

Table 1: Result Categories - Amino Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Amino Acid Disorders	Abnormal	Citrulline	Borderline	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Amino Acid Disorders	Abnormal	Citrulline	Elevated	Possible ASA or Citrullinemia. Recommend ammonia and plasma amino acids. Refer to a metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Leucine Valine	Each analyte may have a result of Elevated, Borderline, or Normal	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Amino Acid Disorders	Abnormal	Leucine Valine	Each analyte may have a result of Elevated, Borderline, or Normal	Possible Maple Syrup Urine Disease. Recommend blood glucose, plasma amino acids and urine organic acids. Refer to a metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Methionine	Borderline	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Amino Acid Disorders	Abnormal	Methionine	Elevated	Possible Homocystinuria. Recommend plasma amino acids, urine organic acids and plasma total homocysteine. Consult with a metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Phenylalanine Phe/Tyr	Each analyte may have a result of Elevated, Borderline, or Normal	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Amino Acid Disorders	Abnormal	Phenylalanine Phe/Tyr	Each analyte may have a result of Elevated, Borderline, or Normal	Possible PKU. Recommend plasma phenylalanine or plasma amino acids. Refer to a metabolic specialist.

Table 1: Result Categories - Amino Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine Succinylacetone	Borderline	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine	Elevated	Possible Tyrosinemia Type I. Recommend plasma amino acids, urine organic acids (including succinylacetone), liver function tests and refer to a metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine Succinylacetone	Each analyte may have a result of Elevated, Borderline, or Normal	Possible Tyrosinemia Type I. Recommend plasma amino acids, urine organic acids (including succinylacetone), liver function tests and refer to a metabolic specialist.
Abnormal	Amino Acid Disorders	Abnormal	Tyrosine Succinylacetone	Each analyte may have a result of Elevated, Borderline, or Normal	Possible Tyrosinemia Type II, III or Transient Tyrosinemia of the Neonate. Recommend plasma amino acids, urine organic acids, liver function tests and refer to a metabolic specialist.
Normal	Amino Acid Disorders	Revised Result			free text
Abnormal	Amino Acid Disorders	Revised Result			free text
Unsatisfactory	Amino Acid Disorders	Revised Result			free text
Abnormal	Amino Acid Disorders	Non-specific			Elevation(s) in a non-diagnostic pattern. Please repeat the newborn screen.
Abnormal	Amino Acid Disorders	TPN			Possible TPN. Please repeat the newborn screen when TPN is discontinued.

Table 2: Result Categories - Fatty Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Fatty Acid Disorders	Abnormal	C0 C3+C16	C0 may be Borderline or Low C3+C16 may be Normal or Low	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen in a week .
Abnormal	Fatty Acid Disorders	Abnormal	C0 C3+C16	C0 will be Low C3+C16 may be Normal or Low	Possible CUD. Recommend blood sugar, plasma (free and total) carnitine and maternal plasma (free and total) carnitine within a week . Refer to a metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	C0 C3+C16	C0 will be Low C3+C16 may be Normal or Low	Possible CUD. Place baby on carnitine immediately and repeat the newborn screen in one week.
Abnormal	Fatty Acid Disorders	Abnormal	C0/(C16 +C18) C0	Each analyte may have a result of Elevated, Borderline, or Normal	Possible CPT1. Recommend plasma carnitine and acylcarnitine profile. Refer to a metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	C14:1 C14 C14:1/C2 C16 C18:1	Each analyte may have a result of Elevated, Borderline, or Normal	Possible VLCAD. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	C16-OH C16:1-OH C18-OH C18:1-OH C18:2-OH	Each analyte may have a result of Elevated, Borderline, or Normal	Possible LCHAD or TFP. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Fatty Acid Disorders	Abnormal	C8 C6 C10:1 C10 C8/C2	Each analyte may have a result of Elevated, Borderline, or Normal	Possible MCAD. Recommend plasma acylcarnitine profile and urine organic acids (including acylglycines). Refer to a metabolic specialist. DNA report to follow.
Normal	Fatty Acid Disorders	Revised Result			free text

Table 2: Result Categories - Fatty Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Fatty Acid Disorders	Revised Result			free text
Unsatisfactory	Fatty Acid Disorders	Revised Result			free text
Abnormal	Fatty Acid Disorders	Non-specific			Elevation(s) in a non-diagnostic pattern. Please repeat the newborn screen.
Abnormal	Fatty Acid Disorders	TPN			Possible TPN. Please repeat the newborn screen when TPN is discontinued.

