

A Ray of Hope Amid the Sorrow

The diagnosis of a rare disease punctuated by the gift of life, from an ECD spouse perspective.

It was Christmas Eve of 2017 when Ish went for his annual physical and his PCP wanted him to follow up with a specialist regarding a lump in his knee that had been there for many years; I had seen it for at least six years (since we started dating). I used to joke that it was his second knee. It never bothered him; he was in good shape. Ish ran two half marathons that year, so we never thought so much into this.

Few weeks passed between trying to get appointments, biopsy, and every other test you can think of, and sometime mid-Jan we found out we were pregnant. Our first pregnancy! Young married couple, madly in love and finally starting our family – most couples dream come true! When we went to our first OBGYN appointment around eight weeks of pregnancy with sky-high excitement, the doctor brought her doppler and couldn't find a heartbeat. In our first shock of 2018, we miscarried and our worlds shattered.

Now looking back, I believe this incident bought Ish and me closer. A shared tragedy can make a relationship stronger or it can break it; ours definitely became stronger. We took solace with the belief that God had a better plan for us, and our baby girl was with Him.

A few weeks later in early February, one Tuesday night, his PET scan results were released through his NYU Patient Portal. Just when we thought things couldn't get worse, Ish opens the report and reads that the scan detected several more tumors across his body. We were shocked to read the report. Without being able to reach my doctor (it was around 10 pm), we sent it to a friend, who was a med student. She responded back saying that the PET scan did detect some more tumors. Ish was scared; I was in shock. I had no time to think, I needed to console Ish. We immediately made an appointment for the following day with the PCP.

To this date, I am not sure how an institution like NYU could release a PET scan result without the doctor reaching out to me first.

The following morning, we walked in, super nervous, heart racing, sweaty palms, and saying my prayers under my breath. The PCP confirmed the PET scan results and our fear – he had cancer. I think my heart might have stopped and now I wanted to scream out loud and cry. It's all a blur now, but I think Ish saw me go pale and held me tight, and kept rubbing my back. Here we were with 1000s of questions racing in our minds - How bad was it? What should we do now? Wasn't losing pregnancy enough? What were we getting tested on? Why us?!?!



“Why us?!?!”

That was the low point, and believe it or not, things started to get better from there.

NYU sent us to Dr. Eli Diamond of Memorial Sloan Kettering Cancer Center (MSK) and we were able to see him that week. I have never felt better talking to a doctor as I did when I spoke to Dr. Diamond. He was extremely knowledgeable, compassionate, and we got the feeling that we were finally speaking to someone who knew what he was talking about. He suspected Ish had Erdheim-Chester Disease (ECD). He explained what ECD was and told us that it was an incredibly rare disease. A part of us was relieved that there was finally a diagnosis. Another part of us couldn't stop thinking about how we got here. It was like winning a lottery in reverse.

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Dr. Diamond did some more tests to confirm his suspicion, and it all came back with a positive ECD diagnosis. He decided to put Ish on oral chemo. Right before he started his treatment, we found out we were pregnant again! The joy and relief that news brought to us could not possibly be expressed in words. There was again a ray of hope amid all the sorrow!

The treatment started off rough, but it was made bearable as our minds were occupied with the incoming new addition to the family. The care team from MSK was always there - super responsive. Anytime Ish felt bad because of his medication, we knew whom to contact. Then around six months after he started the treatment, his PET scan showed dramatic improvement. He stayed on the medication for several more months to make sure that the disease is fully under control. And then in December 2018, we were blessed with a baby boy, who instantly made us forget all the pain and sorrow the year 2018 bought to us!