

Supplementary Materials:

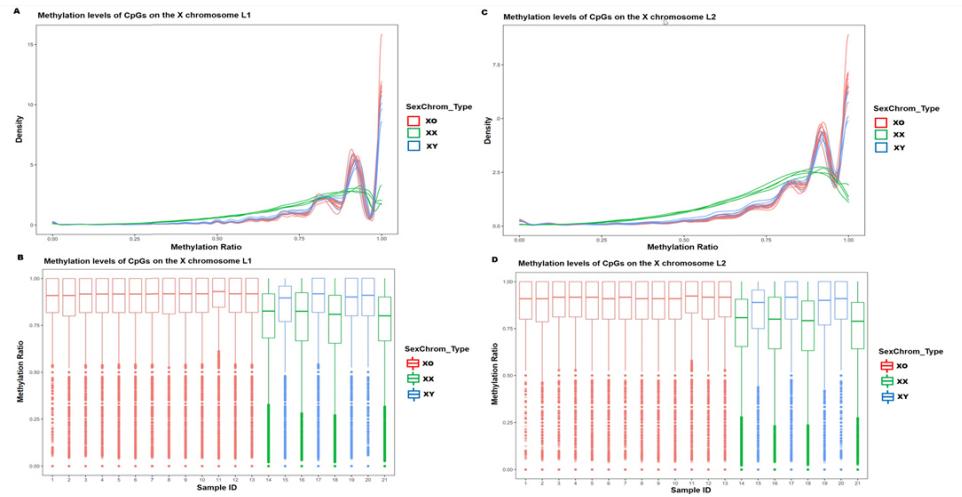


Figure S1. The methylation levels of 13 TS individuals and 8 healthy controls in the L1 and L2 regions on the chrX. The methylation density (A) and methylation ratio (B) of each sample in the L1 regions. The methylation density (C) and methylation ratio (D) of each sample in the L2 regions. (Red: TS, Green: female controls, and Blue: male controls).

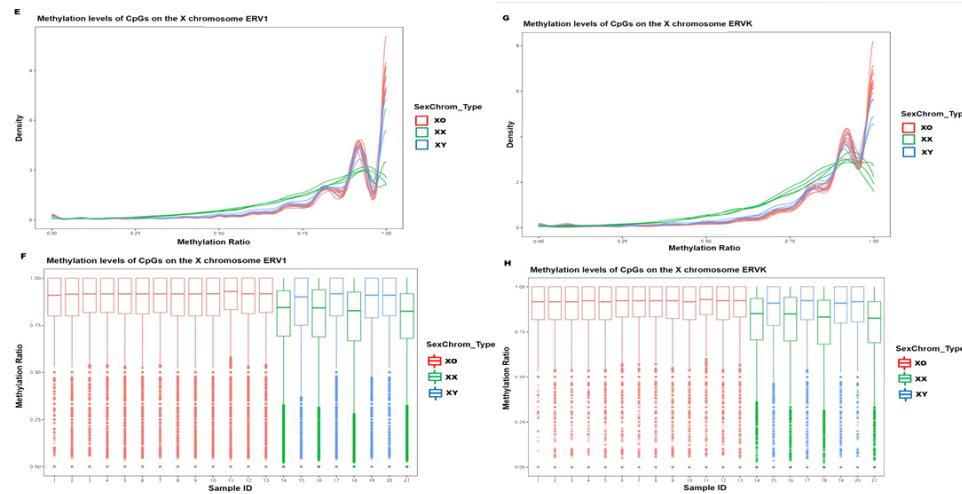


Figure S2. The methylation levels of 13 TS individuals and 8 healthy controls in the ERV1 and ERVK regions on the chrX. The methylation density (E) and methylation ratio (F) of each sample in the ERV1 regions. The methylation density (G) and methylation ratio (H) of each sample in the ERVK regions. (Red: TS, Green: female controls, and Blue: male controls).

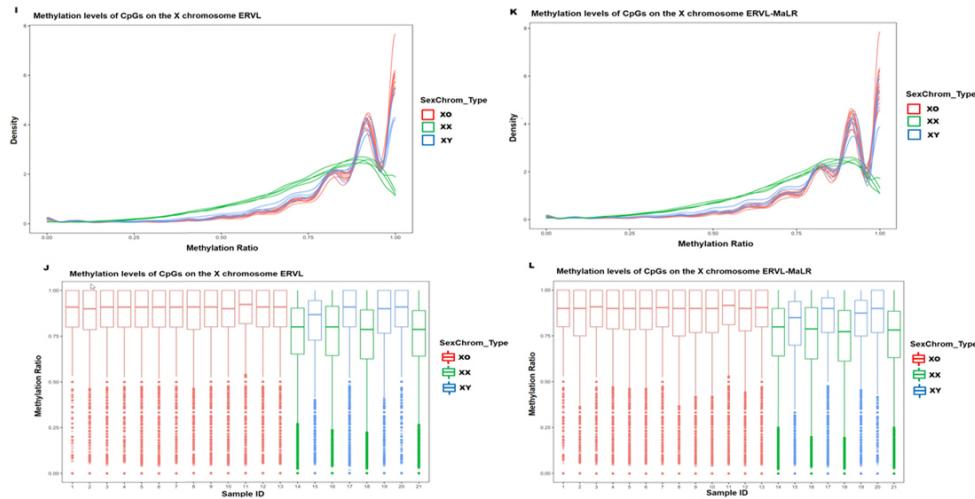


Figure S3. The methylation levels of 13 TS individuals and 8 healthy controls in the ERVL and ERVL–MaLR regions on the chrX. The methylation density (I) and methylation ratio (J) of each sample in ERVL regions. The methylation density (K) and methylation ratio (L) of each sample in the ERVL–MaLR regions. (Red: TS, Green: female controls, and Blue: male controls).

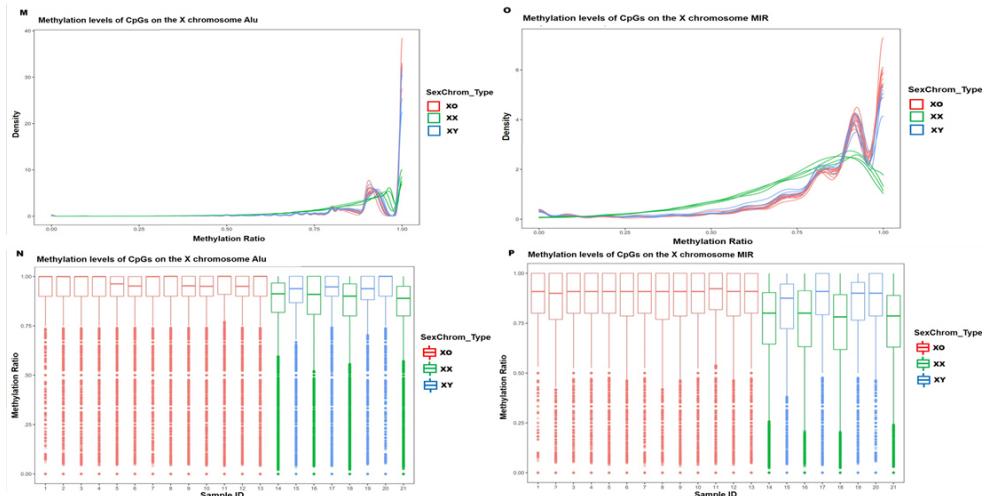


Figure S4. The methylation levels of 13 TS individuals and 8 healthy controls in the Alu and MIR regions on the chrX. The methylation density (M) and methylation ratio (N) of each sample in Alu regions. The methylation density (O) and methylation ratio (P) of each sample in the MIR regions. (Red: TS, Green: female controls, and Blue: male controls).

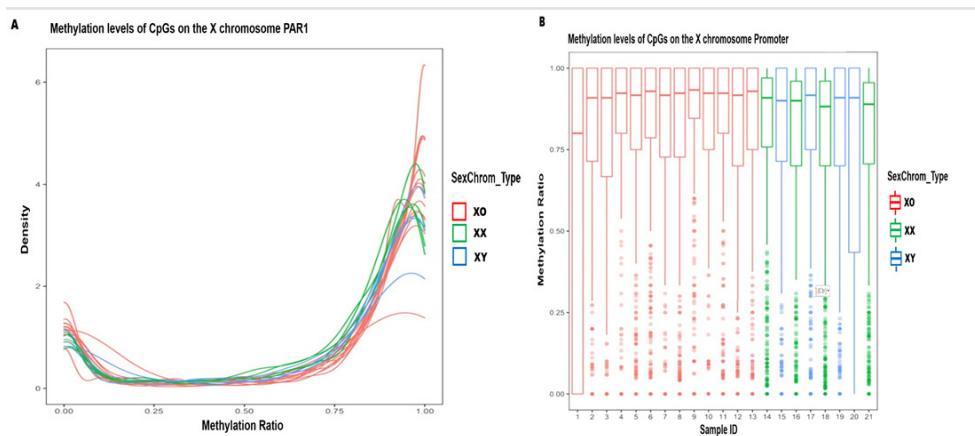


Figure S5. The methylation levels of 13 TS individuals and 8 healthy controls in the PAR1 regions on the chrX. The methylation density (A) and methylation ratio (B) of each sample in the PAR1

regions. (Red: TS, Green: female controls, and Blue: male controls).

