ARUP Laboratories
500 Chipeta Way – Salt Lake City, UT 84108
(800)522-2787 - www.aruplab.com
Julio C. Delgado, M.D. M.S., Director of Laboratories

*****Example Report*****

Patient Age/Gender: Unknown Unknown
Printed: 18-Dec-19 13:01:44

Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/ Verified
Creatinine, Urine	24	mg/dL		19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Alpha-amino butyric acid, Plasma	24	umol/L	[<=40]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Alanine, Plasma	225	umol/L	[160-530]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Allo-isoleucine, Plasma	<2	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Alpha-aminoadipic acid, Plasma	3	umol/L	[<=4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Anserine, Plasma	<5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Arginine, Plasma	103	umol/L	[35-125]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Argininosuccinic Acid, Plasma	2	umol/L	[<=2]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Asparagine, Plasma	65	umol/L	[20-80]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Aspartic Acid, Plasma	12	umol/L	[<=15]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Beta-amino isobutyric acid, Plasma	8	umol/L	[<=10]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Beta-alanine, Plasma	<25	umol/L	[<=25]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Citrulline, Plasma	16	umol/L	[10-45]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Cystathionine, Plasma	5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Cystine, Plasma	15	umol/L	[10-65]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Ethanolamine, Plasma	7	umol/L	[<=15]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Gamma-amino butyric acid, Plasma	5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Glutamic Acid, Plasma	35	umol/L	[15-130]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Glutamine, Plasma	544	umol/L	[380-680]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Glycine, Plasma	354	umol/L	[140-420]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Histidine, Plasma	97	umol/L	[50-130]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Homocitrulline, Plasma	<5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Homocystine, Plasma	<2	umol/L	[<=2]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Hydroxylysine, Plasma	<5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Hydroxyproline, Plasma	24	umol/L	[5-40]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Isoleucine, Plasma	88	umol/L	[30-120]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Leucine, Plasma	110	umol/L	[60-180]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Lysine, Plasma	143	umol/L	[85-230]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Methionine, Plasma	32	umol/L	[15-40]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Ornithine, Plasma	66	umol/L	[25-110]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Phenylalanine, Plasma	47	umol/L	[30-82]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Proline, Plasma	321	umol/L	[90-350]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Sarcosine, Plasma	5	umol/L	[<=5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Serine, Plasma	115	umol/L	[60-170]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Taurine, Plasma	55	umol/L	[30-130]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Threonine, Plasma	78	umol/L	[60-190]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Tryptophan, Plasma	72	umol/L	[25-80]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Tyrosine, Plasma	99	umol/L	[35-110]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Valine, Plasma	189	umol/L	[120-320]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C2, Acetyl	12.65	umol/L	[3.74-16.56]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C3, Propionyl	0.45	umol/L	[0.00-0.83]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C4, Iso-/Butyryl	0.32	umol/L	[0.00-0.45]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50

***** Abnormal, **#** = Corrected, **C** = Critical, **f** = Footnote, **H** = High, **L** = Low, **t** = Interpretive Text, **@** = Reference Lab

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Julio C. Delgado, M.D. M.S., Director of Laboratories

*****Example Report*****

Patient Age/Gender: Unknown Unknown
Printed: 18-Dec-19 13:01:44

Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/ Verified
C5, Isovaleryl/2Mebutyryl	0.22	umol/L	[0.00-0.30]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C5-DC, Glutaryl	0.06	umol/L	[0.00-0.09]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C5-OH, 3-OH Isovaleryl	0.03	umol/L	[0.00-0.07]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C6, Hexanoyl	0.07	umol/L	[0.00-0.12]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C8, Octanoyl	0.19	umol/L	[0.00-0.23]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C8:1, Octenoyl	0.21	umol/L	[0.00-0.61]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C10, Decanoyl	0.21	umol/L	[0.00-0.31]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C10:1, Decenoyl	0.17	umol/L	[0.00-0.31]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C12, Dodecanoyl	0.04	umol/L	[0.00-0.12]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C12:1, Dodecenoyl	0.16	umol/L	[0.00-0.17]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C12-OH, 3-OH-Dodecanoyl	0.01	umol/L	[0.00-0.02]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C14, Tetradecanoyl	0.03	umol/L	[0.00-0.05]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C14:1, Tetradecenoyl	0.11	umol/L	[0.00-0.16]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C14:2, Tetradecadienoyl	0.12	umol/L	[0.00-0.12]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C14-OH, 3-OH-Tetradecanoyl	0.01	umol/L	[0.00-0.02]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C14:1-OH, 3-OH-Tetradecenoyl	0.02	umol/L	[0.00-0.02]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C16, Palmitoyl	0.09	umol/L	[0.00-0.10]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C16:1, Palmitoleyl	0.03	umol/L	[0.00-0.04]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C16-OH, 3-OH-Palmitoyl	0.01	umol/L	[0.00-0.01]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C16:1-OH, 3-OH-Palmitoleyl	0.01	umol/L	[0.00-0.01]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18, Stearoyl	0.02	umol/L	[0.00-0.04]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18:1, Oleyl	0.15	umol/L	[0.00-0.17]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18:2, Linoleyl	0.02	umol/L	[0.00-0.10]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18-OH, 3-OH-Stearoyl	0.01	umol/L	[0.00-0.01]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18:1-OH, 3-OH-Oleyl	0.01	umol/L	[0.00-0.01]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
C18:2-OH, 3-OH-Linoleyl	0.01	umol/L	[0.00-0.01]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Mucopolysaccharides mg/mmol CRT	3.9		[0.0-7.1]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Lactic Acid, Urine	47		[0-50]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Pyruvic Acid, Urine	13		[0-15]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Succinic Acid, Urine	8		[0-20]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Fumaric Acid, Urine	1		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
2-Ketoglutaric Acid, Urine	72		[0-75]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Methylmalonic Acid, Urine	3		[0-5]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
3-OH-Butyric Acid, Urine	1		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Acetoacetic Acid, Urine	2		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
2-Keto-3-methylvaleric Acid, Urine	8		[0-10]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
2-Ketoisocaproic Acid, Urine	3		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
2-Ketoisovaleric Acid, Urine	2		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Ethylmalonic Acid, Urine	3		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Adipic Acid, Urine	27		[0-35]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Suberic Acid, Urine	2		[0-3]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Sebacic Acid, Urine	1		[0-3]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50

