Supplementary Figure 2. Copy number alterations in HER2-amplified breast cancer. (A) Overlap of GISTIC regions in the current and a previous study comprising 200 HER2-amplified cases analyzed by 32K BAC aCGH [5]. Frequency of copy number gain and loss across the 218 HER2-amplified tumors in the current study is shown in grey. Red regions indicate GISTIC regions (n=80) identified from the 218 cases in the current study, while black regions (n=90) indicate GISTIC regions previously identified by BAC aCGH analysis of 200 HER2-amplified tumors [5] (remapped to hg18 coordinates and filtered for q-value < 0.05). Sizes of GISTIC regions are inflated for improved visualization. (B) Frequency of CN gain (black) and loss (black) across 16 HER2-amplified breast cancer cell lines analyzed by SNP microarrays. CN gain and loss are called using log2ratio thresholds of 0.12 (gain) and -0.12 (loss). The frequency of CN gain (red) and loss (green) obtained from GISTIC analysis of the 218 HER2-amplified primary tumors are shown for comparison. HER2-amplified cell lines include MDA-MB-361, MD-MBA-453_Hahn, UACC-893, UACC-812, SKBR3, SM-126O, AU5_65, EFM192A, HCC202, HCC1419, ZR7530, HCC1569, BT474, HCC2218, HCC1008, and HCC1954. (C) Shortest region of amplification including the complete HER2 gene in the 218 tumors +16 cell lines. Y-axis shows percentage of cases with a specific amplicon size. Purple vertical line indicates shortest overlapping region chr17:35087594-35146597. The region was defined in a conservative manner using the start /stop assignments of the first / last probes not included in the HER2 segment plus/minus 1 bp. (D) Sample GSM447303 defining the centromeric limit. Purple line indicates the shortest region of amplification, red line the HER2 gene. Black lines correspond to partitioned segments. (E) Sample GSM511316 defining the telomeric limit. Purple line indicates the shortest region of amplification, red line the HER2 gene. Black lines correspond to partitioned segments. (F) GISTIC regions stratifying HER2-amplified cases based on ER-status: ER-positive (n=59, blue), ER-negative (n=64, red). Regions identified by Fisher’s test, with p-value < 0.01. GISTIC regions are defined in Additional file 3. Significant gain of GISTIC region 1p31.3 in ER-negative tumors in the current study is matched by a similar region of gain also on 1p31.3 in ER-negative tumors in our previous BAC-based aCGH study [5]. Significant loss of GISTIC region 16q24.3 in ER-positive tumors in the current study is matched by a similar region of loss on 16q23.3 in ER-positive tumors in our previous BAC-based aCGH study [5].