Table 1: Amino Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Amino Acid Disorders	Abnormal	Arginine	Borderline	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	Arginine	Elevated	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	Citrulline	Borderline	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	Citrulline	Elevated	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	Leucine Valine	Borderline Normal	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	Leucine Valine	Elevated Elevated or Normal	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	Leucine Valine	Borderline Elevated	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.

Table 1: Amino Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Amino Acid Disorders	Abnormal	Methionine	Borderline	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	Methionine	Elevated	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	Phenylalanine Phe/Tyr	Borderline Normal	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	Phenylalanine Phe/Tyr	Borderline Elevated	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	Phenylalanine Phe/Tyr	Elevated Elevated or Normal	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	Succinylacetone Tyrosine	Elevated Elevated, Borderline, or Normal	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.
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine Succinylacetone	Elevated Normal	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.

Table 1: Amino Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine Succinylacetone	Borderline Normal	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	Tyrosine Succinylacetone	Elevated Normal	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	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Fatty Acid Disorders	Abnormal	C0 C3+C16	Borderline Low or Normal	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	C0 C3+C16	Low Normal	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	C0 C3+C16	Borderline Low	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	C0 C3+C16	Low Low or Normal	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	C0 C3+C16	Low Low	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	C0 C3+C16	Low Low or Normal	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	C0 C3+C16	Low Low	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.

Table 2: Fatty Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Fatty Acid Disorders	Abnormal	C0/(C16 +C18) C0	Elevated Elevated, Borderline, or Normal	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	C10:2	Elevated	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	C14:1 C14 C14:1/C2	Borderline Elevated or Normal Elevated or Normal	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	C14:1 C14 C14:1/C2	Elevated Elevated or Normal Elevated or Normal	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	C16 C18:1 C14 (C16+C18:1)/C2	Borderline Normal Normal Normal	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	C16 C18:1 C14 (C16+C18:1)/C2	Borderline Elevated or Normal Elevated or Normal Elevated or Normal	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	C16 C18:1 C14 (C16+C18:1)/C2	Elevated Elevated or Normal Elevated or Normal Elevated or Normal	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	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Fatty Acid Disorders	Abnormal	C16-OH C16:1-OH C18-OH C18:1-OH C18:2-OH	Elevated Elevated or Normal Elevated or Normal Elevated or Normal Elevated or Normal	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	C4 C4/C2	Borderline Elevated	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	C4 C4/C2	Elevated Normal	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	C4 C4/C2	Elevated Elevated	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	C4 C5	Elevated or Borderline Elevated or Borderline	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	C8 C6 C10:1 C10 C8/C2	Borderline Elevated or Normal Elevated or Normal Elevated or Normal Elevated or Normal	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	C8 C6 C10:1 C10 C8/C2	Elevated Elevated or Normal Elevated or Normal Elevated or Normal Elevated or Normal	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.

Table 2: Fatty Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
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	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Organic Acid Disorders	Abnormal	C3 C3/C2	Borderline or Normal Elevated or Normal	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	C3 C3/C2	Elevated Elevated or Normal	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	C3 C3/C2	Borderline Elevated	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	C3DC+C4OH C3DC+C4OH/C5DC	Borderline Elevated	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	C3DC+C4OH C3DC+C4OH/C5DC	Elevated Elevated or Normal	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	C5	Borderline	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.

Table 3: Organic Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Organic Acid Disorders	Abnormal	C5	Elevated	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	C5DC	Elevated	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	C5-OH C5:1 C6DC	Borderline Elevated, Borderline, or Normal	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	C5:1 C5-OH C6DC	Borderline Elevated, Borderline, or Normal	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	C5-OH C5:1 C6DC	Elevated Elevated, Borderline, or Normal	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	C5-OH C5:1 C6DC	Elevated Elevated Borderline or Normal	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	C5-OH C5:1 C6DC	Borderline Elevated Borderline or Normal	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.

