**Supplementary Table S1. The MMACHC gene mutations of patients (N=64)**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Patient | MMACHC mutation1 | MMACHC mutation2 | Patient | MMACHC mutation1 | MMACHC mutation2 |
| 1 | c.80A>G | c.217C>T | 33 | c.482 G>A | c.609 G>A |
| 2 | c.482 G>A | c.609 G>A | 34 | c.609 G>A | c.658\_660 delAAG |
| 3 | c.80 A>G | c.609G>A | 35 | c.394 C>T | c.482 G>A |
| 4 | c.394 C>T | c.626\_627delTG | 36 | c.365 A>T | c.482 G>A |
| 5 | c.80 A>G | c.609G>A | 37 | c.482 G>A | c.609G>A |
| 6 | c.482G>A | c.658\_660delAAG | 38 | c.482 G>A | c.609 G>A |
| 7 | c.394C>T | c.609 G>A | 39 | c.217C>T | c.609 G>A |
| 8 | c.1 A>G | c.609G>A | 40 | c.609 G>A | c.217C>T |
| 9 | c.609G>A | c.609G>A | 41 | c.80 A>G | c.609 G>A |
| 10 | c.482G>A | c.482G>A | 42 | c.217C>T | c.658\_660 delAAG |
| 11 | c.217C>T | c.609 G>A | 43 | c.609G>A | c.482 G>A |
| 12 | c.80 A>G | c.609G>A | 44 | c.80 A>G | c.609G>A |
| 13 | c.482G>A | c.609G>A | 45 | c.80 A>G | c.658\_660 delAAG |
| 14 | c.609G>A | c.609G>A | 46 | c.80 A>G | c.609 G>A |
| 15 | c.80 A>G | c.609G>A | 47 | c.217C>T | c.609 G>A |
| 16 | / | c.445\_446 delTG | 48 | c.80 A>G | c.609G>A |
| 17 | c.609G>A | c.568 insert T | 49 | c.271dupA | c.482G>A |
| 18 | c.394C>T | c.568 insert T | 50 | c.609G>A | c.609G>A |
| 19 | c.349G>C | c.658\_660 delAAG | 51 | c.394 C>T | c.609G>A |
| 20 | c.567delT | c.609G>A | 52 | c.482G>A | c.609G>A |
| 21 | c.609G>A | c.658\_660 delAAG | 53 | c.394 C>T | c.609G>A |
| 22 | c.365A>T | c.658\_660 delAAG | 54 | c.567dupT | c.637G>T |
| 23 | c.482G>A | c.448A>C/c.449T>C | 55 | c.217C>T | c.609G>A |
| 24 | c.609 G>A | c.609 G>A | 56 | c.217C>T | c.394 C>T |
| 25 | c.609G>A | c.615C>A | 57 | c.609G>A | c.609G>A |
| 26 | c.440 G>A | c.482 G>A | 58 | c.609G>A | c.658\_660delAAG |
| 27 | c.365A>T | c.658\_660 delAAG | 59 | c.394C>T | c.568 insert T |
| 28 | c.467 G>A | c.609 G>A | 60 | c.80 A>G | c.394C>T |
| 29 | c.394C>T | c.568 insert T | 61 | c.609G>A | c.658\_660delAAG |
| 30 | c.80 A>G | c.609 G>A | 62 | c.217C>T | c.482G>A |
| 31 | c.365 A>T | c.609G>A | 63 | c.482G>A | c.568 insert T |
| 32 | c.482G>A | c.609 G>A | 64 | c.609G>A | c.609G>A |