BCM FAMILIES FOUNDATION

28 February 2019

Rare Disease Day

RAREDISEASEDAY.ORG

My passion for BCM

-Corinne

There is one moment that defines us all. Maybe it was that trip to Guatemala. Or the moment you were told your mother had cancer. Maybe it was the first time you held a guitar or a paint brush. Or maybe it was the first time you made the most delicious meal and you wished you could share it with the world.

My defining moment came the moment I had my first son. We named him Ezra and you can find his picture above. His name means help. He has helped me in ways you cannot even begin to imagine. My little boy is now 7 and has been diagnosed with an extremely rare eye disease called Blue Cone Monochromacy. We call it BCM for short. It is an x-linked recessive trait that is expressed in males. He has impaired color discrimination, low visual acuity and photophobia due to the dysfunction of his L and M cones. Ezra is legally blind and cannot decipher colors.

BCM is an extremely rare retinal disease. It is estimated that 1 out of 100,000 males will experience this disease. To put this in perspective, albinism affects primarily 1/17,000 people. BCM is an orphan disease.

I as a mother, am desperate for a cure --and a cure is within our reach. We recently attended the first conference for those families affected by BCM. A donation to the BCM Families Foundation would mean the world to us. A donation for us is hope. Hope that our boys and men would see the world as we see it. They would become able to do some things that we take for granted, like seeing a rainbow. My son has never seen a rainbow in its full glory. I want to see him reach his full potential in life without his vision ever holding him back. I would gladly give all of my vision to him if I could.

This is why I'm passionate about finding a cure for BCM just like you might be passionate about music. Or travel, animals, or people. Or maybe you are passionate about computers. You can be passionate about a lot of things.

I've turned my passion and desire into finding a cure for my family. It's for my little girl who may be a carrier. It's for my future grandchildren and great grandchildren who may be affected by it. We desperately need a cure. For our family. For all BCM families.

I am writing this in support of BCM Families Foundation