Table S1. The phenotypes and genetic information of patients with 45X0 Turner syndrome.

| | Cardiac malformation | | | | | | | | | | | | | 4/13 | |
|--|----------------------|---|---|---|---|---|---|---|---|---|---|---|---|------|------|
| Bicuspid aortic valve (HP:0001647) | - | - | - | - | + | - | - | - | - | - | + | - | - | + | 3/13 |
| Aortic root dilation (HP:0002616) | - | + | - | - | + | - | - | - | - | - | + | - | - | - | 3/13 |
| Renal malformation | | | | | | | | | | | | | | 4/13 | |
| Medullary nephrocalcinosis (HP:0012408) | - | - | - | - | - | - | - | - | - | - | + | - | - | - | 1/13 |
| Horseshoe kidney (HP:0000085) | - | - | - | - | - | - | - | - | - | - | - | - | + | + | 2/13 |
| Hydronephrosis (HP:0000126) | - | - | - | - | - | - | - | - | - | - | - | - | + | - | 1/13 |
| Nephrolithiasis (HP:0000787) | - | + | - | - | - | - | - | - | - | - | - | - | - | - | 1/13 |
| Specific learning disability (HP:0001328) | + | + | - | - | - | - | - | - | - | - | - | - | - | - | 2/13 |
| Hypothyroidism (HP:0000821) | - | + | + | - | - | - | - | - | - | - | - | - | - | - | 2/13 |

* Origin of chrX: M, maternal origin and P, paternal origin.

Table S2. Differentially methylated genes identified among the three groups of Turner syndrome 45X0 and controls 46XY and 46XX.

| Gene | Set | Number |
|-----------------|------------|--------|
| CNTNAP3 | XO/XXhyper | 8 |
| BCOR | XO/XXhyper | |
| AC004870.3 | XO/XXhyper | |
| AK2 | XO/XXhyper | |
| MYT1L | XO/XXhyper | |
| OPCML | XO/XXhyper | |
| GRK1 | XO/XXhyper | |
| RP11-453F18_B.1 | XO/XXhyper | |
| MVB12B | XO/XXhypo | 117 |
| CXorf40A | XO/XXhypo | |
| RPL10 | XO/XXhypo | |
| RP4-537K23.4 | XO/XXhypo | |
| NDUFA1 | XO/XXhypo | |
| VMA21 | XO/XXhypo | |
| KLF8 | XO/XXhypo | |
| RPL36A | XO/XXhypo | |
| RPL36A-HNRNPH2 | XO/XXhypo | |
| HUWE1 | XO/XXhypo | |
| TBC1D25 | XO/XXhypo | |
| AF196779.12 | XO/XXhypo | |
| WDR45 | XO/XXhypo | |
| ZNF630 | XO/XXhypo | |
| PIM2 | XO/XXhypo | |
| F8 | XO/XXhypo | |
| RP11-85L21.4 | XO/XXhypo | |
| SYP | XO/XXhypo | |
| TIMM17B | XO/XXhypo | |
| IKBKG | XO/XXhypo | |
| NUDT10 | XO/XXhypo | |
| TMEM255A | XO/XXhypo | |
| ELK1 | XO/XXhypo | |
| FAM127B | XO/XXhypo | |
| CCDC22 | XO/XXhypo | |
| TMEM185A | XO/XXhypo | |
| BTBD11 | XO/XXhypo | |
| WDR13 | XO/XXhypo | |
| ESX1 | XO/XXhypo | |

| | |
|--------------|-----------|
| PHF8 | XO/XXhypo |
| ZNF674-AS1 | XO/XXhypo |
| PNCK | XO/XXhypo |
| MAGED2 | XO/XXhypo |
| KIAA2022 | XO/XXhypo |
| WNK3 | XO/XXhypo |
| G6PD | XO/XXhypo |
| CHST7 | XO/XXhypo |
| ARHGEF9 | XO/XXhypo |
| TAF9B | XO/XXhypo |
| TM4SF2 | XO/XXhypo |
| TSPAN7 | XO/XXhypo |
| EIF2AK4 | XO/XXhypo |
| BCORL1 | XO/XXhypo |
| PCDH19 | XO/XXhypo |
| RP3-326L13.2 | XO/XXhypo |
| SCML1 | XO/XXhypo |
| CACNA1F | XO/XXhypo |
| WAS | XO/XXhypo |
| GPR50-AS1 | XO/XXhypo |
| RP4-769N13.6 | XO/XXhypo |
| RP4-769N13.7 | XO/XXhypo |
| RAB33A | XO/XXhypo |
| SYN1 | XO/XXhypo |
| ATP6AP1 | XO/XXhypo |
| TRMT2B | XO/XXhypo |
| TRMT2B-AS1 | XO/XXhypo |
| HS6ST2 | XO/XXhypo |
| TIMM8A | XO/XXhypo |
| MORC4 | XO/XXhypo |
| MAGIX | XO/XXhypo |
| IRAK1 | XO/XXhypo |
| DLG3 | XO/XXhypo |
| OTUD5 | XO/XXhypo |
| AR | XO/XXhypo |
| AC115618.1 | XO/XXhypo |
| HAUS7 | XO/XXhypo |
| NHS | XO/XXhypo |
| MAGT1 | XO/XXhypo |
| NUDT11 | XO/XXhypo |
| NDUFB11 | XO/XXhypo |
| PTCHD1 | XO/XXhypo |
| CROCC | XO/XXhypo |
| U1 | XO/XXhypo |
| BHLHB9 | XO/XXhypo |
| HDAC6 | XO/XXhypo |
| EBP | XO/XXhypo |
| LINC00086 | XO/XXhypo |
| LONRF3 | XO/XXhypo |
| ZBTB33 | XO/XXhypo |
| BEX4 | XO/XXhypo |
| ELF4 | XO/XXhypo |
| NKAPP1 | XO/XXhypo |
| TAZ | XO/XXhypo |
| RP11-66N11.8 | XO/XXhypo |
| ZNF41 | XO/XXhypo |
| HMGB3 | XO/XXhypo |
| EMD | XO/XXhypo |
| SLC25A5-AS1 | XO/XXhypo |
| STARD8 | XO/XXhypo |
| MID1IP1 | XO/XXhypo |
| MOSPD1 | XO/XXhypo |
| FLNA | XO/XXhypo |
| LANCL3 | XO/XXhypo |
| CXorf40B | XO/XXhypo |
| MAGEE1 | XO/XXhypo |

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|-----------------|------------|-----|
| TAF7L | XO/XXhypo | |
| RAI2 | XO/XXhypo | |
| SAMD11 | XO/XXhypo | |
| SLC25A5 | XO/XXhypo | |
| ZMYM3 | XO/XXhypo | |
| BEX1 | XO/XXhypo | |
| SRPX | XO/XXhypo | |
| IDS | XO/XXhypo | |
| ARHGAP6 | XO/XXhypo | |
| UBL4A | XO/XXhypo | |
| RBMX2 | XO/XXhypo | |
| PRRG1 | XO/XXhypo | |
| PGK1 | XO/XXhypo | |
| C1GALT1C1 | XO/XXhypo | |
| AFF2 | XO/XXhypo | |
| FMR1 | XO/XXhypo | |
| LINC00629 | XO/XXhypo | |
| SRPK3 | XO/XXhypo | |
| IL13RA1 | XO/XXhypo | |
| DOCK11 | XO/XXhypo | |
| HOOK2 | XO/XXhypo | |
| DNASE1L1 | XO/XXhypo | |
| HOXB3 | XO/XYhyper | 3 |
| HOXB-AS3 | XO/XYhyper | |
| DTX2 | XO/XYhyper | |
| TAF1 | XO/XYhypo | |
| MVB12B | XO/XYhypo | |
| TNS1 | XO/XYhypo | |
| AC093609.1 | XO/XYhypo | 16 |
| MPL | XO/XYhypo | |
| EIF2AK4 | XO/XYhypo | |
| FOXI2 | XO/XYhypo | |
| BTBD11 | XO/XYhypo | |
| HOXA3 | XO/XYhypo | |
| HOXA-AS2 | XO/XYhypo | |
| HOXA4 | XO/XYhypo | |
| RP1-170O19.22 | XO/XYhypo | |
| SPRN | XO/XYhypo | |
| CYP2E1 | XO/XYhypo | |
| NAPRT1 | XO/XYhypo | |
| CTU1 | XO/XYhypo | |
| BCOR | XY/XXhyper | 8 |
| TAF1 | XY/XXhyper | |
| RP11-453F18_B.1 | XY/XXhyper | |
| LMTK3 | XY/XXhyper | |
| AK2 | XY/XXhyper | |
| SPRN | XY/XXhyper | |
| CYP2E1 | XY/XXhyper | |
| AC093609.1 | XY/XXhyper | 121 |
| DNASE1L1 | XY/XXhypo | |
| FAM156B | XY/XXhypo | |
| DOCK11 | XY/XXhypo | |
| AMMECR1 | XY/XXhypo | |
| SSR4 | XY/XXhypo | |
| FZD1 | XY/XXhypo | |
| BEX1 | XY/XXhypo | |
| ARX | XY/XXhypo | |
| EFNB1 | XY/XXhypo | |
| C1GALT1C1 | XY/XXhypo | |
| FAM157C | XY/XXhypo | |
| GPC3 | XY/XXhypo | |
| TSPAN6 | XY/XXhypo | |
| UBL4A | XY/XXhypo | |
| FMR1 | XY/XXhypo | |
| HS6ST2 | XY/XXhypo | |
| FAM156A | XY/XXhypo | |

