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.

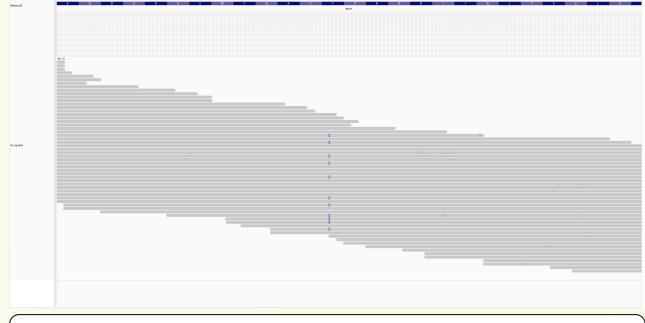
DRAGEN Variants: 0 in raw | 0 in filt.

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
SpliceAl : N/A	CADD : 23.2	REVEL : N/A	SIFT : N/A
Sentieon : N/A	DeepVariant : 🗸	HaplotypeCaller: ✓	DRAGEN : N/A



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

✓ DRAGEN: N/A

