NGS Data Analysis Questionnaire (Tumor/Normal Pair)

## **Study Objectives: Please specify the Aim and** **Brief description of the project**.

This is to understand project, the analysis will include results as per section F-K

## **Please specify appropriate species below**

For Non-human species please provide link to preferred reference genome

|  |  |  |  |
| --- | --- | --- | --- |
| Standard species | Human (Homo sapiens) | hg38 (GRCh38) (the most updated version of human genome) |  |
| hg19 (old version) |  |
| Others (Note Limitations) |  | |  |

## **Please select appropriate sequencing method.** (Must Select One)

Whole Genome Sequencing (WGS)

Whole Exome Sequencing (WES)

Panel Sequencing

1. **Please identify approximate number of samples and experimental design**

**Example Somatic Matched Design**

Total 4 samples, 2 Tumor tissues and 2 matched normal from blood

Total 10 samples, 5 Tumor tissues and 5 Adjacent Tissue normal

Total 30 samples, 10 Tumor tissues and 20 processed matched controls/normal

## **Please specify the source of control/normal sample** (Must Select One)

Germline control from blood or saliva

Adjacent/Nearby Tissue control (There may be False-negative mutations due tumor infiltration)

Processed matched controls (There may be False-positive mutations due to private germline variants)

Other Please explain

## **Standard deliverables**

## QC & Alignment (Common for all sequencing methods and analysis types)

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| --- | --- | --- | --- |
| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| Sequencing QC | FastQC |  |  |
| Sequence alignment Bams | BWA (Dragen Pipeline For WGS) |
| Duplicate Marking | Picard (Dragen Pipeline For WGS) |
| Hybrid Selection Metrics (WES/Panel)  WGS Metrics (WGS) | Picard |
| Matched Somatic Mutation Analysis | Mutect2 (Dragen Pipeline For WGS) |  |  |

## **Detailed Annotations**

Please note only available for certain species and will incur additional cost. Please reach a PM team member for more details.

|  |  |  |  |
| --- | --- | --- | --- |
| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| Functional Annotations | SNPeff |  |  |
| dbSNP (Human Only) | SNPsift |  |  |
| ClinVAR (Human Only) |
| 1000g and gnomAD Allele Frequency (Human Only) |
| dbNSFP (Human Non-syn mutations only) |

## **CNV Analysis** Please note only available for certain species and will incur additional cost. Please reach a PM team member for more details.

|  |  |  |  |
| --- | --- | --- | --- |
| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| WES/Panel Matched Somatic CNV detection (Panel size > 1 mb) | PureCN (Dragen Pipeline For WGS) |  |  |

## **Structural Variation Analysis** Please note only available for certain species and will incur additional cost. Please reach a PM team member for more details.

|  |  |  |  |
| --- | --- | --- | --- |
| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| Somatic Structural Variation (Only available for WES) | Manta |  |  |

## **HLA Typing**

Please note only available for certain species and will incur additional cost. Please reach a PM team member for more details.

|  |  |  |  |
| --- | --- | --- | --- |
| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| HLA Typing | Hisat Genotyper |  |  |

## **Customized deliverables**

If any customized analysis item(s) is needed, please provide the detailed requirements below, our bioinformatics team will evaluate the feasibility.

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|  |

## **References: Please attach and or provide links of the relevant references as per your project**