to correct heart defects, cleft palate and lip repair, abdominal wall hernias, feeding tube placement, and seizure prophylaxis.

Trisomy 18 (Edward syndrome) is a chromosomal disorder caused by an extra copy of chromosome 18 in some (mosaic Trisomy 18), or all, of the body's cells. In mosaic Trisomy 18, clinical expression is less severe, and survival is usually longer. Among liveborn children, Trisomy 18 is the second most common autosomal trisomy after Trisomy 21. High mortality rate is due to cardiac and renal malformation, feeding difficulties, sepsis, and respiratory failure. The disorder is characterized by growth deficiency and distinct physical features: microcephaly, microphthalmia, malformed ears, micrognathia or retrognathia (mandible or maxilla malformation), microstomia, distinctively clenched fingers, and other congenital malformations. Cardiac symptom management with diuretics and digoxin for heart failure and indomethacin for ductal closure may improve symptoms. Enteral nutrition via nasogastric or gastrostomy is provided to supplement feeding difficulties. Because of the prevalence of liver and kidney malignancies (Wilms tumor) with imaging and abdominal ultrasound is recommended for children older than one year.

Trisomy 21 (Down syndrome) is caused by an extra copy of chromosome 21 (95% of cases). As with other trisomies, extra chromosome copies in only some of the body's cells is referred to as a mosaic variant (e.g., mosaic Down syndrome). Down syndrome is associated with intellectual disability, a characteristic facial appearance, and low muscle tone in infancy. The degree of intellectual disability varies from mild to moderate. People with Down syndrome may be born with a variety of health concerns, including congenital heart defects or digestive abnormalities. In addition, they have an increased risk of developing gastroesophageal reflux, celiac disease, hypothyroidism, hearing and vision problems, leukemia, and early onset Alzheimer disease. The primary treatment objective for individuals with Down syndrome is to boost cognition by improving learning, memory, and speech. Late onset Lennox-Gastaut seizure syndrome is prevalent in people with Down syndrome. Multidisciplinary specialist care should be provided to help assess, diagnose, and manage symptoms as they develop.

von Willebrand disease (VWD) is a family of congenital bleeding disorders caused by an abnormality in von Willebrand factor (VWF). Type 1 is the mildest form of VWD, in which the individual has lower than normal levels of VWF and may also have lower than normal levels of clotting factor VIII. In Type 2 VWD, the body makes normal amounts of VWF but the factor is defective. Type 2 has 4 subtypes, 2A (mild to moderate mucocutaneous bleeding), 2B (mild to moderate mucocutaneous bleeding and thrombocytopenia), 2M (mild to moderate or severe mucocutaneous bleeding), and 2N (excessive bleeding with surgery and procedures that mimics hemophilia) depending on the nature of the defect. Type 3 is the most severe form of VWD, in which the individual produces almost no VWF or clotting factor VIII, and frequently includes musculoskeletal bleeding. VWD can produce spontaneous nosebleeds, easy bruising, heavy menstrual bleeding, and longer than normal bleeding after injuries, surgeries, or dental work. Treatments may include desmopressin (hormone that increases production of VWF and factor VIII in the blood), factor replacement therapy, antifibrinolytic medications (aminocaproic acid or tranexamic acid), and oral contraceptives (increase VWF and factor VII and reduce menstrual bleeding). Desmopressin response is poor in Type 2M and has limited benefit in Type 2N, so clotting factor VIII is the treatment of choice in these subtypes. Desmopressin is not effective in Type 3 VWD, but antifibrinolytics or oral contraceptives may be beneficial, clotting factor VIII is treatment of choice for severe bleeding episodes. Desmopressin must be used cautiously in children under 2 years of age due to required fluid restrictions and risk of hyponatremia.

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Prognostic Tips for the Child with Medical Complexity

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Children with rare disease and/or medical complexity present a challenge as the course and the prognosis are often unclear. A recent article listed in the resources has helpfully clarified some of these issues.

1. Accept that prognostication is particularly challenging for children. Prognosis, likely quality of life, probability of survival, and length of life are uncertain. Engaging the family and the child is important for the team to understand preferences and optimal modes of communication for shared understanding.
2. The younger the child, the more challenging it is to predict the course and outcome. It is often difficult to predict the future for fetuses and newborns with potentially life-limiting conditions. "Hope for the best; plan for the worst" is a philosophy that can help optimize quality of life.
3. Cultural norms, economic situation, and needs are among the many things that need to be explored when discussing prognosis. In all cases, review the complete range of likely expected outcomes.
4. For adolescents/young adults, parents may wish less complete than full disclosure. Review with the family the likelihood that the patient has a greater understanding of prognosis than might be expected for their age. Honest communication near end of life facilitates exploration of hopes, concerns, and decisions about future treatment.
5. Especially in advanced pediatric cancer, determining prognosis is difficult with any degree of accuracy.
6. Wide ranges of prognosis can be seen in Trisomy 13 and 18, as well as some other chronic conditions. Decisions about aggressiveness of treatment may impact length of survival.
7. Gene therapy may positively impact the length of life with conditions such as spinal muscular atrophy and other genetic conditions. As this is a relatively new option, the unknown long-term outcomes make it challenging to determine the risks vs. benefits.
8. Declining prognosis may be signaled by feeding intolerance/GI failure, increasing number and length of hospitalizations, lack of returning to baseline function, and diminishing quality of life as determined by the child and family.
9. Sometimes uncertainty prevails and no prognostication should be offered. This is particularly true in rare or undiagnosed conditions. The team should support the family and child in dealing with uncertainty. Planning for more than one trajectory of the condition/disease is one approach that works well in this situation.
10. Expect anticipatory as well as post-death grief. The parents grieve the loss of their hoped for healthy child. But they may find that they can hope for the best at the same time as they might be anticipating the worst.

Discussing with the child and family about potential outcomes and disease trajectory is important. While an exact prognosis and course may be difficult to determine, the discussion supports the family in defining goals and helps them feel as though they have some input and control.

Resources

Bergstraesser, E., Thienprayoon, R., Brook, L. A., et al. Top ten tips palliative care clinicians should know about prognostication in children. *Journal of Palliative Medicine,* 2021;24:1725-1731.

