Table 1: Amino Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Normal		Normal	
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Arginine Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible Argininemia. Arginine Elevated. Recommend plasma ammonia, plasma quantitative amino acids and urine orotic acid within 48 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Citrulline Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible ASA, Citrullinemia or Citrullinemia Type II. Citrulline Elevated. Recommend immediate plasma ammonia, quantitative plasma amino acids, urine organic acids, urine orotic acid and liver function tests within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Leucine Slightly Elevated; Valine Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible Maple Syrup Urine Disease. Leucine Elevated. Recommend plasma quantitative amino acids and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible Maple Syrup Urine Disease. Leucine Slightly Elevated; Valine Elevated. Recommend plasma quantitative amino acids and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Methionine Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible Homocystinuria or Hypermethioninemia. Methionine Elevated. Recommend quantitative plasma amino acids and plasma total homocysteine and telephone consultation with a pediatric metabolic specialist within 24 hours.
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Phenylalanine Slightly Elevated; Phe/Tyr Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible PKU, Benign Hyperphenylalaninemia, Biopterin defect in cofactor biosynthesis or Biopterin defect in cofactor regeneration. Phenylalanine Slightly Elevated; Phe/Tyr Elevated. Recommend plasma quantitative amino acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible PKU, Benign Hyperphenylalaninemia, Biopterin defect in cofactor biosynthesis or Biopterin defect in cofactor regeneration. Phenylalanine Elevated. Recommend plasma quantitative amino acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Possible Tyrosinemia Type I. Succinylacetone Elevated. Recommend plasma quantitative amino acids, succinylacetone and liver function tests within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.

Table 1: Amino Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Tyrosine Elevated; Succinylacetone Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible Amino Acid Disorder. Tyrosine Slightly Elevated; Succinylacetone Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	Abnormal	Possible Tyrosinemia Type II, III or Transient Tyrosinemia of the Neonate. Tyrosine Elevated; Succinylacetone Normal. Recommend quantitative plasma amino acids, urine organic acids, succinylacetone, liver function tests and telephone consultation with a pediatric metabolic specialist within 24 hours.
Abnormal	Amino Acid Disorders	Non-specific	Elevation(s) in a non-diagnostic pattern. Repeat the newborn screen within 7 days.
Abnormal	Amino Acid Disorders	TPN	Possible TPN. Repeat the newborn screen when TPN is discontinued.
Abnormal	Amino Acid Disorders	Revised Result	free text
Normal	Amino Acid Disorders	Revised Result	free text
Unsatisfactory	Amino Acid Disorders	Revised Result	free text

Table 2: Fatty Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Normal		Normal	
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C0 Slightly Low. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C0 Low; C3+C16 Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C0 Slightly Low; C3+C16 Low. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CUD. C0 Low for birthweight less than 1000 grams. Place baby on carnitine immediately. Recommend immediate telephone consultation with a pediatric metabolic specialist. Repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CUD. C0 and C3+C16 Low for birthweight less than 1000 grams. Place baby on carnitine immediately. Recommend immediate telephone consultation with a pediatric metabolic specialist. Repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CUD. C0 Low. Recommend blood sugar, plasma (free and total) carnitine and maternal plasma (free and total) carnitine within 7 days and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CUD. C0 and C3+C16 Low. Recommend blood sugar, plasma (free and total) carnitine and maternal plasma (free and total) carnitine within 7 days and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CPT1. C0/(C16+C18) Elevated. Recommend plasma carnitine and plasma acylcarnitine profile within 7 days and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible DE-RED. C10:2 Elevated. Recommend plasma acylcarnitine profile, plasma quantitative amino acids, plasma carnitine, urine acylglycines, urine organic acids and telephone consultation with a pediatric metabolic specialist within 48 hours.
Abnormal	Fatty Acid Disorders	Abnormal	Possible VLCAD. C14:1 Slightly Elevated. Recommend plasma acylcarnitine profile and plasma (free and total) carnitine within 24 hours and immediate telephone consultation with a pediatric metabolic specialist. DNA report to follow.
Abnormal	Fatty Acid Disorders	Abnormal	Possible VLCAD. C14:1 Elevated. Recommend plasma acylcarnitine profile and plasma (free and total) carnitine within 24 hours and immediate telephone consultation with a pediatric metabolic specialist. DNA report to follow.
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C16 Slightly Elevated; C18:1, C14, and (C16+C18:1)/C2 Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CACT or CPTII. C16 Slightly Elevated. Recommend plasma carnitine, plasma acylcarnitine, and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible CACT or CPTII. C16 Elevated. Recommend plasma carnitine, plasma acylcarnitine, and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.

