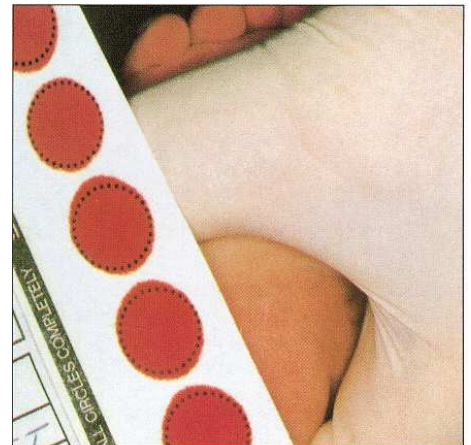


Newborn Screening

The BOL Newborn Screening Laboratory in Jacksonville screens more than 300,000 specimens annually for 35 genetic disorders on blood samples from newborn babies using protein electrophoresis, tandem mass spectrometry (MS) and DNA mutation analyses. Early detection is the key to newborn screening and the laboratory has implemented procedures for early detection and reporting to Children's Medical Services (CMS), hospitals, physicians and birthing centers as part of Florida's effort to ensure that all newborns identified through the screening process receive adequate and prompt medical care and follow up. The laboratory consists of 37 employees and operates on annual budget of approximately 14 million dollars. Results are reported within 24-48 hours.

Highlights/specialties

- 1965 tested for phenylketonuria (PKU)
- 1979 expanded to include hypothyroidism, maple syrup urine disease (discontinued in 1985) and galactosemia
- 1988 added hemoglobinopathies including sickle cell disease
- 1995 added congenital adrenal hyperplasia (CAH)
- 2005 added biotinidase deficiency
- 2006 panel substantially expanded utilizing tandem mass spectrometry (MS/MS technology)
- 2007 added cystic fibrosis as the 35th disorder



The addition of cystic fibrosis to the panel of disorders screened in Florida completes the expansion proposed in November 2004. With cystic fibrosis, Florida is now screening for all disorders recommended by the March of Dimes and the American College of Medical Genetics.

Challenges

- Implementing new methods.
- Expanding the number of disorders for which we test.
- Provide a turn around time for reporting results of 24 hours or less.

