

Newborn Screening Bloodspot Test (16:55)

SAVING BABIES THROUGH SCREENING FOUNDATION

NEWBORN BLOODSPOTS SAVES LIVES...

Amy Gaviglio, Genetic Counselor, Minnesota Department of Health: Everything we do in this lab comes back to the same mission, and that's helping babies and helping their families.

Female Narrator: Amy Gaviglio and her colleagues play an important part in one of the most successful public health programs in U.S. history. Every year, they check blood samples from the 70,000 new Americans born in Minnesota, searching for hidden disorders that can threaten a baby's future.

Amy Gaviglio: From the time that we receive them in the lab to the time that we have a positive result is usually 3 to 5 days. It's very quick turnaround, and it needs to be because these disorders are so serious.

(1:00)

Bradley Therrell, Ph.D., Director, National Newborn Screening Resource Center, Texas: Over my career, we've probably seen 9,000 or 10,000 babies who had some sort of condition that was picked up in the newborn screening program.

Female Narrator: Dr. Brad Therrell supervised this lab in Austin, Texas for almost 30 years. He's seen the newborn screening program grow to help babies with many different challenges.

Dr. Bradley Therrell: When I started, there was one test being done. Now you can do 20 or 30 conditions from one little spot, whereas before we did one test, one spot.

Female Narrator: Scientific advances have come so quickly over the past decade, it's been hard to keep the public up to date. But since newborn screening can mean the difference between life and death for some families, expectant parents need to know the facts. That's why we asked Dr. Therrell and Ms. Gaviglio to tell us more about the work they do. Korissa Olson, a Minnesota mom, who almost skipped newborn screening, also shares her experience.

(2:00)

Korissa Olson, Brooklyn Park, Minnesota: All these diseases and disorders, so rare, that they test for – never thought that my child would ever have something like that. His life would have been completely different had I not taken that test, not had the diagnosis so early on. So, we feel very blessed.

(2:30)

WHY IS NEWBORN SCREENING DONE?

Dr. Bradley Therrell: Newborn screening is set up so we don't wait until the child gets sick because if we wait, sometimes that's too late, and damage will have occurred.

Female Narrator: Most of the disorders covered by newborn screening can be treated and well-controlled if they are caught early enough. That's the challenge because usually there are no visible signs that anything is wrong.

Amy Gaviglio: What makes screening so important is that it really is the only way to pick up these disorders. These babies look beautiful at birth. They often have great Apgar scores, and they're crying, and they get discharged to home just as any other healthy baby.

Female Narrator: That's exactly what happened to the Olson family just outside Minneapolis. Six-year-old Emerson is healthy. But when Everett was five days old, his parents learned that his newborn screening test indicated a rare condition called galactosemia.

Korissa Olson: There's no other galactosemia or metabolic disorders in our family whatsoever. So that wasn't even on my radar.

Amy Gaviglio: And I think it's important for parents to know that all of us have a chance for having one of these disorders. Most of the children we find with this disorder do not have a family history. They're the first case.

Female Narrator: Babies with galactosemia like Everett are unable to digest milk properly, so a byproduct called galactose 1-phosphate builds up in their bodies like a poison. Without newborn screening, the Olsons might not have known Everett was in danger in time to save him.

Korissa Olson: This is a disorder that if it's even in a few days later could have had such horrible ramifications on Everett. Other kids that have it have gone into comas and have had severe liver damage. We just have to adjust his diet. It's not that hard. There's wonderful delicious things that are safe for him.

Dr. Bradley Therrell: We don't see babies who have the conditions because we're picking them up by newborn screening, but if you go into the developing world, you'll see many of these babies who – it breaks your heart – they could have been screened and could have been detected, and they weren't.

Female Narrator: Dr. Therrell has helped develop newborn screening programs in more than 40 countries around the world, but when he's home, he's more than an expert – he's a father and grandfather. Here's how he explained the newborn screening test to his daughter-in-law Emily.

