Fact #2: Newborn screening is a public health success story

Newborn screening has been called a triumph of the 20th century public health system. In short, newborn screening has been a spectacular success because it saves lives, prevents disability and saves money.

Untreated infants with newborn screening conditions, and their families, may suffer enormous burdens. Untreated infants with phenylketonuria (PKU) will have an average IQ of below 40, a severe intellectual deficiency. Untreated infants with medium-chain acyl-CoA dehydrogenase deficiency (MCAD) may suffer sudden death. Untreated infants with biotinidase deficiency may have unstoppable seizures, hearing loss, blindness, and movement difficulties. Yet, babies with these disorders often appear healthy at birth.

Perhaps the most painful burden for families whose children suffer the consequences of untreated early disease is knowing that adverse outcomes could have been prevented. For PKU, the main treatment is a low-protein diet. For MCAD, it is regular feedings. For biotinidase deficiency, it is daily supplements of the vitamin biotin.

By enabling prompt treatment, newborn screening keeps rare congenital conditions from stealing children's lives.

Parent stories...

"Our son was born at the end of December 2007. We were so happy and excited that he was finally here. He was absolutely perfect!

After a week had passed, we received a phone call with unexpected news our son's newborn screen results detected that he had MCAD (medium chain acyl-coA dehydrogenase deficiency). Our world stopped. We were devastated and numb upon learning this news.

Today our son is doing great! Without the newborn screening, we do not know if we would be writing this to explain how grateful we are that the test, which revealed that our son has MCAD, was performed. We truly believe that the newborn screen saved our son's life! We are grateful that the screen was performed."

Parents of a son diagnosed with MCAD