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Parents of Children with Rare Diseases Need a Break

Kasey Kaler,
Communications Strategist
kkaler@ncppch.org

and

Jonathan Cottor, MBA, MPH
Founder/CEO
National Center for Pediatric Palliative Care Homes/
Children's Respite Homes of America
Scottsdale, AZ
jcottor@ncppch.org

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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