Table 13: Lysosomal Diseases

Public Health Laboratory Division
Newborn Screening Laboratory
dshs.texas.gov/lab

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Lysosomal Diseases	Normal	
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Elevated. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Elevated. A homozygous 30KB Deletion was detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Elevated. A heterozygous 30KB Deletion was detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Elevated. No 30KB Deletion Detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Normal. A homozygous 30KB Deletion was detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Possible Krabbe disease. GALC activity Low. Psychosine Normal. A heterozygous 30KB Deletion was detected. Recommend consultation with a Krabbe Referral Center within 72 hours. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Indeterminate	Repeat the newborn screen within 72 hours. GALC activity Low. Insufficient or unsatisfactory sample to complete reflex testing.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. Psychosine Slightly Elevated. A homozygous 30KB Deletion was detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Possible Krabbe disease. GALC activity Low. Psychosine Slightly Elevated. A heterozygous 30KB Deletion was detected. Recommend consultation with a Krabbe Referral Center within 72 hours. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Probable Krabbe disease. GALC activity Low. A homozygous 30KB Deletion was detected. Insufficient or unsatisfactory sample to complete Psychosine testing. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Possible Krabbe disease. GALC activity Low. A heterozygous 30KB Deletion was detected. Insufficient or unsatisfactory sample to complete Psychosine testing. Recommend consultation with a Krabbe Referral Center within 72 hours. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Possible Krabbe disease. GALC activity Low. Psychosine Slightly Elevated. Recommend consultation with a Krabbe Referral Center within 72 hours. Follow recommendations received from Clinical Care Coordination.
Abnormal	Lysosomal Diseases	Abnormal	Possible Krabbe disease. GALC activity Low. Psychosine Slightly Elevated. No 30KB Deletion Detected. Recommend consultation with a Krabbe Referral Center within 72 hours. Follow recommendations received from Clinical Care Coordination.

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Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Lysosomal Diseases	Abnormal	Possible Mucopolysaccharidosis type I (Hurler syndrome). IDUA activity Low. GAGs Elevated. Recommend confirmatory urine glycosaminoglycans (GAGs) quantitative analysis and serum alpha-L-iduronidase enzyme assay in leukocytes and consultation with a pediatric metabolic specialist within 5 days. Follow recommendations received from Clinical Care Coordination. DNA report to follow.
Abnormal	Lysosomal Diseases	Abnormal	Possible Mucopolysaccharidosis type I (Hurler syndrome). IDUA activity Low. GAGs Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Lysosomal Diseases	Abnormal	Possible Mucopolysaccharidosis type II (Hunter syndrome). I2S activity Low. GAGs Elevated. Recommend confirmatory urine glycosaminoglycans (GAGs) quantitative analysis and serum iduronate 2-sulfatase (I2S) enzyme assay in leukocytes and consultation with a pediatric metabolic specialist within 5 days. Follow recommendations received from Clinical Care Coordination. DNA report to follow.
Abnormal	Lysosomal Diseases	Abnormal	Possible Mucopolysaccharidosis type II (Hunter syndrome). I2S activity Low. GAGs Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Lysosomal Diseases	Abnormal	Possible Pompe disease. GAA activity Low. Recommend immediate consultation with a pediatric metabolic specialist, immediate cardiac evaluation including chest x-ray, electrocardiogram and echocardiogram within 24 hours. Follow recommendations received from Clinical Care Coordination. DNA report to follow.
Abnormal	Lysosomal Diseases	Abnormal	Possible Pompe disease. GAA activity Slightly Low. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 72 hours.
Abnormal	Lysosomal Diseases	Abnormal	free text
Abnormal	Lysosomal Diseases	Revised Result	free text
Normal	Lysosomal Diseases	Revised Result	free text
Unsatisfactory	Lysosomal Diseases	Revised Result	free text