

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB 9/2/2005
Sex: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Autism and Intellectual Disability Comprehensive Panel

ARUP test code 2014314

Creatinine, Urine	139 mg/dL	
Alpha-amino butyric acid, Plasma	20 umol/L	(Ref Interval: <=40)
Alanine, Plasma	641 umol/L	H (Ref Interval: 160-530)
Allo-isoleucine, Plasma	<2 umol/L	(Ref Interval: <=5)
Alpha-aminoadipic acid, Plasma	<2 umol/L	(Ref Interval: <=4)
Anserine, Plasma	<5 umol/L	(Ref Interval: <=5)
Arginine, Plasma	98 umol/L	(Ref Interval: 35-125)
Argininosuccinic Acid, Plasma	<2 umol/L	(Ref Interval: <=2)
Asparagine, Plasma	58 umol/L	(Ref Interval: 20-80)
Aspartic Acid, Plasma	<5 umol/L	(Ref Interval: <=15)
Beta-amino isobutyric acid, Plasma	<5 umol/L	(Ref Interval: <=10)
Beta-alanine, Plasma	<25 umol/L	(Ref Interval: <=25)
Citrulline, Plasma	19 umol/L	(Ref Interval: 10-45)
Cystathionine, Plasma	<5 umol/L	(Ref Interval: <=5)
Cystine, Plasma	41 umol/L	(Ref Interval: 10-65)
Ethanolamine, Plasma	7 umol/L	(Ref Interval: <=15)
Gamma-amino butyric acid, Plasma	<5 umol/L	(Ref Interval: <=5)
Glutamic Acid, Plasma	27 umol/L	(Ref Interval: 15-130)
Glutamine, Plasma	668 umol/L	(Ref Interval: 380-680)
Glycine, Plasma	328 umol/L	(Ref Interval: 140-420)

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 25-221-400602
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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Histidine, Plasma	84 umol/L	(Ref Interval: 50-130)
Homocitrulline, Plasma	<5 umol/L	(Ref Interval: <=5)
Homocystine, Plasma	<2 umol/L	(Ref Interval: <=2)
Hydroxylysine, Plasma	<5 umol/L	(Ref Interval: <=5)
Hydroxyproline, Plasma	32 umol/L	(Ref Interval: 5-40)
Isoleucine, Plasma	83 umol/L	(Ref Interval: 30-120)
Leucine, Plasma	136 umol/L	(Ref Interval: 60-180)
Lysine, Plasma	176 umol/L	(Ref Interval: 85-230)
Methionine, Plasma	33 umol/L	(Ref Interval: 15-40)
Ornithine, Plasma	48 umol/L	(Ref Interval: 25-110)
Phenylalanine, Plasma	52 umol/L	(Ref Interval: 30-82)
Proline, Plasma	257 umol/L	(Ref Interval: 90-350)
Sarcosine, Plasma	<5 umol/L	(Ref Interval: <=5)
Serine, Plasma	113 umol/L	(Ref Interval: 60-170)
Taurine, Plasma	38 umol/L	(Ref Interval: 30-130)
Threonine, Plasma	159 umol/L	(Ref Interval: 60-190)
Tryptophan, Plasma	68 umol/L	(Ref Interval: 25-80)
Tyrosine, Plasma	74 umol/L	(Ref Interval: 35-110)
Valine, Plasma	202 umol/L	(Ref Interval: 120-320)
C2, Acetyl	4.57 umol/L	(Ref Interval: 2.93-15.06)
C3, Propionyl	0.25 umol/L	(Ref Interval: <=0.82)
C4, Iso-/Butyryl	0.09 umol/L	(Ref Interval: <=0.42)
C5, Isovaleryl/2Mebutyryl	0.07 umol/L	(Ref Interval: <=0.24)
C5-DC, Glutaryl	0.08 umol/L	(Ref Interval: <=0.23)
C5-OH, 3-OH Isovaleryl	0.03 umol/L	(Ref Interval: <=0.07)
C6, Hexanoyl	0.03 umol/L	(Ref Interval: <=0.12)
C8, Octanoyl	0.05 umol/L	(Ref Interval: <=0.22)
C8:1, Octenoyl	0.15 umol/L	(Ref Interval: <=0.60)