NeuroJourney: Anticipatory Guidance and Resources for Families and Clinicians Caring for a Child with Severe Neurologic Impairment (SNI)

Courageous Parents Network,
courageousparentsnetwork.org

SNI is a phrase used to describe a group of conditions, or disorders, that affect the central nervous system. Categories of SNI include metabolic neurodegenerative disorders, genetic conditions and syndromes, developmental epileptic encephalopathy and seizure disorders, and brain malformations, infections, and injuries. As they progress, these conditions commonly result in problems with motor skills, cognitive skills, and other medical complexity.

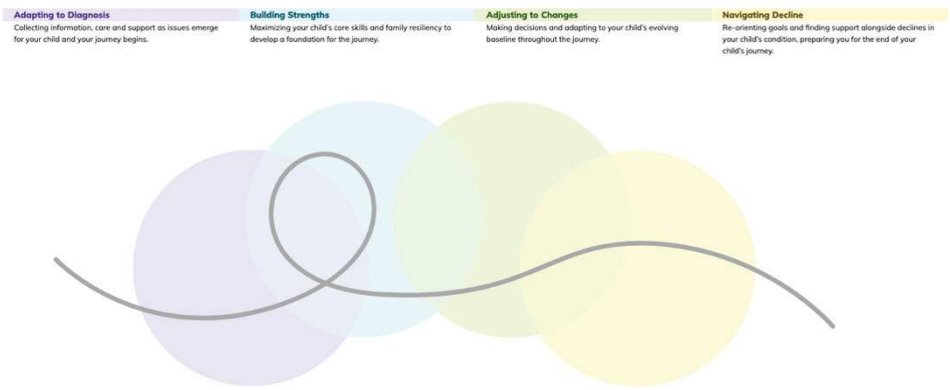
Families of a child with SNI typically encounter both medical issues and daunting physical, social, and emotional challenges—all without a roadmap, an integrated way to understand what is occurring and what might occur. Acknowledging that a child with SNI may have a shorter life, an educational resource from Courageous Parents Network seeks to help parents and other caregivers consider that life as a full arc. NeuroJourney (NeuroJourney.org) presents many aspects of the SNI disease trajectory in a series of phases—*the arc of a life*—to help parents and other caregivers navigate the present and prepare for the future.

Medical considerations are staged along these phases as they commonly occur, discussed through the lens of changes in the central nervous system and how they affect other bodily systems. Psychosocial topics are also represented, along with commentary that brings in a holistic, palliative point of view.

The NeuroJourney Phases

- Adapting to Diagnosis and all it entails
- Building Strengths to establish baseline and care for the child
- Adjusting to Changes as they occur along the arc of a life
- Navigating Decline as end of life approaches

The NeuroJourney Arc of a Life



Medical Considerations and Psychosocial Topics

NeuroJourney was created in partnership with Julie Hauer, MD, FAAHPM; and Rachel Thienprayoon, MD, MSCS, FAAHPM, FAAP, Cincinnati Children's Hospital; with significant contributions from advising medical reviewers, parent authors, and content editors. Medical considerations discussed include Endocrinology; Gastroenterology; Neurology; Nutrition and Growth; Pain and Irritability; Pulmonology; Sleep; Urology; and Musculoskeletal with more to come. Psychosocial topics include Goals of Care; Caregiver Well-being; Anticipatory Grief; Advance Care Planning and End of Life; Partnership and Marriage; Out-of-Home Placement; Extended Family and Friends; Guilt and Regret; Family Planning; Financial Planning; Communicating Diagnosis; Spirituality; Transition to Adulthood; Work; Siblings and Siblings at End of Life.

Using NeuroJourney Content

NeuroJourney can be used to support parents in building their child's care team, understanding interconnected medical considerations, reflecting on goals of care, and communicating about the child's condition with clinicians, family, and others. This resource is also used by clinicians in offering anticipatory guidance, explaining medical considerations, and training clinicians with emphasis on palliative care and palliative-aware practice.

More Information

Courageous Parents Network invites inquires and feedback. To schedule a virtual or in-person presentation, please contact Courageous Parents Network at connect@courageousparentsnetwork.org.

Perinatal Hospice: Case Study and Test Uncertainty

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Background

Trisomy 13 (T13) was first described in 1960. It is one of the least common autosomal trisomies and it encompasses multiple malformations including central nervous system, cardiac, and urogenital anomalies. Characteristic dysmorphic features include micropthalmia, cleft lip and palate, and polydactyly (Peroos, 2012). Here I report on a 5-week-old girl with presumed T13 based on a prenatal amniocentesis result who is currently thriving and exceeding normal expectations.

Case Presentation

The patient is a former 37 5/7-week African American (AA) female born on 11/2/23 to a 24 y/o G2P1 (gravidity or total number of pregnancies, parity is the number of viable infants brought to viable gestational age) female (now G2P2). Prenatal amniocentesis testing was performed at 20 weeks estimated gestational age (EGA) and confirmed the presence of Trisomy 13 (Patau Syndrome). The mother's delivery was induced, and the patient was born via normal spontaneous vaginal delivery (NSVD) without noted complications or Respiratory Distress Disorder (RDS) and was average gestational age (AGA) with normal growth parameters for her age and noted birth weight of 2.5 kilograms. She was stabilized and discharged home to begin hospice services for presumed imminent demise. The patient was examined on day of life (DOL) 2 by the hospice attending and the following was noted on exam: no dysmorphic features appreciated except for questionable microcephaly, no murmur or cyanosis appreciated, tachypnea noted with RR in the 40s-50s otherwise stable. The patient began receiving breast milk (BM) and 20 cal formula via bottle with no associated feeding intolerance and did not exhibit dehydration or jaundice.

