Newborn
Screening Program

Unified State Laboratories: Public Health
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HOSPITAL OF BIRTH
ATTN: NAME
750 W 800 N
OREM UT 84057-3660

BABY

Infant's Name : LAST, FIRST
Sex : Male
Birth Date : 01/01/2001
Birth Record # : UT123A123
Hospital MR # : 99999999
Mother's Name : LAST, FISRT

SPECIMEN INFORMATION

Type : FIRST
Ascn Number : F1110111200111
Date Collected : 01/02/2001
Date Received : 01/03/2001
Date Reported : 01/04/2001
Date Printed : 01/05/2001

NEWBORN SCREENING RESULTS

DISORDER/*TEST*	DATE TESTED	RESULTS	DETERMINATION/ *NORMAL RANGE*
SCID *TREC*	11/16/2018	Normal	Normal *Normal*
Spinal Muscular Atrophy *SMN1*	11/16/2018	Normal	Normal *Normal*
Biotinidase Deficiency *Enzyme activity*	11/16/2018	Normal	Normal *Full enzyme activity*
Congenital Adrenal Hyperplasia * *17-OHP ELISA*	11/16/2018	11.1 ng/mL	Normal *Based on baby's birth weight*
Cystic Fibrosis *Immuno-reactive Trypsinogen ELISA*	11/16/2018	20.5 ng/mL	Normal *< 51 ng/mL*
Galactosemia *G-1-P uridyltransferase activity*	11/16/2018	9.5 U/gHb	Normal *> 3.0 U/gHb*
Hemoglobinopathies *Isoelectric Focusing*	11/16/2018	Normal - FA	Normal *FA (Fetal Adult)*
Congenital Hypothyroidism *TSH*	11/16/2018	13.8 µIU/mL	Normal *< 40 µIU/mL*
Acylcarnitine Disorders *MS/MS Tandem Mass screening*	11/19/2018	Abnormal	ABNORMAL *Based on baby's birth weight*
Amino Acid Disorders *MS/MS Tandem Mass screening*	11/19/2018	Normal	Normal *Based on baby's birth weight*

* **If glucocorticoids administered to infant or mother, use caution when interpreting the CAH result. Acylcarnitines, amino acids, and CAH second tier testing performed by ARUP Laboratories, 500 Chipeta Way, Salt Lake City, UT 84108-1221. Results are validated only for dried blood spot specimens collected using the heelstick method.**

Specimen Comments:
In this sample the concentrations of C14-carnitine (0.71 umol/L; cut-off = 0.60 umol/L) and C14:1-carnitine (0.93 umol/L; cut-off = 0. 60 umol/L) were increased. This is suggestive of VLCAD (Very Long Chain Acyl-CoA Dehydrogenase) deficiency, an inherited disorder of fatty acid oxidation. Plasma acylcarnitine profile and urine organic acids are recommended to confirm this possibility. With this condition, tests obtained when infants are not under metabolic stress (fasting), might be normal, therefore genetic evaluation is recommended to consider the need for additional testing. ACTION: URGENT (CRITICAL) PLASMA ACYLCARNITINE PROFILE, URINE ORGANIC ACIDS, REFERRAL TO METABOLIC CLINIC

A newborn screening result should not be considered diagnostic, a… replace the individualized evaluation and diagnosis of an infant by a well-trained, knowl… care provider.

If you have questions regarding these results, please contact th…
visit our website http://health.utah.gov/newbor…

RPT_FIRST

This is where specifics about results and actions needed will be entered; not all disorders will have a footnote.

Program personnel will call the health care provider noted on the demographic card for all abnormal results. Instructions will be given for follow-up such as collection of the routine 2nd screen, collection of a 3rd screen or additional confirmatory/diagnostic testing. If you have questions, call for clarification – 801-584-8256.