

Why Us? Grappling with the Realities of a Rare Disease Diagnosis

By Kelly Huber

ckhuber99@gmail.com

with Deb Lee Gould, MEd

deb@fodsupport.org

The shock, numbness, tears, sorrow all seem to come with a rare disease diagnosis. The question of “Why Us?” seems to follow as families try to grasp the realities. If you asked, many could tell you what they were doing at that very moment of diagnosis. They could tell you the weather, the color of the room and how it was arranged, as time stands still for that moment. Rare Diseases can feel like a death sentence especially since many that came before them have died from the same diagnosis. Families are in this tug of war with reality of the disease and reality of the world in which they must not only survive but thrive.

Just like death, each individual and family deals with the disease and the emotions from the disease in different ways. There are some realities that are certain for each person and family. Rare Disease Diagnosis, for many, may mean not feeling whole. Something is missing whether it is visible or not. Rare means that only you may know the answer to your own pains and aches as the medical world struggles themselves with funding, politics, and the red tape to find those cures. Being diagnosed can come as a huge blow to the family as many do not see this rare diagnosis coming. More times than not, there is no family history. It is completely shocking and heartbreaking.

There can be so much emotional strain that comes with grasping the rare diagnosis. The worrying can be immeasurable, sometimes unbearable. A rare disease diagnosis is going to come with grief. The grief of “Why Us?” can seem unbearable on that first day and every day that these diseases show more symptoms of struggle.

Again each family deals with this in a different way, and hopefully over time in a proactive empowering way. However, the grasping of not knowing what life will be like, what does this all mean, and what will this family member have to endure is probably one of the hardest and complex issues in our rare communities. These families need to be surrounded with love and compassion to get through their new way of life. Even if you don't understand it, just being there for the family and helping them in any way can bring the community of rare disease and the rest of the world a little closer.

Denial and acceptance are kind of like oil and water - they don't mix well, but if you stir enough you can create a rainbow. In my own family, it was much easier for us to sort of deny anything was wrong. To smile through the clouds and rain. To feel numb and empowered all at the same time takes mastering to a whole new level of life. There is a little bit of denial in all of the 'rare.' We want to believe that we can do anything we put our minds to and the truth is we can! We just have to do it in different ways and that is where the acceptance piece gets mixed in. We have to eventually accept that our lives will be different. Our lives are not going to be the same as our neighbors, our friends, or our other family members. At each milestone, when living with a rare disease, it often starts all over again - the shock, the grief, the denial and acceptance that maybe what I was doing today, I won't be able to do tomorrow. But we keep moving forward as best we can.

If you ever get a chance to get to know someone with a rare disease I am certain your life will never be the same. There is so much appreciation for life and what our real reason for being is. I have watched 'rare' defy all odds with little resources, little help, little understanding and individuals and families still find the strength to carry on each day despite physical, emotional, financial, etc., struggles. They no longer ask "Why us?" - they start to say "Why not us?"

Speaking for many in the rare disease community, I strongly feel we are the perfect people to advocate for these diseases. We have faith in mankind even when the world seems to turn on us. We are the strong, the proud, the resilient, and in my humble opinion, the most amazing humans on the planet! We are in many ways 'super heroes' carrying an array of new bright light and wonder for the world of those that came before us...and for the world that is yet to travel our road with us.