Table 2: Fatty Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Fatty Acid Disorders	Abnormal	Possible LCHAD or TFP. C16-OH, C16:1-OH, C18-OH, C18:1-OH, and C18:2-OH analyzed to determine result. Recommend plasma acylcarnitine profile, urine organic acids and plasma carnitine profile (free and total carnitine) within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C4 Slightly Elevated; C4/C2 Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible Fatty Acid Disorder. C4 Elevated; C4/C2 Normal. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	Abnormal	Possible SCAD or IBG. C4 and C4/C2 Elevated. Recommend plasma acylcarnitine profile, urine organic acids, urine acylglycines and telephone consultation with a pediatric metabolic specialist within 48 hours.
Abnormal	Fatty Acid Disorders	Abnormal	Possible GA2. C4 and C5 analyzed to determine result. Recommend plasma acylcarnitine profile, urine organic acids, and urine acylglycine analysis within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	Possible MCAD or MCAT. C8 Slightly Elevated. Recommend plasma acylcarnitine profile, plasma carnitine levels, urine acylglycines, and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist. DNA report to follow.
Abnormal	Fatty Acid Disorders	Abnormal	Possible MCAD or MCAT. C8 Elevated. Recommend plasma acylcarnitine profile, plasma carnitine levels, urine acylglycines, and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist. DNA report to follow.
Abnormal	Fatty Acid Disorders	Non-specific	Elevation(s) in a non-diagnostic pattern. Repeat the newborn screen within 7 days.
Abnormal	Fatty Acid Disorders	TPN	Possible TPN. Repeat the newborn screen when TPN is discontinued.
Normal	Fatty Acid Disorders	Revised Result	free text
Abnormal	Fatty Acid Disorders	Revised Result	free text
Unsatisfactory	Fatty Acid Disorders	Revised Result	free text

Table 3: Organic Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Normal		Normal	
Abnormal	Organic Acid Disorders	Abnormal	Possible Organic Acid Disorder. C3 and C3/C2 analyzed to determine result. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	Abnormal	Possible Methylmalonic Acidemia or Propionic Acidemia. C3 Elevated. Recommend plasma methylmalonic acid, total plasma homocysteine, plasma acylcarnitine profile and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible Methylmalonic Acidemia or Propionic Acidemia. C3 Slightly Elevated; C3/C2 Elevated. Recommend plasma methylmalonic acid, total plasma homocysteine, plasma acylcarnitine profile and urine organic acids within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible Organic Acid Disorder. C3DC+C4OH Slightly Elevated; C3DC+C4OH/C5DC Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	Abnormal	Possible MAL or M/SCHAD. C3DC+C4OH Elevated. Recommend plasma acylcarnitine profile, plasma insulin, plasma methylmalonic acid, urine organic acids and telephone consultation with a pediatric metabolic specialist within 48 hours.
Abnormal	Organic Acid Disorders	Abnormal	Possible Organic Acid Disorder. C5 Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	Abnormal	Possible Isovaleric Acidemia or 2MBG. C5 Elevated. Recommend plasma acylcarnitine profile, urine organic acids and urine acylglycines within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible GA1. C5DC Elevated. Recommend plasma acylcarnitine profile and urine organic acids within 7 days and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible Organic Acid Disorder. C5-OH Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	Abnormal	Possible Organic Acid Disorder. C5:1 Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	Abnormal	Possible 3MCC, HMG, MCD, 3MGA or 2M3HBA. C5-OH Elevated. Recommend urine organic acids, urine acylglycines and plasma acylcarnitine profile on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible 3MCC, HMG, MCD, BKT, 3MGA or 2M3HBA. C5-OH and C5:1 Elevated. Recommend urine organic acids, urine acylglycines and plasma acylcarnitine profile on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible 3MCC, HMG, MCD, BKT, 3MGA or 2M3HBA. C5-OH Slightly Elevated; C5:1 Elevated. Recommend urine organic acids, urine acylglycines and plasma acylcarnitine profile on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible BKT. C5:1 Elevated; C5-OH and C6DC Normal. Recommend urine organic acids, plasma acylcarnitine profile and urine acylglycines on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.