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|----------------|------------|-----------|
| | IDS | XY/XXhypo |
| | AFF2 | XY/XXhypo |
| | RBMX2 | XY/XXhypo |
| | NHS | XY/XXhypo |
| | SLITRK4 | XY/XXhypo |
| | SLC25A5 | XY/XXhypo |
| | PRRG1 | XY/XXhypo |
| | TM4SF2 | XY/XXhypo |
| | PABPC1L2A | XY/XXhypo |
| | KIAA2022 | XY/XXhypo |
| | SRPX | XY/XXhypo |
| | STARD8 | XY/XXhypo |
| | RAI2 | XY/XXhypo |
| | PGK1 | XY/XXhypo |
| | LANCL3 | XY/XXhypo |
| | IKBKG | XY/XXhypo |
| RP11-66N11.8 | | XY/XXhypo |
| | FLNA | XY/XXhypo |
| | ARHGAP6 | XY/XXhypo |
| | CXorf40B | XY/XXhypo |
| | MOSPD1 | XY/XXhypo |
| | WNK3 | XY/XXhypo |
| | MID1IP1 | XY/XXhypo |
| | NUDT11 | XY/XXhypo |
| | HMGB3 | XY/XXhypo |
| | TAF7L | XY/XXhypo |
| | PCDH19 | XY/XXhypo |
| SLC25A5-AS1 | | XY/XXhypo |
| | ZBTB33 | XY/XXhypo |
| | ZNF41 | XY/XXhypo |
| RP4-537K23.4 | | XY/XXhypo |
| | EMD | XY/XXhypo |
| | SOWAHD | XY/XXhypo |
| | LONRF3 | XY/XXhypo |
| | BCORL1 | XY/XXhypo |
| | ZMYM3 | XY/XXhypo |
| | TAZ | XY/XXhypo |
| | EBP | XY/XXhypo |
| | NKAPP1 | XY/XXhypo |
| | ELF4 | XY/XXhypo |
| | HDAC6 | XY/XXhypo |
| | LINC00086 | XY/XXhypo |
| | GPR50-AS1 | XY/XXhypo |
| | NDUFB11 | XY/XXhypo |
| | PTCHD1 | XY/XXhypo |
| | PHF8 | XY/XXhypo |
| | BHLHB9 | XY/XXhypo |
| | WDR45 | XY/XXhypo |
| RPL36A-HNRNPH2 | | XY/XXhypo |
| | AC115618.1 | XY/XXhypo |
| | HAUS7 | XY/XXhypo |
| | OTUD5 | XY/XXhypo |
| | MAGT1 | XY/XXhypo |
| | DLG3 | XY/XXhypo |
| | MAGIX | XY/XXhypo |
| | IRAK1 | XY/XXhypo |
| | MORC4 | XY/XXhypo |
| | AR | XY/XXhypo |
| | ZNF630 | XY/XXhypo |
| | TIMM8A | XY/XXhypo |
| RP3-326L13.2 | | XY/XXhypo |
| | SYN1 | XY/XXhypo |
| | TRMT2B | XY/XXhypo |
| TRMT2B-AS1 | | XY/XXhypo |
| | ZNF75D | XY/XXhypo |
| | ATP6AP1 | XY/XXhypo |

| | |
|--------------|-----------|
| RP4-769N13.6 | XY/XXhypo |
| RP4-769N13.7 | XY/XXhypo |
| WAS | XY/XXhypo |
| NUDT10 | XY/XXhypo |
| CACNA1F | XY/XXhypo |
| RAB33A | XY/XXhypo |
| SCML1 | XY/XXhypo |
| TSPAN7 | XY/XXhypo |
| CHST7 | XY/XXhypo |
| HUWE1 | XY/XXhypo |
| G6PD | XY/XXhypo |
| ZNF674-AS1 | XY/XXhypo |
| MAGED2 | XY/XXhypo |
| SYP | XY/XXhypo |
| WDR13 | XY/XXhypo |
| TAF9B | XY/XXhypo |
| PNMA6A | XY/XXhypo |
| ARHGEF9 | XY/XXhypo |
| CCDC22 | XY/XXhypo |
| FAM127B | XY/XXhypo |
| TMEM185A | XY/XXhypo |
| ESX1 | XY/XXhypo |
| TMEM255A | XY/XXhypo |
| PNCK | XY/XXhypo |
| ELK1 | XY/XXhypo |
| TIMM17B | XY/XXhypo |
| F8 | XY/XXhypo |
| RP11-85L21.4 | XY/XXhypo |
| PIM2 | XY/XXhypo |
| AF196779.12 | XY/XXhypo |
| TBC1D25 | XY/XXhypo |
| RPL36A | XY/XXhypo |
| RPL10 | XY/XXhypo |
| CXorf40A | XY/XXhypo |
| VMA21 | XY/XXhypo |
| KLF8 | XY/XXhypo |
| NDUFA1 | XY/XXhypo |
| SRPK3 | XY/XXhypo |

Table S3. Enriched GO and pathway lists.

| Category | Category ID | GO | Description | PARENT_GO | LogP | Enrichment | Z-score | #TotalGen eInLibrary | #GeneI nGO | #GeneIn HitList | #GeneIn GOAnd HitList | STDV %InGO | GeneID | Hits | Log(q-value) | Gene List | |
|-------------------------|-------------|------------|--|---|------|------------|---------|-------------------------|---------------|--------------------|-----------------------------|---------------|--------|--|---|--------------|--------|
| GO Biological Processes | 19 | GO:000423 | mitophagy | 19_GO:0008152 metabolic process | -5.2 | 33 | 11 | 30,242 | 28 | 132 | 4 | 3 | 1.5 | 292 6901 10013 10075 | SLC25A5 TAFAZZIN HDAC6 HUWE1 | -0.82 | MyList |
| GO Biological Processes | 19 | GO:009042 | tubulin deacetylation | 19_GO:0008152 metabolic process | -4.6 | 53 | 12 | 30,242 | 13 | 132 | 3 | 2.3 | 1.3 | 2316 10013 5627 1 | FLNA HDAC6 BEX4 | -0.78 | MyList |
| GO Biological Processes | 19 | GO:000422 | autophagy of mitochondrion | 19_GO:0008152 metabolic process | -4.5 | 14 | 7.8 | 30,242 | 81 | 132 | 5 | 3.8 | 1.7 | 292 6901 10013 10075 11152 | SLC25A5 TAFAZZIN HDAC6 HUWE1 WD R45 | -0.78 | MyList |
| GO Biological Processes | 19 | GO:0061726 | mitochondrion disassembly | 19_GO:0009987 cellular process | -4.5 | 14 | 7.8 | 30,242 | 81 | 132 | 5 | 3.8 | 1.7 | 292 6901 10013 10075 11152 | SLC25A5 TAFAZZIN HDAC6 HUWE1 WD R45 | -0.78 | MyList |
| GO Biological Processes | 19 | GO:0051090 | regulation of DNA-binding transcription factor activity | 19_GO:0050789 regulation of biological process | -3.9 | 4.7 | 5.2 | 30,242 | 440 | 132 | 9 | 6.8 | 2.2 | 367 2316 3654 16 872 8321 8517 12 8952 55859 4402 75 | AR FLNA IRAK1 TA F1 FZD1 IKBKG CCDC C22 BEX1 EIF2AK4 | -0.25 | MyList |
| GO Biological Processes | 19 | GO:1903008 | organelle disassembly | 19_GO:0009987 cellular process | -3.8 | 10 | 6.4 | 30,242 | 114 | 132 | 5 | 3.8 | 1.7 | 292 6901 10013 10075 11152 | SLC25A5 TAFAZZIN HDAC6 HUWE1 WD R45 | -0.25 | MyList |
| GO Biological Processes | 19 | GO:0043123 | positive regulation of I-kappaB kinase/NF-kappaB signaling | 19_GO:0048518 positive regulation of biological process | -3.8 | 7.4 | 5.8 | 30,242 | 186 | 132 | 6 | 4.5 | 1.8 | 2316 3654 7105 18 8517 11040 2895 6 10075 11152 22 | FLNA IRAK1 TSPAN 8 IKBKG PIM2 CCDC 22 | -0.25 | MyList |
| GO Biological Processes | 19 | GO:0061912 | selective autophagy | 19_GO:0008152 metabolic process | -3.6 | 13 | 6.8 | 30,242 | 69 | 132 | 4 | 3 | 1.5 | 292 6901 10013 10075 | SLC25A5 TAFAZZIN HDAC6 HUWE1 | -0.16 | MyList |
| GO Biological Processes | 19 | GO:0016236 | macroautophagy | 19_GO:0008152 metabolic process | -3.5 | 5.4 | 5.1 | 30,242 | 295 | 132 | 7 | 5.3 | 2 | 292 4943 6901 18 517 10013 10075 11152 | SLC25A5 TBC1D25 T AC6 HUWE1 WDR45 | -0.08 | MyList |
| GO Biological Processes | 19 | GO:0007005 | mitochondrion organization | 19_GO:0009987 cellular process | -3.4 | 4.1 | 4.6 | 30,242 | 509 | 132 | 9 | 6.8 | 2.2 | 292 1678 4694 16 901 10013 10075 10245 11152 54 | SLC25A5 TIMM8A NDUFB11 DUFA1 TAFAZZIN H DAC6 HUWE1 TIMM 539 17B WDR45 NDUFB11 | -0.056 | MyList |
| WikiPathways | 27 | WP2858 | Ectoderm differentiation | | -3.4 | 8 | 5.5 | 30,242 | 144 | 132 | 5 | 3.8 | 1.7 | 10013 23133 232 29 54880 170302 | HDAC6 PHF8 ARHG EF9 BCOR ARX | -0.056 | MyList |

