A) Before extraction of read depth information

B) After extraction of read depth information

Number of potential de novo SNVs per proband

Frequency

Number of potential de novo SNVs per proband

Frequency
Interactome analysis of USP19

http://string-db.org/
The plot shows the B-allele frequency information (variant/total reads ratio) extracted from exome data VCF files over the genome. Then, we subtracted 0.5 from the signal and then segments with absolute value of the mean signal >0.45 and size >100 kb were detected as AOH regions and displayed in blue. In the presented plot, y axis shows the B-allele frequency (variant/total reads ratio) extracted from exome data VCF files, with each chromosome shown in a separate plot. This B-allele frequency information was then processed using Circular Binary Segmentation (CBS) implemented in the DNAcopy R bioconductor package.