Table 3: Organic Acid Disorders

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Organic Acid Disorders	Abnormal	Possible HMG. C5-OH and C6DC Elevated; C5:1 Normal. Recommend urine organic acids, plasma acylcarnitine profile and urine acylglycines on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	Possible HMG. C5-OH Slightly Elevated; C6DC Elevated; C5:1 Normal. Recommend urine organic acids, plasma acylcarnitine profile and urine acylglycines on infant and mother within 24 hours and immediate telephone consultation with a pediatric metabolic specialist.
Abnormal	Organic Acid Disorders	Non-specific	Elevation(s) in a non-diagnostic pattern. Repeat the newborn screen within 7 days.
Abnormal	Organic Acid Disorders	TPN	Possible TPN. Repeat the newborn screen when TPN is discontinued.
Normal	Organic Acid Disorders	Revised Result	free text
Abnormal	Organic Acid Disorders	Revised Result	free text
Unsatisfactory	Organic Acid Disorders	Revised Result	free text

Table 4: Galactosemia

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Galactosemia	Normal	
Abnormal	Galactosemia	Abnormal	Possible Galactosemia. GALT activity Low. If this is the first screen, recommend serum GALT enzyme within 24 hours and immediate telephone consultation with a pediatric metabolic specialist. Otherwise, follow recommendations received from Clinical Care Coordination. DNA report to follow.
Abnormal	Galactosemia	Abnormal	GALT activity Indeterminate. Result may be due to limitation of the test method and should be considered in context of the child's clinical status. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days. DNA report to follow.
Abnormal	Galactosemia	Revised Result	free text
Normal	Galactosemia	Revised Result	free text
Unsatisfactory	Galactosemia	Revised Result	free text

Table 5: Biotinidase Deficiency

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Biotinidase Deficiency	Normal	
Abnormal	Biotinidase Deficiency	Abnormal	Possible Biotinidase Deficiency. Biotinidase activity Low. Recommend serum biotinidase and telephone consultation with a pediatric metabolic specialist within 7 days. If this is the first screen, repeat the newborn screen within 7 days.
Abnormal	Biotinidase Deficiency	Revised Result	free text
Normal	Biotinidase Deficiency	Revised Result	free text
Unsatisfactory	Biotinidase Deficiency	Revised Result	free text

Table 6: Hypothyroidism

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Hypothyroidism	Normal	
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. TSH Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. TSH Moderately Elevated. Recommend TSH, Free T4, and T4 within 5 days. If this is the first screen, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. TSH Very Elevated. Recommend TSH, Free T4, and T4 and refer to an endocrinologist within 24 hours. If this is the first screen, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. T4 Low. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. T4 Low; TSH Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. T4 Low; TSH Moderately Elevated. Recommend TSH, Free T4, and T4 and refer to an endocrinologist within 5 days. If this is the first screen, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hypothyroidism. T4 Low; TSH Very Elevated. Recommend TSH, Free T4, and T4 and refer to an endocrinologist within 24 hours. If this is the first screen, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible TBG Excess. T4 Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	Hypothyroidism	Abnormal	Possible Hyperthyroidism. T4 and TSH Elevated. Recommend TSH, Free T4, and T4 and refer to an endocrinologist within 5 days.
Abnormal	Hypothyroidism	Revised Result	free text
Normal	Hypothyroidism	Revised Result	free text
Unsatisfactory	Hypothyroidism	Revised Result	free text

