

## Preventing Genetic Defects

(0:00 - 0:12)

We continue our series, In Your Genes, this morning with a look at genetic abnormalities which can threaten the lives of babies born with them. Our Dr. Jennifer Ashton has been looking at some fascinating breakthroughs in genetic technology. Good morning, Jennifer.

(0:12 - 0:24)

Good morning, Maggie. Some scientists estimate that one out of every 200 babies suffer from a single gene disorder, a defect in one particular gene. Getting that news can be devastating for parents.

(0:25 - 0:36)

Sarah, our daughter, was born in January of 2000. We knew things weren't well at all as soon as she was born. She couldn't move her arms or her legs.

(0:37 - 0:51)

She couldn't breathe on her own. Doctors diagnosed Sarah Galloway with spinal muscular atrophy, a fatal form of muscular dystrophy that's genetically inherited. The prognosis was that we would never get her home.

(0:51 - 1:10)

They made the recommendation that we take her off life support. She was three weeks old the day we did that and that held her. Within a month or so after her death, we just kept feeling like this can't be the end.

(1:10 - 1:27)

We just needed another child. It just felt empty. Is there some type of technology out there that would allow us to pre-test for the disorder? So Linda and David turned to the Genetics and IVF Institute in Fairfax, Virginia to help them have another child.

(1:28 - 1:55)

Scientists here have developed many breakthrough genetic technologies, including a way to create viable embryos free of inherited genetic diseases. We can track a segment of DNA from the inheritance through both parents to an embryo and to be able to determine if that embryo is affected with one of a number of genetic disorders. And for the Galloways, it was the answer to their prayers.

(1:55 - 2:10)

They now have a daughter, Shannon. Today she's a healthy and strong seven-year-old. Scientists at the Institute are now working on a test that could detect 18,000 diseases prior to an embryo being implanted.

(2:10 - 2:28)

The utilisation of this test can lead to a probably one and a half to two-fold increase in that success. Could this lead to the creation of a perfect baby? A perfect baby is an unrealistic concept. This is not genetic engineering.

(2:28 - 2:49)

We are not changing the genes that parents will give to their child. And that's what the Galloways had always hoped for. It's never totally filling the hole, but it sure lessens the pain, you know, to be able to get that, you know, hi daddy, I love you.

(2:50 - 3:08)

I'm just being thankful every day. She's our miracle. Scientists at the Genetics and In-Vitro Fertilisation Institute hope they'll be able to offer the new screening tool that tests for thousands of diseases called 24-Chromosome Testing sometime early next year.

(3:08 - 3:16)

And I see that they really want to stress that this is not about engineering the perfect baby. That's correct. And in this case, this was truly a matter of life or death.

(3:16 - 3:39)

The Galloways do have a previously born healthy 13-year-old son, but as you can see, for certain genetic diseases that can be fatal or non-existent with a normal life expectancy, this is really, really critical. So the fact that they have a healthy child also shows that even if you are a carrier of a genetic disease, you don't necessarily pass along the disease, even if you pass along the gene. Absolutely.

(3:39 - 3:55)

And we have to remember that all of these genetic advances and these genetic types of testing, oftentimes the results are not black or white. You might have a genetic defect, which puts you at risk for a disease, or you might be a carrier. It doesn't necessarily mean that the disease will manifest itself in something serious.

(3:56 - 3:57)

All right, Dr. Jennifer Ashton, thank you, Jen.