

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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Consultant/Scientist:	Dr Marli Dercksen / Prof Chris Vorster																											
Telephone number:	018 299 2302 / 018 299 4196																											
E-mail address:	marli.dercksen@nwu.ac.za / chris.vorster@nwu.ac.za																											
Contact for results & other enquiries:	Ansie Mienie																											
Telephone number:	018 299 2312 / 018 285 2652 (leave message)																											
Fax number:	018 299 2316																											
E-mail address:	ansie.mienie@nwu.ac.za																											
Delivery address for samples:	Center for Human Metabolomics (CHM), Sample reception (PLIEM/NBS/CRS) Building F3, Room Number G19, 11 Hoffmann street North West University, Potchefstroom, 2531																											