

## TOP STORIES

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Knowing at birth

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By: Ivanhoe Newswire

### Knowing at Birth

Every newborn undergoes genetic screening at birth for life threatening conditions and diseases, but what they're tested for varies greatly from state to state.

WASHINGTON, D.C. (Ivanhoe Broadcast News) -- Every newborn undergoes genetic screening at birth for life threatening conditions and diseases, but what they're tested for varies greatly from state to state. One family, the Monacos, says that can make a devastating difference.

"Stephen came into the world just like any other healthy baby," Jana Monaco, Stephen's mother, says. But when he was three-and-a-half, he had a stomach virus when his parents put him to bed one night. "That was the last time we said goodnight to each other and ... I love you," Jana says.

When he woke up, he had suffered a metabolic crisis that nearly killed him. "They told us then, 'If he makes it through the weekend, he won't be the same child you knew and loved,'" Jana says. Just 24 hours later, he was left a severely disabled child with complicated medical issues. "It was one day. He went from making his grandmother's birthday cake to being on life support," Tom Monaco, Stephen's father, says.



Some conditions can be dealt with early.

Stephen had an undetected genetic metabolic disorder called Isovaleric Acidemia (IVA). If tested for it as a newborn, medicine can prevent brain damage. However, "At the time, Virginia was screening for eight disorders, and this wasn't one of them," Jana says. The American College of Medical Genetics recommends testing for 29 disorders, but the decision is left up to each state. "Some states are going to screen newborns for just a handful of conditions, whereas other states are doing many, many conditions," Elizabeth Wood, M.S., a Certified Genetics Counselor at Johns Hopkins Medical Center, says.

When Jana got pregnant again, they screened for IVA. Turns out this child, Caroline, had it too, but they treated it at birth. And today, she's a happy, healthy four-year-old who regulates her condition with her diet.

"We consider ourselves now the poster family for newborn screening advocacy," Tom says. The Monacos successfully pushed to get Virginia to screen for the recommended 29 conditions. "The first six months after the expanded screening, 22 babies were picked up with these disorders," Jana says.

They're also dedicated to educating parents about genetic testing and genetic counseling. "Many families don't know if they have a condition in their family, so what we usually recommend is meeting with a genetic counselor to take a family history," Wood says. Experts say you should visit a genetic counselor before or right after getting pregnant. Once you are pregnant, make sure you know what diseases your state screens for. If it's not the full panel, you can get supplemental screening from private firms for an average of \$25.

Stephen can't walk, talk or feed himself. But he inspires his parents everyday. "Yeah, we lost some dreams with him, but his situation has made such a profound effect on so many people," Jana says. "If we can help one family not have to go through this, then we've done our job," says Tom. That's how Stephen is making a difference.

Want more info?

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