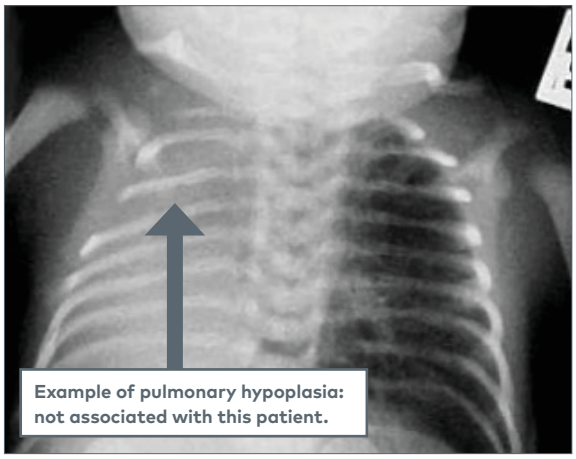
Discussion

Genetic disorders intermittently complicate pregnancies. Approximately 3% to 5% of pregnancies demonstrate birth defects or genetic disorders which are present in 1 to 150 live births. Furthermore, congenital malformations remain the leading cause of childhood death (CDC, 2008). The known prevalence of these disorders promotes prenatal testing by both medical providers and family members. Maternal motivations for pursuing aneuploidy screening or prenatal diagnosis are varied. Some may choose pregnancy termination if the defect is identified at an early enough gestational age. Others may choose to pursue screening or testing to allow them time to prepare for the situation that is to ensue after delivery takes place (Alfirevic, 2003). Considering the fact that decisions are

sometimes based on inaccurate results, it is imperative that parents understand which potential consequences may arise with available testing. First of all, a tendency to overestimate the risks for Trisomies 21, 18, and 13 exists with a noted 95% confidence interval (CI) which means that 5% of those infants tested may receive a false diagnosis (Santorum, 2017). Secondly, deciding on a proper time to test can also present unattended complications. First trimester testing allows for early detection but may result in increased pregnancy loss between 1.9% and 4.7% and laboratory failure varies between 0% to 20% (Alfirevic, 2003). Second trimester testing decreases pregnancy failure risk but may cause unavoidable distress while the families wait for the infant to achieve an EGA of at least 17 weeks before the testing can take place.

Trisomy 13 is one of the most common fetal life-limiting diagnoses with a prevalence of 1.68 per 10,000 pregnancies (Cortezzo, 2021). This disorder is often presumed to be lethal and often "futile" in nature, due to the poor survival rate associated with it. Most of the patients presenting with T13 pass in the first 7-10 days and between 86% and 91% do not survive beyond 1 year of life (Peroos, 2012). However, reports of patients living beyond 7 years of life or more do exist if the child inherits a mosaic pattern. Cautious decision-making, while testing this particular subset of children, seems appropriate in that studies now suggest that increased longevity seems more plausible than what was once thought (Cowen, 1979).

Outcome and Follow Up



Example of pulmonary hypoplasia: not associated with this patient.

The patient is currently 5 weeks old and remains stable. She continues to maintain adequate po intake and weight gain and takes approximately 2-3 oz of BM and 22 cal formula every 2 hours. Intermittent tachypnea remains a permanent finding at this time, yet she does not demonstrate retractions, nasal flaring, or other signs of distress. Her neurologic exam remains normal (NL) and she manifests intact neonatal reflexes and NL tone. No head lag appreciated. Due to the patient's stable condition and lack of familial genetic disorders, the patient's mother appropriately requests that the patient receive a confirmation test. SNP Microarray T13 was recently ordered, and results are pending at the time of this case study. The child will also eventually receive a chest x-ray to evaluate for the presence of pulmonary hypoplasia. This is a condition in which the patient's lung demonstrates decreased size and weight, thus making the lungs limited for respiratory purposes. This might account for the reason this particular patient is persistently tachypneic and the condition may present itself in the face of a T13 diagnosis.

Furthermore, although the patient does not exhibit signs of cardiovascular compromise or demonstrate an audible heart murmur, it also seems justifiable to order an echocardiogram. This will help to evaluate the patient's current heart status noting that 50-85% of all T13 patients have cardiac defects. Furthermore, reports seem to suggest that the children who are free of defects will demonstrate longer survival rates (Peroos, 2012).

Regardless of what information the test outcomes might provide, the parents vow to continue to support their wonderful daughter in any way possible. As long as her stability is maintained she will eventually transition from hospice to palliative care in order to allow my group the opportunity to follow her neurologic development and provide any necessary ancillary services such as speech, occupational, and physical therapies to the patient and her family. This is a viable option noting the fact that these children do usually meet their early development milestones and have meaningful relationships with their families (2012, p. 9).

Conclusion

Genetic testing is now an integral part of medical health care. Although data results seem to suggest that available tests are accurate for the most part, one must take in consideration that false positive rates do exist. How this might affect a patient's status and subsequent medical treatment plan must receive due attention especially as it pertains to a vulnerable population that includes children. For instance, based on one particular test performed in utero at 20 weeks EGA, the particular child in this case study is receiving hospice care with the expectation of dying in less than 6 months if not much sooner. The stress that this places on her family exists and needs its own separate consideration. Repeat testing to confirm the accuracy of a lab result is not advocated for all cases but may help to strengthen the case in a situation in which an infant is thriving and whose physical exam is normal otherwise. Hospice and palliative care is a wonderful field for those needing this type of specialized care, but certainly should reserve itself to self-evaluation in cases of uncertainty.

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Building Concurrent Care Bridges: Navigating Clinical Trials for Pediatric Hospice Patients

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For many families with children facing terminal and life-limiting illness, having access to concurrent care benefits while also being enrolled in a hospice program has been life changing. When President Barak Obama signed Concurrent Care for Children into law in 2010 (NHPCO, 2023), it opened doors for families to both hold space for preparation and acceptance while also maintaining hope and feelings of purpose. As the medical world continues to maintain a curative-focused mindset, such as with clinical trials, pediatric hospice programs advocate for the holistic care of the person inclusive of quality of life, symptom management, and psychosocial support for the family while the child continues concurrent treatment in hopes for a cure and/or life prolongation.

The purpose of clinical trials can be wide-ranging; inclusive of behavioral, diagnostic, prevention, quality-of-life, screening, and treatment with various phases that indicate the anticipated or tested effectiveness of the medicine or treatment (NHLBI, 2022). These varying approaches and involvement levels promote an inevitable sense of unpredictability and uncertainty despite the descriptive and prescriptive methods clinical trials follow. One way to combat these conflicting emotions and experiences is to include consistency in a home-based pediatric hospice team that can provide assessment of progression and symptoms, emotional support through processing the experience, and ongoing education and reinforcement for the family outside of the hospital environment. Operating within the concurrent care model through coordination of care and communication provides positive outcomes that can benefit the pediatric patient, family, and medical teams throughout the entire process. Given that concurrent care is covered under law by those with Medicaid that are under 21 with a hospice-eligible diagnosis (NHPCO, 2023) and with increasing amount of coverage through private insurance, most children likely have access to this additional support while enrolled in clinical trials in all phases.

