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CLINIC  
2 SOUTH 9 EAST  
SALT LAKE CITY UT 84100

## BABY

Infant's Name : Baby  
Sex : F  
Birth Date : 01/01/2018  
Birth Record # : UT999A888  
Hospital MR # : 12345  
Mother's Name : Mom

## SPECIMEN INFORMATION

Type : SECOND  
AscN Number : S2860999201899  
Date Collected : 1/1/2018  
Date Received : 1/1/2018  
Date Reported : 1/1/2018  
Date Printed : 1/1/2018

## NEWBORN SCREENING RESULTS

DISORDER/TEST	DATE TESTED	RESULTS	DETERMINATION/ NORMAL RANGE
Cystic Fibrosis CFTR DNA	10/16/2018	Abnormal	<b>ABNORMAL</b>
Congenital Hypothyroidism TSH	10/15/2018	7.7 $\mu$ IU/mL	Normal < 20 $\mu$ IU/mL
Biotinidase Deficiency Enzyme activity	10/15/2018	Normal	Normal Full enzyme activity
Congenital Adrenal Hyperplasia * 17-OHP ELISA	10/15/2018	5.4 ng/mL	Normal Based on baby's birth weight
Cystic Fibrosis Immuno-reactive Trypsinogen ELISA	10/17/2018	344.9 ng/mL	<b>INDETERMINATE</b> < 42 ng/mL
Acylcarnitine Disorders MS/MS Tandem Mass screening	10/15/2018	Normal	Normal Based on baby's birth weight
Amino Acid Disorders MS/MS Tandem Mass screening	10/15/2018	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:

Cystic Fibrosis DNA result: Positive for two pathogenic mutations: deltaF508/deltaF508

Interpretation: This individual is homozygous for the deltaF508 mutation in the CFTR gene. This result is consistent with the clinical diagnosis of Cystic Fibrosis. Method: DNA isolated from dried blood spot specimen undergoes CFTR mutation detection assay using Luminex xTAG Cystic Fibrosis v2 (CF60) v.02.

ACTION: URGENT SWEAT CHLORIDE TESTING AND REFERRAL TO THE CYSTIC FIBROSIS CENTER (801) 213-3599.

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

If you have questions regarding these results, please contact the Newborn Screening staff or visit our website <http://health.utah.gov/newbornscreening>