Our Families Journey with IVF

by Nicole Shen

Like many of you, our son Roman endured a medical odyssey of tests trying to figure out what was wrong. We are grateful at six months old, he was genetically confirmed with Shwachman-Diamond Syndrome. This seemed lightning fast compared to a lot of stories I have heard. However, at the time, it seemed like an eternity racing against time to save our son. The day he was diagnosed, a genetics counselor came to see us. She explained how SDS is inherited and our chances of having another child with SDS. We learned this was not a spontaneous disease our son had. This was genetically inherited from both of us. Gut punch! Our first thoughts were that we caused this. How could we not know we were carriers of such an awful disease? She went on to explain that SDS carriers usually do not exhibit any symptoms. It is inherited in an autosomal recessive manner meaning approximately 25% of our children may have this. Another gut punch! This could happen again. We originally wanted three children. When we were pregnant with Roman, we built a home for these future children. The hematologist then explained that Roman had a significant chance of needing a bone marrow transplant. His ANC was 0 with another blood line failing. She said she went ahead and ran his HLA (bone marrow type) against all the bone marrow registries in the WORLD. Not one match was found. So what we learned that morning was Roman had a serious disease, we could go through this again if we tried to have another child, AND the “medicine” our son needed could not be found. The grief was intense that day. We went home with a child who we now had confirmation was seriously ill. The family we wanted would be very risky. Would we ever have another child? Would we lose the child we had because he didn’t have a match? How can we find this match for him? It was a very heavy day. However, the

genetics counselor saw we had used fertility medicine to conceive Roman as I have PCOS. She said, "I see you are already somewhat familiar with IVF. Let me explain to you how you may be able to have healthy children in your future and perhaps make your son's bone marrow match." There was hope!

Within that month, we saw another genetics counselor and our reproductive endocrinologist to find out how we can achieve this goal. In hindsight, this was a lot to take on so early. Our insurance did not cover any portion of the IVF or testing. The doctors did believe we could make a case for them to cover this so we started writing letters to insurance companies pleading our case. This process took a year. Appeal after appeal we wrote. Letters from multiple doctors. Nothing worked. We would need to come up with A LOT of money. I was now 38 years old. We needed to figure this out quickly. We spent the next two years saving money, taking out loans, and working at as many bone marrow booths as we could registering bone marrow donors.

Three years later, still not one match was found for Roman. We had to try the IVF as well. It was a longshot with my age. I was now 40 years old. The chances of chromosome issues were about 50% on top of the 25% chance of SDS. Our first goal was to make a healthy sibling for Roman. We also added HLA testing as he needed a bone marrow match. Statistically, one in four siblings should match each other's HLA. The same chance as inheriting SDS. How were we going to make this "unicorn" baby that did not have SDS, did not have chromosome issues, AND also matched Roman's bone marrow type? Also, a reminder to us was my being 40 years old and I might not have been making many healthy eggs to use.

We started the very expensive process of IVF. This process took a few months to get started as the lab had to test the embryos and make a "probe" to test them. This involves saliva samples from multiple family members to make the test. To my luck, I was making an extremely high amount of eggs for my age. Finally, a break! This may work after all. The first round of IVF, we made 11 embryos. We sent them off for testing. Approximately 50% had chromosome issues, 25% had SDS, and one was an HLA match. The one HLA match also had SDS and could not be used. We rolled the dice and tried another round of IVF. This time we made 10 embryos. 50% had chromosome issues, 25% had SDS, and none were HLA matches. Should we stop and transfer a healthy embryo or keep trying for a

match? We rolled the dice one more time for IVF round three. This time we only made 6 embryos. Same results as round two. We were defeated and sad. My body had gone through three back to back IVF cycles. Physically I was exhausted but emotionally I was even more exhausted. The testing takes weeks. Waiting on each call was excruciating. My husband Peter was very stressed about how we would pay for all this.

Peter and I regrouped. We decided the same odd "luck" we had to both carry such a rare gene and have a child with SDS, may be the same luck we need to make this baby. Let's try ONE last round. I was willing to sell my house, my wedding ring, whatever it took. IVF round 4 we produced 16 embryos! Wow. 50% had chromosome issues, 25% had SDS, and…….we made 2 healthy HLA matches - a boy and a girl. We did it! One year of IVF. We made not one but two "unicorns" that passed every test. We transferred the female embryo as her grading was slightly higher.

We are happy to announce Luciana Violet Shen was born this past January. She is now seven months old. Her cord blood was saved for Roman. Her Chinese name is Yu meaning "to heal." Her cord blood will most likely save Roman's life one day. However, she has healed our family in more ways than one. We have the family we wanted. The grief is lifting. Years of writing appeals letters, working at bone marrow booths, saving money, four consecutive IVF cycles - she is here. We are so beyond thankful.