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Newborn Metabolic Screening
Adequate Medical Care Begins Here
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Anyone who has had a baby in the past 40 years probably remembers the quick “heel prick” the baby gets before leaving the hospital. Most people do not give this a second thought, but that little sample of blood can prevent a world of hurt.

This routine lab study often referred to as simply the “PKU” test, checks for a number of inborn errors of metabolism that may be present in the newborn. In addition to Phenylketonuria (PKU), newborn screening (NBS) tests for a variety of metabolic abnormalities, including disorders relating to the metabolism of organic acids, fatty acids, amino acids, as well as hemoglobinopathies, such as sickle cell disease, and other disorders ranging from congenital hypothyroidism to galactosemia to cystic fibrosis. Amazingly, that one drop of blood can be used to screen for 55 or more illnesses.

With the advent of mandatory newborn metabolic screening and optional supplemental screening, parents are armed with the information that may eventually save their child’s life. Without these tests, a child suffering from an unknown metabolic disorder may experience complications resulting in brain injury or even death.¹

TESTS DEPEND ON JURISDICTION

Unfortunately, the newborn screening tests that are administered on any given child depends entirely on where the baby is born. By law, each state or region operates its own newborn screening program, and the disorders included in the mandatory panel are determined on a state-by-state basis.

All states test for phenylketonuria, hypothyroidism, and galactosemia; and most states screen for sickle cell anemia, congenital adrenal hyperplasia (CAD), and hearing loss. However, beyond those few there is a wide variation in the disorders covered by law. In a few states, a parent may “opt out” of mandatory screening for religious or other reasons. Some states test for as many as 30 disorders and others for only four.²

Whether a state requires extensive screening or not, a hospital may still offer, or require, comprehensive newborn screening to cover additional disorders. Certainly, the research justifies the expanded approach and a facility’s failure to provide both mandated and optional comprehensive screening may provide the basis for liability.

Looking at the numbers, the incidence of newborn errors of metabolism is significant. For example, PKU affects about one in 25,000 newborns. The most common disorder, congenital hypothyroidism, affects at least one in 5,000. Others are less common, such as biotinidase deficiency, which occurs once in between 72,000 and 126,000 births; and maple syrup urine disease, which has an incidence of approximately one in 250,000.

Even where the incidence is less frequent, the consequence of failing to diagnose these conditions early justifies the insignificant costs of testing for even the rarest of disorders. For example, galactosemia, which affects only about one in 30,000-50,000 babies, frequently causes death in infancy.

Galactosemia usually shows no symptoms at birth, but jaundice, diarrhea, and vomiting soon develop and the baby fails to gain weight. If not detected immediately, it results in liver disease, cataracts, mental retardation, and even death. Death can occur as early as one to two weeks of age from severe escherichia (*E. coli*) bacteria infections.

As with many other metabolic disorders, preventing these catastrophic results is simple, if the parents and caregivers are merely aware of the condition sufficiently early in life. A child with galactosemia is unable to convert galactose – a sugar in milk -- into glucose, for energy. Treatment amounts solely to eliminating milk or lactose from the individual's diet for life.

Similarly, a child with PKU cannot process phenylalanine, a part of protein that is found in almost all types of food, which causes it to build up in the blood resulting in brain damage and mental retardation. Treatment is simple, but only when implemented early in life: a low-phenylalanine diet, including special formula at infancy. Fortunately, since PKU has been the subject of testing from the inception of newborn screening programs, adverse events related to the disorder are now rarely seen.

Another example of a disorder that screening has affected significantly is Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency. It is estimated that one in 5,000-12,000 children are born with this disorder. Because testing for many metabolic disorders has not occurred until recently, it is difficult to know exactly how many infants would have been diagnosed, had screening been available at birth.

MCAD is one of the inherited disorders that have only recently been added to some state panels. Children with MCAD have an inactive enzyme in the blood meant to break down fats in the body. There is virtually no indication of this deficiency until the child experiences a period of fasting. Because children with MCAD are unable to use stored fat for energy, the body uses all of its glucose reserves instead, resulting in hypoglycemia, lethargy, and if left untreated, ultimately encephalopathy, respiratory arrest, cardiac arrest, coma and death.

Unfortunately, the early symptoms of MCAD mirror those of common gastrointestinal illnesses, and therefore the underlying problem is not ascertained before the child becomes critically ill and dies. Without prior indication of the disease, 20 to 25 percent of children with this disorder will die with their first occurrence of illness. Fortunately, however, when the child's deficiency is known, care is taken to avoid periods of fasting and when the child becomes sick with a stomach virus or similar innocuous illness, intravenous glucose can prevent further complications.

