ITD75__VJ1020_S4_R1_001

Overall QC Status: PASS
Sample QC Status: PASS
Variations QC Status: PASS
Anomaly QC Status: PASS
Job:
Type: DNA Structural VariationDNA CNVDNA SNP/InDel Detection: Somatic Mutation Targeted Mutations: Archer Comprehensive Targets
Software Version:
Analysis Date:
Report Creator
Report Date:

Statistics

Molecular Barcode Statistics

Total Fragments	Fragments with Complete Adapter	Number of Reads After Trimming Adapters
8,000,000	7,598,193	5,964,669

Read Statistics

Type	Total Fragments (# / %)	Mapped (# / %)	Pass Alignment Filter (%)	On Target (%)
All Fragments	5,964,669 / 100.0	5,964,213 / 100.0	100.0	98.6
Unique Fragments	2,983,000 / 50.0	2,982,625 / 100.0	100.0	97.5

DNA/RNA Statistics

Type	DNA Reads (# / %)	RNA Reads (# / %)	Ambiguous Reads (# / %)
All Fragments	5,878,351.0 / 100.0	0.0 / 0.0	0.0 / 0.0
Molecular Bins	2,907,501.0 / 100.0	0.0 / 0.0	0.0 / 0.0
Average Molecular Bins per GSP2	3,887.03	0.0	0.0
Unique Start Sites	178,000.0 / 100.0	0.0 / 0.0	0.0 / 0.0
Average Unique Start Sites per GSP2	382.32	0.0	0.0
Average Unique Start Sites per GSP2 Control	0.0	0.0	0.0

QC Statistics

Avg. Unique DNA And Ambiguous Start Sites Per GSP2
382.32

Miscellaneous Statistics

On Target Deduplication Ratio
2.02:1

DNA/RNA Fragment Lengths

DNA Median Fragment Length	DNA Mean Fragment Length	RNA Median Fragment Length	RNA Mean Fragment Length
153.0	166.2	No Fragments	No Fragments

Reportable Variants

NP_056153.2:p.Gly646T rpfsTer12	
Gene: ASXL1	**Location:** chr20:31022441
Ref./ Mutation: A / AG (. PRESENT)	**Depth:** 7466
Allele Fraction: 0.0344	**HGVSp:** NP_056153.2
Mutation Classification: Undefined	
Is Artifact: no	

EZH2	
Gene: EZH2	**Location:** chr7:148543693
Ref./ Mutation: TAA / T (.)	**Depth:** 4497
Allele Fraction: 0.0503	**Clinical Sig.** Likely benign
Mutation Classification: Undefined	**Disease:** Weaver_syndrome
Is Artifact: no	

NP_001745.2:p.Ser424A la	
Gene: RUNX1	**Location:** chr21:36164605
Ref./ Mutation: A / C (.)	**Depth:** 2131
Allele Fraction: 0.0446	**HGVSp:** NP_001745.2
Mutation Classification: Undefined	**Sift:** deleterious(0)
Is Artifact: no	**PolyPhen:** possibly_damaging(0.453)

NP_000537.3:p.Arg248T rp	
Gene: TP53	**Location:** chr17:7577539
Ref./ Mutation: G / A (. PRESENT)	**Depth:** 5841
Allele Fraction: 0.2017	**Clinical Sig.** Pathogenic
Mutation Classification: Undefined	**Disease:** Li-Fraumeni_syndrome_1
Is Artifact: no	**HGVSp:** NP_000537.3
	Sift: deleterious(0)
	PolyPhen: probably_damaging(1)

Reportable CNVs

None Found

Reportable Isoforms

- Passed all strong-evidence filters
- Likely off-target mispriming event
- Exact breakpoint known
- Cross contamination
- User-annotated false positive
- Known fusion partners in Archer Quiver™
- Percent GSP2 reads below threshold
- Fusion expression imbalance
- Low confidence
- User-annotated true positive
- Intronic fusion
- Not enough unique start sites
- Transcriptional readthrough event
- Known ensembl paralogue

DNA Structural Variation: FLT3		
Filters: ☑ **GSP2:** FLT3_chr13_28608285_27_-_A1_GSP2 **Mutation Classification:** Undefined **Is Artifact:** no	**Reads**: 9266 (70.54%) **Start Sites:** 513	**Segments** chr13:28608351→28608255 FLT3(-) NM_004119.2, exon:14 chr13:28608284→28608219 FLT3(-) NM_004119.2, exon:14