Table 3: Organic Acid Disorders

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Organic Acid Disorders	Abnormal	C5:1 C5-OH C6DC	Elevated Normal Normal	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.
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C6DC C5:1	Elevated Elevated Normal	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	C5-OH C6DC C5:1	Borderline Elevated Normal	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	Analyte(s)	Analyte Result	Screening Result Note
Normal	Galactosemia	Normal			
Abnormal	Galactosemia	Abnormal	GALT	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	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	Analyte(s)	Analyte Result	Screening Result Note
Normal	Biotinidase Deficiency	Normal			
Abnormal	Biotinidase Deficiency	Abnormal	Biotinidase	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	Analyte(s)	Analyte Result	Screening Result Note
Normal	Hypothyroidism	Normal			
Normal	Hypothyroidism	Normal Normal	T4 TSH	Normal Normal	
Abnormal	Hypothyroidism	Abnormal	TSH	TSH Slightly Elevated	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	TSH	TSH Moderately Elevated	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	TSH	TSH Very Elevated	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	T4	T4 Low	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	T4/TSH	T4 Low,TSH Slightly Elevated	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	T4/TSH	T4 Low,TSH Moderately Elevated	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	T4/TSH	T4 Low,TSH Very Elevated	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	T4	T4 Elevated	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.

Table 6: Hypothyroidism

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hypothyroidism	Abnormal	T4/TSH	T4 Elevated,TSH Elevated	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	Analyte(s)	Analyte 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.

Table 7: CAH

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
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	Analyte(s)	Analyte Result	Screening Result Note
Normal	Hemoglobinopathies	Normal			
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,A	Probable Normal. Only Hemoglobin A detected. If result is due to transfusion, repeat in three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,C	Probable C Trait. Hemoglobin A,C detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,C,F	Probable C Trait. Hemoglobin A,C,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,D	Probable D Trait. Hemoglobin A,D detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,D,F	Probable D Trait. Hemoglobin A,D,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,E	Probable E Trait. Hemoglobin A,E detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,E,F	Probable E Trait. Hemoglobin A,E,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F	Probable Normal. Hemoglobin A,F detected. If result is due to transfusion, repeat in three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,C	Probable C Trait. Hemoglobin A,F,C detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,D	Probable D Trait. Hemoglobin A,F,D detected. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,G	Probable G Trait. Hemoglobin A,F,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,Other	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	Hemoglobin	A,F,Other,Barts	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	Hemoglobin	A,F,S	Probable S Trait. Hemoglobin A,F,S detected. Notify family of test results. DNA report to follow.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,S,Barts	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	Hemoglobin	A,G	Probable G Trait. Hemoglobin A,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,G,F	Probable G Trait. Hemoglobin A,G,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,Other	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	Hemoglobin	A,S	Probable S Trait. Hemoglobin A,S detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,S,F	Probable S Trait. Hemoglobin A,S,F detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,A	Probable Hemoglobin C/Beta Thalassemia Disease. Hemoglobin C,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,C	Probable CC Disease. Only Hemoglobin C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,F	Probable CC Disease. Hemoglobin C,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	E,E	Probable EE Disease. Only Hemoglobin E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	E,F	Probable EE Disease. Hemoglobin E,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F Only Detected	Possible Beta Thalassemia Major. Only Hemoglobin F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,Barts	Probable Alpha Thalassemia Trait. Hemoglobin F,A,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C	Probable C Trait. Hemoglobin F,A,C detected. Notify family of test results.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,Barts	Probable C Trait and Alpha Thalassemia Trait. Hemoglobin F,A,C,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,G	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	Hemoglobin	F,A,C,G,Barts	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	Hemoglobin	F,A,C,Other	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	Hemoglobin	F,A,D	Probable D Trait. Hemoglobin F,A,D detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,D,Barts	Probable D Trait and Alpha Thalassemia Trait. Hemoglobin F,A,D,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,E	Probable E Trait. Hemoglobin F,A,E detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,E,Barts	Probable E Trait and Alpha Thalassemia Trait. Hemoglobin F,A,E,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,elevated Barts	Probable H Disease. Hemoglobin F,A,elevated Barts detected. Refer to a pediatric hematologist within one month.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,G	Probable G Trait. Hemoglobin F,A,G detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,G,Barts	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	Hemoglobin	F,A,O-Arab	Probable O-Arab Trait. Hemoglobin F,A,O-Arab detected. Notify family of test results.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,Other	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	Hemoglobin	F,A,Other,Barts	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	Hemoglobin	F,A,S	Probable S Trait. Hemoglobin F,A,S detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S Other	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	Hemoglobin	F,A,S,Barts	Probable S Trait and Alpha Thalassemia Trait. Hemoglobin F,A,S,Barts detected. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S,G	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	Hemoglobin	F,A,S,G,Barts	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	Hemoglobin	F,C	Probable CC Disease. Hemoglobin F,C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,C (A Questionable)	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	Hemoglobin	F,C,A	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	Hemoglobin	F,C,Barts	Probable CC Disease and Alpha Thalassemia Trait. Hemoglobin F,C,Barts detected. Refer to pediatric hematologist within one month. DNA report to follow.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,C,E	Probable CE Disease. Hemoglobin F,C,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,D	Probable DD Disease. Hemoglobin F,D detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,E	Probable EE Disease. Hemoglobin F,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,E (A Questionable)	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	Hemoglobin	F,E,Barts	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	Hemoglobin	F,G	Probable GG Disease. Hemoglobin F,G detected. Refer to pediatric hematologist within one month.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,Other	Probable Unidentified Hb Variant. Hemoglobin F,Other detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S	Probable SS Disease. Hemoglobin F,S detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S (A Questionable)	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	Hemoglobin	F,S,A	Probable Sickle Beta Thalassemia Disease. Hemoglobin F,S,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S,Barts	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	Hemoglobin	F,S,C	Probable SC Disease. Hemoglobin F,S,C detected. Refer to pediatric hematologist within one month. DNA report to follow.