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Julio C. Delgado, M.D. M.S., Director of Laboratories

*****Example Report*****

Patient Age/Gender: Unknown Unknown
Printed: 18-Dec-19 13:01:44

Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/ Verified
4-OH-phenylacetic Acid, Urine	11		[0-25]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
4-OH-phenyllactic Acid, Urine	1		[0-4]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
4-OH-phenylpyruvic Acid, Urine	Not Detected		[0-2]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Succinylacetone, Urine	Not Detected		[0-0]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Creatine, Urine	30	mmol/mol CRT	[10-370]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Guanidinoacetic acid, Urine	22	mmol/mol CRT	[7-130]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Creatinine, Urine	2122.0	umol/L		19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Creatine, Serum/Plasma	14.2	umol/L	[9.0-90.0]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
Guanidinoacetic acid, Serum/Plasma	1.41	umol/L	[1.10-3.80]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50
FRAG X Specimen	Whole Blood			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	18-Dec-19 11:43:23
Fragile X Allele 1	> 200			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	18-Dec-19 11:43:23
Fragile X Allele 2	Not Applicable	CGG repeats		19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	18-Dec-19 11:43:23
Fragile X Methylation Pattern	**Full ***			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	18-Dec-19 11:43:23
Fragile X Interpretation	See Note f			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	18-Dec-19 11:43:23
Cytogenomic SNP Microarray	Normal f		[Normal]	19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	13-Dec-19 10:27:32
EER Cytogenomic SNP Microarray	EERUnavailable			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	13-Dec-19 10:27:32
Autism/Intellectual Interp	See Note f			19-343-900152	09-Dec-19 12:29:00	09-Dec-19 16:43:00	10-Dec-19 12:07:50

09-Dec-19 12:29:00 Fragile X Interpretation:

This male fetus has a loss-of-function FMR1 allele (typically greater than 200 CGG repeats that is fully methylated), thus, is affected with fragile X syndrome. Genetic consultation is recommended.

The expanded allele is fully methylated and predicted to be nonfunctional.

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

09-Dec-19 12:29:00 Cytogenomic SNP Microarray:

Test Performed: Cytogenomic SNP Microarray (CMA SNP)
Specimen Type: Peripheral blood
Indication for Testing: Speech delay

RESULT SUMMARY
Normal Microarray Result (Male)

RESULT DESCRIPTION
No clinically significant copy number changes or regions of homozygosity were detected.

INTERPRETATION
This analysis showed normal results.

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

Cytogenetic Nomenclature (ISCN):
arr(1-22)x2,(X,Y)x1

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 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 these 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 Copy Number Variant Assertion Criteria, which can be found on ARUP’s Genetics Resources website at www.aruplab.com/genetics/resources
- 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 Resources website at www.aruplab.com/genetics/resources.

This result has been reviewed and approved by Patricia A. Mowery-Rushton, PhD, ABMGG

[redacted]

09-Dec-19 12:29:00 Autism/Intellectual Interp:
No 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.

09-Dec-19 12:29:00 Mucopolysaccharides mg/mmol CRT:
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).

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS

09-Dec-19 12:29:00 Creatine, Urine:

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS

09-Dec-19 12:29:00 Fragile X Interpretation:
Background Information for Fragile X (FMR1)
Characteristics: Fragile X syndrome, the most common heritable form of mental retardation, is characterized by moderate mental retardation in males and mild mental retardation in females, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders behavioral phenotype, and

connective tissue anomalies. Adult males may have physical findings including: macroorchidism, a long narrow face, prominent ears and jaw, and a single palmar crease.
Incidence: 1 in 4,000 Caucasian males and 1 in 8,000 Caucasian females; unknown in other ethnicities.
Inheritance: X-linked dominant.
Penetrance: Reduced in females.
Cause: Expansion of the FMR1 gene CGG triplet repeat.
Full mutation: >200-230 CGG repeats (methylated)
Premutation: 55-200 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 100 or greater. Methylation analysis is used to distinguish between premutation and full mutation alleles.
Analytic Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations.

See Compliance Statement C: www.aruplab.com/CS

09-Dec-19 12:29:00 Cytogenomic SNP Microarray:
INTERPRETIVE INFORMATION: CYTOGENOMIC SNP MICROARRAY

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

09-Dec-19 12:29:00 Autism/Intellectual Interp:
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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS