

Supplementary Figure 2. Genomic sequence of the deletion breakpoint in family ASD 1 and prediction of quadruplex forming G-rich sequences (QGRS) at the terminal end of chromosome 22q13. The breakpoint identified in family ASD 1 is similar to those previously identified in probands A, B, C from Bonaglia et al.⁵ The sequence surrounding the breakpoint is recognized as a QGRS and has the best prediction score (143, indicated by a red circle) out of the 1700 GQRS identified. This structure may represent a substrate for telomere formation, thereby increasing the risk of recurrent deletions in this region. Sequence was extracted from the Human March 2006 (hg18) assembly. Prediction performed QGRS was using the mapper software (http://bioinformatics.ramapo.edu/QGRS/index.php).