

| CP 3.5 Mailer Wording for Metabolic Disorders (1/16/08) | |
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| Result | Wording/Follow-Up |
| General | |
| General Disclaimer on all Mailers | Test interpretations are based on the Birth/Collection Information provided above and subject to disclaimer below. |
| General Disclaimer on all Mailers | Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does <u>not</u> rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. |
| Multiple Positives | Attention: This child has tested positive for more than one test. |
| Contact Coordinator | If you have questions regarding these results, please contact the Newborn Screening staff at [xxx] Area Service Center [phone #]. |
| Inadequate | |
| Inadequate Specimen | This specimen was inadequate for testing for the following reason: [Inadeq. Code#] [Reason] |
| Galactosemia and Variants | |
| Follow Up Rider - Galactosemia Positive | Galactosemia: Additional testing is required to rule out disorder. Call your Newborn Screening Coordinator for assistance (see below). |
| Genotype Interpretation - Genotype N | Interpretation: The results are within normal limits. |
| Genotype Interpretation - Genotype NN | Interpretation: The results suggest normal Transferase homozygote (NN). |
| Genotype Interpretation - Genotype GG | Interpretation: The results suggest Galatosemia homozygote (GG) . |
| Genotype Interpretation - Genotype GN | Interpretation: The results suggest Galatosemia-Normal heterozygote (GN) . |
| Genotype Interpretation - Genotype DG | Interpretation: The results suggest Duarte-Galatosemia heterozygote (DG) . |
| Genotype Interpretation - Genotype DD | Interpretation: The results suggest Duarte homozygote (DD) . |
| Genotype Interpretation - Genotype DN | Interpretation: The results suggest Duarte-Normal heterozygote (DN) . |
| Genotype Interpretation - Genotype LAG | Interpretation: The results suggest Los Angeles - Galactosemia heterozygote (LA/G) . |
| Genotype Interpretation - Genotype LAN | Interpretation: The results suggest Los Angeles - Normal heterozygote (LA/N) . |
| Genotype Interpretation - Genotype LALA | Interpretation: The results suggest Los Angeles - Los Angeles homozygote (LA/LA) . Needs to be changed in SIS somehow...SIS says "heterozygote" |
| Genotype Interpretation - Genotype LAD | Interpretation: The results suggest Los Angeles - Duarte heterozygote (LA/D). |
| Genotype Interpretation - Genotype UNK | Interpretation: The results are indeterminate. |
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| MS/MS - General | |
| Out of Range | No Follow-Up Required. This pattern of flags is not known to be associated with any disorder detectable by MS/MS |
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| Acyl Carnitine Panel | |
| Acyl Carnitine Panel Positive - Complex Pattern Interpretation | This complex pattern of elevations require further evaluation. |
| Acyl Carnitine Positive | This pattern of elevations is consistent with [] |
| Follow Up Rider - Acyl Carnitine panel is positive | Acyl Carnitine Panel: An immediate referral to a CCS Metabolic Center is strongly recommended. Call your Newborn Screening Coordinator for assistance (See below). |
| "Too Old" Interpretation - (Acyl panel "negative" and age at collection > time based parameter (10 days) | Acyl Carnitine Panel: Reference Ranges for acylcarnitines are based on specimens collected on newborns who are less than 10 days old. |
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| Amino Acid Panel | |
| Amino Acid Panel Positive Interpretation - incl. PKU (single or multiple elevations) | This pattern of elevations is consistent with [] |
| Amino Acid Positive - Complex Pattern Interpretation | This complex pattern of elevations require further evaluation. |
| Follow Up Rider - Amino Acid Panel. Non-PKU Positive. When the panel is positive, but not because of PKU. | Amino Acid Panel: An immediate referral to a CCS Metabolic Center is strongly recommended. Call your Newborn Screening Coordinator for assistance (See below). |
| Follow Up Rider - PKU positive . Amino Acid panel is positive because of PKU. | PKU: This screen for phenylkentonuria (PKU) was positive. Another specimen must be collected on this infant. Call your Newborn Screening Coordinator for assistance (See below). |
| Follow Up Rider - TPN feeding type positive. Amino Acid panel positive and feeding type TPN. | Amino Acid Panel: TPN feedings can produce amino acid elevations that are not clinically significant. |
| Amino Acid Results, if "Early" or "Missing" and "Negative" Interpretation | Collected Early - No Interpretation |
| < 12 hour collection (Early or Missing Data) | |
| Follow Up Rider - Amino Acid Panel reported < 12 hours because of missing data | Amino Acid Panel: There was insufficient data to determine whether or not this newborn was at least 12 hours old when this specimen was collected. Testing of amino acids at less than 12 hours of age is not reliable for detecting certain metabolic disorders. |
| Follow Up Rider - Amino Acid Panel collected early (<12 of age) | Amino Acid Panel: This specimen was collected at less than 12 hours of age. Testing of amino acids at less than 12 hours of age is not reliable for detecting certain metabolic disorders. Another specimen MUST be collected. |

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| Transfused | |
| Galactosemia Interpretation | Transfused. No Interpretation. |
| Rider - Transfused - No Gal or Hb. Sent when Transfused flag is "Yes" and Transfusion date < specimen collection date | Because of transfusion, further testing on whole blood is required to rule out hemoglobinopathies. If this baby has symptoms or family history of galactosemia, parent testing is recommended. Call your Newborn Screening Coordinator for assistance (See below). |
| Acyl Carnitine Panel Interpretation | Transfusion may, in rare occasions, give misleading results that fail to detect some of these disorders. If a metabolic disorder is suspected, consult a metabolic specialist. |
| Amino Acid Panel Interpretation | Transfusion may, in rare occasions, give misleading results that fail to detect some of these disorders. If a metabolic disorder is suspected, consult a metabolic specialist. |
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| Biotinidase Deficiency | This screen for Biotinidase was positive. Another specimen must be collected in this infant. |