IRCCS-GC: mutation - Illumina NovaSeq, WES

Molecular Methods Description:

DNA extracted from PDX models along with a sample of normal germline DNA from each patient were utilized for whole exome sequencing (Agilent SureSelect XT Human All Exon V6 library).

Analysis Description:

Data analysis has been performed by Silvia Giordano's group at the Candiolo Cancer Institute (Turin, Italy).

NGS was performed on an Illumina NovaSeq sequencer. 150-bp read pairs were aligned to concatenated mm10-hg38 reference genomes using BWA-MEM and human mapping reads were sorted and duplicate-marked using Picard tools. The alignments were further refined using the Genome Analysis Toolkit (GATK) for recalibration of quality scores.

Somatic events were identified with MuTect2 and annotated using Variant Effect Predictor v95 (VEP). Only non-synonymous mutations were considered (missense, nonsense, splice donor, splice acceptor, start/stop lost, frameshift mutations, in-frame insertions/deletions).