

Supplementary Figure 2. Comparison of v1 and v2.1 methodologies against MLPA for determining copy number. 16 samples were processed by both methods and 20 regions in common with the MLPA data were compared. Correlation coefficients for each of the 20 regions were calculated for both versions and plotted against each other. The correlation coefficient was $R^2=0.689$. The largest discrepancy was with one datapoint at exon 1 of WWOX (red dot) which had a low correlation coefficient between v1 and MLPA (0.77) compared to v2.1 and MLPA (0.91). This improvement in correlation for v2.1 copy number calling at the first exons was seen across many genes, and is likely due to increased hybridization efficiency in GC-rich regions of the genome with the newer HyperCap reagents. After removing WWOX exon 1, the correlation coefficient between v1 and v2.1 was $R^2=0.92$. The average correlation between MLPA and the panels for the 20 regions was 0.91 (v1) and 0.92 (v2.1).

