|  |
| --- |
| Table 1. Imprinted loci in MLID BWS patients detected by HumanMethylation450 array (Illumina) |
| **Hypomethylated**  |
|   | MLID BWS1 | MLID BWS2 | MLID BWS3 | MLID BWS4 | MLID BWS5 | Number of patients |
| **KCNQ1OT1** | **●** | **●** | **●** | **●** | **●** | 5 |
| **DIRAS3** | ○ | **●** | ○ |  - | ○ | 4 |
| DIRAS3\_Ex2 | ○ | ○ | ○ |  - | ○ | 4 |
| **GNAS-AS1** | **●** | **●** | **●** |  - | **●** | 4 |
| GNAS-XL | ○ | ○ | ○ |  - | ○ | 4 |
| WRB |  - | ○ | ○ | ○ | ○ | 4 |
| IGF1R |  - |  - | ○ | ○ | ○ | 3 |
| FAM50B |  - | ○ | ○ |  - | ○ | 3 |
| FANCC |  - | ○ | ○ |  - | ○ | 3 |
| GNAS-A/B | ○ | ○ | ○ |  - |  - | 3 |
| NHP2L1 |  - | ○ | ○ |  - | ○ | 3 |
| **MEST** | **●** |  - |  - | **●** |  - | 2 |
| ERLIN2 |  - | ○ |  - |  - | ○ | 2 |
| MAGEL2 | ○ |  - |  - |  - | ○ | 2 |
| MCTS2P |  - |  - | ○ |  - | ○ | 2 |
| PPIEL |  - |  - |  - |  - | ○ | 1 |
| **GRB10** |  - |  - |  - |  - | **●** | 1 |
| PEG10 |  - |  - |  - | ○ |  - | 1 |
| RB1 | ○ |  - |  - |  - |  - | 1 |
| NDN |  - |  - |  - |  - | ○ | 1 |
| SNRPN\_5'DMR4 |  - |  - |  - |  - | ○ | 1 |
| SNRPN\_variant4 |  - |  - |  - |  - | ○ | 1 |
| **SNRPN/SNURF** |  - |  - |  - |  - | **●** | 1 |
| L3MBTL1 | ○ |  - |  - |  - |  - | 1 |
| **Hypermethylated**  |
| ZDBF2 | ○ | ○ | ○ | - | ○ | 4 |
| GNAS-NESP  |  - |  - | ○ |  - |  - | 1 |
| ● and ○ aberrant methylation detected by HumanMethylation array (Illumina)● previously known to be hypomethylated |  |  |