| | | | | | | | | | | | | | | | | |
|-------------------------|----|---------------|---|--|------|-----|-----|--------|-----|-----|----|-----|-----|--|--------|--------|
| | | | | | | | | | | | | | | | | |
| GO Biological Processes | 19 | GO:0046777 | protein autophosphorylation | 19_GO:0008152 metabolic process | -3.3 | 6.1 | 5.1 | 30,242 | 227 | 132 | 6 | 4.5 | 1.8 | 3654 6011 6872 11040 65267 440275 IRAK1 GRK1 TAF1 PI M2 WNK3 EIF2AK4 | -0.056 | MyList |
| GO Biological Processes | 19 | GO:0061919 | process utilizing autophagic mechanism | 19_GO:0009987 cellular process | -3.3 | 3.9 | 4.5 | 30,242 | 528 | 132 | 9 | 6.8 | 2.2 | 292 4943 6901 8 SLC25A5 TBC1D25 T 406 8517 10013 AFAZZIN SRPX IKBK 10075 11040 111 G HDAC6 HUWE1 PI 52 M2 WDR45 | -0.056 | MyList |
| GO Biological Processes | 19 | GO:0006914 | autophagy | 19_GO:0008152 metabolic process | -3.3 | 3.9 | 4.5 | 30,242 | 528 | 132 | 9 | 6.8 | 2.2 | 292 4943 6901 8 SLC25A5 TBC1D25 T 406 8517 10013 AFAZZIN SRPX IKBK 10075 11040 111 G HDAC6 HUWE1 PI 52 M2 WDR45 | -0.056 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9609507 | Protein localization | | -3.1 | 7 | 5.1 | 30,242 | 163 | 132 | 5 | 3.8 | 1.7 | 1678 2010 6901 8266 10245 TIMM8A EMD TAFA ZZIN UBL4A TIMM17B | 0 | MyList |
| GO Biological Processes | 19 | GO:0043122 | regulation of I-kappaB kinase/NF-kappaB signaling | 19_GO:0023052 signaling | -3.1 | 5.5 | 4.7 | 30,242 | 249 | 132 | 6 | 4.5 | 1.8 | 2316 3654 7105 FLNA IRAK1 TSPAN 8517 11040 2895 6 IKBKG PIM2 CCDC 2 22 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9012999 | RHO GTPase cycle | | -3.1 | 4.1 | 4.4 | 30,242 | 449 | 132 | 8 | 6.1 | 2.1 | 395 537 2010 48 10 7454 9754 23 229 139818 ARHGAP6 ATP6AP1 EMD NHS WAS STA RD8 ARHGEF9 DOC K11 | 0 | MyList |
| KEGG Pathway | 24 | ko03022 | Basal transcription factors | | -3 | 15 | 6.3 | 30,242 | 45 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 TAF1 TAF9B TAF7L | 0 | MyList |
| GO Biological Processes | 19 | GO:0051091 | positive regulation of DNA-binding transcription factor activity | 19_GO:0050789 regulation of biological process | -3 | 5.3 | 4.6 | 30,242 | 260 | 132 | 6 | 4.5 | 1.8 | 367 3654 6872 8 AR IRAK1 TAF1 FZD 321 8517 55859 1 IKBKG BEX1 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-194315 | Signaling by Rho GTPases | | -3 | 3.2 | 4 | 30,242 | 707 | 132 | 10 | 7.6 | 2.3 | 367 395 537 201 0 2316 4810 745 AP1 EMD FLNA NH 4 9754 23229 13 S WAS STARD8 ARH 9818 GEF9 DOCK11 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-76042 | RNA Polymerase II Transcription Initiation And Promoter Clearance | | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 TAF1 TAF9B TAF7L | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-73779 | RNA Polymerase II Transcription Pre-Initiation And Promoter Opening | | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 TAF1 TAF9B TAF7L | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-73776 | RNA Polymerase II Promoter Escape | | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 TAF1 TAF9B TAF7L | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-75953 | RNA Polymerase II Transcription Initiation | | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 TAF1 TAF9B TAF7L | 0 | MyList |

| | | | | | | | | | | | | | | | | | |
|-------------------------|----|---------------|--|---------------|------|-----|--------|--------|-----|-----|-----|-----|---|---|--|--------|--------|
| Reactome Gene Sets | 6 | R-HSA-167162 | RNA Polymerase II HIV Promoter Escape | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 | TAF1 TAF9B TAF7L | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-167161 | HIV Transcription Initiation | -2.9 | 15 | 6.2 | 30,242 | 47 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 | TAF1 TAF9B TAF7L | 0 | MyList | |
| KEGG Pathway | 24 | hsa0302 | Basal transcription factors | -2.9 | 14 | 6.1 | 30,242 | 48 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 | TAF1 TAF9B TAF7L | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-9716542 | Signaling by Rho GTPases, Miro GTPases and RHOBTB3 | -2.9 | 3.2 | 3.9 | 30,242 | 723 | 132 | 10 | 7.6 | 2.3 | 3671 3951 5371 201 012316 4810 745 419754 123229 13 9818 | AR ARHGAP6 ATP6 AP1 EMD FLNA NH S WAS STARD8 ARH GEF9 DOCK11 | 0 | MyList | |
| GO Biological Processes | 19 | GO:0043433 | negative regulation of DNA-binding transcription factor activity | 19_GO:0050789 | -2.9 | 6.2 | 4.7 | 30,242 | 185 | 132 | 5 | 3.8 | 1.7 | 2316 3654 6872 28952 440275 | FLNA IRAK1 TAF1 C CDC22 EIF2AK4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-1793185 | Chondroitin sulfate/dermatan sulfate metabolism | -2.9 | 14 | 6 | 30,242 | 50 | 132 | 3 | 2.3 | 1.3 | 2719 3423 56548 | GPC3 IDS CHST7 | 0 | MyList | |
| GO Biological Processes | 19 | GO:0071478 | cellular response to radiation | 19_GO:0050896 | -2.9 | 6.1 | 4.7 | 30,242 | 187 | 132 | 5 | 3.8 | 1.7 | 2002 2332 6011 6872 440275 | ELK1 FMR1 GRK1 T AF1 EIF2AK4 | 0 | MyList |
| GO Biological Processes | 19 | GO:0009896 | positive regulation of catabolic process | 19_GO:0048518 | -2.8 | 3.7 | 4 | 30,242 | 493 | 132 | 8 | 6.1 | 2.1 | 292 2332 2719 6 872 8517 10075 11040 28952 | SLC25A5 FMR1 GPC3 TAF1 IKBKG HUWE 1 PIM2 CCDC22 | 0 | MyList |
| GO Biological Processes | 19 | GO:0007249 | I-kappaB kinase/NF-kappaB signaling | 19_GO:0023052 | -2.8 | 4.9 | 4.3 | 30,242 | 281 | 132 | 6 | 4.5 | 1.8 | 2316 3654 7105 8517 11040 28956 2 | FLNA IRAK1 TSPAN IKBKG PIM2 CCDC 22 | 0 | MyList |
| GO Biological Processes | 19 | GO:0000300 | regulation of synaptic vesicle exocytosis | 19_GO:0051179 | -2.8 | 13 | 5.7 | 30,242 | 54 | 132 | 3 | 2.3 | 1.3 | 2332 6853 6855 | FMR1 SYN1 SYP | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-1638091 | Heparan sulfate/heparin (HS-GAG) metabolism | -2.7 | 12 | 5.7 | 30,242 | 55 | 132 | 3 | 2.3 | 1.3 | 2719 3423 90161 | GPC3 IDS HS6ST2 | 0 | MyList | |
| GO Biological Processes | 19 | GO:0006360 | transcription by RNA polymerase I | 19_GO:0008152 | -2.7 | 12 | 5.7 | 30,242 | 55 | 132 | 3 | 2.3 | 1.3 | 2316 6872 23133 | FLNA TAF1 PHF8 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-71387 | Metabolism of carbohydrates | -2.7 | 4.7 | 4.2 | 30,242 | 295 | 132 | 6 | 4.5 | 1.8 | 2539 2719 3423 5230 56548 9016 1 | G6PD GPC3 IDS PGK 1 CHST7 HS6ST2 | 0 | MyList | |