Table 7: CAH

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	CAH	Normal	
Abnormal	CAH	Abnormal	Possible CAH. 17-Hydroxyprogesterone Elevated for birth weight less than 2500 grams. Insufficient specimen to complete reflex testing. Repeat the newborn screen within 7 days.
Abnormal	CAH	Abnormal	Possible CAH. 17-Hydroxyprogesterone (17-OHP) Very Elevated for birth weight less than 2500 grams. Insufficient specimen to complete reflex testing. Recommend serum 17-OHP, sodium and potassium and refer to an endocrinologist within 24 hours.
Abnormal	CAH	Abnormal	Possible CAH. 17-Hydroxyprogesterone Slightly Elevated for birth weight greater than or equal to 2500 grams. Insufficient specimen to complete reflex testing. If this is the second newborn screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	CAH	Abnormal	Possible CAH. 17-Hydroxyprogesterone (17-OHP) Moderately Elevated for birth weight greater than or equal to 2500 grams. Insufficient specimen to complete reflex testing. Recommend serum 17-OHP, sodium and potassium within 24 hours.
Abnormal	CAH	Abnormal	Probable CAH. 17-Hydroxyprogesterone (17-OHP) Very Elevated for birth weight greater than or equal to 2500 grams. Insufficient specimen to complete reflex testing. Recommend serum 17-OHP and daily sodium and potassium and refer to an endocrinologist within 24 hours.
Abnormal	CAH	Abnormal	Probable CAH. 17-Hydroxyprogesterone (17-OHP) Elevated and reflex panel Abnormal. Recommend serum 17-OHP within 24 hours. Follow additional recommendations received from Clinical Care Coordination.
Abnormal	CAH	Abnormal	Possible CAH. 17-Hydroxyprogesterone Elevated and reflex panel Abnormal for birth weight less than 2500 grams. Repeat the newborn screen within 7 days. Follow additional recommendations received from Clinical Care Coordination.
Abnormal	CAH	Revised Result	free text
Normal	CAH	Revised Result	free text
Unsatisfactory	CAH	Revised Result	free text

