

A newborn screening

By CARRIE KIRSCHNER - *The Independent*

ASHLAND - Recently expanded newborn screening in Kentucky will save lives and prevent a range of severe disabilities in newborns diagnosed with certain disorders, officials say.

Since Dec. 5, every newborn in Kentucky has been tested for 29 different diseases and metabolic disorders. That number is up from just four.

The expansion of newborn screening brings Kentucky up to newborn screening levels recommended by the March of Dimes and the American College of Medical Genetics.

Early in 2005, the General Assembly passed Senate Bill 24, at Governor Fletcher's urging. The bill approved the expanded screening and secured permanent funding for the program.

Area doctors are excited by the expanded testing. "This is a tremendous advance for babies in Kentucky. We're thrilled that Kentucky can do this," said Cheryl Cook, chief of pediatrics at Our Lady of Bellefonte Hospital, who also practices at Tri-State pediatrics.

"This has the potential to improve outcomes and improve lives," she added.

"Some of these diseases are very rare, but it's not rare if your child has it," Cook said, adding her practice does treat area children with some of these rare diseases.



Mark Anthony Williamson, a newborn at King's Daughters Medical Center, gets some sleep after undergoing a blood sample to screen for possible disorders. Photo by Kevin Goldy / *The Independent*

The test requires a small amount of blood normally taken from the baby's heel after it is at least 24 hours old. The blood sample is placed on a "post card" and sent to the State Public Health Lab in Frankfort, which processes the samples.

According to Scott Knight, a neonatologist and director of the newborn intensive care unit at King's Daughters Medical Center, many of the diseases now being screened for are caused by the excess accumulation of normal body toxins. This happens because of the absence of a specific enzyme, which breaks down the toxins and helps the body dispose of them.

By screening an infant's blood for the absence of these vital enzymes doctors can catch a disorder and begin treating it well before symptoms begin to show. Early diagnosis can prevent a range of side effects from mental retardation to death.

Although some of the diseases screened for have no cure - cystic fibrosis or sickle cell disease, for example - there are treatments for nearly all the disorders that, if administered early enough, can help prevent many of the more serious side effects.

If not diagnosed many of these serious illnesses go untreated and undetected until the child becomes very ill or dies.

According to Cook, time is of the essence in treating many of these diseases, and stressed that parents need to bring their child in immediately if a test needs to be repeated. "Sometimes babies just need a change in formula," she said, to prevent side effects like blindness.

Knight said he thought one disease in particular where the screening would make a large difference was on cases of medium-chain acyl-coA dehydrogenase deficiency, or MCAD. MCAD leads to an accumulation of fatty acids, and when a child with it gets sick it often has problems with hypoglycemia, Knight said,

adding that undiagnosed MCAD is probably the culprit in many cases of Sudden Infant Death Syndrome. Since the expansion of the screening program KDMC has already had one newborn test positive for MCAD, he said.

According to the Kentucky Cabinet of Health and Family Services it is estimated that up to 5 percent of childhood deaths attributed to sudden infant death syndrome may have actually been caused by a treatable metabolic condition. Over two years (2002-03), Kentucky lost 91 infants to SIDS. It is estimated that 55,000 babies are born in Kentucky each year.

In addition to saving lives, Knight said the new screening program has the potential to save the health care system money as well. "The frequency of diseases are incredibly low but the payoffs are incredibly big," he said. "If we can catch just one case it's a huge benefit to the parents and to the health care system."

According to Steve Davis, a pediatrician and deputy commissioner of the Department for Public Health, the expanded program is more than a blood test, it's a comprehensive "circle of newborn screening," implemented through "a broad based effort of all the different folks in Kentucky who put their arms around the needs of children in Kentucky."

Davis spearheaded the creation of the program, which includes the mass screening of all newborns, enhanced education of physicians and parents in addition to diagnosis and treatment options. Children who test positive for disorders need to be able to be referred to physicians with a greater knowledge of the rare disorders, to have additional diagnostic evaluations and know how to treat the disorders. As a result of the state's program the University of Kentucky and University of Louisville Departments of Pediatrics have specialty physicians in place around the clock to diagnose and treat the disorders, he said.

Davis said all the components of the circle are required to have “ a program that is really solid for your children.” He added, “That's what we have in place in Kentucky.”

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