

Centre for Human Metabolomics (CHM)

| Test: | Quantitative Isotope Specific CYSTINE URINE | | | | | | | | | | | | | | | | | | | | | | | | | | | |
|---|--|--------------------|-------------------------|--------------------|------------|-------|------|------------|------|------|-------------|------|------|-----------|------|------|-----------|------|------|-----------|------|------|------------|------|------|---------------|------|--------|
| Test Code: | 4335 + 4221 + 4321 + 4238 | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Tariff (including VAT): | R 42.47 + R 42.47 + R 17.59 + R 453.27 = R 555.80 | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Description: | LC-MS Isotope dilution Assay – Sulphur containing amino acid. | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Turnaround time: | 35 work days from receipt of sample at our laboratory (part of full screen). Single analysis: 10 work days from receipt of sample at our laboratory. | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Transit stability / Sample viability: | Keep frozen, send on dry ice. Viability: 12 months – kept frozen | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Comments: | 1. NO preservatives added. 2. Total volume information for 24 hour period required for calculation of excretion per day. | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Sample required: | 10 ml urine - aliquot from 24 hour urine sample / freeze and only send 10 ml aliquot, frozen, on dry ice for analyses. | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Method: | TANDEM – MS [isotope specific assay] | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Reference range & units: | <p>New reference ranges implemented from 1 February 2016</p> <table border="1"> <thead> <tr> <th>Age group</th> <th>Random (mmol/mol creat)</th> <th>24 hour (µmol/day)</th> </tr> </thead> <tbody> <tr> <td>0-1 months</td> <td>12-39</td> <td>2-48</td> </tr> <tr> <td>1-6 months</td> <td>7-24</td> <td>2-41</td> </tr> <tr> <td>6-12 months</td> <td>6-15</td> <td>2-41</td> </tr> <tr> <td>1-2 years</td> <td>5-13</td> <td>4-54</td> </tr> <tr> <td>2-4 years</td> <td>4-15</td> <td>4-54</td> </tr> <tr> <td>4-7 years</td> <td>4-11</td> <td>4-54</td> </tr> <tr> <td>7-13 years</td> <td>4-12</td> <td>4-54</td> </tr> <tr> <td>Over 13 years</td> <td>3-17</td> <td>42-245</td> </tr> </tbody> </table> <p>Reference: Blau et al. 2005. Inborn Errors of metabolic disease diagnosis and treatment. Springer-Verlag. Heidelberg.</p> | Age group | Random (mmol/mol creat) | 24 hour (µmol/day) | 0-1 months | 12-39 | 2-48 | 1-6 months | 7-24 | 2-41 | 6-12 months | 6-15 | 2-41 | 1-2 years | 5-13 | 4-54 | 2-4 years | 4-15 | 4-54 | 4-7 years | 4-11 | 4-54 | 7-13 years | 4-12 | 4-54 | Over 13 years | 3-17 | 42-245 |
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