Education

Opening doors with other clinicians allows for information sharing to become more accessible and inherent. Hospice providers and clinicians have unique access to community resources that may differ from what can be offered through hospital systems. By combining forces through a concurrent model of care, patients and families have a greater pool of potential resources to pull from and thus greater supports to assist them in navigating their journey. This may span from emotional supports to financial and social supports that are designed to aid families faced with possible terminal illness including food/living expenses, sibling and child-based resources, and/or funeral assistance should it be needed in the future.

The constantly changing medical world indicates a just as constant change in treatment methodologies. Although clinical trials must be rigid in treatment so as to not disrupt the process and result, there are opportunities for education to be provided on behalf of palliative and hospice professionals to decrease symptom burden inclusive of pain and anxiety that may not interfere with the integrity of the trial. Just as specialists seek out ongoing

education on treatments pertaining to their field, so do palliative and hospice physicians in the specialized field of palliative medicine. Pediatric patients seeking out concurrent hospice care have an increased chance of managing unwanted symptoms when providers have the ability to share intellect, ideas, philosophies, and approaches. In addition, the education afforded to hospice and palliative physicians by providers working with cutting-edge technologies and medicine through clinical trials can continue to grow and flourish while also best supporting patient and families.

Communication

The benefit of consistent communication between providers is essential for all cases, no matter the complexity. However, the nuances and specifics that clinical trials require for participation of families make this collaboration crucial for success and ongoing management of needs. Rare diseases create an inherent sense of the unknown and another world away from home pulling everyone in the family in different directions. A concurrent model for care provides a sense of stability in the home while offering a consistent point of contact between the family, the existing specialists/hospital system, and the clinical trial itself. Symptoms can be better assessed, treated, and logged when a consistent team is available for support in the home that can then be communicated to the clinical trial team. This promotes better adherence to protocols and strategies to allow the trial to be as successful as possible while hopefully limiting the amount of hospitalization and follow up visits required for specialists.

An imperative outcome of communication between providers through concurrent care is collaboration and the multiplication of provider experience, intellect, and skill that can lead to idea-sharing and problem solving. Collaboration can look like open lines for communicating via phone or technology, sharing of appropriate and necessary medical records as agreed to by the patient/family, and collaborative and interdisciplinary team meetings. The hospice paradigm thrives on communication and interdisciplinary teamwork, which can then pave the way for example with other medical teams so there can be a full circle of support surrounding the patients and families.

Coordination of Care

A possible misconception of concurrent care is that the addition of another team, no matter the type, can lead to "too many cooks in the kitchen." As previously indicated, there are benefits to the sharing of experience and skill in potential areas of concern. The hospice team is clinically trained and specializes in relief of pain and distressing symptoms. As previously mentioned, the ongoing education and sharing of medical treatment and philosophy can have an extremely positive effect on the entire family. This coordination of care between all providers is essential to sharing information in a timely and effective manner.

Insurance can become a barrier for many families facing illness. Providers are often faced with the need to constantly advocate for coverage and authorization. However, given the ACA guidelines, concurrent care has become increasingly accessible to pediatric patients even beyond the age of 21. Those working in the concurrent care space often have the ears of the insurance case managers and can provide further advocacy for families navigating clinical trials. For example, there are inherent financial and programmatic benefits through partnership with concurrent care that can potentially impact coverage for clinical trials. Hospice organizations can provide the majority of medication and treatment related to symptom management (inclusive of medication, oxygen, and durable medical equipment), which takes pressure and cost off of the insurance provider. This may allow more coverage to be dedicated to ongoing treatments or help facilitate conversations. By working together collaboratively, insurance and medical teams are able to support each other and the family in a mutually beneficial way.

The regulatory guidelines that hospices abide by, regardless of patient age, have created a foundation for care coordination to thrive. One aspect includes patient travel outside of the service area, which is often part of a child's experience as they travel for treatment, pleasure, and memory-making. When a child is enrolled in concurrent care through hospice, they then have access to support in finding agencies that can continue to support them on their

travels. Admittedly, there are sometimes barriers to finding other pediatric programs in the desired travel destination; however, the comfort level of hospices in caring for pediatrics continues to increase with opportunities to advocate for more access when travel is explored. If a child is planning to travel to another area, the hospice agency can establish a contract with another program to provide care as it is needed and thus offering more comfort for families away from home whether they're going to participate in a new clinical trial or going to Disney.

Conclusion

Rather than navigating the "wild west" of clinical trials alone, a collaborative and concurrent model of care that involves a home-based hospice provider and team offers consistency and more opportunities for support. Having providers work together in a concurrent model helps lift the weight off families to navigate and coordinate the clinical trial paradigm alone and can lead to increased tools to add to their "toolbelt" moving forward. Clinically, the ongoing collaboration and assessment of physical status in the home can lead to a better reaction time to symptom needs as they arise and limit the potential need for ED visits. The hospice team benefits from the education and collaboration with the clinical trial team to best understand how to address and medicate symptoms in a way that maintains the integrity of the trial parameters. Goals of care can be continually addressed and honored as changes occur without creating silos where conversations are repeated unnecessarily.

The Capital Caring Kids pediatric hospice program has prioritized concurrent care for families in the DC metro area that are seeking ongoing curative treatments. This ongoing relationship-building in the medical community helps promote wrap-around support for those facing the medical unknowns in their journey. By building these relationships through education, collaboration, communication, and coordination of care, families have the benefit of maintaining consistency in their care teams while also having additional resources at their disposal for questions, psychosocial support, and management of the illness. Additionally, providers that have partnered with the team report having better awareness of home conditions, clinical responses/changes, and a better understanding of the family's goals for care. Positive, didactic relationships in the concurrent care and clinical trial space have facilitated more conversation at providing what all individuals deserve—consistent support.