**Results in red added/modified February 2014

Table 3: Result Categories - Organic Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal		Normal			
Abnormal	Organic Acid Disorders	Abnormal	C3 C3/C2	Each analyte may have a result of Elevated, Borderline, or Normal	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Organic Acid Disorders	Abnormal	C3 C3/C2	Each analyte may have a result of Elevated, Borderline, or Normal	Possible Methylmalonic Acidemia or Propionic Acidemia. Recommend plasma amino acids, plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C3 C3/C2	Each analyte may have a result of Elevated, Borderline, or Normal	Possible Methylmalonic Acidemia. Recommend plasma amino acids, plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C5	Borderline	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Organic Acid Disorders	Abnormal	C5	Elevated	Possible Isovaleric Acidemia. Recommend plasma acylcarnitine profile, urine organic acids (including acylglycines). Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C5DC	Elevated	Possible GA1. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C5:1 C6DC	Each analyte may have a result of Elevated, Borderline, or Normal	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C5:1 C6DC	Each analyte may have a result of Elevated, Borderline, or Normal	Possible 3MCC, HMG, MCD, BKT. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.

Table 3: Result Categories - Organic Acid Disorders

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C5:1 C6DC	Each analyte may have a result of Elevated, Borderline, or Normal	Possible 3MCC, HMG, MCD. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C5:1 C6DC	Each analyte may have a result of Elevated, Borderline, or Normal	Possible BKT. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Abnormal	Organic Acid Disorders	Abnormal	C5-OH C5:1 C6DC	Each analyte may have a result of Elevated, Borderline, or Normal	Possible HMG. Recommend plasma acylcarnitine profile and urine organic acids. Refer to a metabolic specialist.
Normal	Organic Acid Disorders	Revised Result			free text
Abnormal	Organic Acid Disorders	Revised Result			free text
Unsatisfactory	Organic Acid Disorders	Revised Result			free text
Abnormal	Organic Acid Disorders	Non-specific			Elevation(s) in a non-diagnostic pattern. Please repeat the newborn screen.
Abnormal	Organic Acid Disorders	TPN			Possible TPN. Please repeat the newborn screen when TPN is discontinued.

Table 4: Result Categories - Galactosemia

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	Galactosemia	Normal			
Abnormal	Galactosemia	Borderline	GALT	Borderline	Possible Galactosemia. DNA report to follow.
Abnormal	Galactosemia	Abnormal	GALT	Abnormal	Possible Galactosemia. DNA report to follow.
Abnormal	Galactosemia	Revised Result			free text
Normal	Galactosemia	Revised Result			free text
Unsatisfactory	Galactosemia	Revised Result			free text

Table 5: Result Categories - Biotinidase Deficiency

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	Biotinidase Deficiency	Normal			
Abnormal	Biotinidase Deficiency	Abnormal	Biotinidase	Abnormal	Possible Biotinidase Deficiency. Recommend enzyme assay for biotinidase. Refer to metabolic specialist.
Abnormal	Biotinidase Deficiency	Borderline	Biotinidase	Borderline	Possible Partial Biotindase Deficiency. Please repeat the newborn screen.
Abnormal	Biotinidase Deficiency	Revised Result			free text
Normal	Biotinidase Deficiency	Revised Result			free text
Unsatisfactory	Biotinidase Deficiency	Revised Result			free text

Table 6 - Result Categories - Hypothyroidism

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Normal	Hypothyroidism	Normal			
Normal	Hypothyroidism	Normal Normal	T4 TSH	Normal Normal	
Abnormal	Hypothyroidism	Abnormal	T4	T4 Elevated	Possible TBG Excess. Recommend thyroid profile.
Abnormal	Hypothyroidism	Abnormal	T4	T4 Low	Possible Hypothyroidism. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Hypothyroidism	Abnormal	T4/TSH	T4 Elevated,TSH Elevated	Possible Hypothyroidism. Recommend thyroid profile and refer to an endocrinologist.
Abnormal	Hypothyroidism	Abnormal	T4/TSH	T4 Low,TSH Moderately Elevated	Possible Hypothyroidism. Recommend thyroid profile and refer to an endocrinologist.
Abnormal	Hypothyroidism	Abnormal	T4/TSH	T4 Low,TSH Slightly Elevated	Possible Hypothyroidism. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Hypothyroidism	Abnormal	T4/TSH	T4 Low,TSH Very Elevated	Possible Hypothyroidism. Recommend thyroid profile and refer to an endocrinologist.
Abnormal	Hypothyroidism	Abnormal	TSH	TSH Moderately Elevated	Possible Hypothyroidism. Recommend thyroid profile.
Abnormal	Hypothyroidism	Abnormal	TSH	TSH Slightly Elevated	Possible Hypothyroidism. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	Hypothyroidism	Abnormal	TSH	TSH Very Elevated	Possible Hypothyroidism. Recommend thyroid profile and refer to an endocrinologist.
Normal	Hypothyroidism	Revised Result			free text
Abnormal	Hypothyroidism	Revised Result			free text
Unsatisfactory	Hypothyroidism	Revised Result			free text