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|-------------------------|----|---------------|--|---|------|-----|-----|--------|-----|-----|---|-----|-----|---|--|---|--------|
| GO Biological Processes | 19 | GO:0010257 | NADH dehydrogenase complex assembly | 19_GO:0009987 cellular process | -2.7 | 12 | 5.5 | 30,242 | 57 | 132 | 3 | 2.3 | 1.3 | 4694 6901 54539 | NDUFA1 TAFAZZIN NDUFB11 | 0 | MyList |
| GO Biological Processes | 19 | GO:0032981 | mitochondrial respiratory chain complex I assembly | 19_GO:0009987 cellular process | -2.7 | 12 | 5.5 | 30,242 | 57 | 132 | 3 | 2.3 | 1.3 | 4694 6901 54539 | NDUFA1 TAFAZZIN NDUFB11 | 0 | MyList |
| GO Biological Processes | 19 | GO:0071482 | cellular response to light stimulus | 19_GO:0050896 response to stimulus | -2.7 | 7.4 | 4.7 | 30,242 | 124 | 132 | 4 | 3 | 1.5 | 2332 6011 6872 440275 | FMR1 GRK1 TAF1 EI F2AK4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-1630316 | Glycosaminoglycan metabolism | | -2.7 | 7.4 | 4.7 | 30,242 | 124 | 132 | 4 | 3 | 1.5 | 2719 3423 56548 | GPC3 IDS CHST7 HS 190161 6ST2 | 0 | MyList |
| GO Biological Processes | 19 | GO:0050808 | synapse organization | 19_GO:0009987 cellular process | -2.6 | 3.8 | 3.8 | 30,242 | 426 | 132 | 7 | 5.3 | 2 | 1741 2316 6853 8321 10013 139065 | DLG3 FLNA SYN1 F 8321 10013 8082 ZD1 HDAC6 BHLHB9 3 SLTRK4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-1268020 | Mitochondrial protein import | | -2.6 | 11 | 5.2 | 30,242 | 64 | 132 | 3 | 2.3 | 1.3 | 1678 6901 10245 | TIMM8A TAFAZZIN TIMM17B | 0 | MyList |
| Canonical Pathways | 11 | M34 | PID TCR PATHWAY | | -2.6 | 11 | 5.2 | 30,242 | 64 | 132 | 3 | 2.3 | 1.3 | 2316 7454 8517 | FLNA WAS IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-450294 | MAP kinase activation | | -2.6 | 11 | 5.2 | 30,242 | 64 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList |
| GO Biological Processes | 19 | GO:0009416 | response to light stimulus | 19_GO:0050896 response to stimulus | -2.5 | 4.3 | 3.9 | 30,242 | 321 | 132 | 6 | 4.5 | 1.8 | 778 2002 2332 011 6872 440275 | CACNA1F ELK1 FMR 1 GRK1 TAF1 EIF2AK 4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-162599 | Late Phase of HIV Life Cycle | | -2.5 | 6.6 | 4.4 | 30,242 | 139 | 132 | 4 | 3 | 1.5 | 6872 51616 5445 | TAF1 TAF9B TAF7L 7 89853 MVB12B | 0 | MyList |
| Canonical Pathways | 11 | M153 | PID P75 NTR PATHWAY | | -2.5 | 10 | 5 | 30,242 | 68 | 132 | 3 | 2.3 | 1.3 | 3654 8517 55859 | IRAK1 IKBKG BEX1 | 0 | MyList |
| GO Biological Processes | 19 | GO:0051129 | negative regulation of cellular component organization | 19_GO:0048519 negative regulation of biological process | -2.5 | 3 | 3.5 | 30,242 | 691 | 132 | 9 | 6.8 | 2.2 | 292 367 395 231 6 7454 10013 10 075 23133 58526 | SLC25A5 AR ARHGA P6 FLNA WAS HDA C6 HUWE1 PHF8 MI D1IP1 | 0 | MyList |
| GO Biological Processes | 19 | GO:0016241 | regulation of macroautophagy | 19_GO:0050789 regulation of biological process | -2.5 | 6.5 | 4.3 | 30,242 | 141 | 132 | 4 | 3 | 1.5 | 292 4943 8517 1 0013 | SLC25A5 TBC1D25 IK BKG HDAC6 | 0 | MyList |
| GO Biological Processes | 19 | GO:0071214 | cellular response to abiotic stimulus | 19_GO:0050896 response to stimulus | -2.5 | 4.1 | 3.8 | 30,242 | 333 | 132 | 6 | 4.5 | 1.8 | 2002 2332 6011 6872 65267 4402 | ELK1 FMR1 GRK1 T AF1 WNK3 EIF2AK4 | 0 | MyList |
| GO Biological Processes | 19 | GO:0104004 | cellular response to environmental stimulus | 19_GO:0050896 response to stimulus | -2.5 | 4.1 | 3.8 | 30,242 | 333 | 132 | 6 | 4.5 | 1.8 | 2002 2332 6011 6872 65267 4402 | ELK1 FMR1 GRK1 T AF1 WNK3 EIF2AK4 | 0 | MyList |

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|-------------------------|----|---------------|---|---|------|-----|-----|--------|-----|-----|---|-----|-----|---|---|--------|
| | | | | | | | | | | | | | | | | |
| GO Biological Processes | 19 | GO:0045732 | positive regulation of protein catabolic process | 19_GO:0048518 positive regulation of biological process | -2.4 | 4.9 | 4 | 30,242 | 232 | 132 | 5 | 3.8 | 1.7 | 2332 2719 6872 10075 28952 FMR1 GPC3 TAF1 H UWE1 CCDC22 | 0 | MyList |
| GO Biological Processes | 19 | GO:0022411 | cellular component disassembly | 19_GO:0009987 cellular process | -2.4 | 3.6 | 3.6 | 30,242 | 448 | 132 | 7 | 5.3 | 2 | 292 4943 6901 10013 10075 11152 58526 SLC25A5 TBC1D25 TAF4 AFAZZIN HDAC6 H UWE1 WDR45 MID1IP1 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-162906 | HIV Infection | | -2.4 | 4.9 | 4 | 30,242 | 234 | 132 | 5 | 3.8 | 1.7 | 292 6872 51616 54457 89853 SLC25A5 TAF1 TAF9B TAF7L MVB12B | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-448424 | Interleukin-17 signaling | | -2.4 | 9.5 | 4.8 | 30,242 | 72 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-167172 | Transcription of the HIV genome | | -2.4 | 9.4 | 4.8 | 30,242 | 73 | 132 | 3 | 2.3 | 1.3 | 6872 51616 54457 TAF1 TAF9B TAF7L | 0 | MyList |
| GO Biological Processes | 19 | GO:0010639 | negative regulation of organelle organization | 19_GO:0048519 negative regulation of biological process | -2.4 | 4 | 3.7 | 30,242 | 348 | 132 | 6 | 4.5 | 1.8 | 292 395 7454 10013 10075 58526 SLC25A5 ARHGAP6 WAS HDAC6 HUWE1 MID1IP1 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-162587 | HIV Life Cycle | | -2.3 | 6 | 4.1 | 30,242 | 152 | 132 | 4 | 3 | 1.5 | 6872 51616 54457 TAF1 TAF9B TAF7L MVB12B | 0 | MyList |
| GO Biological Processes | 19 | GO:0031346 | positive regulation of cell projection organization | 19_GO:0048518 positive regulation of biological process | -2.3 | 3.9 | 3.6 | 30,242 | 353 | 132 | 6 | 4.5 | 1.8 | 2316 2332 8321 FLNA FMR1 FZD1 C9696 80823 1398 ROCC BHLHB9 DOC18 K11 | 0 | MyList |
| WikiPathways | 27 | WP4666 | Hepatitis B infection | | -2.3 | 6 | 4.1 | 30,242 | 154 | 132 | 4 | 3 | 1.5 | 537 2002 3654 8 ATP6AP1 ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9013148 | CDC42 GTPase cycle | | -2.3 | 5.9 | 4.1 | 30,242 | 155 | 132 | 4 | 3 | 1.5 | 7454 9754 23229 WAS STARD8 ARHGEF9 DOCK11 139818 | 0 | MyList |
| KEGG Pathway | 24 | hsa05230 | Central carbon metabolism in cancer | | -2.3 | 8.8 | 4.6 | 30,242 | 78 | 132 | 3 | 2.3 | 1.3 | 2539 8266 29911 G6PD UBL4A HOOK2 | 0 | MyList |
| GO Biological Processes | 19 | GO:0051494 | negative regulation of cytoskeleton organization | 19_GO:0048519 negative regulation of biological process | -2.2 | 5.6 | 3.9 | 30,242 | 163 | 132 | 4 | 3 | 1.5 | 395 7454 10013 ARHGAP6 WAS HDAC6 MID1IP1 58526 | 0 | MyList |
| GO Biological Processes | 19 | GO:0007420 | brain development | 19_GO:0032502 developmental process | -2.2 | 2.7 | 3.2 | 30,242 | 753 | 132 | 9 | 6.8 | 2.2 | 2316 2334 2539 6872 8321 23133 57526 139411 1 FLNA AFF2 G6PD TAF1 FZD1 PHF8 PCDH19 PTCHD1 ARX 70302 | 0 | MyList |

