



Additional file 1

Depletion and RNA exome capture protocols were examined using FFPE and FFzn pairs for seven BBD patients:

- a). Boxplot of alignment statistics, showing total number of reads (orange), number of mapped reads (green), number of reads mapped to exon-exon junctions (blue) and number of reads mapped to genes (purple).
- b). Boxplot of number of captured canonical or novel exon-exon junctions, annotated (orange), completely novel (blue), partially novel (purple).
- c). SNP confirmation rate based on DNA exome-seq samples, showing the six nucleotide substitution categories for FFzn samples with each sequencing protocol
- d). Heatmap of gene expression correlation matrix between 5 BBD FFzn samples processed with either Ribo-Depletion protocol or TrueSeq protocol with polyA selection. The top column annotation color bar indicates library preparation protocol. The middle column annotation color bar indicates tissue type. The lower column annotation color bar indicates patient ID. The rectangle boxes indicate samples clustered by patient ID rather than technology type.
- e). Heatmap of gene expression correlation matrix between 5 BBD FFzn samples processed with either RNA exome capture protocol (no polyA selection) or TrueSeq protocol with polyA selection. The top column annotation color bar indicates library preparation protocol. The middle column annotation color bar indicates tissue type. The lower column annotation color bar indicates patient ID. The rectangle boxes indicate samples clustered by patient ID rather than technology type.