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Hemoglobinopathies	Normal	
Abnormal	Hemoglobinopathies	Abnormal	Probable Normal. Only Hemoglobin A detected. If result is due to transfusion, repeat in three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait. Hemoglobin A,C detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait. Hemoglobin A,C,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable D Trait. Hemoglobin A,D detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable D Trait. Hemoglobin A,D,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable E Trait. Hemoglobin A,E detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable E Trait. Hemoglobin A,E,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable Normal. Hemoglobin A,F detected. If result is due to transfusion, repeat in three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait. Hemoglobin A,F,C detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable D Trait. Hemoglobin A,F,D detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable G Trait. Hemoglobin A,F,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant Trait. Hemoglobin A,F,Other detected. Notify family of test results. For additional information see (http://www.dshs.texas.gov/newborn/pdf/fAOther.pdf).
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant Trait and Alpha Thalassemia Trait. Hemoglobin A,F,Other,Barts detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait. Hemoglobin A,F,S detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait and Alpha Thalassemia Trait. Hemoglobin A,F,S,Barts detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable G Trait. Hemoglobin A,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable G Trait. Hemoglobin A,G,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant Trait. Hemoglobin A,Other detected. Notify family of test results. For additional information see (http://www.dshs.texas.gov/newborn/pdf/fAOther.pdf).
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait. Hemoglobin A,S detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait. Hemoglobin A,S,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable Hemoglobin C/Beta Thalassemia Disease. Hemoglobin C,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable CC Disease. Only Hemoglobin C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable CC Disease. Hemoglobin C,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable EE Disease. Only Hemoglobin E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable EE Disease. Hemoglobin E,F detected. Refer to pediatric hematologist within one month. DNA report to follow.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Possible Beta Thalassemia Major. Only Hemoglobin F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable Alpha Thalassemia Trait. Hemoglobin F,A,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait. Hemoglobin F,A,C detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait and Alpha Thalassemia Trait. Hemoglobin F,A,C,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait and G Trait. Hemoglobin F,A,C,G detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait, G Trait, and Alpha Thalassemia Trait. Hemoglobin F,A,C,G,Barts detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable C Trait and Unidentified Hb Variant Trait. Hemoglobin F,A,C,Other detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable D Trait. Hemoglobin F,A,D detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable D Trait and Alpha Thalassemia Trait. Hemoglobin F,A,D,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable E Trait. Hemoglobin F,A,E detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable E Trait and Alpha Thalassemia Trait. Hemoglobin F,A,E,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable H Disease. Hemoglobin F,A,elevated Barts detected. Refer to a pediatric hematologist within one month.
Abnormal	Hemoglobinopathies	Abnormal	Probable G Trait. Hemoglobin F,A,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable G Trait and Alpha Thalassemia Trait. Hemoglobin F,A,G,Barts detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable O-Arab Trait. Hemoglobin F,A,O-Arab detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant Trait. Hemoglobin F,A,Other detected. Notify family of test results. For additional information see (http://www.dshs.texas.gov/newborn/pdf/fAOther.pdf).
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant Trait and Alpha Thalassemia Trait. Hemoglobin F,A,Other,Barts detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait. Hemoglobin F,A,S detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait and Unidentified Hb Variant. Hemoglobin F,A,S,Other detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait and Alpha Thalassemia Trait. Hemoglobin F,A,S,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait and G Trait. Hemoglobin F,A,S,G detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable S Trait, G Trait, and Alpha Thalassemia Trait. Hemoglobin F,A,S,G,Barts detected. Notify family of test results. Consult with pediatric hematologist between 6 and 12 months of age.
Abnormal	Hemoglobinopathies	Abnormal	Probable CC Disease. Hemoglobin F,C detected. Refer to pediatric hematologist within one month. DNA report to follow.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Possible CC or Hemoglobin C/Beta Thalassemia Disease. Hemoglobin F,C (A Questionable) detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable Hemoglobin C/Beta Thalassemia Disease. Hemoglobin F,C,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable CC Disease and Alpha Thalassemia Trait. Hemoglobin F,C,Barts detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable CE Disease. Hemoglobin F,C,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable DD Disease. Hemoglobin F,D detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable EE Disease. Hemoglobin F,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Possible EE or Hemoglobin E/Beta Thalassemia Disease. Hemoglobin F,E (A Questionable) detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable EE Disease and Alpha Thalassemia Trait. Hemoglobin F,E,Barts detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable GG Disease. Hemoglobin F,G detected. Refer to pediatric hematologist within one month.
Abnormal	Hemoglobinopathies	Abnormal	Probable Unidentified Hb Variant. Hemoglobin F,Other detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SS Disease. Hemoglobin F,S detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Possible Sickle Cell or Sickle Beta Thalassemia Disease. Hemoglobin F,S (A Questionable) detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable Sickle Beta Thalassemia Disease. Hemoglobin F,S,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SS Disease and Alpha Thalassemia Trait. Hemoglobin F,S,Barts detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SC Disease. Hemoglobin F,S,C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SC Disease and Alpha Thalassemia Trait. Hemoglobin F,S,C,Barts detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SD Disease. Hemoglobin F,S,D detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SE Disease. Hemoglobin F,S,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable Sickle Beta Thalassemia Disease. Hemoglobin S,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SC Disease. Hemoglobin S,C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SC Disease. Hemoglobin S,C,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SS Disease. Hemoglobin S,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Probable SS Disease. Only Hemoglobin S detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	free text

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Hemoglobinopathies	Revised Result	free text
Normal	Hemoglobinopathies	Revised Result	free text
Unsatisfactory	Hemoglobinopathies	Revised Result	free text