Table 7 - Result Categories - CAH

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	CAH	Normal			
Abnormal	CAH	Abnormal	17-Hydroxy-Progesterone	Abnormal	17-OH Progesterone Abnormal for birth weight less than 2500 grams. Possible CAH. Please repeat the newborn screen.
Abnormal	CAH	Abnormal	17-Hydroxy-Progesterone	Slightly Elevated	17-OH Progesterone Slightly Elevated for birth weight greater than or equal to 2500 grams. Possible CAH. If this is the second newborn screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Abnormal	CAH	Abnormal	17-Hydroxy-Progesterone	Moderately Elevated	17-OHP Moderately Elevated for birth weight greater than or equal to 2500 grams. Possible CAH. Recommend serum electrolytes and 17-OHP.
Abnormal	CAH	Abnormal	17-Hydroxy-Progesterone	Very Elevated	17-OH Progesterone Very Elevated for birth weight greater than or equal to 2500 grams. Possible CAH. Recommend serum electrolytes and 17-OHP and refer to an endocrinologist.
Abnormal	CAH	Revised Result			free text
Normal	CAH	Revised Result			free text
Unsatisfactory	CAH	Revised Result			free text

Table 8: Result Categories - Hemoglobinopathies

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Normal	Hemoglobinopathies	Normal			
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,A	Probable Normal. If result is due to transfusion, repeat in one to three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,C	Probable C Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,C,F	Probable C Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,D	Probable D Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,D,F	Probable D Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,E	Probable E Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,E,F	Probable E Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F	Probable Normal. If result is due to transfusion, repeat in one to three months post transfusion.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,C	Probable C Trait. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,D	Probable D Trait. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,G	Probable G Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,Other	Probable Unidentified Hb Variant Trait. Notify family of test results. Recommend consultation with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,Other,Barts	Probable Unidentified Hb Variant Trait and Alpha Thalassemia Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,S	Probable S Trait. Notify family of test results. DNA report to follow.

Table 8: Result Categories - Hemoglobinopathies

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,F,S,Barts	Probable S Trait and Alpha Thalassemia Trait. Notify family of test results. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,G	Probable G Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,G,F	Probable G Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,Other	Probable Unidentified Hb Variant Trait. Notify family of test results. Recommend consultation with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,S	Probable S Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	A,S,F	Probable S Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,A	Probable Hemoglobin C/Beta Thalassemia Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,C	Probable CC Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	C,F	Probable CC Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	E,E	Probable EE Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	E,F	Probable EE Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F Only Detected	Possible Beta Thalassemia Major. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,Barts	Probable Alpha Thalassemia Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C	Probable C Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,Barts	Probable C Trait and Alpha Thalassemia Trait. Notify family of test results.

Table 8: Result Categories - Hemoglobinopathies

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,Other	Probable C Trait and Unidentified Hb Variant Trait. Consult with pediatric hematologist. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,D	Probable D Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,D,Barts	Probable D Trait and Alpha Thalassemia Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,E	Probable E Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,E,Barts	Probable E Trait and Alpha Thalassemia Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,elevated Barts	Probable H Disease. Refer to a pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,G	Probable G Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,G,Barts	Probable G Trait and Alpha Thalassemia Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,O-Arab	Probable O-Arab Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,Other	Probable Unidentified Hb Variant Trait. Notify family of test results. Recommend consultation with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,Other,Barts	Probable Unidentified Hb Variant Trait and Alpha Thalassemia Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S	Probable S Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S Other	Probable S Trait and Unidentified Hb Variant. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S,Barts	Probable S Trait and Alpha Thalassemia Trait. Notify family of test results.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,C	Probable CC Disease. Refer to pediatric hematologist. DNA report to follow.

