Table 1: Demographics and biochemical results of the three patients around the time of diagnosis (Pre-T, prior to treatment; Post-T, after commencement of targeted treatment for MTHFR deficiency for at least one week).

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Demographics** | **Case 1** | | **Case 2** | | **Case 3** | |
| **Gender** | female | | male | | female | |
| **Age at metabolic diagnosis** | 2 years | | 12 years | | 10 years | |
| **Biochemical results** | Pre-T | Post-T | Pre-T | Post-T | Pre-T | Post-T |
| **Total Homocysteine**  (umol/L);  reference 3-8 | 149 | 55 | 86 | 97 | 330 | 72 |
| **Free Homocystine**  (umol/L); reference <5 | 30 | <5 | 5 | 5 | 36 | 5 |
| **Cystine**  (umol/L); reference 27 - 107 | <5 | 32 | 38 | 53 | 21 | 37 |
| **Methionine**  (umol/L); reference 5-77 | 5 | 32 | 34 | 35 | 7 | 35 |