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Cystic Fibrosis	Normal	
Normal	Cystic Fibrosis	Normal	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Normal. None of the CFTR variants in the DSHS panel were detected. However, the presence of other variants not included in the panel cannot be ruled out.
Abnormal	Cystic Fibrosis	Indeterminate	Repeat the newborn screen within 72 hours. Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level on the first specimen. The second screening specimen is required to determine if result is significant.
Abnormal	Cystic Fibrosis	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. Elevated IRT level is consistent with the previous newborn screening result. None of the CFTR variants in the DSHS panel were detected in the previous specimen.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. This result is consistent with the previous newborn screening specimen results for the same baby. CF cannot be ruled out.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. This result is consistent with the previous newborn screening specimen results for the same baby. The previous specimen results are indicative of CF.
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	Revised Screening Result for Cystic Fibrosis (CF). Additional testing using a CFTR Mutation Panel has been performed. *(Note: Result notes vary depending on the results applied for CFTR Mutation Panel)* [The specimen was originally reported as Indeterminate for CF. The original screening result note read "Repeat the newborn screen within 72 hours. Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level on the first specimen. The second screening specimen is required to determine if result is significant."]
Abnormal	Cystic Fibrosis	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected. However, there is a minimal risk for Cystic Fibrosis due to variants not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Very Elevated. None of the CFTR variants in the DSHS panel were detected. Although there is a minimal risk for CF in the absence of detected variants, a very elevated IRT result may be indicative of CF due to variants not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1078delT (c.948delT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1717-1G>A (c.1585-1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1898+1G>A (c.1766+1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2183AA>G (c.2051_2052delAAinsG), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2184delA (c.2052delA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2789+5G>A (c.2657+5G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3120+1G>A (c.2988+1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3659delC (c.3528delC), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3849+10kbC>T (c.3717+12191C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3849+4A>G (c.3717+4A>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3876delA (c.3744delA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3905insT (c.3773_3774insT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 394delTT (c.262_263delTT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 621+1G>T (c.489+1G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 711+1G>T (c.579+1G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, A455E (c.1364C>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, D1152H (c.3454G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, DF508 (c.1521_1523delCTT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, DI507 (c.1519_1521delATC), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant which is not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, E60X (c.178G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G542X (c.1624G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G551D (c.1652G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G85E (c.254G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, N1303K (c.3909C>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Q493X (c.1477C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1162X (c.3484C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-9T/9T (c.[350G>A;1210-12[9]/1210-12[9]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R334W (c.1000C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R347H (c.1040G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R347P (c.1040G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R553X (c.1657C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R560T (c.1679G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549N (c.1646G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549R A>C (c.1645A>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549R T>G (c.1647T>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, V520F (c.1558G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, W1282X (c.3846G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y1092X C>A (c.3276C>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y1092X C>G (c.3276C>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y122X (c.366T>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 406-1G>A (c.274-1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 935delA (c.803delA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1677delTA (c.1545_1546delTA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1898+5G->T (c.1766+5G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2055del9>A (c.1923_1931del9insA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2143delT (c.2012delT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2307insA (c.2175_2176insA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3791delC (c.3659delC), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3199del6 (c.3067_3072delATAGTG), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, A559T (c.1675G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, CFTRdele2,3 (c.54-5940_273+10250del21kb), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G178R (c.532G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G330X (c.988G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, K710X (c.2128A>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, L206W (c.617T>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, M1101K (c.3302T>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Q890X (c.2668C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R75X (c.223C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H (c.350G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-5T/5T (c.[350G>A;1210-12[5]/1210-12[5]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-5T/9T (c.