For more information on concurrent hospice care programs for pediatrics that may service your area, please utilize the **"Find a Care Provider"** function through NHPCO at www.nhpco.org/find-a-care-provider/.

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Patient Advocacy Organizations: Community, Connection, and Support

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Families facing a life-limiting illness with their child often feel alone or adrift at sea. Connecting with a palliative care team and patient advocacy organizations can be a port in the storm by offering information, compassion, and support from diagnosis through bereavement.

One of the most important things a provider can do is to be well-educated about a patient's condition at the moment of diagnosis. Families often seek out medical information because they are already alarmed by their child's symptoms. Acknowledging this, validating their concerns and making every effort to rule out any potential diagnoses is key to a family's ability to confidently move forward in seeking appropriate and trustworthy medical care. While rare diseases are just that, rare, they should never be dismissed without proper consideration and testing. While the medical community may be trained to think horses when they hear hoofbeats, zebras do still exist.

Providers can support a family from the moment of diagnosis by cultivating an awareness of resources for families that go beyond the clinical setting to support their child's everyday needs. Understanding their potential medical journey as it encompasses various therapies, equipment, and care for the emotional needs of the family is imperative to the family's feelings of support and trust in their providers. Providing information about palliative and patient care groups will help families expand their care team and to have a support system to make difficult care choices.

As one of the oldest patient advocacy organizations, National Tay-Sachs and Allied Diseases (NTSAD) empowers families living with the rare diseases of Tay-Sachs, Sandhoff, Gm1 gangliosidosis, and Canavan to find their voice and advocate for themselves and their children. Adults diagnosed with the Late Onset form of Tay-Sachs and Sandhoff also receive support when their long roads to diagnosis lead them to NTSAD. (Oftentimes, their "clumsiness" as children is labeled as just that—clumsy, two left feet, distracted, etc.) NTSAD gives adults and their loved ones caring for them the tools to navigate the issues that may arise because of these diseases.

NTSAD customizes support to meet families' individual needs when they're ready to connect. In doing so the following is offered to all families:

- Support for the whole family
- Peer connection
- Internal and external resources regarding therapies, equipment, and/or navigating clinical trial and natural history studies

- Disease specific information
- 1:1 Zoom calls
- Monthly newsletters
- Research updates
- Annual Family Conference
- Regional meetups
- Family support funds
- Bereavement support

As with many patient advocacy organizations, there is no endpoint to the support NTSAD offers. Families find a sense of belonging and connection to, and derive support from those living similar lives with similar conditions, from diagnosis through bereavement. No matter where a family is on their journey, NTSAD is committed to meeting them where they are and walking this path together. As NTSAD's mission states, *"Supporting families is the center of everything we do."*

Resources For Families Living With Rare Diseases

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Families grappling with a rare disease diagnosis for their children face a myriad of practical, emotional, and support needs. On a practical level, navigating the complex healthcare system becomes a primary concern, involving frequent medical appointments, specialized treatments, and potential financial strains. They experience a rollercoaster of emotions, from shock and grief to anxiety about the uncertain future and often feel isolated and alone. The need for emotional support becomes paramount and connecting to others facing similar challenges is a vital source of understanding and strength. A robust support system that includes healthcare professionals, community organizations, and peer support becomes crucial in helping families navigate the unique challenges that come with caring for a child with a rare disease.

Presented here is a compilation of resources specifically tailored for the rare disease community. The organizations listed focus on providing rare disease families the information, assistance, and support they need while navigating their journey with their child. Please note that the following list of resources is not exhaustive but rather a selection of a few key options.

General Rare Disease Resources

The **National Organization for Rare Diseases (NORD)** is dedicated to fostering practical, meaningful, and lasting change to empower individuals with rare diseases to lead fulfilling lives. Through daily efforts, NORD strives to enhance care, propel research forward, and influence policy in a purposeful and comprehensive manner, all aimed at uplifting the rare disease community. Their website includes a section for patients to access a variety of assistance programs. For more information, visit https://rarediseases.org/

The **EveryLife Foundation for Rare Diseases,** found at https://everylifefoundation.org/, is a nonprofit organization that is committed to empowering the rare disease patient community through advocacy for impactful, science-driven legislation and policies. The foundation does not speak for patients but rather provides training, education, resources, and opportunities to amplify their voices. The website's resources page, https://everylifefoundation.org/financial, is a valuable hub offering information on organizations and foundations that provide medical support, nonprofit grants, scholarships, and travel assistance for patients and their families dealing with rare diseases.

Global Genes is a non-profit organization that is dedicated to alleviating the burdens of rare diseases globally. Through initiatives such as Rare Disease Day, observed annually on the last day of February, Global Genes raises awareness and advocates for the unique needs of individuals and families affected by rare conditions. The organization fosters a sense of community, amplifies patient voices, and works towards increased research and funding. Global Genes provides practical support through resources and educational programs. To find more information, visit their website at https://www.globalgenes.org

The **Genetic and Rare Diseases Information Center (GARD),** accessible at https://rarediseases.info.nih.gov/, is dedicated to supporting individuals living with rare diseases and their caregivers. Its mission is to provide accessible, free, and reliable information in an easy-to-understand format. By offering valuable resources, GARD aims to empower those affected by rare diseases by facilitating informed decision-making and, thus, enhancing the overall well-being of individuals and their caregivers. The center plays a crucial role in bridging the knowledge gap, ensuring that individuals facing rare diseases have the necessary information and support to navigate their unique healthcare journeys.

The **B Brave Foundation** is a non-profit dedicated to enhancing the lives of children and families affected by rare, incurable neurological disorders. Their mission focuses on providing practical support, fostering improved communication between families and medical providers, and offering caregiving resources. By addressing the unique challenges posed by rare, neurological conditions, the foundation is a valuable resource for both families and healthcare professionals. Visit the B Brave website at https://bbravefoundation.org/

Disease-Specific Resources

The **Batten Disease Support and Research Association (BDSRA)** provides invaluable support and resources for individuals and families affected by Batten disease, a rare and fatal inherited disorder of the nervous system. Their website, https://bdsrafoundation.org/, offers educational resources, support services such as peer networks, and information about ongoing research and clinical trials.

