1. Bioinformatics Analysis

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| **Data quality control** |
| Distribution of Sequencing Quality |
| Distribution of Sequencing Error Rate |
| Distribution of A/T/G/C Base |
| Statistic Summary of Sequencing Quality. Including amount of data output, error rate, Q30 and GC content, etc. |

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| **Standard Analysis** |
| Data Quality Control: filtering reads containing adapter or with low quality |
| Alignment to Reference Genome; Statistics of Sequencing Depth and Coverage |
| Variant (SNP, CNV, InDel and SV) Calling, Annotation and Statistics |
| Somatic Variant (paired tumor samples) Detection |
| -SNP calling, annotation and statistics |
| -CNV calling, annotation and statistics |
| -InDel calling, annotation and statistics |
| -SV calling, annotation and statistics |
| Display of Genomic Variants with Circos |

**Note: For advanced analysis, sales/technical support should consult product specialist at first. Advanced analysis includes but not limited to following contents.**

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| **A. Package of Tumorigenesis Analysis-for Cancer Samples** (Tumor-Normal paired) |
| Intra-Tumor Heterogeneity Analysis |
| Tumor Purity & Ploidy Estimation |
| Tumor Evolution Analysis (One normal and at least 3 tumor samples from the same patient are needed) |
| **B. Package of Driver Gene/Mutation Analysis-for Cancer Samples** (Tumor-Normal paired) |
| Identification of Driver Somatic CNVs Based on Frequency (More than 20 pairs of “case/control” samples are needed) |
| Identification of Driver Mutations in Noncoding Regions |
| Identification of Driver Genes Based on Mutation Clustering Bias |
| Drive Gene Pathway Analysis-DO/GO/KEGG |
| Screening for Predisposing Genes (feasible if only normal samples are provided) |
| Prioritization of Candidate Genes |
| **C. Package of Mutation Profile Analysis-for Cancer Samples** (>20 pairs of “case/control” samples are needed) |
| Mutational Spectrum & Mutational Signature |
| Significantly Mutated Gene & Pathway Analysis: |
| -Tumor significantly mutated genes |
| -Heatmap of significantly mutated genes |
| -Significantly mutated pathways |
| Mutation Relation Test of Significantly Mutated Genes |
| **D. Independent Advanced Analysis-for Cancer Samples** (Tumor-Normal paired) |
| Tumor Neoantigen Identification |
| Xenograft Tumor Analysis (PDX) |
| Fusion Gene Detection |
| Integration Site Identification |
| CRISPR/Cas9 On-Target and Off-Target Detecting (gRNA sequence is required) |
| **E. Package Advanced Analysis-for Disease Samples** |
| Candidate Variant Identification |
| Prioritization of Candidate Genes |
| Regions of Homozygosity (ROH) Analysis (**Monogenic disorder only**) |
| *De novo* Mutation Analysis (Trio/Quartet) (**Complex/Multifactorial disorder**) |
| Linkage Analysis (family-based) (**Monogenic disorder only**) |
| Integration Site Identification |
| CRISPR/Cas9 On-Target and Off-Target Detecting (gRNA sequence is required) |