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C10, Decanoyl	0.06 umol/L	(Ref Interval: <=0.33)
C10:1, Decenoyl	0.08 umol/L	(Ref Interval: <=0.27)
C12, Dodecanoyl	0.02 umol/L	(Ref Interval: <=0.13)
C12:1, Dodecenoyl	0.02 umol/L	(Ref Interval: <=0.13)
C12-OH, 3-OH-Dodecanoyl	<0.01 umol/L	(Ref Interval: <=0.02)
C14, Tetradecanoyl	0.01 umol/L	(Ref Interval: <=0.06)
C14:1, Tetradecenoyl	0.02 umol/L	(Ref Interval: <=0.15)
C14:2, Tetradecadienoyl	0.01 umol/L	(Ref Interval: <=0.08)
C14-OH, 3-OH-Tetradecanoyl	<0.01 umol/L	(Ref Interval: <=0.01)
C14:1-OH, 3-OH-Tetradecenoyl	<0.01 umol/L	(Ref Interval: <=0.04)
C16, Palmitoyl	0.05 umol/L	(Ref Interval: <=0.12)
C16:1, Palmitoleyl	0.01 umol/L	(Ref Interval: <=0.04)
C16-OH, 3-OH-Palmitoyl	0.01 umol/L	(Ref Interval: <=0.02)
C16:1-OH, 3-OH-Palmitoleyl	0.01 umol/L	(Ref Interval: <=0.02)
C18, Stearoyl	0.03 umol/L	(Ref Interval: <=0.06)
C18:1, Oleyl	0.04 umol/L	(Ref Interval: <=0.18)
C18:2, Linoleyl	0.04 umol/L	(Ref Interval: <=0.10)
C18-OH, 3-OH-Stearoyl	<0.01 umol/L	(Ref Interval: <=0.02)
C18:1-OH, 3-OH-Oleyl	<0.01 umol/L	(Ref Interval: <=0.02)
C18:2-OH, 3-OH-Linoleyl	<0.01 umol/L	(Ref Interval: <=0.02)
Mucopolysaccharides mg/mmol CRT	4.9 REFERENCE INTERVAL: Mucopolysaccharides mg/mmol CRT Access complete set of age- and/or gender-specific reference intervals for this test in the ARUP Laboratory Test Directory (aruplab.com). This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.	(Ref Interval: 0.0-7.1)
Lactic Acid, Urine	17	(Ref Interval: 0-50)
Pyruvic Acid, Urine	9	(Ref Interval: 0-15)

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Succinic Acid, Urine	5	(Ref Interval: 0-20)
Fumaric Acid, Urine	Not Detected	(Ref Interval: 0-4)
2-Ketoglutaric Acid, Urine	9	(Ref Interval: 0-75)
Methylmalonic Acid, Urine	Not Detected	(Ref Interval: 0-5)
3-OH-Butyric Acid, Urine	Not Detected	(Ref Interval: 0-4)
Acetoacetic Acid, Urine	Not Detected	(Ref Interval: 0-4)
2-Keto-3-methylvaleric Acid, Urine	Not Detected	(Ref Interval: 0-10)
2-Ketoisocaproic Acid, Urine	Not Detected	(Ref Interval: 0-4)
2-Ketoisovaleric Acid, Urine	Not Detected	(Ref Interval: 0-4)
Ethylmalonic Acid, Urine	1	(Ref Interval: 0-4)
Adipic Acid, Urine	1	(Ref Interval: 0-35)
Suberic Acid, Urine	1	(Ref Interval: 0-3)
Sebacic Acid, Urine	Not Detected	(Ref Interval: 0-3)
4-OH-phenylacetic Acid, Urine	5	(Ref Interval: 0-25)
4-OH-phenyllactic Acid, Urine	Not Detected	(Ref Interval: 0-4)
4-OH-phenylpyruvic Acid, Urine	Not Detected	(Ref Interval: 0-2)
Succinylacetone, Urine	Not Detected	(Ref Interval: 0-0)
Creatine, Urine	37 mmol/mol CRT	(Ref Interval: 10-370)
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Guanidinoacetic acid, Urine	57 mmol/mol CRT	(Ref Interval: 7-130)
Creatinine, Urine	10050.7 umol/L	
Creatine, Serum/Plasma	37.6 umol/L	(Ref Interval: 9.0-90.0)
Guanidinoacetic acid, Serum/Plasma	1.96 umol/L	(Ref Interval: 1.10-3.80)
FRAG X Specimen	whole Blood	
Fragile X Allele 1	30 CGG repeats	
Fragile X Allele 2	29 CGG repeats	
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Fragile X Methylation Pattern

