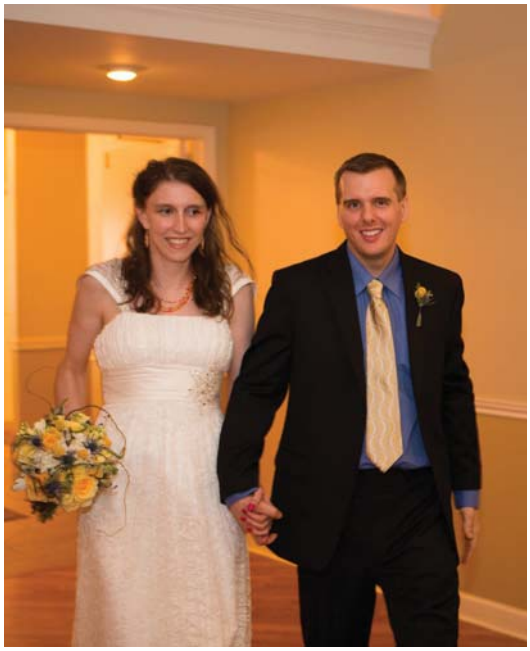
Personal Experience with Pregnancy and HCU

by Danae' Bartke

From as early as I can remember, I knew I always wanted to have my own family. I grew up in a large family; the third of 8 kids. There always seemed to be a baby around, literally!

Growing up, my cousin and I had dreams of moving to Australia and starting our families there. We were going to each have 4 kids and of course 2 sets of twins. So practical, I know. We would practice our fantastic 7 year old mothering skills on our cabbage patch dolls and my younger brothers. We were sure we were going to be fantastic mothers; my brothers might have begged to differ.

In 1995, at the age of 10, I was diagnosed with homocystinuria. We weren't sure exactly what that would mean in the long run for us; we just knew it came with deadly consequences if untreated. As puberty found its way into my life, my geneticists told me I'd likely never be able to have my own kids because of the risks that pregnancy posed to me. Homocystinuria causes an elevated risk for blood clots and so does pregnancy. It was not the news I really wanted to hear because I knew I wanted to have my own family. I eventually accepted that while I may not be able to give birth to my own kids, adoption was still a strong option. By the time I was in my early 20s, the science had started to change and my doctor told me if I wanted to have my own kids I would have to get better control of my homocysteine levels. Since I was still in college, having kids wasn't on my radar and I just brushed it off, knowing if I got my act together I could have kids one day.

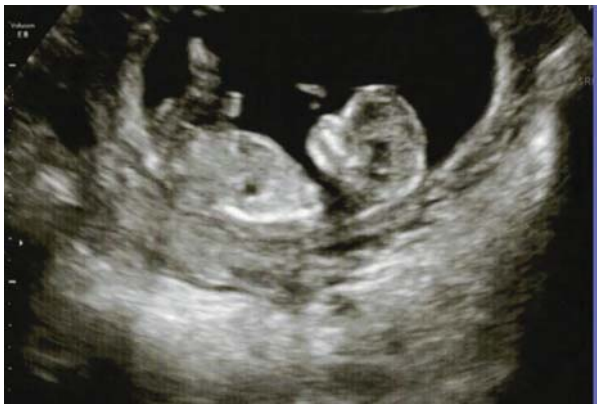


I pursued a bachelor's degree while working full time, not leaving much time to take care of myself. Because of my neglect I ended up having a blood clot. While most people don't view a blood clot as a good thing, I view mine as a life altering experience that put my life on the right path. Not long after my blood clot I started attending low protein cooking classes and metabolic meetings. It really helped me feel part of a community. I met people who had much more strict protein restrictions than myself, and I told myself if they can do it, then I can too! It was the first time I was following the diet and taking my formula the correct way.

In 2011, a couple years after my blood clot, I met the man who would become my husband. In 2014, we got married and shortly after my geneticist requested we see them to get my husband tested to see if he was a carrier. I assured them children were not on our schedule anytime soon, but they insisted – "just in case".

Regardless of the results we knew we would want children of our own, one day but we were a bit relieved when his results came back that he was not a carrier.

In May 2017, well before I was pregnant, I met with maternal fetal medicine specialist and a hematologist to discuss the possibility of pregnancy for a patient with homocystinuria. In August of that year I also started making the necessary steps to switch clinics. The genetics clinic I had been at did not have a team, rather it was just a geneticist. I knew in order to have a successful pregnancy I'd also need a dietitian. I'm very lucky to live in an area with a few options; I ultimately ended up at Ann & Robert H. Lurie Children's Hospital of Chicago. There I had a much more complete team; not only did I have a geneticist, but I had a social worker, advanced nurse practitioner, metabolic dietitian, and genetic counselor.



By the end of October, we found out we were pregnant and my first appointment with the new genetics team was in the middle of November. At my first genetics appointment with the new clinic they reviewed my health history, current diet and formula regime. They increased my formula intake by 15 grams a day as it would not be enough regardless of pregnancy. They took blood samples and got back to me with recommended changes. Because diet and pregnancy are so different, I will not go into detail about the changes they made with vitamins. They did suggest increasing my caloric intake to 2,000 calories from 1,500, which later proved to be too much.

Even with such low total homocysteine numbers the hematologist felt it was important to follow the guidelines for HCU patients and patients who previously had blood clots. At my first appointment at 10 weeks pregnant, they started me on Lovenox injections. About 1 month before my delivery they switched me to Heparin, and then discontinued it 3 days before I was induced. The day following my delivery, they started me back on Lovenox injections, and I continued them for 8 weeks post pregnancy.