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|-------------------------|----|---------------|--|---------------|------|-----|--------|--------|-----|-----|-----|-----|---|--|-----------------------------------|--------|--------|
| Reactome Gene Sets | 6 | R-HSA-674695 | RNA Polymerase II Pre-transcription Events | -2.2 | 8.2 | 4.4 | 30,242 | 84 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 | TAF1 TAF9B TAF7L | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-168142 | Toll Like Receptor 10 (TLR10) Cascade | -2.2 | 8.1 | 4.3 | 30,242 | 85 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-975871 | MyD88 cascade initiated on plasma membrane | -2.2 | 8.1 | 4.3 | 30,242 | 85 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-168176 | Toll Like Receptor 5 (TLR5) Cascade | -2.2 | 8.1 | 4.3 | 30,242 | 85 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| GO Biological Processes | 19 | GO:1902904 | 19_GO:0048519 negative regulation of supramolecular fiber organization | -2.2 | 5.5 | 3.9 | 30,242 | 167 | 132 | 4 | 3 | 1.5 | 395 7454 10013 58526 ARHGAP6 WAS HDA C6 MID1IP1 | | 0 | MyList | |
| GO Biological Processes | 19 | GO:0046928 | regulation of neurotransmitter secretion | 19_GO:0051179 | -2.2 | 8 | 4.3 | 30,242 | 86 | 132 | 3 | 2.3 | 1.3 | 2332 6853 6855 | FMR1 SYN1 SYP | 0 | MyList |
| GO Biological Processes | 19 | GO:0030901 | midbrain development | 19_GO:0032502 | -2.1 | 7.6 | 4.2 | 30,242 | 90 | 132 | 3 | 2.3 | 1.3 | 2539 6872 8321 | G6PD TAF1 FZD1 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-975138 | TRAF6 mediated induction of NFkB and MAP kinases upon TLR7/8 or 9 activation | -2.1 | 7.6 | 4.1 | 30,242 | 91 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| GO Biological Processes | 19 | GO:0034644 | 19_GO:0050896 cellular response to UV | -2.1 | 7.6 | 4.1 | 30,242 | 91 | 132 | 3 | 2.3 | 1.3 | 2332 6872 44027 5 | FMR1 TAF1 EIF2AK4 | 0 | MyList | |
| GO Biological Processes | 19 | GO:0042176 | regulation of protein catabolic process | 19_GO:0050789 | -2.1 | 3.5 | 3.3 | 30,242 | 391 | 132 | 6 | 4.5 | 1.8 | 2316 2332 2719 6872 10075 2895 2 | FLNA FMR1 GPC3 T AF1 HUWE1 CCDC22 | 0 | MyList |
| KEGG Pathway | 24 | hsa05161 | Hepatitis B | -2.1 | 5.2 | 3.7 | 30,242 | 177 | 132 | 4 | 3 | 1.5 | 537 2002 3654 8 517 | ATP6AP1 ELK1 IRAK 1 IKBKG | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-975155 | MyD88 dependent cascade initiated on endosome | -2.1 | 7.5 | 4.1 | 30,242 | 92 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| Reactome Gene Sets | 6 | R-HSA-6804756 | Regulation of TP53 Activity through Phosphorylation | -2.1 | 7.5 | 4.1 | 30,242 | 92 | 132 | 3 | 2.3 | 1.3 | 6872 51616 5445 7 | TAF1 TAF9B TAF7L | 0 | MyList | |
| GO Biological Processes | 19 | GO:0051668 | 19_GO:0051179 localization within membrane | -2.1 | 2.8 | 3.1 | 30,242 | 651 | 132 | 8 | 6.1 | 2.1 | 367 1678 1741 2 316 2719 8266 2 8952 65267 | AR TIMM8A DLG3 F LNA GPC3 UBL4A C CDC22 WNK3 | 0 | MyList | |

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|-------------------------|----|---------------|--|---|------|-----|--------|--------|-----|-----|-----|-----|----------------|--|-------------------------|--------|--------|
| Reactome Gene Sets | 6 | R-HSA-168181 | Toll Like Receptor 7/8 (TLR7/8) Cascade | -2.1 | 7.4 | 4.1 | 30,242 | 93 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList | |
| GO Biological Processes | 19 | GO:0033108 | mitochondrial respiratory chain complex assembly | 19_GO:0009987 cellular process | -2.1 | 7.4 | 4.1 | 30,242 | 93 | 132 | 3 | 2.3 | 1.3 | 4694 6901 54539 | NDUFA1 TAFAZZIN NDUFB11 | 0 | MyList |
| GO Biological Processes | 19 | GO:0044089 | positive regulation of cellular component biogenesis | 19_GO:0048518 positive regulation of biological process | -2.1 | 3.1 | 3.2 | 30,242 | 520 | 132 | 7 | 5.3 | 2 | 2316 2332 7454 FLNA FMR1 WAS CR9696 80823 1390 OCC BHLHB9 SLITR65 139818 K4 DOCK11 | | 0 | MyList |
| GO Biological Processes | 19 | GO:0019693 | ribose phosphate metabolic process | 19_GO:0008152 metabolic process | -2.1 | 3.5 | 3.3 | 30,242 | 397 | 132 | 6 | 4.5 | 1.8 | 204 2539 5230 6 AK2 G6PD PGK1 TA901 55190 17068 FAZZIN NUDT11 NU5 DT10 | | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-168164 | Toll Like Receptor 3 (TLR3) Cascade | | -2.1 | 7.3 | 4.1 | 30,242 | 94 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9013423 | RAC3 GTPase cycle | | -2.1 | 7.3 | 4.1 | 30,242 | 94 | 132 | 3 | 2.3 | 1.3 | 395 2010 4810 | ARHGAP6 EMD NHS | 0 | MyList |
| GO Biological Processes | 19 | GO:0060322 | head development | 19_GO:0032502 developmental process | -2.1 | 2.6 | 3 | 30,242 | 796 | 132 | 9 | 6.8 | 2.2 | 2316 2334 2539 6872 8321 23133 57526 139411 170302 FLNA AFF2 G6PD TAF1 FZD1 PHF8 PCDH19 PTCHD1 ARX | | 0 | MyList |
| GO Biological Processes | 19 | GO:1903827 | regulation of cellular protein localization | 19_GO:0051179 localization | -2.1 | 3 | 3.1 | 30,242 | 527 | 132 | 7 | 5.3 | 2 | 367 2010 2316 2 AR EMD FLNA GPC3719 9696 10075 CROCC HUWE1 W65267 NK3 | | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-168138 | Toll Like Receptor 9 (TLR9) Cascade | | -2.1 | 7.2 | 4 | 30,242 | 96 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9013149 | RAC1 GTPase cycle | | -2 | 5 | 3.6 | 30,242 | 185 | 132 | 4 | 3 | 1.5 | 2010 4810 7454 EMD NHS WAS DOC139818 K11 | | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-166166 | MyD88-independent TLR4 cascade | | -2 | 6.9 | 3.9 | 30,242 | 99 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-937061 | TRIF(TICAM1)-mediated TLR4 signaling | | -2 | 6.9 | 3.9 | 30,242 | 99 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 | ELK1 IRAK1 IKBKG | 0 | MyList |
| GO Biological Processes | 19 | GO:0060996 | dendritic spine development | 19_GO:0032502 developmental process | -2 | 6.9 | 3.9 | 30,242 | 99 | 132 | 3 | 2.3 | 1.3 | 2332 10013 8082 FMR1 HDAC6 BHLH3 B9 | | 0 | MyList |
| GO Biological Processes | 19 | GO:0051588 | regulation of neurotransmitter transport | 19_GO:0051179 localization | -2 | 6.9 | 3.9 | 30,242 | 100 | 132 | 3 | 2.3 | 1.3 | 2332 6853 6855 FMR1 SYN1 SYP | | 0 | MyList |