Not Applicable

Fragile X Interpretation

See Note

Interpretation: This individual has two FMR1 alleles with CGG sizes in the normal range. This patient is predicted to be neither affected with nor a carrier of an FMR1-associated disorder, including fragile X syndrome (FXS), fragile-X associated primary ovarian insufficiency (FXPOI), fragile X-associated tremor/ataxia syndrome (FXTAS), and fragile X-associated neuropsychiatric disorders (FXAND). This test does not detect rare FMR1 variants causing less than 1% of FXS.

Recommendation: Genetic counseling is recommended.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

BACKGROUND INFORMATION: Fragile X (FMR1) with Reflex to Methylation Analysis

CHARACTERISTICS OF FRAGILE X SYNDROME (FXS): Affected males have moderate intellectual disability, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders and connective tissue anomalies. Females are usually less severely affected than males. FXS is caused by FMR1 full mutations.

CHARACTERISTICS OF FRAGILE X-ASSOCIATED TREMOR ATAXIA SYNDROME (FXTAS): Onset of progressive ataxia and intention tremor typically after the fifth decade of life. Cognitive impairment and behavioral features may also develop. FXTAS is caused by FMR1 premutations.

CHARACTERISTICS OF FRAGILE X-ASSOCIATED PRIMARY OVARIAN INSUFFICIENCY (FXPOI): Primary ovarian insufficiency or hypergonadotropic hypogonadism before 40 years of age. FXPOI is associated with FMR1 premutations.

CHARACTERISTICS OF FRAGILE X-ASSOCIATED NEUROPSYCHIATRIC DISORDERS (FXAND): Symptoms may include anxiety, depression, adult ADHD, or addictive behavior. FXAND is associated with FMR1 premutations.

PREVALENCE OF FXS: Approximately 1 in 4,000-7,000 males and 1 in 8,000-11,000 females.

PREVALENCE OF PREMUTATION ALLELE: Approximately 1 in 150-300 females and 1 in 300-800 males.

INHERITANCE: X-linked.

PENETRANCE OF FXS: Complete in males; 50 percent in females.

PENETRANCE OF FXTAS: For individuals greater than 50 years of age, approximately 40 percent in males and 16-20 percent in females.

PENETRANCE OF FXPOI: Approximately 20 percent in females.

CAUSE: Expansion of the FMR1 gene CGG triplet repeat.

Full mutation: Typically greater than 200 CGG repeats (methylated). Premutation: 55 to approximately 200 CGG repeats (unmethylated). Intermediate: 45-54 CGG repeats (unmethylated). Normal: 5-44 CGG repeats (unmethylated).

CLINICAL SENSITIVITY: 99 percent.

METHODOLOGY: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis. Methylation-specific PCR analysis is performed for CGG repeat lengths of greater than 100 to distinguish between premutation and full mutation alleles.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent; estimated precision of sizing for intermediate and premutation alleles is within 2-3 CGG repeats.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Rare FMR1 variants unrelated to trinucleotide expansion will not be detected. A specific CGG repeat size

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estimate is not provided for full mutation alleles. AGG trinucleotide interruptions within the FMR1 CGG repeat tract are not assessed.

