



NEWBORN SCREENING PROGRAM OVERVIEW – JANUARY 2012

It is the stated policy of the state of Washington “...to make every effort to detect as early as feasible and to prevent where possible phenylketonuria and other preventable heritable disorders leading to developmental disabilities or physical defects” (Revised Code of Washington, RCW 70.83.010; Declaration of policy and purpose). The law gives the State Board of Health authority to determine which disorders to look for and directs the board to adopt regulations needed to carry out the intent of the law.

Board of Health regulations (chapter 246-650 WAC) direct hospitals throughout the state to collect a small specimen of blood from each infant’s heel before they are discharged. The specimens are absorbed on a special type of paper, dried, and then sent to the Newborn Screening Program at the Department of Health’s Public Health Laboratories facility in Shoreline, Washington. The program monitors every birth to assure that a specimen is received from every baby. Highly sensitive and specific tests are performed on these specimens by the program’s laboratory for each of the disorders. The laboratory employs a range of technologies including immunoassay, high performance liquid chromatography, iso-electric focusing, mass spectrometry, and DNA/polymerase chain reaction.

Positive test results are immediately communicated to the child’s health care provider along with links to appropriate diagnostic and treatment services. The program tracks all positive results to assure that the children receive the necessary care as quickly as possible. The program also tracks the long-term outcome of infants found to be affected by the disorders to evaluate the effectiveness of the system.

Over two and a half million infants have been screened since 1966 and over a thousand children have been given the opportunity to live healthy, productive lives because their conditions were detected and treated in time through this screening. These children have been spared permanent disability, mental retardation, or death that would otherwise result from their disorders.

Currently 27 of the 30 conditions recommended by US Department of Health and Human Services are on the screening panel specified by the Board of Health. Exceptions are:

1. Infant hearing screening is not mandated but is universally offered through Washington hospitals. The Department of Health operates tracking and surveillance activities. Federal funding is central to these functions.
2. A condition named 3MCC was determined to not meet the Board’s criteria for inclusion; however, some cases are detected incidental to screening for the other conditions.
3. Severe combined immunodeficiency (SCID) has not yet been formally considered by the Board.

The screening program is self-sustaining through a charge of \$60.90 per child that is collected through the hospital where the child is born. Diagnostic and treatment care is funded through many sources including government and private insurance, federal grants and self-pay. An \$8.40 fee collected at the same time as the screening charge helps support specialty care clinics for the disorders (RCW 70.83.040).

For more information:

Visit our web site at www.doh.wa.gov/nbs; or call 206-418-5410 (1-866-660-6050 toll free)