TYPICAL TESTING PROCEDURE

The blood for the screen is usually drawn while the baby is still in the hospital, within 24 to 48 hours of birth. Because some of the disorders are not adequately picked up within the first 24 hours of life, if the baby is discharged before 48 hours, the test may be repeated one to two weeks later at the pediatrician's office.

The hospital should have a policy and procedure in place to draw the blood, and typically will also have standing orders related to NBS. Nevertheless, the order must come from a medical doctor, usually the pediatrician. Therefore, any claim for the failure to perform the test usually involves the discharging pediatrician and the hospital.

Typically, the blood is sent to a state public health laboratory for testing and the results are forwarded to the baby's pediatrician. For supplemental screening, the analysis may be performed in the hospital or through a private lab that has contracted with the facility. All of these entities are potential defendants in the event the screening is not performed appropriately, or the results transmitted timely, or the information is not acted on accordingly.

Even when the testing is not mandated, and the hospital does not offer supplemental screening as an option, parents may wish to obtain the testing on their own. By 2002, several well-regarded laboratories offered supplemental/comprehensive newborn metabolic screening to the public.

For example, Pediatrix Analytical (formerly NeoGen Screening) first offered private testing in 1993. The Mayo Clinic was on board as of 2002, and Baylor, and the University of Colorado Biochemical Genetics Laboratory in Denver in 1998 and 2002, respectively. Many are aware of the recent interest in having cord blood taken and stored for possible treatment of diseases later in life, but might be unaware that the Cord Blood Registry also offered parents the option of supplemental comprehensive NBS through private labs.

Whether the testing is conducted at a public or private lab, however, the blood must still be drawn at the hospital at the time of birth, or shortly thereafter in the pediatrician's office. The hospital or physician's refusal to do so may raise issues of liability, as was the case recently when a Georgia family was denied the opportunity to have their child tested despite requests to do so.

RESPONSIBLE PARTIES

The standard of care for newborn metabolic screening is a little different than in other areas of medicine because the primary responsibility for newborn screening in the United States rests with public health agencies, not the private sector. Thus, individual hospitals and pediatricians will refer to state guidelines in an effort to avoid responsibility for alleged deficiencies in their own policies and procedures for screening. This, of course, does not absolve entities from liability for the failure to perform state-mandated screening, and claims arising from such omissions have the added force of negligence per se.

Further, where a baby suffers because he was not screened for a metabolic disorder later shown to exist, the inquiry surrounding whether the disorder should have been picked up at birth, and the party responsible for that discovery, should continue beyond the state program. Where the birth facility involved is located in a large metropolitan area, with a high number of annual deliveries, access to sophisticated equipment and research facilities, there is no justification for its failure to offer full-scale supplemental screening even in the absence of state mandate.

This is especially true given the range of disorders that can now be covered by screening with the use of tandem mass spectrometry (MS/MS). As of 2005, newborn screening by MS/MS was required or offered in 60 percent of the states to cover many additional amino acid, organic acid and fatty acid oxidation disorders.

Keep in mind that every disorder detectable by MS/MS is treatable – and all of these disorders are tested through the same sample of blood! It just makes sense to use the technology to its full advantage to save more lives. Regrettably, however, most parents are not even told that comprehensive MS/MS screening is available.

As one might expect, the incidence of metabolic disorders increases in families where there is a history of inherited disorders. A parent with another child who is affected by a disorder should definitely be counseled to test in the event of a subsequent birth. Thus, even where the

disorder is extremely rare and not part of the regular panel, testing should be standard for that patient.

If a family comes to you with the sad news that they have lost a child as a result of an undiagnosed metabolic disorder, consider what information was known within the pediatric and genetics community about the disorder. Determine whether state or national organizations previously proposed that the particular disorder be added to the mandatory NBS panel prior to the date of delivery. Establish the ease with which the hospital could have added it, which was likely already screening for other disorders with MS/MS. And demonstrate statistically the number of babies likely to be affected by the disorder based on the number of births in the state and at the particular facility.

The bottom line is that a family should not have to learn the hard way that their child suffers from a metabolic disorder. Simple, easy, and cost-effective methods are available, and doctors and hospitals should offer them to detect these orders before they become apparent only when the child dies. When the system fails these families, those responsible should be held accountable.

¹ The Save Babies Through Screening Foundation is dedicated to raising awareness of newborn screening and related disorders. Its website, www.savebabies.org, is an excellent source for information regarding all issues and advances related to NBS.

² To determine which disorders are included for each state, go to www.genes-r-us.uthscsa.edu/.

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