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Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Cytogenomic SNP Microarray

See Note (Ref Interval: Normal)

Test Performed: Cytogenomic SNP Microarray (CMA SNP)
Specimen Type: Peripheral blood
Indication for Testing: Autism, intellectual disability

RESULT SUMMARY

Copy Number Variant Detected- Clinical Significance Uncertain (Female)

16p11.2 Distal Duplication (BP2 to BP3 Region)

Classification: Variant of Uncertain Significance (VUS)
Copy number change: 16p11.2 gain
Size: 219 kb

RESULT DESCRIPTION

This analysis showed an interstitial duplication (3 copies present) involving chromosome 16, within 16p11.2. This region contains at least 12 genes (listed below), including the gene SH2B1.

This is a duplication of the 16p11.2 distal (SH2B1) region, involving recurrent breakpoints (BPs) within flanking low-copy repeat regions, BP2 and BP3. The reported size of this duplication may vary across studies due to variability in breakpoints within flanking repeat regions. Please note this region is distinct from the recurrent 16p11.2 proximal (TBX6) region, which involves breakpoints BP4-BP5.

INTERPRETATION

The clinical significance of this duplication is uncertain. Evidence for a clinical association with duplication of the 16p11.2 (SH2B1) distal region is limited at this time. Although duplications of this region have been reported in association with neurodevelopmental phenotypes, the clinical features are variable, nonspecific, and sometimes inconsistent. Features reported include microcephaly, reduced BMI, scoliosis, autism, ADHD, and differences in brain morphology that are measurable but of uncertain clinical significance. The 16p11.2 distal duplication has also been observed in both unaffected relatives of probands and individuals in the general population from studies of natural genomic variation. Case-control studies have found this duplication to be enriched in patients versus control populations. It is possible this duplication is unrelated to the indication for testing.

Surveillance of the literature for new information concerning this duplication is recommended before definitive conclusions are made regarding its pathogenicity, as additional information should become available over time.

Copy number variants (CNVs) of uncertain significance are often observed in the context of nonspecific and/or variable clinical

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phenotypes. Studies of recurrent CNVs have shown both phenotypic variability as well as incomplete penetrance, in which a variant is inherited from an unaffected parent. Therefore, parental testing for the CNV may be uninformative, as its presence or absence in a clinically unaffected parent or sibling may not rule out or confirm causality.

Recommendations:

- 1) Genetic counseling
- 2) Surveillance of the literature for new information concerning this duplication

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

References:

- 1) Loviglio et al. Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. *Mol Psychiatry*. 2017 Jun;22(6):836-849. PMID: 27240531.
- 2) Bachmann-Gagescu et al. Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. *Genet Med*. 2010 Oct;12(10):641-7. PMID: 20808231.
- 3) Vos et al. Evaluation of 100 Dutch cases with 16p11.2 deletion and duplication syndromes; from clinical manifestations towards personalized treatment options. *Eur J Hum Genet*. 2024 Nov;32(11):1387-1401. PMID: 38605127.
- 4) Sadler et al. Distal chromosome 16p11.2 duplications containing SH2B1 in patients with scoliosis. *J Med Genet*. 2019 Jul;56(7):427-433. PMID: 30803986.
- 5) Sonderby et al. Dose response of the 16p11.2 distal copy number variant on intracranial volume and basal ganglia. *Mol Psychiatry*. 2020 Mar;25(3):584-602. Erratum in: *Mol Psychiatry*. 2019 Jan 31. PMID: 30283035.

