



Bureau of Public Health Laboratories (BPHL) Jacksonville - Miami - Tampa

NEWBORN SCREENING



The Newborn Screening (NBS) Laboratory in Jacksonville receives about 275,000 specimens annually from babies born in Florida to test for genetic disorders using tandem mass spectrometry (MSMS), high performance liquid chromatography (HPLC), fluorometric analyzers and DNA mutation analyses. Early detection is the primary objective of newborn screening in Florida's effort to ensure that all newborns are screened. BPHL-Jacksonville has implemented procedures for immediate detection and timely reporting of presumptive abnormal results to Children's Medical Services (CMS), hospitals, physicians and birthing centers. NBS testing helps to ensure affected babies receive immediate follow-up and adequate medical care. Most results are reported within 24 to 48 hours of receipt. The NBS laboratory operates 6 days a week, 52 weeks a year. The NBS Laboratory tests for 35 core conditions and 22 secondary conditions (a total of 57 conditions, 53 of which are included in the Recommended Uniform Screening Panel approved by the US Secretary of the Department of Health and Human Services)



Highlights / Specialties

- 1965 – Started testing for Phenylketonuria (PKU)
- 1979 – Added Congenital Hypothyroidism, Galactosemia, and Maple Syrup Urine Disease (later discontinued in 1985)
- 1988 – Added Hemoglobinopathy testing (Sickle Cell Disease)
- 1995 – Congenital Adrenal Hyperplasia
- 2005 – Biotinidase deficiency
- 2006 – Expansion of screening panel to 34 disorders using the tandem mass spectrometry (MSMS)
- 2007 – Cystic Fibrosis
- 2012 – Severe Combined Immunodeficiency (SCID)
- 2018 – X-Linked Adrenoleukodystrophy (XALD)
- 2020 – Pompe, Mucopolysaccharidosis type I (MPS I), Spinal Muscular Atrophy (SMA)

