Carnitine Profile, Quantitative, Plasma

Test Code: CN
Turnaround time: 7 days
CPT Codes: 82379 x1

Condition Description

Carnitine deficiency can be from a primary defect or can be a secondary effect. Primary carnitine deficiency can be the result of several metabolic conditions including Carnitine Uptake Deficiency (CUD), Carnitine Acylcarnitine Transporter (CACT) Deficiency, Carnitine Palmitoyltransferase I (CPT I) Deficiency, Carnitine Palmitoyltransferase II (CPTII) Deficiency. Primary carnitine deficiency can present with:

- hypoketotic hypoglycemia encephalopathy
- hepatomegaly
- cardiomyopathy
- muscle weakness
- gastrointestinal dysmotility
- hypochromic anemia

Secondary carnitine deficiency results from another metabolic disorder, such as another fatty acid oxidation disorder, or an organic acidemias leading to carnitine depletion secondary to the formation of acylcarnitines for excretion of accumulating by products. Secondary carnitine deficiency can present with symptoms of:

- encephalopathy
- hypotonia
- hepatomegaly
- cardiac hypertrophy
- failure to thrive
- hypoglycemia
- ketoacidosis
- hyperammonemia

Indications

This test is indicated for patients with:

- lethargy/hypotonia
- myopathy (cardiac/skeletal)
- clinical suspicion of a fatty acid oxidation defect and an organic acidemia
- renal dysfunction
- positive newborn screening result with decreased free carnitine and/or abnormal acylcarnitine profile

The test is most commonly performed in serum or plasma. A urine carnitine profile is recommended in addition to a plasma carnitine profile in patients suspected of having a primary disorder of carnitine transport.

Methodology

Electrospray Tandem Mass Spectrometry (MS/MS).

Detection

Detection of carnitine deficiency can be sensitive to the age and eating status of the patient.

Reference Range

<table>
<thead>
<tr>
<th>Condition</th>
<th>Total Carnitine (umol/L)</th>
<th>Free Carnitine (FC) (umol/L)</th>
<th>Acylcarnitine (AC) (umol/L)</th>
<th>AC / FC</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Day</td>
<td>23 - 68</td>
<td>12 - 36</td>
<td>7 - 37</td>
<td>0.4 - 1.7</td>
</tr>
<tr>
<td>2-7 Days</td>
<td>17 - 41</td>
<td>10 - 12</td>
<td>3 - 24</td>
<td>0.2 - 1.4</td>
</tr>
<tr>
<td>8-31 Days</td>
<td>19 - 59</td>
<td>12 - 46</td>
<td>4 - 15</td>
<td>0.1 - 0.7</td>
</tr>
<tr>
<td>32 Days - 12 Months</td>
<td>38 - 68</td>
<td>27 - 49</td>
<td>7 - 19</td>
<td>0.2 - 0.5</td>
</tr>
<tr>
<td>13 Months - 5 Years</td>
<td>35 - 84</td>
<td>24 - 63</td>
<td>4 - 28</td>
<td>0.1 - 0.8</td>
</tr>
<tr>
<td>6 - 10 Years</td>
<td>28 - 83</td>
<td>22 - 66</td>
<td>3 - 32</td>
<td>0.1 - 0.9</td>
</tr>
</tbody>
</table>

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Specimen Requirements

Submit only 1 of the following specimen types

Type: Plasma

Specimen Requirements:
Sodium Heparin (Green Top)
1-2 ml
Sample should be collected while fasting or 2-4 hours post prandial. Centrifuge to separate plasma and freeze.

Specimen Collection and Shipping:
Ship frozen sample on dry ice with overnight delivery.

Special Instructions

Please indicate any medications or dietary changes on the test requisition form.

Related Tests

- Organic Acids Profile (OA)
- Acylcarnitine Profile (AR) - Plasma