Cytogenomic Nomenclature (ISCN):

arr[GRCh37] 16p11.2(28824491_29043972)x3

Genes in the 16p11.2 duplicated region:

ATXN2L, TUFM, MIR4721, SH2B1, ATP2A1, ATP2A1-AS1, RABEP2, CD19, NFATC2IP, MIR4517, SPNS1, LAT

Technical Information

- This assay was performed using the CytoScan(TM) HD Suite (Thermo Fisher Scientific) according to validated protocols within the Genomic Microarray Laboratory at ARUP Laboratories
- This assay is designed to detect alterations to DNA copy number state (gains and losses) as well as copy-neutral alterations (regions of homozygosity; ROH) that indicate an absence- or loss-of-heterozygosity (AOH or LOH)
- AOH may be present due to parental relatedness (consanguinity) or uniparental disomy (UPD)
- LOH may be present due to acquired UPD (segmental or whole chromosome)
- The detection sensitivity (resolution) for any particular genomic region may vary dependent upon the number of probes (markers), probe spacing, and thresholds for copy number and ROH determination
- The CytoScan HD array contains 2.67 million markers across the genome with average probe spacing of 1.15 kb, including 750,000 SNP probes and 1.9 million non-polymorphic probes
- In general, the genome-wide resolution is approximately 25-50 kb for copy number changes and approximately 3 Mb for ROH (See reporting criteria)
- The limit of detection for mosaicism varies dependent upon the size and type of genomic imbalance. In general, genotype mixture due to mosaicism (distinct cell lines from the same individual) or chimerism (cell lines from different individuals) will be

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detected when present at greater than 20-30 percent in the sample
- Genomic coordinates correspond to the Genome Reference Consortium human genome build 37/human genome issue 19 (GRCh37/hg19)

Variant Classification and Reporting Criteria

- Copy number variant (CNV) analysis is performed in accordance with recommendations by the American College of Medical Genetics and Genomics (ACMG), using standard 5-tier CNV classification terminology: pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign
- CNVs classified as pathogenic, likely pathogenic, or variant of uncertain significance are generally reported, based on information available at the time of review
- Known or expected pathogenic CNVs affecting genes with known clinical significance but which are unrelated to the indication for testing will generally be reported
- Variants that do not fall within the standard 5-tier CNV classification categories may be reported with descriptive language specific to that variant
- In general, recessive disease risk and recurrent CNVs with established reduced penetrance will be reported
- For a list of databases used in CNV classification, please refer to ARUP Constitutional CNV Assertion Criteria, which can be found on ARUP's Genetics website at www.aruplab.com/genetics
- CNVs classified as likely benign or benign that are devoid of relevant gene content or reported as common findings in the general population, are generally not reported
- CNV reporting (size) criteria: losses greater than 50 kb and gains greater than 400 kb are generally reported, dependent on genomic content
- ROH are generally reported when a single terminal ROH is greater than 3 Mb and a single interstitial ROH is greater than 10-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder) or when total autosomal homozygosity is greater than 3 percent (only autosomal ROH greater than 3 Mb are considered for this estimate)

Limitations

This analysis cannot provide structural (positional) information associated with genomic imbalance. Therefore, additional cytogenetic testing by chromosome analysis or fluorescence in situ hybridization (FISH) may be recommended.

Certain genomic alterations may not or cannot be detected by this technology. These alterations may include, but are not limited to:

- CNVs below the limit of resolution of this platform
- Sequence-level variants (mutations) including point mutations and indels
- Low-level mosaicism (generally, less than 20-30 percent)
- Balanced chromosomal rearrangements (translocations, inversions and insertions)
- Genomic imbalance in repetitive DNA regions (centromeres, telomeres, segmental duplications, and acrocentric chromosome short arms)

Data Sharing

In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, de-identified (cannot be traced back to the patient) genetic test results and health information to public databases. The confidentiality of each sample is maintained. If you prefer that your test result not be shared, call ARUP Laboratories at (800) 242-2787 ext. 3301. Your de-identified information will not be disclosed to public databases after your request is received, but a separate request is required for each genetic test. Additionally, patients have the opportunity to participate in patient registries and research. To learn more, visit ARUP's Genetics website at

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www.aruplab.com/genetics.