Table 8: Result Categories - Hemoglobinopathies

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

Table 8: Result Categories - Hemoglobinopathies

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,C,F	Probable SC Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,F	Probable SS Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	S,S	Probable SS Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S,G	Probable S Trait and G Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,S,G,Barts	Probable S Trait, G Trait, and Alpha Thalassemia Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,G	Probable C Trait and G Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,A,C,G,Barts	Probable C Trait, G Trait, and Alpha Thalassemia Trait. Notify family of test results. Consult with pediatric hematologist.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,E,Barts	Probable EE Disease and Alpha Thalassemia Trait. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,C,Barts	Probable CC Disease and Alpha Thalassemia Trait. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S,D	Probable SD Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,C,E	Probable CE Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	F,S,E	Probable SE Disease. Refer to pediatric hematologist. DNA report to follow.
Abnormal	Hemoglobinopathies	Abnormal	Hemoglobin	Abnormal	free text
Abnormal	Hemoglobinopathies	Revised Result			free text
Normal	Hemoglobinopathies	Revised Result			free text

Table 8: Result Categories - Hemoglobinopathies

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Unsatisfactory	Hemoglobinopathies	Revised Result			free text

Table 9: Result Categories - Cystic Fibrosis

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	Cystic Fibrosis	Normal			
Normal	Cystic Fibrosis	Normal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Normal 0 Mutations Detected	No further evaluation necessary unless clinically indicated. The immunoreactive trypsinogen (IRT) result was normal. Tests for a 40-mutation panel in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were negative, however, the presence of other mutations cannot be ruled out.
Abnormal	Cystic Fibrosis	Indeterminate	Immunoreactive Trypsinogen	Elevated	Many unaffected infants have an elevated immunoreactive trypsinogen (IRT) level on the first specimen. The second screening specimen (collected after 7 days of age) is required to determine if result is significant. Please repeat the newborn screen.
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen	Elevated	No further evaluation necessary unless clinically indicated. Elevated immunoreactive trypsinogen (IRT) level is consistent with the previous newborn screening specimen results for the same baby. Recommend sweat testing and possible genetic evaluation only if clinically indicated.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen	Elevated	Elevated immunoreactive trypsinogen (IRT) level is consistent with the previous newborn screening specimen results for the same baby. Cystic Fibrosis cannot be ruled out. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen	Elevated	Elevated immunoreactive trypsinogen (IRT) level is consistent with the previous newborn screening specimen results for the same baby. The previous specimen results are indicative of Cystic Fibrosis. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	IRT Elevated CFTR 40 Mutation Panel may be 0, 1, or 2 Mutations Detected	Revised Screening Result for Cystic Fibrosis. Additional testing using a CFTR 40 Mutation Panel has been performed. <i>(Note: Result notes vary depending on the results applied for CFTR 40 Mutation Panel. See list below.)</i> [The specimen was originally reported as Indeterminate for Cystic Fibrosis showing Immunoreactive Trypsinogen as Elevated. The original screening result note read “Many unaffected infants have an elevated immunoreactive trypsinogen (IRT) level on the first specimen. The second screening specimen (collected after 7 days of age) is required to determine if result is significant. Please repeat the newborn screen.”]

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated 0 Mutations Detected	No further evaluation necessary unless clinically indicated. Although there is a minimal risk for Cystic Fibrosis (CF) in the absence of detected mutations, an elevated immunoreactive trypsinogen (IRT) result may be indicative of CF due to a mutation not included in the 40-mutation panel. Recommend sweat testing and possible genetic evaluation only if clinically indicated.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Very Elevated 0 Mutations Detected	Although there is a minimal risk for Cystic Fibrosis (CF) in the absence of detected mutations, a very elevated immunoreactive trypsinogen (IRT) result may be indicative of CF due to mutations not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 1078delT, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 1717-1G>A, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 1898+1G>A, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 2183AA>G, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 2184delA, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 2789+5G>A, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3120+1G>A, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3659delC, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3849+10kbC>T, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3849+4A>G, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3876delA, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 3905insT, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 394delTT, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 621+1G>T, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, 711+G>T, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, A455E, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, D1152H, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, deltaF508, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, deltaI507, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, E60X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, G542X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, G551D, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, G85E, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, N1303K, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, Q493X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R1162X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R117H (IVS8-5T/7T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R117H (IVS8-7T/7T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R117H (IVS8-7T/9T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R117H (IVS8-9T/9T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R334W, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R347H, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R347P, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R553X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, R560T, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, S549N, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, S549R A>C, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, S549R T>G, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, V520F, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, W1282X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, Y1092X C>A, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, Y1092X C>G, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutation Detected	One mutation, Y122X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis can not be ruled out due to a possibility of a second mutation which is not included in the 40-mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 1 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	A homozygous Cystic Fibrosis-causing mutation, deltaF508, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	free text

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and 621+1G>T, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and G542X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and G551D, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and N1303K, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and R117H (IVS8-5T/7T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and R117H (IVS8-7T/7T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and R117H (IVS8-7T/9T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and R117H (IVS8-9T/9T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.

