

Peace of Mind: A Patient's Story

Julie Fernandes

The question of genetic testing was never far from our minds when my husband and I started on our adventure into parenthood. Before becoming pregnant, we discussed at length the question of whether we believed ourselves to be emotionally capable of having a child with a severe disability and our feelings about abortion. At the time, we were both 36, so we knew that we had a significantly increased risk. We concluded that we wanted to know more about any fetus that we might create, in order to make the best decisions for our future children and ourselves.

During our first appointment, our OB/GYN, asked whether we were interested in genetic testing, given that I was over 35. We told him that we had discussed it and were considering the choice between amniocentesis and CVS. He then informed us of a third option – extensive blood tests, along with a sonogram. This combination would provide us with more specific information on the odds that our child had a severe genetic abnormality. But that was still odds. We both wanted facts, not odds. So, while the blood test/sonogram option was appealing because it is non-invasive, we concluded that it would not provide us the peace of mind that we both craved. So it was amniocentesis or CVS. But which one?

Our OB/GYN referred us to a doctor who specializes in genetic testing. When we met with her and her genetic counselor a few weeks later, they patiently and thoroughly explained to us how genetics works (how traits are inherited, etc.) and what can go wrong in the procreation process. They also walked us through the options for testing, including amniocentesis, CVS, and the blood test/sonogram option our OB had discussed. We then talked about risks, and the doctor gave us concrete, honest, and very helpful information about the miscarriage rate of her patients after amniocentesis or CVS. At the end of the 2 hours with the doctor and the counselor, we felt we had the facts – but not the answer.

The answer, for us, really came down to time. With CVS, I could find out much sooner in the pregnancy whether there was a problem with the fetus. For me, those 5 or 6 weeks meant everything. Every day that I was pregnant, I felt more connected to the fetus. I decided that a miscarriage at 12 weeks would be terrible, but termination at 18 weeks would be devastating. I decided that I was not willing to take the risk of getting really bad news well into my second trimester.

The CVS procedure itself was uncomfortable, but painless: it felt like the longest pap smear of my life. Our doctor was excellent, professional, and patient throughout.

Afterwards, we just had to wait. I was lucky that I had no spotting or discomfort at all following the procedure. Just anxiety.

One week later, on the day before Christmas, when I was just about 12 weeks pregnant, the genetic counselor called to tell us the good news: no chromosomal abnormalities, and It's a Boy!

Of course, many risks remain and there are no guarantees. But we were happy to have an option for early peace of mind: to know what was knowable as soon as it could be known. Would we do it again? Absolutely.

Julie Fernandes and Avner Shapiro are expecting their first child this Summer.

The Genetics and Public Policy Center is part of the Phoebe R. Berman Bioethics Institute at the Johns Hopkins University and is funded through a grant from The Pew Charitable Trusts.