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 with reference, statistics of sequencing depth and coverage |
| SNP and InDel calling, annotation and statistics |
| Somatic SNP/InDel/CNV calling, annotation and statistics (paired tumor samples) |

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

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| **Advanced Analysis (Disease)** |
| Candidate Variant Identification |
| -Variant filtering using known database |
| -Variant filtering based on genetic model (Pedigree information is needed) |
| Linkage analysis (family-based) (**Monogenic disorder only**) |
| Regions of homozygosity (ROH) analysis (**Monogenic disorder only**) |
| *De novo* mutation analysis (Trio/Quartet) (**Complex/Multifactorial disorder**) |
| **Advanced Analysis (Cancer: Tumor-Normal paired samples)** |
| Screening for Predisposing Genes (feasible if only normal samples are provided) |
| Mutational Spectrum & Mutational Signature |
| Driver Genes analysis |
| -Significantly mutated gene test |
| - Identification of Driver Genes Based on Mutation Clustering Bias |
| - Mutation Relation Test of Significantly Mutated Genes |
| - Significantly mutated pathway/gene set analysis |
| -Driver somatic copy-number alterations |
| Tumor Heterogeneity analysis |
| -Cancerous cell purity |
| -Tumor Ploidy |
| -Clonal Architecture |
| - Tumor Evolution Analysis (One normal and at least 3 tumor samples from the same patient are needed) |
| Tumor Neoantigen Identification |