Overall my health was very good during the pregnancy. The issues I did have were not believed to be caused by having Homocystinuria. In my second trimester I was told that I had a condition called Placenta Previa. I was very anxious about this because if the condition continued I would need to have a C-section. After my appointment I turned to good old Google and found out the condition was fairly common and 1 in 200 women experienced it and about 75% of the time it corrected itself, so it was very likely I would not need to have a C-section. They continued to monitor the amniotic fluid and position of the placenta until she was born. It did correct itself with time so a C-Section was not needed.

Until our gender reveal ultrasound I was very convinced we were going to have a boy, and my husband was so sure it would be a girl. In February, with our immediate families we announced we were going to have a baby girl. Regardless of gender we had the name picked out, Dana Leah Bartke. Dana after my father, for whom I was named after, and Leah after my grandfather (Lee). In April, between family being in and out of town and my busy conference schedule we planned our baby shower – it was so nice to have everyone together to celebrate the soon to be arrival our of baby girl.

In May, about 1 and a half months before Dana's due date, I was at a genetics appointment and I mentioned to them how swollen and itchy my feet had become. They were so enlarged I could no longer fit in my regular shoes and the only comfortable shoes I could wear were sandals, mostly flip flops. The swelling wasn't so bad, but the itchiness felt unbearable! Nothing I could do or take helped. I tried soaking them in hot water (later found out that made it worse) then iced them – that helped for a little bit. I tried Benadryl cream, that would help, but ultimately would end up itching again just minutes later. My genetics team found this very concerning and ran some test. While I was at my other doctor appointment later that day I got a call telling me I needed to go to the hospital for further blood work and to be monitored overnight, that they suspected a condition called Intrahepatic cholestasis of pregnancy, or just commonly known as cholestasis of pregnancy. The next day I received confirmation that I did indeed have this condition. This meant being induced at 37 weeks, because anything past that was dangerous to the health of the baby. They sent me home with medication to take daily and then had me followed by Maternal Fetal Medicine more frequently (twice a week instead of once).

At exactly 37 weeks to the day, they had me check into the hospital at 6 am and started the process of being induced. The first day was pretty non-eventful, by 10 pm I hadn't made it past 4 cm so I went to bed hooked up to a plethora of monitors and I Vs. The next day continued to also be non-eventful. At about 9 pm they came in and broke my water, which quickly sped up the process. With each passing minute the contractions picked up getting me closer and closer to having her. At 10:38 pm Dana finally was born!

This next part may be a bit scary for some, but I feel it's an important part to share. Minutes after Dana was born, I started to have additional issues. They could not get the placenta to detach, and along with the difficulty I started hemorrhaging; my uterus was not contracting properly. They immediately started me on medication to stop the hemorrhaging and started a blood transfusion. I was in a pretty weakened state of things, so the first few hours I spent under a heating blanket and Dana was in the nursery – not how any mother anticipates the first moments/hours of their child's life. Most importantly though, Dana was healthy – even at 37 weeks she weighed just under 8 lbs. I later learned that roughly 18-25 percent of births involve a postpartum hemorrhage – something they do not talk about at all in any birthing or pregnancy class.

Four days after Dana was delivered both of us were cleared to go home. We finally got to be a family of three in our own home. Life has definitely been an adjustment. My homocysteine levels continue to stay down around 15-17 and immediately after Dana was born they had me go back to 20 grams of protein a day. I had to reestablish new routines for my formula and eating that worked with having a baby – I didn't anticipate that being so difficult, but it's doable.

Today, Dana is a now a very typical 2 year old. She is talking, running, and into everything. She loves baby

dolls, bunny rabbit stuffed animals and loves to sing. Some people have asked despite the cholestasis and my delivery experience would I do it again, yes – and eventually we do plan to have a second.

My advice if you have Homocystinuria and want to have children is this:

1. Get your levels down into safe range – whatever your doctor deems that is. Some might say 50, some might say 25 – it's doable!
2. Get your partner tested to see if they are a carrier. As Katie shared in the previous article there are a lot of options – in our case we were going to have our own children either way.
3. Make sure you have a team you feel can help guide you through your pregnancy – not just a geneticist, but also a dietitian and a MFM that is familiar with Homocystinuria.
4. Enjoy the time you are pregnant! Things may come up regardless if you have Homocystinuria or not.

September is Newborn Screening Awareness Month!

Ways to get involved:

- Share how newborn screening (or lack of) has impacted your life!
  How do I share my story?
  - Include the basics.
  - Be authentic.
  - Use images or video.
  - Choose one story to tell.
  - Try and include a call to action.
- Where do I share?
  - With HCU Network America
  - Facebook and Facebook Live
  - Baby First Test (https://www.babysfirsttest.org/newborn-screening/family-experiences)
  - Twitter
  - Instagram Live

Raise Awareness on Facebook –

- Post, share, and comment using the hashtag #2020NBS
- Add the #2020NBS frame to your profile picture
  a. Update your profile picture
  b. Click add frame
  c. Search for #2020NBS
  d. Reposition your profile picture accordingly
  e. Determine how long the frame will be displayed

Raise Awareness on Twitter

- See example Tweets: https://www.babysfirsttest.org/newborn-screening/2020-newborn-screening-awareness-month
- Attend the twitter chat!