


Urine Amino Acid Analysis:

Unique advantages provided by the IUBGL approach

 GENETIC TESTING LABORATORIES							
Biochemical Genetics Laboratory				02/21/2020			
Ordered By Physician Name: Wilson, D Reason for Referral: kidney stones				Patient Name: Arginine, Ursella Accession #: R3032 Specimen #: X3032 Specimen: Urine Birthdate: 02/12/2009 Age: 11 Gender: Female MRN #: 8675309 Collected: 02/20/2020 Ethnicity: Caucasian Received: 02/21/2020			
Urine Amino Acid Analysis - Quantitative							
RESULTS							
ANALYTE	REFERENCE RANGE*	RESULT*	FLAG	ANALYTE	REFERENCE RANGE*	RESULT*	FLAG
3-Methyl-histidine	0-511	2		Glycine	0-4280	12068	H
Alanine	0-661	1971	H	Guanidinoacetate	30-1200	7.5	L
Alloisoleucine	0-8	6		Histidine	0-3043	1338	
Alpha-aminoadipate	0-214	3		Homocitrulline	0-78	0	
Alpha-amino-n-butyrate	0-112	20		Homocystine	0-7	0.0	
Anserine	0-369	1		Hydroxyproline	0-93	5570	H
Arginine	0-112	104		Isoleucine	0-59	50	
Argininosuccinate	0-121	0		Leucine	0-141	132	
Asparagine	0-652	266		Lysine	0-546	489	
Aspartate	0-194	51		Methionine	0-58	28	
Beta-alanine	0-313	12		Ornithine	0-58	95	H
Beta-Aminoisobutyrate	0-388	0		Phenylalanine	0-264	70	
Citrulline	0-64	38		Proline	0-128	1948	H
Creatine/Creatinine Ratio	0-0.7	0.50		Sarcosine	0-27	8	
Creatinine	15.8-320.0	20.3		Serine	0-1320	1852	H
Cystathionine	0-81	5		Sulfocysteine	0-87	77	
Cystine	0-197	54		Taurine	0-2158	5512	H
Delta-aminolevulinate	0-42	1		Threonine	0-410	681	H
Gamma-amino-n-butyrate	0-14	14	H	Tryptophan	0-191	64	
Glutamate	0-167	64		Tyrosine	0-230	309	H
Glutamine	0-1097	1111	H	Valine	0-99	106	H

Expanded Scope of Detections- *The IUBGL urine amino acid panel contains multiple clinically relevant analytes not traditionally included in urine amino acid panels.*

- Creatine/Creatinine Ratio - ↑ X-linked creatine transporter deficiency (OMIM# 300352)
- Delta-aminolevulinate - ↑ Tyrosinemia Type 1 (OMIM# 276700) and n-↑ Porphyrrias (e.g., OMIM# 176000, 176200, 121300)
- Guanidinoacetate - ↑ GAMT deficiency (OMIM# 612736) and ↓AGAT deficiency (OMIM# 612718)
- Sulfocysteine - ↑ Sulfite oxidase deficiency (OMIM# 272300) and ↑ Molybdenum cofactor deficiencies A-C (OMIM# 252150, 252160, 615501)

Improved Accuracy- *We have built a stable isotope dilution assay for creatinine quantification into our LC-MS/MS analysis of urine amino acids. This optimizes accuracy and precision not only for creatinine but for all amino acids, which are normalized to the specimen's creatinine concentration.*