



Patient ID <b>SA00054289</b>	Patient Name <b>SAMPLEREPORT, SNS A</b>	Birth Date <b>2013-02-24</b>	Gender <b>F</b>	Age <b>2 D</b>
Order Number <b>SA00054289</b>	Client Order Number <b>SA00054289</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>26 Feb 2013 00:00</b>		

## Supplemental Newborn Screen, BS

### Supplemental Newborn Screen Result

MCR

In this blood spot, the following result was observed and confirmed by repeat analysis:

Isovaleryl-/2-methylbutyrylcarnitine  
4.1 nmol/mL (abnormal >0.50)

This finding is suggestive of a biochemical diagnosis of isovaleric acidemia or, less likely, 2-methylbutyryl-CoA dehydrogenase deficiency (see Matern et al. Pediatrics 112:74-78, 2003). A drug artifact (pivalic acid) cannot be excluded on the sole basis of this result. We recommend prompt confirmatory testing by plasma acylcarnitine and urine acylglycine analyses.

Please contact the genetics/metabolic specialist on call at Mayo Clinic (1-800-533-1710) for further assistance.

### Reviewed By

MCR

Jennifer Peterson

**Received:** 27 Feb 2013 14:44

**Reported:** 27 Feb 2013 15:21

### Performing Site Legend

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