



Date: September 19, 2018

To: Oregon Newborn screening
partners and clients

From: Christianne Biggs, Newborn Screening Manager

Subject: Addition of Lysosomal Storage Disorders (LSDs) to the Newborn Screening Panel

Beginning with specimens received at the Oregon State Public Health Laboratory (OSPHL) on October 1, 2018, the Newborn Screening Program will add testing for enzymatic activity for the following Lysosomal Storage Disorders (LSDs) to the newborn screening panel for infants born in Oregon:

<u>Disorder</u>	<u>Enzyme</u>	<u>Abbreviation</u>
Pompe	Alpha-D-glucosidase	GAA
MPS I	Alpha-L-iduronidase	IDUA
Fabry	Alpha-D-galactosidase A	GLA
Gaucher	Beta-glucocerebrosidase	GBA

What is changing?

All first specimens received for newborn screening at the OSPHL on or after October 1, 2018 will be tested for Pompe, MPS I, Gaucher, and Fabry in addition to the other disorders already on the screening panel.

There will be no changes to fees for newborn screening collection kits at this time. The fee change implemented April 1, 2018 supports this testing change.

How will the result report change?

For first screen specimens, the results will be expanded to include Lysosomal Storage Disorders. If the infant has normal results for all four disorders the result will be "Normal". If the infant is positive for an LSD, One of the following results will be listed with a value, the Disorder Evaluation will be "Abnormal", and the Reference range will be listed:

Pompe:	"GAA"
MPS I:	"IDUA"
Fabry:	"GLA"
Gaucher:	"GBA"

Additional comments will be included on the report to provide supporting information.

The test for LSDs will not be performed on second screen specimens unless the first specimen is unsatisfactory or abnormal. Therefore, there will be no change to the report for second screen unless another specimen has been requested for LSD testing.

How do partners and facilities implement this change?

Partners and facilities should continue to collect and submit first and second specimens as normal and continue to review newborn screening result reports for each infant. If there is an abnormal result you will be given instructions for additional actions that should be taken on the report or via fax or phone if the result is significant.

What are LSDs?

LSDs are a group of over 40 genetic disorders that result in enzyme deficiencies within the lysosomes of the body's cells, causing irreversible damage to the muscles, nerves, and organs in the body over time.

Lysosomes are spherical vesicles that contain hydrolytic enzymes and function to break down and recycle large molecules in cells. The presence of a LSD results in the accumulation of these large molecules in the lysosomes, causing the build-up and storage of certain compounds. The build-up of these compounds is what causes damage to the body's tissues.

Treatments may be available for these disorders if they are identified early, before symptoms begin to appear in the infant.

Why this change?

The U.S. Secretary of Health and Human Services approved the addition of Pompe and MPS I to the Recommended Uniform Screening Panel for Newborn Screening in 2015 and 2016 respectively. The testing method the OSPHL will use also includes Gaucher and Fabry, so these conditions will also be included. If identified early enough, treatment for these conditions can often significantly improve the health outcome for the infant.

If you have questions about these changes you may contact me at 503-693-4172 or Christianne.biggs@dhsosha.state.or.us.