Dr. Bradley Therrell: I told them that the babies will be tested at birth for some disorders that you may not know you're a carrier for. If a test comes back positive and diagnosed, then let's get the treatment right away, and if we do that, that early treatment and diagnosis can make a world of difference.

(5:30)

WHAT HAPPENS AT THE LAB?

Dr. Bradley Therrell: When they arrive at the health department, the samples are submitted with identifying information. They're given numbers so they can be analyzed in the laboratory. All the laboratory uses after the initial check-in is just the blood spot with this identifying number. There are federal laws that require the states to handle this data in a certain way, so it remains private. So they go to extremes to make sure that privacy is protected.

Female Narrator: After the cards are checked into the lab and assigned a tracking number, these machines punch small samples from the blood spots into trays.

Dr. Bradley Therrell: Six different 3-mm discs have been punched out, and there's a seventh one for the seventh test here, and out of those we get the 28 different tests that they're doing in this laboratory.

Female Narrator: From here, the trays go to various sections of the lab for different tests. These technicians in Austin are testing for sickle cell diseases. This lab worker in Minneapolis is screening for galactosemia.

Dr. Bradley Therrell: Conditions like galactosemia, maple syrup urine disease – those babies may die in the first few days because their systems cannot metabolize foods correctly.

Korissa Olson: It happens so incredibly fast. He became lethargic. I couldn't wake him up. He looked yellow.

Female Narrator: Based on the results of Everett's newborn screen, his doctor knew the milk he'd digested during his first few days of life could be causing his problems. He was rushed to the hospital.

Korissa Olson: If we didn't have a diagnosis, they'd probably want to feed him more milk because when you're jaundiced, you want to get those liquids in. There's rice milks. There's almond milks. Coconut milk is okay. Now we've gotten used to it. He's gotten used to it. Sometimes we don't even think about it. But it would have been very different, had we not had that diagnosis. 75% infant mortality rate. He might not have been with us.

(7:33)

WHAT IS A FALSE POSITIVE RESULT?

Female Narrator: About 1% of all babies screened in the U.S. need further testing after the first newborn screen.

Amy Gaviglio: We have to cast our net wider than a normal diagnostic test. We want to make sure we don't miss anyone. So in order to make sure that you don't miss anyone, you also are going to pick up people who aren't affected.

Female Narrator: Newborn screening works well, but some babies systems are still developing, and occasionally their tests come back with what is known as a false positive result. When their bodies settle into life outside the womb, and the test is repeated, the good news is that it usually comes back normal. Still, there's no way to know for certain without doing this follow-up.

Female Medical Personnel: Given his levels, this may be a false positive, but we'll let you know.

Female Narrator: In the end, only about 1 out of 700 babies will be diagnosed with a condition included in the blood spot screening program.

Dr. Bradley Therrell: It's rare conditions, but it's a condition if you have it, you're the 1 in 700. It's very important to you.

Amy Gaviglio: For those children and their families, it changes their life. It gives them a life that they might not otherwise have. So, even though you may pick up one or two a month, you know you've made such a big difference, and it's such a great feeling.

(9:05)

WHAT HAPPENS AFTER THE TEST?

Female Narrator: When the newborn screening tests are finished, usually one or two dried blood spots about the size of a dime are left. Most states keep these leftover specimens in storage. They are used to improve the current test and to develop new ones.

Amy Gaviglio: That new test might not help your child, but it could help a lot of children down the road. When newborn screening started in 1965, it screened for PKU, one disorder. Today in Minnesota, we screen for 53 disorders, and that wouldn't be possible without using newborn dried blood spots. So many more babies are now benefitting.

Female Narrator: Scientists consider newborn blood spots a unique research tool. Over the past 50 years, samples like these have contributed to public health studies investigating everything from the effect of smoking during pregnancy to the cause of hearing loss in infants. But in the past few years, some groups have expressed concern about possible misuse of this sensitive material.