Table 8: Hemoglobinopathies

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S,C,Barts	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	Hemoglobin	F,S,D	Probable SD Disease. Hemoglobin F,S,D detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S,E	Probable SE Disease. Hemoglobin F,S,E detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,A	Probable Sickle Beta Thalassemia Disease. Hemoglobin S,A detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,C	Probable SC Disease. Hemoglobin S,C detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,C,F	Probable SC Disease. Hemoglobin S,C,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,F	Probable SS Disease. Hemoglobin S,F detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,S	Probable SS Disease. Only Hemoglobin S detected. Refer to pediatric hematologist within one month. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	Abnormal	free text
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	Analyte(s)	Analyte Result	Screening Result Note
Normal	Cystic Fibrosis	Normal			
Normal	Cystic Fibrosis	Normal	Immunoreactive Trypsinogen CFTR Mutation	Normal 0 Mutations Detected	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	Immunoreactive Trypsinogen	Elevated	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	Immunoreactive Trypsinogen	Elevated	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	Immunoreactive Trypsinogen	Elevated	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	Immunoreactive Trypsinogen	Elevated	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	Immunoreactive Trypsinogen CFTR Mutation	IRT Elevated CFTR Mutation Panel may be 0, 1, or 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 0 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Very Elevated 0 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	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	Inconclusive	Immunoreactive Trypsinogen	Elevated	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	Immunoreactive Trypsinogen	Very Elevated	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	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 1 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 2 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 3 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated Abnormal	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated Unsatisfactory	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	Analyte(s)	Analyte Result	Screening Result Note
Normal	SCID	Normal			
Abnormal	SCID	Abnormal	TREC	Low TREC	Possible Severe Combined Immunodeficiency or other T-cell lymphopenia. T-cell receptor excision circles (TREC) number Slightly Low. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
Abnormal	SCID	Abnormal	TREC	Very Low TREC	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	Analyte(s)	Analyte 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	Analyte(s)	Analyte 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.

SMA disorder added effective June 1, 2021

Table 12: SMA

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
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			free text
Normal	SMA	Revised Result			free text
Abnormal	SMA	Revised Result			free text
Unsatisfactory	SMA	Revised Result			free text

SMA disorder added effective June 1, 2021

Table 13: Unsatisfactory for All Tests

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte 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 14: Partially Unsatisfactory

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte 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.

Table 14: Partially Unsatisfactory

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
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.