



BAYLOR
Institute of Metabolic Disease

Baylor Research Institute

3812 Elm Street, Dallas, TX 75226
(214) 820-4533 Fax (214) 820-4853
www.baylorhealth.edu/imd

Charles R. Roe, M.D., Medical Director

Patient Name: Last, Baby Girl

Patient ID #:

Age: 1 Day(s)

Date of Birth: 03/16/01

Print Date: 03/27/01

Reprint Date: 5/16/03

SNBS Client's Name

Attn: Send results to the attention of

Mailing Address

City, State, Zip Code

IMD Order #: #####

Date received: 03/21/01

Specimen Type: Dried Blood Spot

Accession Number: SS#####

Date of Collection: 3/17/01 **Time of Collection:**

Client Specimen ID:

Specimen Integrity:

Tandem MS Supplemental Screening

The presence of all these disorders was evaluated by Tandem Mass Spectrometry (MS/MS):

- | | |
|-----------------------------------------------------------------------|-----------------------------------------------------------------|
| Argininemia: arginase deficiency (ARG) | Malonic aciduria (MAL) |
| Argininosuccinate lyase deficiency (ASA) | Maple syrup urine disease (MSUD) |
| Beta ketothiolase deficiency (BKT or T2) | Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) |
| Carnitine palmitoyltransferase I deficiency (CPT-I or CPT-IA) | 2-Methylbutyryl-CoA dehydrogenase deficiency (2-MBCD or 2MBG) |
| Carnitine palmitoyltransferase II deficiency (CPT-II) | 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC) |
| Carnitine/acylcarnitine translocase deficiency (CACT) | 3-Methylglutaconic aciduria type I (3-MGA type I) |
| Carnitine transport defect (CTD or CUD) | Methylmalonic acidemia (MMA or MUT) |
| Citrullinemia type I or II (CIT I or CIT II) | Mitochondrial trifunctional protein deficiency (TFP or MTP) |
| Ethylmalonic encephalopathy (EMA ENC) | Multiple acyl-CoA dehydrogenase deficiency (MADD or GA-II) |
| Glutaric aciduria Type I (GA I) | Nonketotic hyperglycinemia (NKH) |
| Holocarboxylase synthetase deficiency (HCS or MCD) | 5-Oxoprolinuria (5OXOPRO) or 5-Oxoprolinase deficiency |
| Homocystinuria: cystathionine synthetase deficiency (HCY or HCYS) | Phenylketonuria (PKU) or Hyperphenylalaninemia (H-PHE) |
| 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG) | Propionic acidemia (PA or PROP) |
| Hyperammonemia, hyperornithinemia, homocitrullinuria (HHH or ORNT2) | Short-chain acyl-CoA dehydrogenase deficiency (SCAD) |
| Hypermethioninemia (MET or MAT) | Tyrosinemia Type I, II, III (TYR-I, TYR-II, TYR-III) |
| Isobutyryl-CoA dehydrogenase deficiency (IBCD or IBG) | Very long-chain acyl-CoA dehydrogenase (VLCAD) |
| Isovaleric acidemia (IVA) | Some Cobalamin or Vitamin B12 disorders |
| Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) | |

** Although the aforementioned list of metabolic diseases is an accurate list of the diseases that are detectable through the screening processes being performed, under no circumstances can it be guaranteed that the screening process will for each patient tested detect the existence of each of the aforementioned listed diseases. Like many screening processes, the screening process being conducted is a tool to be utilized by health care providers to assist them in attempting to detect the existence of a number of diseases whose detection is dependent upon a number of factors, some of which are outside the parameters of the Screening Service being performed.*

Result / Interpretation: Screening results negative, no evidence of any of the above disorders.

REPORT FORMAT FOR A SINGLE NORMAL RESULT

Mass Spectrometry Laboratory

Director - Larry Sweetman, Ph.D.
Asst. Director - Xiaowei Fu, M.D., Ph.D.

E-mail: larrys@baylorhealth.edu
E-mail: xiaoweif@baylorhealth.edu

Patient Name: Last1, Girl

Patient ID:

Page 1 of 1