



I ask myself how I can best help a future parent who will be in my shoes at some point down the road

In anticipation of [Rare Disease Day](#) on February 29, we are honored to share this guest post by Terri Granard. She is a caregiver for her son, Eli, who was diagnosed with the rare chromosomal abnormality Ring 14, and one of the authors of the book [Uncommon Challenges; Shared Journeys](#).

We all have something to deal with in our lifetime and you do the best you can with what you've got. It's just life; that's how it is. I'm one of many moms around the world who's the caregiver of a child with a rare disease or disorder. My son, Eli, was diagnosed with Ring 14 (chromosome ring 14 deletion syndrome) when he was six months old after suffering a stroke and seizure. There are fewer than 300 people worldwide who have this rare condition.

Eli can suffer from one to 10 or more daily seizures that may include body tremors, major body thrashing and apneic episodes that cause him to stop breathing and require oxygen. A vagus nerve stimulator implanted in his chest sends waves to the brain to "interrupt" a major seizure. Because of the strokes and seizures, Eli has had to learn and relearn basic skills like walking and sign language.

There is no cure or drug therapy for Ring 14. The greatest spark of hope comes from a research project in Rome launched in 2006. Using the child's own stem cells rather than embryonic cells, we're hoping to find a way for stem cells or bone marrow to duplicate normal cells, or find a way for stem cells to rejuvenate.

Knowing what I know and having experienced all that I've tackled, *I ask myself how I can best help a future parent who will be in my shoes at some point down the road.* I've decided that the biggest contribution I can make is to help increase awareness of Ring 14, and let people know that it's one of many diseases that can be detected through newborn screening.

Newborn screenings

Newborn screenings are done when the baby is two days old. A nurse pricks the baby's heel to collect a few drops of blood, which are analyzed to detect various diseases and disorders. Since individual states regulate newborn screening, the diseases that are screened vary considerably from state to state in the US. All 50 states screen for congenital hypothyroidism, galactosemia, and phenylketonuria (PKU). Organizations such as the March of Dimes and the American College of Medical Genetics suggest more than two dozen additional tests. The most thorough screening panel checks for about 40 disorders.

Newborn screening would have shown that Eli would develop Ring 14. Although it wouldn't have changed the inevitable, it would have provided my husband and me with peace of mind and helped us anticipate what we would be facing. I would have been better prepared to care for him sooner.

Sharing support

We first learned about this disorder after searching the internet and reaching out to other parents of children with similar illnesses. I've become an advocate for children diagnosed with Ring 14 and other metabolic problems. I counsel parents about what to expect and where to find available resources. Some of my closest relationships are with people we've met online. Eli and I have flown from Los Angeles to Tennessee twice to visit a family that has a child with Ring 14. It's like meeting someone who has not only the same blood type as you, but the same fingerprints. I talk with people from all over the world—and I use Google Translator a lot!

Ring 14 outreach

We recently established a non-profit, Ring 14 USA Outreach, to help raise awareness, research funds and provide support for children affected with Ring 14. Plus, we just launched our new web site (<http://Ring14usa.net>). I'm partnering with five other mom caregivers across the country from Virginia, Washington, D.C., Pennsylvania, Oklahoma and Florida, to promote it. We have a few sponsors and we're looking for more.

Eli is an angel. He's happy. He enjoys his life. Like any mom, I want the best for my son. I'm doing whatever I can to provide the best care for Eli, help others by sharing my experience, and support research to find a treatment or cure for Ring 14. It's about giving people hope, even if it's a shot in the dark. There are advances in medicine every day. That's why Eli is alive. You have to look to other parents, and you have to be there for each other.

(Image courtesy of Terri Granard).

About Wendy White



Since founding Siren Interactive in 1999, Wendy has been recognized as a thought leader at the intersection of niche pharma brands, patient empowerment and online marketing. Her vision for how the internet can facilitate interactions and provide crucial information that patients, caregivers and their healthcare providers previously struggled to find has propelled Siren to the forefront of relationship marketing for rare disorder therapies.

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