

Newborn Screening and Follow-up

Program Summary:

Newborn Screening and Follow-up refers to a public health population based screening system designed to detect and treat infants with special needs who have certain hematologic, endocrine or embolic disorders. Immediate medical management of an infant with one of these disorders will prevent many, and in some disorders all the serious clinical sequelae.¹

Since the diseases on the newborn screening panel are relatively rare and require a substantial level of knowledge of confirmatory testing and treatment, follow-up by the public health program is critical to ensuring that affected newborns are provided appropriate care within the proper time frames. Thus, newborn screening is seen as more than just a laboratory test, but a much larger system involving the following components: 1) Screening 2) Follow-up 3) Diagnosis 4) Management 5) Evaluation

Louisiana's Experience

Louisiana's Board of Health had begun a program of case finding for the detection and treatment of phenylketonuria (PKU) as early as 1960, but this was based solely on the ferric chloride test on urine of infants and children served at local parish health units. Dr. Robert Guthrie's development of an effective blood test for mass screening in the early 1960's set the stage for a new standard of care. Act 269 passed by the Louisiana State Legislature during the 1964 session expanded the PKU program by requiring laboratory testing of all newborns for PKU. The testing panel has expanded over the years to now include 27 diseases as listed below by disorder and the year the disease(s) addition was made:

- PKU 1964
- Sickle Cell Disease 1972 (although not universal on the newborn filter paper specimen until 1992)
- Congenital Hypothyroidism 1979
- Biotinidase Deficiency 1999
- Galactosemia 2002
- Five additional metabolic diseases detected through tandem mass spectrometry: Argininosuccinic Aciduria (ASA), Citrullinemia, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) 2004
- Adoption of the panel recommended by the American College of Medical Genetics (AMCG) except for Cystic Fibrosis 2006
- Cystic Fibrosis (proposed for July 1, 2007)

The history of legislation pertaining to this program (R.S. 40:1299 et seq) is found under State Mandates.

¹ U.S Newborn Screening System Guidelines II, Follow-up of Children, Diagnosis, Management and Evaluation (1999) Council of Regional Networks for Genetic Services (CORN) supported in part by project #MCJ-131006 of the Health Resources Services Administration