

Cognitive AA pilot scheme – 1st Circulation Participants Report

Sample A

Sample details:

These results were from a patient with classical homocystinuria due to cystathionine synthase deficiency. Plasma amino acid concentrations and a chromatogram showing a clearly detectable peak of homocystine were provided.

Correct findings / abnormalities:

Elevated homocystine and methionine scored 2 points. Several participants included low cystine and the presence of methionine sulphoxide but these were not scored.

Correct Diagnosis and further tests:

Classical homocystinuria (CBS deficiency) as certain or most likely, scored 1 point. The possibility of MAT I/III should also be mentioned although this was not scored. Further testing of total homocysteine scored one point but mention of pyridoxine response test and confirmatory tests such as enzyme or mutation analysis are appropriate and could be made.

Comments on overall performance:

Very good with 95% overall proficiency.

Sample B

Sample details:

These results were from a patient with LPI (lysineric protein intolerance). Plasma amino acids with selected urinary levels were provided.

Correct findings / abnormalities:

There was slightly elevated plasma glutamine with low levels of arginine and lysine together with elevated lysine, cystine and arginine in urine. This was considered correct. Mention of some, if not all of these abnormalities scored two points.

Correct Diagnosis and further tests:

LPI was correct (1 point). Further testing should include plasma ammonia, urinary orotic acid and confirmation by mutation analysis of the SLC7A7 gene (1 point). LDH/ferritin measurement was correctly recommended by ten labs.

Comments on overall performance:

Excellent with 99% overall proficiency.

Sample C

Sample details:

These results were from a patient with NKHG (non ketotic hyperglycinaemia). Plasma and urinary amino acid concentrations were provided.

Correct findings / abnormalities:

Glycine elevation in plasma and urine was scored with two points.

Correct Diagnosis and further tests:

NKHG was considered to be correct and scored one point but since the findings are not conclusive, valproic acid treatment or ketotic hyperglycinaemia are also possible. The key follow up is measurement of the ratio of glycine in CSF/ plasma. Urine organic acids are also helpful to exclude an organic aciduria. Confirmation by mutation analysis is appropriate

Comments on overall performance:

Excellent with 97% overall proficiency

Comments on the whole of the first circulation results

The overall proficiency was excellent but it must be borne in mind that each set of sample results was straightforward.

Although there was good agreement in scoring by all evaluators, especially for the analytical abnormalities, there was a good deal of discussion regarding scoring of diagnosis and recommendations for further testing. The need for more flexibility in scoring by using half points was considered and this will be tried out in the next circulation. Also we will endeavour to include more difficult cases

Also we are fortunate in that we will be able to use the ERNDIM website for distribution of case details and results submission.

We encourage participants to send us comments and suggestions regarding this scheme.

Date: 19.12.2017

The Scientific evaluators:

Rachel Carling, Maryanne Preece, Sabine Scholl and Brian Fowler

Lab I.D.	Sample 1-Scores			Sample 2-Scores			Sample 3-Scores		
	Abnormalities (2 pts)	Diag/further tests (2 pts)	Total	Abnormalities (2 pts)	Diag/further tests (2 pts)	Total	Abnormalities (2 pts)	Diag/further tests (2 pts)	Total
1	2	2	4	2	2	4	2	2	4
2	1	1	2	2	2	4	2	2	4
3	2	2	4	2	2	4	2	2	4
4	2	2	4	2	2	4	2	2	4
5	2	2	4	2	2	4	2	2	4
6	2	2	4	2	2	4	2	2	4
7	2	2	4	2	2	4	2	2	4
8	2	2	4	2	2	4	2	1	3
9	2	2	4	2	2	4	2	2	4
10	2	2	4	2	2	4	2	2	4
11	2	2	4	2	2	4	2	2	4
12	2	2	4	2	2	4	2	2	4
13	2	2	4	2	2	4	2	2	4
14	2	2	4	2	2	4	2	2	4
15	1	2	3	2	2	4	2	4	4
16	2	2	4	2	2	4	2	4	4
17	2	2	4	2	2	4	2	2	4
18	2	2	4	2	2	4	2	1	3
19	1	2	3	2	2	4	2	2	4
20	2	2	4	2	2	4	2	2	4
21	2	2	4	2	2	4	2	2	4
22	2	2	4	2	2	4	2	2	4
23	2	2	4	2	2	4	2	2	4
24	2	2	4	2	2	4	2	2	4
25	2	2	4	2	2	4	2	2	4
26	2	1	3	2	1	3	2	1	3
Overall proficiency			95%			99%			97%