Table 1 : Clinical and investigation findings of the patients

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| --- | --- | --- | --- | --- |
| Patient | A | B | C | D |
| Age (months) / Sex | 12 /Female | 11 /Female | 18 /Male | 11/ Female |
| Consanguinity | - | 3rddegree | - | - |
| Presenting complaints | Acute gastroenteritis  Moderate dehydration  Acidotic breathing | Acute gastroenteritis  Moderate dehydration  Acidotic breathing Altered sensorium  Respiratory failure | Acute gastroenteritis  Moderate dehydration  Acidotic breathing Altered sensorium  Respiratory failure | Acute gastroenteritis  Moderate dehydration  Acidotic breathing Altered sensorium  Respiratory failure |
| Metabolic parameters | Increased anion gap metabolic acidosis  Ketonuria  No hypoglycemia  Lactate, Ammonia Normal | Increased anion gap metabolic acidosis  Ketonuria  Hypoglycemia  Lactate, Ammonia Normal | Increased anion gap metabolic acidosis  Ketonuria  No hypoglycemia  Lactate, Ammonia Normal | Increased anion gap metabolic acidosis  Ketonuria  No hypoglycemia  Lactate, Ammonia Normal |
| Other organ dysfunction | Nil | Acute renal injury  Optic atrophy | Acute renal injury | Acute renal injury |
| Acylcarnitines | C4OH elevated | C4OH elevated | Normal | C4OH, C5OH elevated |
| Urine GCMS | Elevated  Lactate  Ketone metabolites  2-hydroxy isovaleric acid  Tiglylglycine  3-OH Glutaric acid  4-hydroxy phenyllactic acid | Elevated  Lactate  Ketone metabolites  3-hydroxy isovaleric acid | Elevated  Lactate  Ketone metabolites  Glutarate | Elevated  Lactate  Ketone metabolites  2-hydroxy isovaleric acid |
| MRI | Not done | Abnormal | Abnormal | Not done |
| Genetics | Likely pathogenic Compound/Double heterozygous variant in ACAT1 gene | Likely pathogenic Homozygous variant in ACAT1 gene | Likely pathogenic Compound heterozygous variant in ACAT1 gene | Not done |
| Outcome | Neurologically preserved  On follow up since 1 yr | Severe neurological impairment Tracheostomy  Oxygen dependent  Death post discharge | Severe neurological impairment Tracheostomy  Oxygen dependent  Death in the hospital | Death following cardiac arrest |