Amy Gaviglio: Parents concerned about whether the government's storing their baby DNA, or what we're doing with it. I think part of it is likely the media. You see a lot more of the uses of genetics in *Gray's Anatomy* and *CSI*, and there's a lot more fears, I think, surrounding genetics, most of which are not based so much in truth.

Female Narrator: True or not, some maintain that these blood samples are private property, and their use without parent's knowledge or permission is an intrusion into family life by big government. The public debate has scared some people like Korissa Olson.

Korissa Olson: I did hear something at church that was controversial. I kind of filed it in the back of my head; didn't really have time or didn't research it too much, but it was something that struck a negative bone in my body.

Female Narrator: If not for the gentle persistence of a nurse in the maternity ward, Korissa would have left the hospital without having Everett tested.

Korissa Olson: I wasn't sure fully what they do in newborn screening. I wasn't aware how un-invasive it really is. It's simply taking a drop of blood. They're not putting anything into your child's body, and that message needs to get out. I feel like there should be more educating before you have your child, so that mothers come into the hospital and say, "I need this test. I need these blood spots preserved for future babies to be saved."

Female Narrator: States like Texas and Minnesota are now working overtime to make sure 21st century families are well-informed and involved in the newborn screening process.

(12:00)

Dr. Bradley Therrell: Just a little bit of knowledge upfront about what's going on and what the program entails is enough information for most parents to say, "Fine. I want that specimen in there because I know that this would improve our life as a society in the future."

(12:20)

WHAT EVERY PARENT SHOULD KNOW

Amy Gaviglio: The important part about screening is that they can find the problem before it starts to cause problems. So, we don't screen for things that a physician could look at the child and tell that they have this disorder. We only screen for conditions where screening is really the only way to pick it up before symptoms begin.

Dr. Bradley Therrell: I can understand paranoia about the baby's blood being kept by the government because, after all, I had babies myself, and my grandbabies are there, and their data is there. On the other hand, DNA is embedded in the blood, just like it's embedded in your hair. So, for instance, if you go to your beautician, or if you go to a barber and they cut your hair, they got your DNA as well. There's no recorded misuse of newborn screening specimens, and they've been around since the early 1960s.

Amy Gaviglio: It's not easy for a researcher to get newborn dried blood spots. It goes through very vigorous regulation to make sure that the privacy and respect for the subjects is maintained throughout the study. So, we really want to see that there's going to be a benefit for the public. Can doing this project, will doing this project help?

(13:40)

Female Narrator: When baby blood spots are given for research, they are anonymous. No name is attached.

Dr. Bradley Therrell: There's a lot of different safeguards in place. There are institutional review boards that review research. They look at the process to make sure they're legally okay, ethically okay. The government in this case is doing a bang-up job of protecting your privacy and your baby's privacy.

Korissa Olson: I really feel strongly that it's important not only taking the test, but actually preserving those blood spots. I want them to be able to find cures. It helped our son. It saved our son's life. I'm thankful for that – that blood spot.

Female Narrator: One thing everyone agrees on is that public knowledge and understanding of newborn screening must be improved. That's the mission of the Save Babies Through Screening Foundation. The Save Babies website run by parents with the assistance of medical experts shares families experiences and provides up-to-date information. Each states decides which disorders it will test for, and has different rules about storing blood spots after testing is completed. To learn more about the policies in your area, just click on your state. It provides a quick link to your local newborn screening program.

Dr. Bradley Therrell: All parents should know that these are most important tests. They will detect diseases that can lead to death, mental retardation, growth disorders, and other sorts of severe problems.

Amy Gaviglio: I want expectant parents to know that newborn screening is a great program that is really looking out for their child. Everyone who works here is so committed because we know that at the end of a result is a family.

Korissa Olson: You know, Everett's doing really well, and a lot of people are really surprised when I tell them that he has a pretty serious, rare, genetic disorder, but it was caught through newborn screening. So we're very passionate about getting the message out about how it can save a life.

Female Narrator: For more information and support, visit [Save the Babies](#) or call us toll-free.

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(End 16:04)