



STATE OF WEST VIRGINIA
DEPARTMENT OF HEALTH AND HUMAN RESOURCES
Bureau for Public Health
Office of Maternal, Child and Family Health

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Interim Cabinet Secretary

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MEMORANDUM

To: All West Virginia Pediatricians, Family Physicians, Midwives and Neonatologists

From: The WV Newborn Screening System

Date: July 7, 2023

RE: Addition of Lysosomal Storage Disorders to Newborn Screening

Title 64, Series 91 of the W.Va. Code of State Rules authorizes newborn blood spot screening for the following Lysosomal Storage Disorders (LSD): Pompeii disease (glycogen storage disease II), Fabry disease (alpha-galactosidase-A deficiency), Hurlers syndrome (Mucopolysaccharidosis Type I) Hunter's Syndrome (Mucopolysaccharidosis type II) and Morquio syndrome (Mucopolysaccharidosis type IVa).

On March 7, 2023, the West Virginia Office of Laboratory Services (WVOLS) began screening for LSDs using the newborn blood spot specimen. LSDs are a disorder group of nearly 50 genetically acquired diseases. These disorders result from dysfunction, deficiency, or absence of a lysosomal enzyme. Affected individuals exhibit a broad variety of disease severities with widespread symptoms and ages of onset. Because symptoms are absent at birth, early diagnosis of these diseases by clinical observation can be difficult. Enzyme replacement therapy or hematopoietic stem cell transplantation are available treatments for some LSDs, but if an available therapy is delayed, irreversible tissue damage may result. Some LSDs may be fatal within the first year of life without treatment.

The infant's physician listed on the blood spot specimen card, West Virginia University (WVU) Genetics/Metabolics, and the WV Newborn Screening System follow-up nurse will be notified of an abnormal screen. WVU Genetics/Metabolics will work with the infant's physician and family to schedule additional genetic testing. WVU Genetics/Metabolics can be reached at 304-293-7334.

For more information on these newborn screening disorders, visit <https://babysfirsttest.org>.

For additional information on WV Newborn Screening System, visit www.wvdhhr.org/nbms or contact Deborah Templin at 304-558-5388 or deborah.s.templin@wv.gov.