



NC DEPARTMENT OF
**HEALTH AND
HUMAN SERVICES**

ROY COOPER • Governor

KODY H. KINSLEY • Secretary

SUSAN GALE PERRY • Chief Deputy Secretary for Opportunity and Well-Being

MARK T. BENTON • Deputy Secretary for Health

SUSAN KANSAGRA MD, MBA • Assistant Secretary for Public Health, Division of Public Health

YVONNE COPELAND • Director, Division of Child and Family Well-Being

Memo

To: North Carolina Newborn Screening Partners

From: Scott M. Shone, PhD, HCLD(ABB), Director, NC State Laboratory of Public Health
Anne O. Odusanya, DrPH, MPH, Assistant Director, Division of Child and Family Well-Being,
Whole Child Health Section

Date: February 13, 2023

Re: Newborn Screening for Mucopolysaccharidosis Type I and Pompe Disease

Beginning today, February 13, 2023, the North Carolina Newborn Screening Program will begin screening all newborn screening specimens submitted to the North Carolina State Laboratory of Public Health (NCSLPH) for Mucopolysaccharidosis Type I (MPS I) and Pompe Disease (Pompe). MPS I and Pompe are lysosomal storage diseases (LSDs), genetic diseases that are characterized by an abnormal build-up of various toxic materials in the body's cells. Symptoms and onset of LSDs vary and may affect different parts of the body, including the skeleton, brain, skin, heart, and central nervous system.

Newborn screening for MPS I and Pompe will be integrated into the standard newborn screening panel and results will be included on all NCSLPH Newborn Screening reports issued starting February 14, 2022. Reports are provided in hardcopy form and are also available via the NCSLPH Clinical and Environmental Lab Results (CELR) online portal.

Screening results that suggest elevated risk for MPS I will be communicated to the newborn's health care provider by the UNC Division of Genetics and Metabolism Follow-up Program. The UNC Follow-up Program will provide consultation services and recommendations for follow-up testing.

Screening results that suggest elevated risk for Pompe Disease will be communicated to the newborn's health care provider by the North Carolina Department of Health and Human Services Division of Child & Family Well-Being (DCFW). The DCFW Follow-up Program will provide consultation services and recommendations for follow-up testing.

For questions about newborn screening laboratory testing for MPS I or Pompe, please contact the NCSLPH Newborn Screening Laboratory at 919-807-8938.

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES • DIVISION OF PUBLIC HEALTH • STATE LABORATORY OF PUBLIC HEALTH

LOCATION: 4312 District Drive, Raleigh, NC 27607

MAILING ADDRESS: 1918 Mail Service Center, Raleigh, NC 27699-1918

www.ncdhhs.gov • <http://publichealth.nc.gov> • slph.dph.ncdhhs.gov • TEL: 919-733-7834 • FAX: 919-733-8695

AN EQUAL OPPORTUNITY / AFFIRMATIVE ACTION EMPLOYER