Uniting to help families of children with rare diseases

by Nina Verin

Finding out that your grandchild has been diagnosed with a rare disease is devastating. After months of uncertainty and multiple health challenges, my one-year-old grandson, Malcolm, was diagnosed with Koolen-de Vries Syndrome (KDVS). Soon after this, I turned to the Web to learn all I could it. The first and most critical information I gleaned is that as disparate as rare diseases are, patients share many common concerns and frustrations. For example, for one-third of the people with rare disorders, getting an accurate diagnosis can take one to five years. Patients must often travel long distances to visit the few doctors knowledgeable about their illnesses, and the costs involved with diagnoses, treatment and other related expenses are exorbitant.

Over 6,000 different rare diseases have been identified to date, affecting the daily life of more than 30 million people in the U.S. alone. In this country, a condition is considered rare if it affects fewer than 200,000 persons combined in a particular disease group. The condition, KDVS, affecting Malcolm is almost 10 times as rare as the standard. To date, only 250 people have been diagnosed with Koolen-de Vries Syndrome.

I live 1,500 miles away from my first-born grandson and his parents. Upon learning about the nature of Malcolm's condition, I immediately sensed that anything I offered to his family, in the way of long distance support would have minimal impact on their day-to-day life. I felt helpless and adrift. The more digging I did on the Web, the more aware I became of the struggles of all people with rare conditions. I felt compelled to try to "DO SOMETHING" ! I reached out to the foundation, Supporting Families with Koolen-de Vries Syndrome (501c3), which was formed to support families and fund research for KDVS. Recently, through my work on their fundraising committee, I discovered that in 2014, over 60 countries participated in a worldwide Rare Disease Day. The overall objective of this consequential day is to raise awareness amongst the general public about rare diseases and their impact on patients' lives. There is much comfort knowing that people all over the world are advocating for more research funding, orphan drug trials and other pressing matters.

Support groups such as National Organization for Rare Diseases (NORD) have worked aggressively in the past to draw attention to people with rare diseases, with a focus on the lack of treatment options.

Still, momentum must be built along many fronts even as new web-based support groups continue to proliferate. Patients and families cannot do this critical public health advocacy alone.

All of us know someone with a Rare Disease. The objective of our first annual Rare Disease Day Awareness Event is to bring people together to share resources and information, provide hope and support for families in the Rare Disease community. Additionally, we hope to broaden the profile of the Supporting Families with Koolen-de Vries Syndrome organization.

We are fortunate to live in a community with extensive medical resources, and yet there are some among us who feel alone. Rare Disease Day is an opportunity to share messages of hope and stand together to raise widespread awareness.

For more information about local Rare Disease Day events, contact Nina Verin ninaverin@gmail.com 651.230.5225

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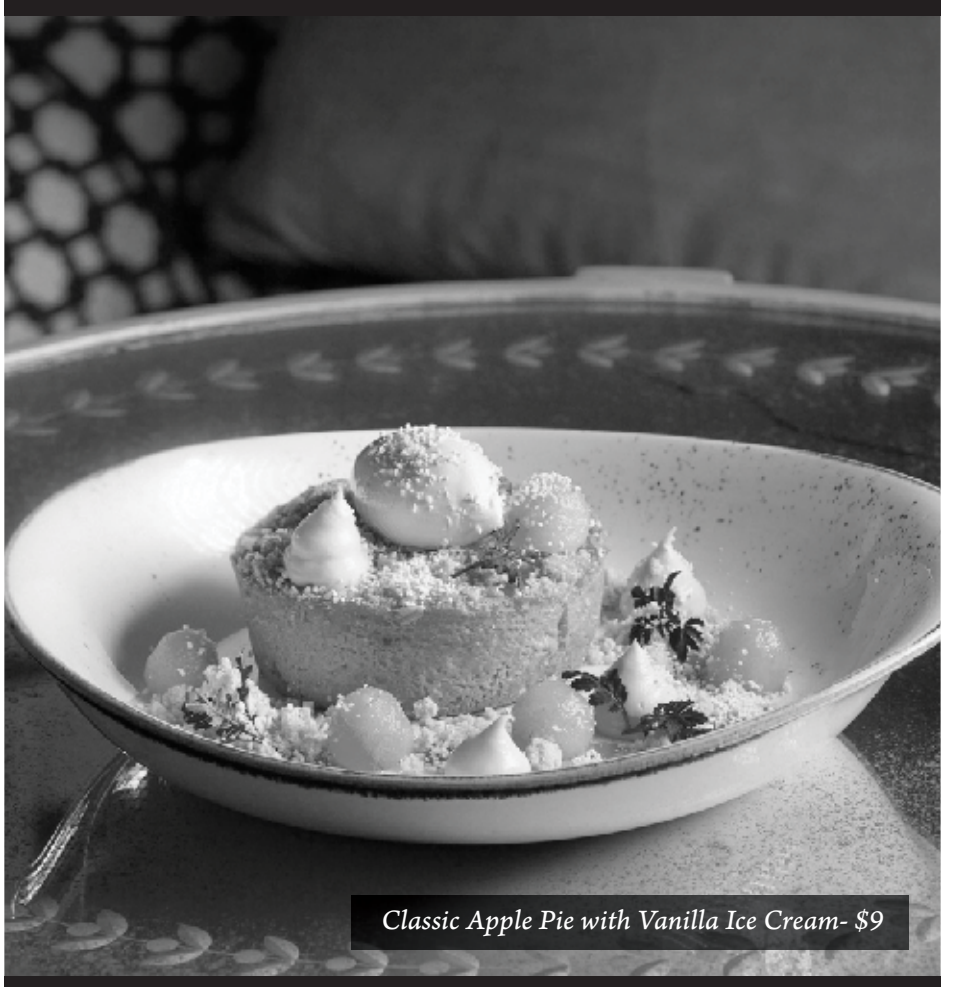

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