OLR RESEARCH REPORT

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CONNECTICUT NEWBORN SCREENING REQUIREMENTS

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You asked for information on the state's newborn screening requirements. Specifically, you wanted to know if the legislature has considered adding Krabbe Leukodystrophy, a degenerative neurological disorder, to the list of genetic and metabolic diseases and conditions for which Connecticut newborns are screened.

SUMMARY

Connecticut law requires all health care institutions that care for newborn infants to test them for over 40 genetic and metabolic diseases and conditions, including phenylketonuria, HIV, and sickle cell disease. Screening occurs primarily through the Department of Public Health's (DPH) newborn screening program. In addition to the initial screening test, the program directs parents of identified infants to appropriate counseling, education, treatment, and follow-up services.

Separate from the newborn screening program, the law also requires these institutions to test infants for (1) critical congenital heart disease, (2) cystic fibrosis, (3) severe combined immunodeficiency disease, and (4) under certain conditions, adrenoleukodystrophy (ALD) (CGS § 19a-55).

In 2009, the legislature considered Proposed Bill 6044, which would have added Krabbe Leukodystrophy to the list of newborn screening requirements. The bill was referred to the Public Health Committee, which did not hold a public hearing or take further action on the bill.

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NEWBORN SCREENING REQUIREMENTS

DPH Newborn Screening Program

DPH administers a newborn screening program that requires all health care institutions caring for newborn infants to test them for certain conditions, including those in Table 1. A complete list of screened conditions can be found on DPH's website (http://www.ct.gov/dph/cwp/view.asp?a=3122&q=387742)

Table 1: Conditions Screened For By DPH's Newborn Screening Program

Phenylketonuria (also called PKU)	HIV	Hypothyroidism
Galactosemia	Sickle cell disease	Maple syrup urine disease
Homocystnuria	Biotinidase deficiency	Congenital adrenal hyperplasia
Fatty Acid Oxidation Disorders	Amino Acid Disorders	Other tests for inborn metabolic errors DPH prescribes

The screening must be performed as soon as medically appropriate, unless a parent objects on religious grounds. The program's goal is to screen all infants before they are discharged from the hospital or within their first four days of life.

DPH labs perform follow-up testing on positive screens and report any abnormal results to the department's tracking unit. The tracking unit reports the results to the newborn's primary care provider and ensures the newborn's parents are referred to state-funded regional treatment centers. These centers provide comprehensive testing, counseling, education, treatment and follow-up services. DPH charges institutions a $28 fee for each newborn to cover the program's costs.

Other Newborn Screening Requirements

Separate from the newborn screening program, the law also requires these health care institutions to test infants for (1) critical congenital heart disease, (2) cystic fibrosis, and (3) severe combined immunodeficiency disease. They must also begin testing infants for ALD once the following conditions are met:

1. (a) a reliable ALD screening method is developed and validated that uses dried blood spots and quality assurance testing methods or (b) the federal Food and Drug Administration approves an ALD test that uses dried blood spots and

2. any reagents necessary for the screening test are available (the screening method and reagents are not currently available).

As under the newborn screening program, the testing must be performed as soon as medically appropriate.

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