**Supplementary Table 2**. **CNV Calling Criteria.** Criteria used for calling CNVs (following our previous report, Kendall et al, 2016). CNVs at *EHMT1* and *SHANK3* were required to intersect at least 1Mbp distance, as small deletions and duplications were found to be common in samples with poor QC criteria, indicating that small CNVs in these telomeric regions were likely to be false-positives.

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| **CNV** | **Criteria** |
| 1p36 del/dup | Size >50% of critical region, affecting *GABRD* |
| TAR del/dup | Size >50% of critical region |
| 1q21.1 del/dup | Size >50% of critical region |
| *NRXN1* del | Exonic deletions |
| 2q11.2 del/dup | Size >50% of critical region, affecting both *LMAN2L* and *ARID5A* |
| 2q13 del/dup | Size >50% of critical region |
| 2q13 del/dup (*NPHP1*) | Size >50% of critical region, affecting *NPHP1* |
| 2q21.1 del/dup | Size >50% of critical region |
| 2q37 del/dup (*HDAC4*) | Size >50% of critical region, affecting *HDAC4* |
| 3q29 del/dup | Size >50% of critical region |
| Wolf-Hirschhorn del/dup | Size >50% of critical region |
| Sotos Syn/5q35 dup | Size >50% of critical region |
| 6q16 del/dup (*SIM1*) | Exonic deletions; whole gene duplications |
| Williams Beuren Syn del/dup | Size >50% of critical region |
| 7q11.23 distal del/distal dup | Size >50% of critical region |
| 8p23.1 del/dup | At least 1Mbp of critical region |
| 9q34 del/dup (*EHMT1*) | At least 1Mbp CNVs, including *EHMT1* |
| 10q11.21q11.23 del/dup | Size >50% of critical region |
| 10q23 del/dup | At least 1Mbp, including *NRG3* and *GRID1* |
| Potocki-Shaffer Syn del/11p11.2 dup (*EXT2*) | Size >50% of critical region, including *EXT2* |
| 13q12 del/dup (*CRYL1*) | Exonic deletions; whole gene duplications |
| 13q12.12 del/dup | Size >50% of critical region |
| 15q11.2 del/dup | Size >50% of critical region |
| PWS del/dup | Full critical region, ~4Mbp |
| 15q11q13 del/dup BP3-BP4 | Size >50% of critical region |
| 15q11q13 del/dup BP3-BP5 | Size >50% of critical region |
| 15q13.3 del/dup | Size >50% of critical region |
| 15q13.3 del/dup (*CHRNA7*) | Size >50% of critical region, affecting *CHRNA7* |
| 15q24 del/dup | At least 1Mbp between the A-E intervals |
| 15q25 del/dup | At least 1Mbp between the A-D intervals |
| Rubinstein-Taybi del/dup (*CREBBP*) | Exonic deletions; whole gene duplications |
| 16p13.11 del/dup | Size >50% of critical region |
| 16p12.1 del/dup | Size >50% of critical region |
| 16p12.2-p11.2 del/dup (7.1-8.7 Mb) | Size >50% of critical region |
| 16p11.2 distal del/distal dup | Size >50% of critical region |
| 16p11.2 del/dup | Size >50% of critical region |
| 17p13.3 del/dup (*YWHAE*) | Exonic deletions; whole gene duplications |
| 17p13.3 del/dup (*PAFAH1B1*) | Exonic deletions; whole gene duplications |
| 17p12 del (HNPP)/dup (CMT1A) | Size >50% of critical region, affecting *PMP22* |
| Smith-Magenis/Potocki-Lupski Syn | Size >50% of critical region |
| 17q11.2 del/dup (*NF1*) | Size >50% of critical region, affecting *NF1* |
| 17q12 del/dup | Size >50% of critical region |
| 17q21.31 del/dup | Size >50% of critical region |
| 17q23.1q23.2 del/dup | Size >50% of critical region |
| 22q11.2 del/dup | Size >50% of critical region |
| 22q11.2 distal del/dup | Size >50% of critical region |
| *SHANK3* del/dup | At least 1Mbp CNVs, including *SHANK3* |
| “Large” CNVs | Size > 20Mbp + >50 genes |