|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **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  | Borderlineor normal  | Normal  | High homocysteineLow b12  | Normal  | Normal  | Excretion of malonic acid  | MTHFR def  |
| MSUD  | 1  | acidosis  | High  | Normal  | Normal  | Normal  | High leucine,isoleucine and valine  |  |  |