



Patient ID M0002190	Patient Name SAMPLEREPORT, OAU A	Birth Date 1971-09-28	Gender F	Age 40
Order Number M0002190	Client Order Number M0002190	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jun 2012 00:09		

Organic Acids Scrn, U

MCR

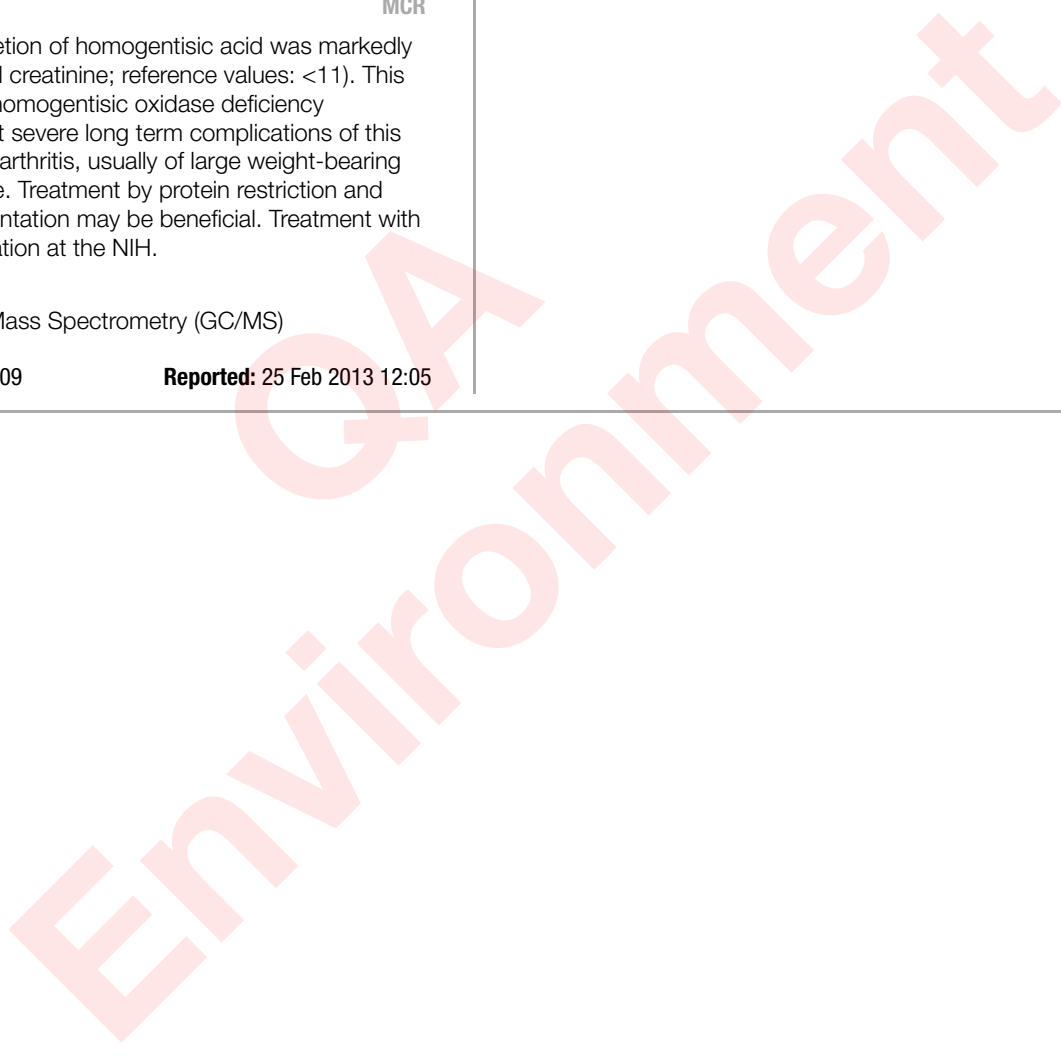
In this sample, the excretion of homogentisic acid was markedly elevated (125 mmol/mol creatinine; reference values: <11). This finding is diagnostic of homogentisic oxidase deficiency (Alkaptonuria). The most severe long term complications of this disorder are ochronotic arthritis, usually of large weight-bearing joints, and heart disease. Treatment by protein restriction and ascorbic acid supplementation may be beneficial. Treatment with NTBC is under investigation at the NIH.

ADDITIONAL INFORMATION

Gas Chromatography-Mass Spectrometry (GC/MS)

Received: 27 Jun 2012 00:09

Reported: 25 Feb 2013 12:05



Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905