[350G>A;1210-12[5]/1210-12[9]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1066C (c.3196C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1158X (c.3472C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S1196X (c.3587C>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S1255X (c.3764C>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, W1089X (c.3266G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, DF508 (c.1521_1523delCTT), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G542X (c.1624G>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G551D (c.1652G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and N1303K (c.3909C>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and W1282X (c.3846G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R553X (c.1657C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 621+1G>T (c.489+1G>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-9T/9T (c.[350G>A;1210-12[9]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, 935delA (c.803delA), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, 1078delT (c.948delT), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, G551D (c.1652G>A), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, Q493X (c.1477C>T), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, 3876delA (c.3744delA), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, L206W (c.617T>G), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, D1152H (c.3454G>C), in the CFTR gene was identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 1717-1G>A (c.1585-1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 2184delA (c.2052delA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 2307insA (c.2175_2176insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 2789+5G>A (c.2657+5G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3120+1G>A (c.2988+1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3876delA (c.3744delA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and D1152H (c.3454G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and DI507 (c.1519_1521delATC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G85E (c.254G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508(c.1521_1523delCTT) and Q493X (c.1477C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R1162X (c.3484C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-5T/9T (c.[350G>A;1210-12[5]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and S1255X (ex. 20) (c.3764C>A), in the CFTR gene were identified. A variant of uncertain significance, S1255X (ex. 19) (I1203V, c.3607A>G), was also identified. While S1255X (ex. 19) and S1255X (ex. 20) are two separate mutations, they are generally considered a haplotype with the two mutations occurring together (Cutting G, et al. 1992. Am. J. Hum. Genet. 50:1185-1194).
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and S549N (c.1646G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3791delC (c.3659delC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G330X (c.988G>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 2055del9>A (c.1923_1931del9insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 1898+1 G>A (c.1766+1 G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3659delC (c.3528delC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3849+10kbC>T (c.3717+12191C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 3905insT (c.3773_3774insT), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 406-1G>A (c.274-1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and A455E (c.1364C>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and CFTRdele2,3 (c.54-5940_273+10250del21kb), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R1158X (c.3472C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R347H (c.1040G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R347P (c.1040G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DI507 (c.1519_1521delATC) and D1152H (c.3454G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, G542X (c.1624G>T) and 2055del9>A (c.1923_1931del9insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, G542X (c.1624G>T) and L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, L206W (c.617T>G) and Q493X (c.1477C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, L206W (c.617T>G) and S549N (c.1646G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, 2789+5G>A (c.2657+5G>A) and R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, D1152H (c.3454G>C) and R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, R347H (c.1040G>A) and R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, R560T (c.1679G>C) and R117H with IVS8-7T/7T (c.[350G>A;1210−12[7]/1210−12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, 3199del6 (c.3067_3072delATAGTG) and R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, 2307insA (c.2175_2176insA) and R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, G542X (c.1624G>T) and R117H with IVS8-7T/9T (c.[350G>A; 1210-12[7]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, G551D (c.1652G>A) and R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, D1152H (c.3454G>C) and L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Galactosemia	Abnormal	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-5T/5T (c.[350G>A;1210-12[5]/1210-12[5]]), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Normal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Normal. One variant, DF508 (c.1521_1523delCTT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected except a “No Call” result for both the wild type and variant R75X loci. Possible reason for a “No Call” result includes a variant or polymorphism within or around the primer annealing sites. There is a minimal risk for Cystic Fibrosis due to a variant not included in the panel. Clinical evaluation not necessary unless symptomatic.
Abnormal	Cystic Fibrosis	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected except a “No Call” result for both the wild type and variant R1162X loci. Possible reason for a “No Call” result includes a variant or polymorphism within or around the primer annealing sites. There is a minimal risk for Cystic Fibrosis due to a variant not included in the panel. Clinical evaluation not necessary unless symptomatic.
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level. Additional testing for a panel of mutations in the CFTR gene is in progress to determine if result is significant. Final report with CFTR Mutation panel results to follow.
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Very Elevated. Although there is a minimal risk for CF in the absence of detected variants, a very elevated IRT result may be indicative of CF. Additional testing for a panel of mutations in the CFTR gene is in progress. Final report with CFTR Mutation panel results to follow.
Abnormal	Cystic Fibrosis	Abnormal	free text
Normal	Cystic Fibrosis	Revised Result	free text
Abnormal	Cystic Fibrosis	Revised Result	free text
Unsatisfactory	Cystic Fibrosis	Revised Result	free text