The **International Rett Syndrome Foundation** is a non-profit organization dedicated to supporting families affected by Rett Syndrome, a rare genetic neurological disorder primarily affecting girls. Providing a wealth of resources for families, caregivers, and healthcare providers, the foundation also facilitates connections among individuals living with Rett Syndrome through initiatives like an annual family conference and scientific meeting. For more information and support, visit https://www.rettsyndrome.org/

The **National MPS Society,** supports families impacted by the rare, genetic diseases of mucopolysaccharidosis (MPS) and mucolipidosis (ML). For more information, visit their website at https://mpssociety.org/

The **National Tay-Sachs and Allied Diseases Organization (NTSAD)** provides support to families affected by Tay-Sachs, Gm1 gangliosidosis, Canavan, and Sandhoff diseases. For more information, visit their website at https://ntsad.org

The **Trisomy 18 Foundation** is dedicated to providing resources for families, health care professionals, and researchers to promote understanding of the diagnosis so that families can make informed decisions about their child's healthcare. For more information, visit their website at https://trisomy18.org

The **United Leukodystrophy Foundation (ULF),** was established in 1982 to address the needs of families who are newly diagnosed or living with leukodystrophy. Their website, https://ulf.org/, provides disease information and education for families and health care professionals caring for children with various types of leukodystrophy. The foundation offers an annual family conference, virtual caregiver support groups, and links to Facebook pages for families to connect with others with similar leukodystrophy diagnoses.

Parents of Children with Rare Diseases Need a Break

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When Jonathan and Holly Cottor welcomed their second-born son in 2001, they felt more prepared to be parents again. Ryan's birth was smooth: a beautiful baby boy who received a perfect Apgar score.

Over the first few months, Ryan was a happy baby who was easy to care for, but he didn't seem as active as their firstborn. The family chalked it up to Ryan being a late bloomer.

A general practitioner first introduced the phrase "Spinal muscular atrophy," and after appointments and tests were arranged with specialists, the diagnosis came back. In February 2002, Spinal muscular atrophy (SMA), type 1.9, was confirmed.

According to Cure SMA, SMA is a rare genetic disorder affecting the spinal cord's motor neurons, leading to muscle weakness and atrophy. SMA is caused by the mutation or deletion of the survival motor neuron 1 (SMN1) gene, which is responsible for producing a protein called SMN. Without sufficient SMN protein, the motor neurons progressively degenerate, resulting in muscle weakness and loss of movement control.

At the time of Ryan's diagnosis, the Cottors were living in London, England, for Jonathan's career. There were no cures or treatments for SMA. The Cottors were told to "take him home, love him, and don't expect him to celebrate his second birthday." Upon their return to Arizona, they celebrated Ryan's second birthday with family, friends, and love. Then, Ryan celebrated his third birthday, then his fourth, then his fifth, and continued to celebrate until his death in December 2018 at 17.

Ryan had an infectious outlook on life, with a cheeky smile describing himself as "moldy cheese," way past his expiration date. His life was beautiful but much too short.

Today, science and medicine have made significant strides in treating SMA. When Ryan was 15, the first FDA-approved SMA treatment, Spinraza, became available. Spinraza required three initial "load-in" doses and a maintenance dose every four months thereafter.

When Spinraza first came onto the market, the initial cost was $750,000, and then each subsequent dose would be $175,000. The Cottor family found themselves balancing excitement with the question of how they would pay for this. Thankfully, the Cottor's insurance approved the treatment, and Ryan showed immediate benefits, even sitting on the floor, unassisted, and stable for the first time in his life.

Today, a child born with SMA will have a very different life trajectory than Ryan. Because of the tireless work of the SMA community, every U.S. State now includes SMA as part of its newborn screening program. If identified, families can immediately begin another FDA-approved treatment like Zolgensma.

Through infusion, Zolgesma delivers a new, working copy of a human SMA1 gene to children under two.

While there is still no cure, these treatments have given hope to the families and parents who are affected by this rare disease.

There are thousands of different rare diseases, and treatment options can be limited, which adds to the stress and suffering children and their families endure.1 Given that most rare diseases occur in childhood, caregivers are faced with immense pressures and challenges throughout an entire child's journey.2 And according to a 2020 systematic review of studies, parents of children with rare diseases experience a reduced Quality of Life compared to parents whose children are healthy.[1]

"I used to say that I wouldn't wish our life on any family," Jonathan said. "But I stopped thinking that way because our son had an amazing life."

Jonathan Cottor knows firsthand that a family's first wish for their child affected by a rare disease or a medically complex diagnosis is a cure. At the same time, they commit themselves to providing the often complex and intensive care required to ensure their child's health.

This reality can leave parents and caregivers exhausted, with no place or person to turn to for rest or sleep. Relatives and friends who step up to help do not typically have the skills or training to provide the required care. This harsh reality and trying to balance the more mundane day-to-day life tasks is a near impossible task.

"That's how I know the second wish parents and caregivers have is for sleep," Jonathan said. "If parents could just get a moment's rest to recharge, it would be a tremendous help to keep them going and prepare for the next challenge, and the one after that."

That mindset helped fuel Jonathan to co-found Ryan House in Phoenix, AZ., one of the first dedicated pediatric palliative care homes to open in the U.S. Ryan House is an extension of "home"–providing safe and essential support for children, so their parents and caregivers can take respite, knowing their child is safe and well cared for.

Intimately familiar with the glaring need and being led by his north star in Ryan, Jonathan founded the National Center for Pediatric Palliative Care Homes (NCPPCH) to establish a national collaborative effort to scale, strengthen, and sustain children's respite, palliative, and hospice home programs across the U.S.

The NCPPCH organization, https://www.ncppch.org/, works within the professional community to solve education strategies on the more complicated concepts of palliative care and the often misunderstood hospice language. NCPPCH aims to create a collaborative national center for shared learning, understanding existing solutions, and addressing Federal and State legislation and policy gaps. Its mission is to champion practical needs to scale, strengthen, and sustain community-based pediatric respite, palliative, and hospice home programs around business model optimization, licensing, and reimbursement methods.

