

BLANK, BABY BOY

Negative

Health Card Number/Type: XXXXXXXXXX Sex: M Birth: 2023-12-01 00:20 Collection: 2023-12-02 00:40 Receipt: 2023-12-04

Guardian's Name: BLANK, PARENT

Submitting Location: HOSPITAL

Other Names:

Location Fax: 11234567890 Submitter #: XXXXXXXXXXX

Ordering HCP:EXAMPLE, DOCTOROrdering HCP Fax:1234561234Infant's HCP:EXAMPLE2, DOCTORInfant's HCP Fax:1234561234

Amino Acidemias:

Phenylketonuria and Variants / Biopterin Defects

Maple Syrup Urine Disease

Homocystinuria (Hypermethioninemias)

Citrullinemias / Argininosuccinic Aciduria

Tyrosinemias

Negative

Amino Acidopathies, other

Organic Acidemias:

Propionic / Methylmalonic Acidemias Negative
Isovaleric Acidemia / 2 Methylbutyric Acidemia Negative
Glutaric Acidemia Type 1 Negative
Guanidinoacetate Methyltransferase Deficiency Negative
Organic Acidemias, other Negative

Fatty Acid Oxidation Defects:

Medium Chain Acyl Dehydrogenase Deficiency

Very Long Chain Acyl Dehydrogenase Deficiency

Long Chain Hydroxyl Acyl Dehydrogenase /Trifunctional Protein Deficiencies

Negative

Carnitine Uptake Defect

Fatty Acid Oxidation Disorders, other

Negative

Galactosemia Negative

Biotinidase Deficiency UNSATISFACTORY

Endocrine Disorders:

Congenital Hypothyroidism
Congenital Adrenal Hyperplasia
Negative
Sickle Cell and other Hemoglobinopathies
Negative

Cystic Fibrosis UNSATISFACTORY *

Severe Combined Immune Deficiency UNSATISFACTORY

Spinal Muscular Atrophy UNSATISFACTORY *

Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease) UNSATISFACTORY

The sample received from your patient was not adequate for the following reason(s):

Quantity of blood insufficient (filter paper not completely saturated).

Version June 2019

Page 1 of 2

o NSO's complete screening panel is available at <u>www.newbornscreening.on.ca.</u> Results for diseases not reflected above are pending.

o Screen negative means that this infant is at decreased risk for the disease(s). Screen positive means that this infant is at increased risk for the disease(s). It does not mean that a disease is present, but further testing is indicated. If a test is positive and you and / or your patient have not already been contacted, please call NSO at (613) 738-3222.

o This report does NOT contain information about this infant's carrier status for hemoglobinopathies. Families wishing to learn their child's carrier result should contact NSO or visit www.newbornscreening.on.ca

o Infants with meconium ileus are at risk of having Cystic Fibrosis (CF) but will often have a screen negative result for CF; it is therefore recommended that they be clinically evaluated for CF regardless of the newborn screening results.



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A REPEAT SAMPLE IS REQUIRED AS SOON AS POSSIBLE

N20231204-9001

Version June 2019

Page 2 of 2

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