Table 10: SCID

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	SCID	Normal	
Abnormal	SCID	Abnormal	Possible Severe Combined Immunodeficiency or other T-cell lymphopenia. T-cell receptor excision circles (TREC) number Very Low. Follow recommendations received from Clinical Care Coordination.
Abnormal	SCID	Revised Result	free text
Normal	SCID	Revised Result	free text
Unsatisfactory	SCID	Revised Result	free text

Table 11: X-ALD

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	X-ALD	Normal	
Abnormal	X-ALD	Abnormal	Possible X-ALD. C26:0 LPC Elevated. Recommend confirmatory very long chain fatty acids and consultation with a pediatric metabolic specialist or pediatric neurogeneticist within 7 days. DNA report to follow.
Abnormal	X-ALD	Abnormal	Possible X-ALD. C26:0 LPC Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	X-ALD	Revised Result	free text
Normal	X-ALD	Revised Result	free text
Unsatisfactory	X-ALD	Revised Result	free text

Table 12: SMA

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	SMA	Normal	
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Zero copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. One copy of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Two copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Three copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Four copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 72 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Four or more copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 72 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Five or more copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number within 24 hours and referral to a neurologist or neurogeneticist within 7 days.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. SMN2 copy number cannot be determined. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. This result is consistent with the previous newborn screening specimen results for the same baby. If not already completed, recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	free text
Normal	SMA	Revised Result	free text
Abnormal	SMA	Revised Result	free text
Unsatisfactory	SMA	Revised Result	free text

Table 13: Lysosomal Diseases

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.

Table 13: Lysosomal Diseases

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

Table 14:
Unsatisfactory for All Tests

Overall Result	Disorder	Screening Result	Screening Result Note
Unsatisfactory	ALL	Unsatisfactory	Blood did not completely fill specimen circles. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Blood did not soak through paper due to incomplete saturation. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Filter paper is scratched from the possible use of capillary tubes. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen appears contaminated or discolored. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Blood was caked, clotted, or layered onto the filter paper. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Patient information incomplete or invalid (e.g. date of collection missing). Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Incomplete, invalid, or no patient identification on specimen. The requirement for two patient-specific identifiers on the specimen was not met. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Serial number on demographic form does not match number on specimen filter paper. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen too old upon receipt. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unable to analyze specimen due to laboratory accident. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	No blood samples received with request form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen submitted on improper collection form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen submitted on expired collection form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Serum separation due to improper drying or specimen collection. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen damaged during transport to laboratory. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Information on demographic form does not match electronically submitted information. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Specimen received in hermetically sealed container. Resubmit within 7 days.

Table 15:
Partially Unsatisfactory

Overall Result	Disorder	Screening Result	Screening Result Note
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Blood did not completely fill specimen circles. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Blood did not soak through paper - Incomplete saturation. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Filter paper is scratched from the possible use of capillary tubes. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen appears contaminated or discolored. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Blood was caked, clotted, or layered onto the filter paper. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Assay interference due to EDTA/Citrate anticoagulant contamination. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Assay interference. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Incomplete elution of blood from filter paper. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Patient information incomplete or invalid (e.g. date of collection missing). Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Incomplete, invalid, or no patient identification on specimen. The requirement for two patient-specific identifiers on the specimen was not met. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Serial number on demographic form does not match number on specimen filter paper. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen too old upon receipt. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Unable to analyze specimen due to laboratory accident. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: No blood samples received with request form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen submitted on improper collection form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen submitted on expired collection form. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen results inconsistent. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Serum separation due to improper drying or specimen collection. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen damaged during transport to laboratory. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Information on demographic form does not match electronically submitted information. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Specimen received in hermetically sealed container. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Testing of this specimen indicates more than one source of blood is present on the filter paper card. Results are inconsistent and do not appear to be due to transfusion. Resubmit within 7 days.
Unsatisfactory	Cystic Fibrosis	Unsatisfactory	Unsatisfactory: Specimen too old to test for immunoreactive trypsinogen (IRT). Resubmit within 7 days.
Unsatisfactory	SCID	Unsatisfactory	Unsatisfactory: Unable to evaluate for detection of TREC (T-cell receptor excision circles) possibly due to low DNA quantity. Resubmit within 7 days.
Unsatisfactory	SMA	Unsatisfactory	Unsatisfactory: Unable to evaluate for detection of SMN1 gene. Resubmit within 7 days.
Unsatisfactory	ALL	Unsatisfactory	Unsatisfactory: Insufficient specimen to complete testing. Repeat the newborn screen within 7 days.

Table 15:
Partially Unsatisfactory

Overall Result	Disorder	Screening Result	Screening Result Note
Unsatisfactory	Lysosomal Diseases	Unsatisfactory	Unsatisfactory: Unable to evaluate for Lysosomal Diseases. All enzymatic activity low. Resubmit within 7 days.