NCPPCH has created Children's Respite Homes of America (CRHA), https://childrensrespitehomes.org/, as the public-facing brand to focus on a simplified message for the general public around the need for respite services. CRHA also operates as a fundraising arm of NCPPCH, telling the stories of the unmet needs of medically fragile children and their families while fundraising to support opening more pediatric palliative care homes nationwide.

Another critical element of NCPPCH is to openly collaborate with other associations, coalitions, and organizations that share a common purpose in enhancing the quality of life for medically fragile children and families, recognizing that together, there's a stronger voice to improve healthcare delivery systems.

The need for dedicated children's respite services and support for medically fragile children and their families is clear. By taking action, a more inclusive and supportive healthcare system that meets the needs of all children, including those with rare diseases and significant medical complexities, can be a reality. A community-based children's respite home is an extension of a family's own home and a temporary haven. If you want to get involved and learn more about existing programs, emerging programs, or where talks of additional homes are happening, please visit https://childrensrespitehomes.org/ or https://www.ncppch.org/.

"We invite all parents and caregivers of children with rare diseases and professionals supporting them to learn more about our work and join us in our vision of growing pediatric palliative care homes to every state in the U.S.," said Jonathan Cottor. "This is not an easy journey, but we know the rewards will improve the lives of so many children and families who do not have the care and support they need for their difficult journey."

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Items of Interest!

Please help us keep the items of interest up-to-date. Share your news, upcoming conferences, or webinars. Are there particular podcasts that may be of interest to our readers? Send any items of interest to Christy at Christy.Torkildson@gcu.edu**. Thank you.**

1. **Pediatric Palliative Care Webinar Series for 2024 has been announced. Calendar and more information, including how to register can be found at** www.ppcwebinars.org

2. **UPCOMING Conferences:**

   - **AAHPM & HPNA Annual Assembly 2024** will be in Phoenix, Arizona in March. Register for more information.
   - **The 2024 NHPCO Interdisciplinary Team Conference** will be held in April Virtually. More information can be found on the NHPCO website.
   - **The 2024 NHPCO Leadership Conference** will be in September 2024. The call for proposals opens in March! More information can be found on the NHPCO website.
   - **Have a conference to submit/share – send us the information to** Christy.Torkildson@gcu.edu.

3. **Subjects and Contributors for Future Issues of this E-Journal**
   Our future issues will be centered on the following main themes. If you have any thoughts about these or any other topics, contributors, or future issues, please contact **Christy** at Christy.Torkildson@gcu.edu or **Melissa Hunt** at melissa.hunt@optum.com.

   Issue Topics: 2024

   - **Let us know your ideas!** What topics would you like to see addressed in the Pediatric e-Journal? Issues will be distributed in February, May, August, and November.
   - **Issue 75** is our celebration issue! Please share your wins in pediatric palliative and hospice care. What is the state of pediatric palliative and hospice care in your region? In your state? Any new programs? Ongoing challenges? Distribution will be May, 2024 and articles are due by the end of March, 2024.
   - **Issue #76:** Social media, technology, communication
   - **Issue #77:** Home Care

4. **Our 75th Issue will be distributed in May of 2024!** Please share your news! What is happening in your region, or in your state? How are children with serious illnesses able to access services in your area? Any new programs? Any programs celebrating special anniversaries? We welcome your contributions and your suggestions. Questions, please contact co-Chairs **Christy Torkildson** or **Melissa Hunt** at Christy.Torkildson@gcu.edu or Melissa.Hunt@optum.com

5. **NHPCO Pediatric Website Pages have been updated for easier searching!**
   NHPCO Palliative Care Online Resources: NHPCO has a variety of pediatric hospice and palliative care resources available at www.nhpco.org/pediatrics. Also, more palliative care resources are available at www.nhpco.org/palliativecare, including:

   - Community–Based Palliative Care
   - Legal and Regulatory Resources
   - Webinars and Courses
   - Brochures in English and Spanish for families
   - Plus, more for NHPCO members

6. **Questions about Concurrent Care?** Dr. Lisa Lindley and her team have created a wonderful website full of resources and information. You can access all the information for **Pediatric End-of-Life Care Research** at https://pedeolcare.utk.edu/

7. **Palliative Care Programs and Professionals:** Founded in 1978, National Hospice and Palliative Care Organization (NHPCO) is the world's largest and most innovative national membership organization devoted exclusively to promoting access to hospice and palliative care and to maintaining quality care for persons facing the end of life and their families. Join NHPCO Today!
Individual Palliative Care Membership
Palliative Care Group Application - Save by registering your entire team

8. **Hospice Action Network blog:** The Choice to Keep Running: Advocating with Grief is a two part blog about Trey Gibson, who lost his daughter to Diffuse Intrinsic Pontine Glioma (DIPG). Trey is a member of the Louisiana Palliative Care Advisory Council and has turned his grief into action by advocating for better palliative care for seriously-ill children and adults.

Note: *Many of the pediatric resources are open access as a community service by NHPCO and membership is not required. However, we would love to have you join our community of vested professionals focused on quality palliative and hospice care throughout the lifespan!*

Do you have a resource that would be helpful for others to know about? Please send the information to **Christy** at Christy.Torkildson@gcu.edu and we will add it to the Items of Interest.

Previous Items of Interest:

9. **Did you know that the State Coalitions from Pennsylvania, Illinois and California, with support from the Shiley Haynes Institute for Palliative Care and the HAP Foundation,** host monthly, affordable webinars with continuing education units available? You can register for one or the entire series, with discounts for multiple registrations. For more information, review the PPC website.

10. **On that same note, did you know there was a network of state coalitions and folks interested in helping with or starting a state coalition for Pediatric Palliative Care?** For more information, contact **Betsy** betsy@ppcc-pa.org

11. **End-of-Life Nursing Education Consortium (ELNEC) project** has several upcoming courses; if you are faculty, you can get free access to the curriculum for your program/courses you teach.

12. Recently, the **California Advocacy Network for Children with Special Health Care Needs** announced their foundation had committed to increasing access to journal articles that may be difficult for family members and non-profit staff to access to improve "effective and equitable systems" as "access to scholarly work is essential to system improvement." More information can be found on the California Advocacy Network for Children website. It may be helpful to contact your state's chapter to determine what resources they may have!

