

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.

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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.

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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