

Ordered By Physician Name: Physician, Test	Patient Name: UOA test, UOA	
	Accession #: R5006	Specimen #: X5006
Reason for Referral:	Specimen: Urine	
	Birthdate: 06/12/1990	Age: 30
	Gender: Unknown	
	MRN #: 4654889	Collected: 08/05/2020
	Ethnicity:	Received: 08/05/2020

Urine Organic Acid Screen

INTERPRETATION

Numerous abnormalities are present in this urine specimen including gross elevations of fumaric acid. Overall, this urine organic acid profile is consistent with fumarase deficiency (OMIM 606812). This finding requires immediate clinical correlation and genetic counseling. The following includes further description of the abnormalities detected in this specimen.

The most marked abnormality detected in this specimen is a gross elevation of fumaric acid. This compound is found at levels well above any seen to date in our unaffected reference population and is consistent with the levels seen in patients with fumarase deficiency. Maleic acid is also elevated (>99th percentile). Maleic acid is the cis stereoisomer of fumaric acid.

Numerous additional analytes are mildly elevated in this specimen including 3-methylglutaconic acid, 3-hydroxypropionic acid, alpha-ketoglutaric acid, 3-methylglutaric acid and malic acid.

Collectively, this pattern of perturbations is consistent with fumarase deficiency (PMID 20549362). Gross elevations of fumaric acid are the key urine organic acid finding in this disorder. Multiple other secondary abnormalities have been reported in the urine of patients with this disorder but these findings are more heterogenous/sporadic in this patient population (PMID: 20549362). Many of these secondary abnormalities are present in this specimen.

Immediate clinical correlation and genetic counseling are recommended.

ASSAY INFORMATION

Method

Urine specimens are diluted based on creatinine concentration, acidified, subjected to an organic extraction and then derivitized using a silylation reagent. Prepared samples are analyzed using gas chromatography mass spectrometry.

Limitations/Disclaimer

False negative results can occur in rare situations when diet and/or clinical condition masks or normalizes disease relevant analyte perturbations. In addition, false negatives may occur when disease presentation is intermittent or the result of a mild defect. Results should always be viewed in the context of clinical presentation and concurrent laboratory studies.

This test was developed and its performance characteristics determined by Indiana University Biochemical Genetics Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88) as qualified to perform high complexity clinical laboratory testing. CLIA# 15D0647198 • CAP# 1678930

ELECTRONICALLY SIGNED BY

Marcus J. Miller, Director of the Biochemical Genetics Laboratory, 08/05/2020