13. **Courageous Parent's Network** has a wealth of resources for parents, caregivers, and providers. The list is too long to add here so please check out CPN's website.

14. **The Pediatric Palliative Care Coalition of Pennsylvania, the Greater Illinois Pediatric Palliative Care Coalition, and the Funeral Service Foundation have created a community resource to guide families through the funeral/memorialization planning process:**

    When a Child Dies: Planning Acts of Love & Legacy

    This resource is available in both English and Spanish and is **FREE,** thanks to generous funding from the Funeral Service Foundation. You pay only a nominal shipping fee. More information can be found at When A Child Dies.

15. ***Pediatric Go Wish Together:*** A conversation game for parents and pediatric caregivers; developed by Meghan Potthoff, Ph.D., APRN-NP, PPCNP-BC, CPNP-AC in collaboration with Coda Alliance. This game is "developed to help parents navigate the unimaginable journey of their child's illness." "It is a tool that provides parents and providers a way to think and talk about what's most important to the child". More information can be found at Pediatric Go Wish Together

16. **Have you heard of the new organization PallCHASE: Palliative Care in Humanitarian Aid Situations and Emergencies?** Their primary ambition is the relief of suffering, and their purpose "To work in partnership through a visible and effective network to advocate for palliative care integration in humanitarian situations or emergencies...". Please visit their website for more information, healthcare professional training and resources in a variety of languages! Check out their website!

17. **Another great new group is the Child Life in Hospice and Palliative Care Network, which provides child life specialists working in hospice or palliative care access to resources, education, research, and networking opportunities to establish and provide best practice care for patients and families experiencing a serious illness.** They are requesting that interested members fill out a brief survey, sign up today at CLHPN

18. **A Toolkit of Autism, Grief, and Loss Resources by Hospice Foundation of America**
The toolkit will include a variety of materials and resources, such as:

    - suggestions for responding to the grief experiences of autistic adults;
    - ways to provide for choice and inclusion in rituals;
    - tips for communicating the news of death;
    - social stories on grief for adults;
    - videos about grief, including interview clips with autistic adults and their families;
    - two complimentary continuing education (CE) programs for professionals; and much more!

19. **A resource for pregnancy or infant loss is Share: Pregnancy & Infant Loss Support.** Share was started in 1977 in response to the urging of one bereaved family by Sr. Jean Marie Lamb, OSF. Initially providing support groups, they now offer online support groups, education, and support for families and caregivers.

20. **The Pediatric Palliative Care Coalition of Pennsylvania (PPCC) has made a new resource available – a Sibling Grief and Bereavement Toolkit.** This Toolkit has been developed to address the needs and concerns of children and teens who have experienced the death of their sibling with medical complexities. Please see the associated article in this edition! Below is a link to the toolkit and one of the activities – **"Make a Feelings Chart"**.
View the PPCC Sibling Grief and Bereavement Toolkit.

    Toolkit Activity

    For children who are grieving the loss of their siblings, returning to school can be a difficult transition. Check out this month's highlighted activity from the toolkit that may help children and teens in the upcoming school year.

    

    Make a Feelings Chart by downloading the activity. *PPCC invites you to share this information with parents, caregivers, medical professionals, providers, therapists, etc.*

21. **Pediatric Hospice and Palliative Care Resources:**

    - **CaringInfo,** a program of the National Hospice and Palliative Care Organization, provides free resources to help people make decisions about end-of-life care and services before a crisis. www.caringinfo.org

NHPCO's Palliative Care Resource Series includes pediatric palliative resources like:

- Communication Between Parents and Health Care Professionals Enhances
- Satisfaction Among Parents of the Children with Severe Spinal Muscular Atrophy
- Consideration for Complex Pediatric Palliative Care Discharges
- Songs of the Dying: The Case for Music Therapy in Pediatric Palliative and
- Hospice Care
- Nonpharmacological Pain Management for Children
- Sibling Grief
- Pediatric Pain Management Strategies
- Communicating with a Child Experiencing the Death of a Loved One: Developmental Considerations
- In an effort to standardize the medication coverage process for children receiving concurrent care, the NHPCO Pediatric Advisory Council developed a new resource for providers titled
- Determination of Hospice Medication Coverage in CHILDREN.

22. **Trends in Pediatric Palliative Care Research**
Every month, PedPalASCNET collects new pediatric palliative care research. For past lists visit their blog, browse in their library, or join the Zotero group. View the New Citation List in their library.

23. **Palliative Care Resources for Nurses, Patient Care Support Staff, and Families of Patients by Life and Death Matters,** https://lifeanddeathmatters.ca/ offers texts, workbooks and resources for providers and family members. Although primarily focused on adults they reference across the lifespan with sound principles that are useful no matter the age of your patients.

The text, workbook and companion resources support nurses and nursing students (in Canada and USA) to develop the knowledge, skills, and attitudes for integrating a palliative approach and providing excellent end-of-life care.

Essentials in Hospice and Palliative Care: A Practical Resource for Every Nurse
Textbook: 978-1-926923-11-6 | Workbook: 978-1-926923-11-6
https://lifeanddeathmatters.ca/product/palliative-care-nurse/

The text, workbook and resources, based on national competencies, will help nurses:

- Develop best practice interactions
- Decrease fears and increase confidence and competence in caring for the dying person and family
- Develop ethically and culturally competent practices with touchstones and by relating experiences

Also available for this title: Videos, Podcasts, PowerPoint™ Presentations and NCLEX-style questions

Palliative Care Resources for Care Aids and Family

- *Integrating a Palliative Approach: Essentials for Personal Support Workers, 2nd Edition; 1926923162*
- *Integrating a Palliative Approach: Essentials for Personal Support Workers 2nd Edition – Workbook; 9781926923178*

This textbook is a rare text that engages you with its warmth and heart—an essential resource for all frontline caregivers and family members supporting loved ones. The companion workbook engages learners through reflective activities, crossword puzzles, worksheets and interactive projects. Video and podcast libraries available on the Life and Death Matters website. https://lifeanddeathmatters.ca/

Please note the archived issues are available as a community-service by NHPCO and can be found at www.nhpco.org/pediatrics or by reaching out to Pediatrics@nhpco.org.

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