|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **IEM** | **No. Of pts**  **Table 1** | **acidosis** | | **ammonia** | **lactate** | **Homocysteine/**  **vit.b12** | **Csf/RBS ratio** | **TMS** | **GCMS** | **Exome sequencing** |
| Pyridoxine def. | 3 | | -  NO | Normal | Normal | Normal | Normal | Normal | Normal |  |
| GLUT1 DEFECT | 2 | | NO | Normal | Normal | Normal | <0.35 | Normal | Normal | Negative for slc1a2 |
| Biotinidase deficiency | 4 | | acidosis |  |  | Normal | Normal | High C5OH | Normal |  |
| Multiple Carboxylase def. | 1 | | acidosis |  |  | Normal | Normal | High C5OH | Abnormal |  |
| MMA | 4 | | acidosis | Borderline  or normal | Normal | High homocysteine  Low b12 | Normal | Normal | Excretion of malonic acid | MTHFR def |
| MSUD | 1 | | acidosis | High | Normal | Normal | Normal | High leucine,isoleucine and valine |  |  |