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|-------------------------|----|---|---|----|-----|-----|--------|-----|-----|---|-----|-----|---|---|--------|
| GO Biological Processes | 19 | GO:0006626 protein targeting to mitochondrion | 19_GO:0051179 localization | -2 | 6.9 | 3.9 | 30,242 | 100 | 132 | 3 | 2.3 | 1.3 | 1678 10,075 1024 TIMM8A HUWE1 TIMM17B | 0 | MyList |
| GO Biological Processes | 19 | GO:0050890 cognition | 19_GO:0032501 multicellular organismal process | -2 | 3.9 | 3.3 | 30,242 | 296 | 132 | 5 | 3.8 | 1.7 | 2334 80823 8406 AFF2 BHLHB9 MAGT1 139411 4402751 PTCHD1 EIF2AK4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-9633012 | Response of EIF2AK4 (GCN2) to amino acid deficiency | -2 | 6.8 | 3.9 | 30,242 | 101 | 132 | 3 | 2.3 | 1.3 | 6134 6173 44,027 RPL10 RPL36A EIF2AK4 | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-168188 | Toll Like Receptor TLR6:TLR2 Cascade | -2 | 6.8 | 3.9 | 30,242 | 101 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 ELK1 IRAK1 IKBKG | 0 | MyList |
| Reactome Gene Sets | 6 | R-HSA-166058 | MyD88:MAL(TIRAP) cascade initiated on plasma membrane | -2 | 6.8 | 3.9 | 30,242 | 101 | 132 | 3 | 2.3 | 1.3 | 2002 3654 8517 ELK1 IRAK1 IKBKG | 0 | MyList |
| GO Biological Processes | 19 | GO:0050821 protein stabilization | 19_GO:0065007 biological regulation | -2 | 4.8 | 3.5 | 30,242 | 191 | 132 | 4 | 3 | 1.5 | 2316 6872 11040 FLNA TAF1 PIM2 TA F9B 51616 | 0 | MyList |

Table S4. Candidate genes of X0_Hypomethylated which were also detected as hypomethylated in TSvs46XX in previous research.

| PNAS2020 | CLINEPI2015 | X0hypo | PNAS2020 CLINEPI201 5 | PNAS2020 X0hypo CLINEPI2015 X0hypo | PNAS2020 CLINEPI2015 X0hyp o |
|---------------|-------------|---------------|--------------------------|------------------------------------|---------------------------------|
| RP11-1148L6.9 | 38961 | MVB12B | RBM3 | NUDT10 | CYP2E1 |
| PFKFB1 | 40057 | RP4-537K23.4 | FTSJ1 | TAF9B | HOXA4 |
| XX- | | | | | |
| FW83563B9.5 | TLE1 | AF196779.12 | EFNB1 | TIMM17B | EMD |
| SNORA70 | MGC3207 | ZNF630 | IGBP1 | ZNF674-AS1 | CXorf40A |
| AL049564.1 | FAM83A | RP11-85L21.4 | APEX2 | FAM127B | SYP |
| LINC00894 | GLUD1 | IKBKG | CSTF2 | RP4-769N13.6 | OTUD5 |
| AIFM1 | CST9L | BTBD11 | RNF113A | TRMT2B | UBL4A |
| AF196970.3 | CCL26 | TM4SF2 | PQBP1 | PCDH19 | WNK3 |
| FAM127A | NPR2 | EIF2AK4 | LDOC1 | RAI2 | ZMYM3 |
| SLC25A53 | HLA-DQB2 | CACNA1F | RBM10 | NKAPP1 | AFF2 |
| ERCC6L | SEC31L2 | GPR50-AS1 | SLC25A14 | RPL36A-HNRNPH2 | ARHGEF9 |
| FAM104B | SH3BP2 | TRMT2B-AS1 | NKAP | F8 | MOSPD1 |
| HNRNPH2 | LYPD3 | AC115618.1 | ATG4A | TMEM255A | BCORL1 |
| OGT | ZNF593 | HAUS7 | PSMD10 | NUDT11 | RBMX2 |
| FANCB | NAV1 | CROCC | DKC1 | VMA21 | NHS |
| ZNF674 | HP | U1 | DACH2 | RP11-66N11.8 | PNCK |
| OOEP | DHODH | LINC0086 | SOX3 | HUWE1 | PGK1 |
| RLIM | AIF1 | TAF7L | YIPF6 | HS6ST2 | TSPAN7 |
| DPPA5 | STMN1 | SAMD11 | PRAF2 | RP3-326L13.2 | TAZ |
| ZNF81 | CIAS1 | DOCK11 | GPR101 | MORC4 | ARHGAP6 |
| AL022578.1 | ZFYVE19 | HOOK2 | GLA | TBC1D25 | WAS |
| RP11-308D16.2 | GPR109B | TNS1 | UTP14A | LINC00629 | MAGEE1 |
| GPKOW | FYB | MPL | MOSPD2 | KIAA2022 | HDAC6 |
| RP13-228J13.9 | VAMP5 | FOXI2 | DDX43 | TMEM185A | TIMM8A |
| PDZD11 | C20orf75 | HOXA3 | TFE3 | AC093609.1 | BHLHB9 |
| RP11-348F1.2 | TACSTD1 | HOXA-AS2 | ARMCX5 | BEX4 | SCML1 |
| RP11-348F1.3 | GPR109A | RP1-170O19.22 | RPL39 | MAGIX | ZBTB33 |
| SPACA5 | HTRA4 | SPRN | KIF4A | PRRG1 | EBP |
| ZNF182 | AMT | NAPRT1 | ATRX | PTCHD1 | SRPX |
| THOC2 | P2RY2 | CTU1 | TSPYL2 | SLC25A5-AS1 | ZNF41 |
| CLCN5 | VENTX | | | RP4-769N13.7 | HMGB3 |
| STK26 | FLJ30046 | | ZCCHC12 | SRPK3 | IRAK1 |
| TAB3-AS2 | F12 | | NR0B1 | | IL13RA1 |
| SMIM10 | STK31 | | ZNF449 | | LANC3 |
| KRBOX4 | ZNF541 | | OPHN1 | | AR |
| ZNF75D | C14orf162 | | SLC35A2 | | DLG3 |
| PIH1D3 | TRPM2 | | MAGEH1 | | DNASE1L1 |
| NUP62CL | DEFA1 | | IDH3G | | PHF8 |
| AL590410.1 | EPN3 | | RBMX | | RAB33A |
| BTK | CCIN | | PIGA | | SLC25A5 |
| MAMLD1 | MAN2A2 | | COX7B | | ELF4 |
| FAM127C | LMO2 | | ARMCX3 | | WDR13 |
| SNORD61 | CLEC4C | | FUNDG2 | | MAGED2 |
| SMIM10L2B | EPAS1 | | RAB39B | | MID1IP1 |
| AC234775.1 | ARS2 | | NSDHL | | RPL10 |
| ARMCX3-AS1 | BRD4 | | CETN2 | | STARD8 |
| ZCCHC18 | C6orf68 | | NKRF | | FMR1 |
| NCL | DYRK4 | | WDR44 | | ATP6AP1 |
| HSPB1P2 | HPS4 | | SUV39H1 | | BEX1 |
| H3F3AP1 | TMEM102 | | ZDHHC15 | | |
| RP1-161N10.1 | LTBR | | LAS1L | | |
| SMAD5-AS1_2 | POR | | MSN | | |
| RHOXF1P1 | ZNF124 | | FHL1 | | |
| CXorf57 | RABGEF1 | | ACSL4 | | |
| ZNF280C | AMDHD1 | | ARAF | | |
| CELSR3-AS1 | PPP4R2 | | MCTS1 | | |
| OVCH1-AS1 | HMGN1 | | HDAC8 | | |
| AMER1 | CIDEB | | CXorf56 | | |
| DGKK | NFE2 | | PRDX4 | | |