Table 9: Result Categories - Cystic Fibrosis

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and R553X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, deltaF508 and W1282X, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated or Normal 3 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated Abnormal	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	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: Result Categories - SCID

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	SCID	Normal			
Abnormal	SCID	Borderline	TREC	Low TREC	Borderline low number of T-cell receptor excision circles (TREC). Please repeat the newborn screen.
Abnormal	SCID	Abnormal	TREC	Very Low TREC	Very low number of T-cell receptor excision circles (TREC). Please follow recommendations received from the DSHS newborn screening Clinical Care Coordination team.
Abnormal	SCID	Revised Result			free text
Normal	SCID	Revised Result			free text
Unsatisfactory	SCID	Revised Result			free text

Table 11: Global Unsatisfactory Result Categories

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Unsatisfactory	ALL	Unsatisfactory			Blood did not completely fill specimen circles.
Unsatisfactory	ALL	Unsatisfactory			Blood did not soak through paper due to incomplete saturation.
Unsatisfactory	ALL	Unsatisfactory			Filter paper is scratched from the possible use of capillary tubes.
Unsatisfactory	ALL	Unsatisfactory			Specimen appears contaminated or discolored.
Unsatisfactory	ALL	Unsatisfactory			Blood was caked, clotted, or layered onto the filter paper.
Unsatisfactory	ALL	Unsatisfactory			Patient information incomplete or invalid (e.g. date of collection missing).
Unsatisfactory	ALL	Unsatisfactory			Serial number on patient information form does not match number on specimen filter paper.
Unsatisfactory	ALL	Unsatisfactory			Specimen too old upon receipt.
Unsatisfactory	ALL	Unsatisfactory			Please Resubmit. Unable to analyze specimen due to laboratory accident.
Unsatisfactory	ALL	Unsatisfactory			No blood samples received with request form.
Unsatisfactory	ALL	Unsatisfactory			Specimen submitted on improper collection form.
Unsatisfactory	ALL	Unsatisfactory			Specimen submitted on expired collection form.
Unsatisfactory	ALL	Unsatisfactory			Serum separation due to improper drying or specimen collection.
Unsatisfactory	ALL	Unsatisfactory			Specimen damaged during transport to laboratory.
Unsatisfactory	ALL	Unsatisfactory			Information on demographic form does not match electronically submitted information.
Unsatisfactory	ALL	Unsatisfactory			Specimen Received in Hermetically Sealed Container.

Unsatisfactory for All Tests

Table 12: Laboratory Unsatisfactory Result Categories

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Blood did not Completely Fill Specimen Circles.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Blood did not Soak Through Paper - Incomplete saturation.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Filter paper is scratched from the possible use of capillary tubes.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Appears Contaminated or Discolored.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Blood was Caked, Clotted, or Layered onto the Filter Paper.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Assay Interference due to EDTA/Citrate anticoagulant contamination.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Assay interference.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Incomplete elution of blood from filter paper.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Patient information incomplete or invalid (e.g. date of collection missing).
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Serial Number on Patient Information form does not match number on specimen filter paper.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Too Old Upon Receipt.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Unable to Analyze Specimen due to Laboratory Accident.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: No Blood Samples Received with Request Form.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Submitted on Improper Collection Form.

Partially Unsatisfactory

Table 12: Laboratory Unsatisfactory Result Categories

Overall Specimen Result	Disorder	Screening Result	Analyte	Analyte Result	Screening Result Note
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Submitted on Expired Collection Form.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Results Inconsistent.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Serum Separation due to Improper Drying or Specimen Collection.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Damaged During Transport to laboratory.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Information on demographic form does not match electronically submitted information.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen Received in Hermetically Sealed Container.
Unsatisfactory	ALL	Unsatisfactory			Unsatisfactory - Please Resubmit: 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.
Unsatisfactory	Cystic Fibrosis	Unsatisfactory			Unsatisfactory - Please Resubmit: Specimen too old to test for immunoreactive trypsinogen (IRT).
Unsatisfactory	SCID	Unsatisfactory			Unsatisfactory - Please resubmit: Specimen inadequate for accurate detection of TREC (T-cell receptor excision circles).

Partially Unsatisfactory