This result has been reviewed and approved by M. Katharine Rudd, PhD, FACMG

A portion of this analysis was performed at the following location(s):
ARUP Laboratories Site CG-NC#2

INTERPRETIVE INFORMATION: CYTOGENOMIC SNP MICROARRAY

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Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

EER Cytogenomic SNP Microarray	See Note
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Autism/Intellectual Interp	See Note
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Mildly elevated plasma alanine. Would repeat plasma amino acids and exclude lactic acidosis. No other metabolic abnormalities identifiable by this panel were detected. Genetic evaluation is recommended to assess the need for additional testing to exclude other rare metabolic disorders associated with autism and/or intellectual disability.

Results reviewed and interpreted by Marzia Pasquali, PhD, FACMG

INTERPRETIVE INFORMATION: Autism and Intellectual Disability Comprehensive Panel
MPS Screen, Urine: Mucopolysaccharides (Glycosaminoglycans) include: Keratan Sulfate, Heparan Sulfate, Dermatan Sulfate, and Chondroitin Sulfates 4 and 6. The excretion of Heparan Sulfate is variable. A normal mucopolysaccharides screen does not exclude Sanfilippo Syndrome (Mucopolysaccharidosis Type III).

Organic Acids, Urine: Results are reported in mmol/mol creatinine.

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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Creatinine, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-amino butyric acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alanine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Allo-isoleucine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

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Alpha-aminoadipic acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Anserine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Arginine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Argininosuccinic Acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Asparagine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Aspartic Acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta-amino isobutyric acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta-alanine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Citrulline, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystathionine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ethanolamine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Gamma-amino butyric acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glutamic Acid, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glutamine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Histidine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Homocitrulline, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Homocystine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hydroxylysine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hydroxyproline, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Isoleucine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Leucine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lysine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Methionine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ornithine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Phenylalanine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Proline, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Sarcosine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Serine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Taurine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Threonine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Tryptophan, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Tyrosine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Valine, Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C2, Acetyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C3, Propionyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C4, Iso-/Butyryl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C5, Isovaleryl/2Mebutyryl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C5-DC, Glutaryl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C5-OH, 3-OH Isovaleryl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 25-221-400602
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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C6, Hexanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C8, Octanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C8:1, Octenoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C10, Decanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C10:1, Decenoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C12, Dodecanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C12:1, Dodecenoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C12-OH, 3-OH-Dodecanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C14, Tetradecanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C14:1, Tetradecenoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C14:2, Tetradecadienoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C14-OH, 3-OH-Tetradecanoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C14:1-OH, 3-OH-Tetradecenoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C16, Palmitoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C16:1, Palmitoleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C16-OH, 3-OH-Palmitoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C16:1-OH, 3-OH-Palmitoleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18, Stearoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18:1, Oleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18:2, Linoleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18-OH, 3-OH-Stearoyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18:1-OH, 3-OH-Oleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C18:2-OH, 3-OH-Linoleyl	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Mucopolysaccharides mg/mmol CRT	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lactic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Pyruvic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Succinic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fumaric Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
2-Ketoglutaric Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Methylmalonic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
3-OH-Butyric Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Acetoacetic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
2-Keto-3-methylvaleric Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
2-Ketoisocaproic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
2-Ketisovaleric Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ethylmalonic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Adipic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Suberic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Sebacic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
4-OH-phenylacetic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
4-OH-phenyllactic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

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Visit Number (FIN): 01234567890ABCD
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4-OH-phenylpyruvic Acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Succinylacetone, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Creatine, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Guanidinoacetic acid, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Creatinine, Urine	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Creatine, Serum/Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Guanidinoacetic acid, Serum/Plasma	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
FRAG X Specimen	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic SNP Microarray	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Cytogenomic SNP Microarray	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Autism/Intellectual Interp	25-221-400602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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Patient: Patient, Example
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