

Supplemental Table2

Results of urine analysis by GC-MS at the time of diagnosis.

| Patient | GC-MS (Urine) | |
|---------|----------------|--|
| | Age at sampled | Results (normal range) |
| PA1 | 1 month | 3-Hydroxypropionic acid: 3.5 (0.0–1.1) |
| | | Phosphoric acid: 145.6 (0.0–43.0) |
| | | Hexanoylglycine: 2.3 (0.0) |
| | | Methylcitric acid (1): 6.5 (0.0–1.1) |
| | | Methylcitric acid (2): 5.8 (0.0–1.0) |
| PA2 | 6 months | Glycolic acid: 5.9 (0.0–2.2) |
| | | 3-Hydroxypropionic acid: 7.6 (0.0–1.1) |
| | | 3-Hydroxybutyric acid: 14.0 (0.0–3.7) |
| | | Benzoic acid: 70.9 (0.0–18.7) |
| | | 3-Hydroxyglutaric acid: 0.9 (0.0) |
| | | Hexanoylglycine: 2.5 (0.0) |
| | | Vanillic acid: 6.9 (0.0) |
| | | Citric acid: 1180.9 (31.4–572.0) |
| | | Methylcitric acid (1): 1.3 (0.0–1.1) |
| | | Methylcitric acid (2): 2.2 (0.0–1.0) |
| PA3 | NA | NA |
| PA4 | 1 month | Oxalic acid: 5.8 (0.0) |
| | | Fumaric acid: 12.1 (0.0–7.3) |
| | | Malic acid: 2.1 (0.0–0.7) |
| | | 5-Hydroxymethyl-2-furoic acid: 81.8 (0.0) |
| | | 2-Ketoglutaric acid (1): 184.0 (3.0–102.9) |
| | | Hexanoylglycine: 8.6 (0.0) |
| | | Methylcitric acid (1): 2.5 (0.0–1.1) |
| | | Methylcitric acid (2): 2.8 (0.0–1.0) |
| PA5 | 18 years | Methylcitric acid |
| | | 3-Hydroxypropionic acid |
| | | Propionylglycine |
| | | Tiglylglycine |
| MMA1 | 1 month | Methylmalonic acid |
| | | Pyruvic acid |
| | | Lactic acid |
| | | Ethylmalonic acid |
| MMA2 | 20 days | 3-Hydroxypropionic acid: 1.8 (0.0–1.1) |
| | | Pyruvic acid: 36.0 (0.0–24.1) |
| | | Methylmalonic acid: 135.7 (0.2–3.6) |
| | | Fumaric acid: 61.0 (0.0–7.3) |
| | | Malic acid: 7.0 (0.0–0.7) |
| | | Hexanoylglycine: 2.9 (0.0) |
| | | Vanillic acid: 12.3 (0.0) |
| | | Methylcitric acid (1): 3.2 (0.0–1.1) |
| | | Methylcitric acid (2): 3.0 (0.0–1.0) |
| MCG1 | NA | NA |