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Babies benefitted greatly from one doctor's research

by Luther Granquist // December 10th, 2011

In October the Minnesota Supreme Court ruled that the Minnesota Department of Health could not retain blood samples from the newborn baby screening program indefinitely or provide them to other researchers without consent from a baby's parents. This ruling, however, did not affect the newborn screening requirement itself. This program was established in 1965 after Dr. Robert Guthrie from the University of Buffalo Children's Hospital developed a simple and effective screening test for phenylketonuria (PKU).

Guthrie, who grew up in Minneapolis, received both his M.D. and his Ph. D. degree from the University of Minnesota. After his son John was diagnosed with developmental disabilities or "mental retardation" as was the term used back then, Guthrie concentrated his research efforts on ways to prevent that condition. And after his niece, Margaret Doll from Minneapolis, was diagnosed with PKU in 1959 as a one-year-old, he sought to find an effective way for early screening and diagnoses for PKU.

A person with PKU lacks an enzyme needed to break down an amino acid called phenylalanine. If this condition is detected shortly after birth and if a strict low-phenylalanine diet is followed, the baby would develop normally. If not, the baby would likely have brain damage and have cognitive delays. But the delayed development caused by PKU does not appear immediately, so many babies up to the early 1960s, including Margaret, were not tested for PKU until I was too late. The standard test used then, applying ferric chloride to urine in a freshly wet diaper, was not effective until the baby was about four weeks old, again too late to prevent significant harm.

The Minnesota health departments and Minnesota physicians recognized the need for early testing for PKU. Before 1960, Well Baby Clinics in St. Paul and Minneapolis provided urine screening. In May 1961 the Minnesota State Medical Association and the Minnesota Department of Health recommended that physicians in the state test all infants for PKU using the urine screening test.

Guthrie's test simplified the process. A few drops of blood from a pin prick on the heel of the baby were applied to a filter paper, dried, and sent to a lab for testing. This test proved effective when a child was only four or five days old.

Guthrie was an advocate as well as a scientist. He publicized his test in a letter to the editor in the Journal of the American Medical Association in October 1961 and stressed the need for nationwide testing for all newborn babies in a press conference at the annual meeting of the National Association for Retarded Children. After Guthrie presented information about his new test at the American Public Health Association annual meeting, also in October 1961, the U. S. Children's Bureau awarded him a grant to try it on 400,000 infants, including an offer to pay the cost of sending personnel from every state's health department to Buffalo for training.

Twenty-nine states accepted this offer, including Minnesota. In May 1962, the Minnesota Department of Health announced that 12,000 babies would be tested between June 1, 1962 and June 30, 1963, primarily in St. Paul hospitals.

In August 1964, the Minnesota Department of Health started a voluntary statewide PKU screening program, including follow-up nutrition and genetic consultation. By April 1, 1965, 157 of 179 hospitals in the state with maternity units participated in this program. It is not surprising that the bill making the program mandatory (with an exception for parental objection based on religious beliefs) passed both houses of the legislature unanimously and was signed by Gov. Karl Rolvaag on April 15, 1965.

Until Guthrie died in 1995, he devoted his professional life to expanding the scope of newborn baby screening. Today, with further advances in screening technology, Minnesota's screening program tests for 54 different conditions. Although Margaret Doll was born too soon to benefit from her uncle's test, she did benefit from the PKU diet started when she was diagnosed. That diet was discontinued, for a time when the premise was that the brain had already fully developed. Some years later she started again on the PKU diet and continues to benefit from it.

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