|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| 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 | | | | |  |  |