NGS Data Analysis Questionnaire (Tumor Only)

## **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 E-I

## **Please specify appropriate species below**

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

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| --- | --- | --- | --- |
| 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 Tumor Only Design**

Total 15 Tumor tissues samples with 10 unmatched controls

## **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 |
| Tumor Only Somatic Mutation Analysis\* | Mutect2 (Dragen Pipeline For WGS) |  |  |

\* Please note, in Tumor Only mode, Mutect2 (or any other variant caller) will contain high number of false positive somatic calls (due to private germline variants) and it is up to researcher to carefully apply downstream filters as per the project requirements.

## **Detailed Annotations (Recommended for Filtering in Tumor Only Analysis)**

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

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| --- | --- | --- | --- |
| **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.

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| **Analysis Item** | **Software** | **Y/N** | **Specific requirements?** |
| WES/Panel Somatic CNV detection (Panel size > 1 mb) \* | PureCN |  |  |

## \* **Only possible if 10 or more processed matched controls are available. Not available for WGS.**

## **HLA Typing**

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

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| **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**