RECALL Variant Report (C2)

Summary

Sentieon Variants: 0 in raw | 0 in filt.

DeepVariant Variants: 30435 in raw | 18641 in filt.

HaplotypeCaller Variants: 21138 in raw | 17990 in filt.

DRAGEN Variants: 0 in raw | 0 in filt.

Mode : HQ (15 GQ; 20 DP; 0.3-0.7 VAF(het); VAF(hom))

Intronic: 24981 in raw | 15224 in filt.	Intergenic: 2902 in raw | 1842 in filt.	ncRNA_intronic: 1124 in raw | 700 in filt.
UTR3: 705 in raw | 466 in filt.	Exonic: 289 in raw | 201 in filt.	UTR5: 180 in raw | 78 in filt.
Upstream: 133 in raw | 43 in filt.	ncRNA_exonic: 56 in raw | 41 in filt.	Downstream: 56 in raw | 41 in filt.
Upstream;Downstream: 2 in raw | 2 in filt.	Splicing: 5 in raw | 1 in filt.	

Variant Details

Gene : **MLH1**	Variant : 3:37028857:->C	Type : Exonic	Consequence : Frameshift Insertion
ClinSig : Pathogenic	InterVar : N/A	ClinDN : Lynch Syndrome	gnomAD AF : 6.195e-7
SpliceAI : N/A	CADD : 23.2	REVEL : N/A	SIFT : N/A
Sentieon : N/A	DeepVariant : ✅	HaplotypeCaller : ✅	DRAGEN : N/A

Gene : **FANCC**	Variant : 9:95249225:C>-	Type : Exonic	Consequence : Frameshift Deletion
ClinSig : Pathogenic	InterVar : N/A	ClinDN : Hereditary cancer-predisposing syndrome	gnomAD AF : 0.0002119
SpliceAI : 0.02	CADD : 23.2	REVEL : N/A	SIFT : N/A

Gene : **FANCC**	Variant : 9:95249225:C>-	Type : Exonic	Consequence : Frameshift Deletion
Sentieon : N/A	DeepVariant : ✅	HaplotypeCaller : ✅	DRAGEN : N/A