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| RAC1P4 | NR1I2 | ABCB7 |
| ARHGAP36 | SFTPД | SLITRK4 |
| LINC01201 | HSPA2 | KLHL34 |
| GPRASP1 | BRCA2 | RRAGB |
| RP11-314A15.2 | HAL | XKRX |
| FAM122B | SLC22A3 | XK |
| FGF13-AS1 | FBXL5 | MECP2 |
| AC131011.1 | ITGB1BP1 | PGRMC1 |
| MBNL3 | RPS6KC1 | IL1RAPL2 |
| HDX | ENTPD1 | FAM58A |
| PBDC1 | ARID3A | PHKA1 |
| APOOL | APOB | CNKSR2 |
| TSR2 | P2RY13 | MBTPS2 |
| RP1-274L7.1 | LPO | PCSK1N |
| RP6-24A23.3 | BTBD6 | HTATSF1 |
| MYCLP1 | SERPINF1 | FAM122C |
| TSPAN6 | ZNF266 | NONO |
| RP3-389A20.5 | CTBP1 | FGD1 |
| LAGE3 | BIRC4BP | RAP2C |
| SAT1 | KLF1 | POU3F4 |
| RP13-314C10.5 | RAB8A | SNX12 |
| FAM156A | CD244 | SH3BGRL |
| ZC4H2 | HSPC159 | SMS |
| HMGN5 | TIGD5 | FAM50A |
| KANTR | LGP1 | UPF3B |
| RP11-236P24.3 | IL27 | INGX |
| PRICKLE3 | CACYBP | CHM |
| NXT2 | CTSG | SYTL4 |
| MORF4L2-AS1 | RPS6KA1 | PIN4 |
| LINC00893 | FLJ25084 | ATP6AP2 |
| RP1-137H15.2 | IFITM3 | ARX |
| PCYT1B-AS1 | SCMH1 | UXT |
| LINC01285 | GPR21 | PHKA2 |
| SMIM10L2A | AQP9 | GNL3L |
| WDR53 | LYAR | MTM1 |
| ZXDB | PRTN3 | GABRQ |
| AL158069.1 | FLJ40919 | MTMR8 |
| FAM155B | GSG1 | TMLHE |
| Y_RNA | LYZ | AMOT |
| snoU13 | H2BFS | NAP1L2 |
| MID1IP1-AS1 | SEPT10 | ZIC3 |
| CENPI | ELA2 | SLITRK2 |
| LINC01381 | UNQ830 | GPC4 |
| UXT-AS1 | CEACAM6 | MAGEE2 |
| TLE1P1 | NKG7 | ATP7A |
| RNU6-800P | IER2 | UBE2A |
| RP13-43E11.1 | SPAG4 | PRPS2 |
| AL035088.1 | SGK2 | EDA |
| LINC01560 | LAIR1 | TCEAL1 |
| RPS26P11 | SLC44A2 | PRPS1 |
| VPS28 | PABPN1 | MTMR1 |
| MSL3 | PARVG | USP51 |
| ZDHHC9 | ACYP2 | CDX4 |
| MIR718 | CLEC5A | TCEAL8 |
| RP11-234P3.2 | BTN1A1 | RPGR |
| RP5-1091N2.9 | KLHDC7B | GSPT2 |
| C17orf98 | GALNT3 | LAMP2 |
| DDX26B-AS1 | GPR97 | HIST1H3E |
| RP13-210D15.8 | RBM35B | GLUD2 |
| BTF3P8 | KCNE1 | RP2 |
| RAP2C-AS1 | GATAD2B | RPS6KA3 |
| CDR1-AS | C18orf22 | PAK3 |
| BX842568.4 | HIST1H2AJ | TCEAL2 |
| RPS6KA6 | ATP6V1E2 | GPC3 |
| CXorf58 | KCNQ1 | SLC10A3 |
| APOO | FUT7 | STAG2 |

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| MAP3K15 | CYP27B1 | GRIA3 |
| ENOX2 | LOC158160 | TMSB4X |
| SASH3 | FCGR3B | MAOA |
| MIR5003 | P518 | PDK3 |
| KIF13A | CLEC9A | TBX22 |
| FCF1P4 | CSF3R | ARMCX6 |
| PJA1 | KNS2 | HPRT1 |
| RP11-287D1.4 | KIAA0427 | DYNLT3 |
| NRK | GNS | SLC9A6 |
| RP5-1000K24.2 | GLRX2 | SLC7A3 |
| CFP | C6orf105 | SCML2 |
| NCBP2L | LGALS4 | USP11 |
| FGF13 | CLEC12A | ARMCX1 |
| DDX26B | MGC26963 | TMEM47 |
| DUSP9 | CCT6A | IL1RAPL1 |
| RP13-188A5.1 | GPR84 | ZNF185 |
| CENPV3 | ZNF696 | ABCD1 |
| BRCC3 | SLPI | SLC16A2 |
| CMC4 | MGC11257 | UBQLN2 |
| GRIPAP1 | FLJ33860 | PORCN |
| LINC01278 | TSPAN16 | ARHGAP4 |
| APLN | H2AFY | ZXDA |
| UPRT | FLJ35773 | PHF6 |
| AL772400.1 | LOC92345 | ERAS |
| DERL2 | C14orf58 | PLP2 |
| POLA1 | CIZ1 | PDHA1 |
| XIAP | BRE | CLCN4 |
| POU5F1 | SLC22A18 | |
| AC234031.1 | PILRA | |
| GPR50 | ECRG4 | |
| BX842568.1 | LCE3A | |
| RP11-641J8.1 | FLJ35530 | |
| ZNF711 | LRG1 | |
| AC003001.1 | FKBP11 | |
| SLC25A43 | SCRL | |
| RNF128 | RNF14 | |
| RP11-521C20.5 | FXYD1 | |
| SOCS5P4 | LYCAT | |
| FOXO4 | SLC45A1 | |
| AC005041.11 | PNPLA2 | |
| CTD-2269E23.4 | CSTA | |
| RP1-164F3.8 | MYL9 | |
| TUBGCP3 | HIPK3 | |
| AL691477.1 | TATDN1 | |
| TREX2 | IL21R | |
| RNU6-894P | PREX1 | |
| RP11-21G20.3 | SH3BP5 | |
| RP1-169K13.3 | ANKMY1 | |
| KLHL13 | DOPEY2 | |
| AMMECR1-IT1 | LOC132321 | |
| RNU6-481P | VNN3 | |
| CITED1 | AGXT | |
| MIR3202-2 | C15orf5 | |
| RP5-972B16.2 | MGC41945 | |
| BX842568.2 | APOA4 | |
| NHSL2 | CENPH | |
| GPR45 | P2RX1 | |
| GHc-857G6.7 | PIP3-E | |
| MIR106A | SMPD3 | |
| ZNF157 | TETRAN | |
| CHD4 | LRP8 | |
| WBP5 | C1orf150 | |
| RP11-68I3.4 | CLTCL1 | |
| CCDC160 | H1F0 | |
| CHIC1 | SEC61G | |
| MAP2K4P1 | GPR25 | |

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| RBM41 | HK2 |
| TMEM263 | DEFB1 |
| RP11-412D9.4 | NDUFS2 |
| LINC00630 | ART3 |
| RP4-613B23.1 | CLCA2 |
| LINGO3 | CYP7B1 |
| RP11-149B9.2 | GRB7 |
| SPIN3 | MS4A6A |
| COL4A5 | DLK1 |
| RP11-62J1.4 | IFNGR2 |
| RP11-548B3.3 | FOLR3 |
| SOWAHD | FCN1 |
| FZD2 | LCN2 |
| RP11-320G24.1 | XYLB |
| AF196972.9 | IL19 |
| FMR1-AS1 | URP2 |
| JADE3 | ZNF710 |
| OR11N1P | RPUSD3 |
| RP1-149M18.4 | THSD3 |
| CFAP47 | FFAR3 |
| FMR1-AS1_2 | KCNQ1DN |
| RNF5P1 | SPATC1 |
| PRDM9 | EIF2B3 |
| PNMA5 | SYCP1 |
| CH17-340M24.3 | ABTB1 |
| BEX5 | RNASE3 |
| TMSB15A | PTGFRN |
| ARHGEF6 | FUT3 |
| OSBPL5 | NOL3 |
| RP5-998H6.2 | SLC26A8 |
| CSNK1G2P1 | USP10 |
| MYCBPAP | NAGPA |
| RP11-94C24.6 | TRIM50C |
| | CALML4 |
| | TIGD2 |
| SERPINB10 | |
| | C3orf18 |
| | SPI1 |
| | GABRR1 |
| | EPHA2 |
| | TFAP2E |
| | ACPP |
| | LEP |
| SLC37A1 | |
| | LOC201164 |
| CEBPE | |
| GP5 | |
| PRR3 | |
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| KCNA6 | |
| SLC22A12 | |
| K6IRS2 | |
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| TACSTD2 | |
| MPO | |
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| CASP6 | |
| C19orf24 | |

CPM
PSMD5
DNAH3
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VTN
GNPDA1
HIVEP3
LYK5
1-Mar
CAMP
NPFFR2
CEACAM3
SLC19A1
LRPAP1
SERGEF
UCK1
FCGRT
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PC
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EDG3
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RALGPS2
CBFA2T3

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LAIR2
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NCOR2
ELMO3
SNN
PHB2
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CNGB1
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TIMM13
INPP5D
GLT1D1
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ASGR2
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SYNE2
JAK2
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NDUFAF1
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PLXDC1
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PODXL2
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MGMT
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MRPL28
POU3F1
TRPM6
HSPBP1
UNQ3033
HNRPF
SEMA3B
MPST
CDH5
ANKRD13C
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SUMO3
TP53TG3
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TYROBP
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CXorf26
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CXorf41
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GABRA3
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PCYT1B
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CLDN2
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BMX
FSHPRH1
KIAA1166
ZCCHC5
NSBP1
GAB3
COVA1
NOX1
RBBP7
MORF4L2
FRMPD4
MAGED1
PNPLA4
L1CAM
CXorf21
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PCTK1
CLIC2
GLRA2
PLXNB3
AGTR2
TLR7
BGN
RENBP
PRRG3
SMC1L1
KCND1
CXorf12
