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 gastroenteritisModerate dehydrationAcidotic breathing | Acute gastroenteritisModerate dehydrationAcidotic breathing Altered sensoriumRespiratory failure | Acute gastroenteritisModerate dehydrationAcidotic breathing Altered sensoriumRespiratory failure | Acute gastroenteritisModerate dehydrationAcidotic breathing Altered sensoriumRespiratory failure |
| Metabolic parameters | Increased anion gap metabolic acidosisKetonuriaNo hypoglycemiaLactate, Ammonia Normal | Increased anion gap metabolic acidosisKetonuriaHypoglycemia Lactate, Ammonia Normal  | Increased anion gap metabolic acidosisKetonuriaNo hypoglycemiaLactate, Ammonia Normal | Increased anion gap metabolic acidosisKetonuriaNo hypoglycemiaLactate, Ammonia Normal |
| Other organ dysfunction | Nil | Acute renal injuryOptic atrophy | Acute renal injury | Acute renal injury |
| Acylcarnitines  | C4OH elevated | C4OH elevated | Normal  | C4OH, C5OH elevated |
| Urine GCMS | Elevated LactateKetone metabolites2-hydroxy isovaleric acidTiglylglycine3-OH Glutaric acid 4-hydroxy phenyllactic acid | Elevated LactateKetone metabolites3-hydroxy isovaleric acid | Elevated LactateKetone metabolitesGlutarate | Elevated LactateKetone metabolites2-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 preservedOn follow up since 1 yr | Severe neurological impairment TracheostomyOxygen dependent Death post discharge | Severe neurological impairment TracheostomyOxygen dependent Death in the hospital | Death following cardiac arrest |