Sequencing Test Report

2019 NOV 07 09:04:11

Sample ID	**Sample ID 1**
Type	**Unknown**
Genome Build	**hg19**
Sex	**Unknown**
Age	**35**
Phenotypes	

CLINICALLY SIGNIFICANT VARIANTS

L1 - National Guidance

Small Variants

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
12:25398281	KRAS	Other	Missense Variant XM_005253365.1 Exon 2 c.38G>A p.(Gly13Asp)	Passed - NGS
1:115256530	NRAS	Predictive	Missense Variant NM_002524.4 Exon 3 c.181C>A p.(Gln61Lys)	Passed - NGS

L2 - Professional Society Guidance

Small Variants

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
3:178936091	PIK3CA	Molecular Classification	Missense Variant NM_006218.2 Exon 10 c.1633G>A p.(Glu545Lys)	Passed - NGS
5:112175323	APC	Molecular Classification	Synonymous Variant XM_005271976.1 Exon 15 c.3855A>G c.3855A>G(p.(Ser1285=))	Passed - NGS
7:55241707	EGFR	Predictive	Missense Variant NM_001346900.1 Exon 18 c.1996G>A p.(Gly666Ser)	Passed - NGS

L3 - Clinical Trial Available

Small Variants

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
2:48030640	MSH6	Prognostic	Frameshift Variant XM_005264271.1 Exon 5 c.2964dupC p.(Phe989LeufsTer5)	Passed - NGS
3:178952085	PIK3CA	Prognostic	Missense Variant NM_006218.2 Exon 21 c.3140A>G p.(His1047Arg)	Passed - NGS
7:55259515	EGFR	Molecular Classification	Missense Variant NM_001346900.1 Exon 21 c.2414T>G p.(Leu805Arg)	Passed - NGS

L4 - Other Reportable Variant

Small Variants

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
4:55599321	KIT	Germline Susceptibility	Missense Variant XM_005265741.1 Exon 17 c.2447A>T p.(Asp816Val)	Passed - NGS
5:112175770	APC	Other	Synonymous Variant XM_005271976.1 Exon 15 c.4302G>A c.4302G>A(p.(Thr1434=))	Passed - NGS
7:140453136	BRAF	Prognostic	Non Coding Transcript Exon Variant XR_242190.1 Exon 15 n.1807T>A	Passed - NGS

L1 - National Guidance

SNV / INDEL

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
12:25398281	KRAS	Other	Missense Variant XM_005253365.1 Exon 2 c.38G>A p.(Gly13Asp)	Passed - NGS

Variant Summary

No data available.

SNV / INDEL

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
1:115256530	NRAS	Predictive	Missense Variant NM_002524.4 Exon 3 c.181C>A p.(Gln61Lys)	Passed - NGS

Variant Summary

No data available.

L2 - Professional Society Guidance

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
3:178936091	PIK3CA	Molecular Classification	Missense Variant NM_006218.2 Exon 10 c.1633G>A p.(Glu545Lys)	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
5:112175323	APC	Molecular Classification	Synonymous Variant XM_005271976.1 Exon 15 c.3855A>G c.3855A>G(p.(Ser1285=))	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
7:55241707	EGFR	Predictive	Missense Variant NM_001346900.1 Exon 18 c.1996G>A p.(Gly666Ser)	Passed - NGS

Variant Summary

No data available.

L3 - Clinical Trial Available

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
2:48030640	MSH6	Prognostic	Frameshift Variant XM_005264271.1 Exon 5 c.2964dupC p.(Phe989LeufsTer5)	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
3:178952085	PIK3CA	Prognostic	Missense Variant NM_006218.2 Exon 21 c.3140A>G p.(His1047Arg)	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
7:55259515	EGFR	Molecular Classification	Missense Variant NM_001346900.1 Exon 21 c.2414T>G p.(Leu805Arg)	Passed - NGS

Variant Summary

No data available.

L4 - Other Reportable Variant

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
4:55599321	KIT	Germline Susceptibility	Missense Variant XM_005265741.1 Exon 17 c.2447A>T p.(Asp816Val)	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION
5:112175770	APC	Other	Synonymous Variant XM_005271976.1 Exon 15 c.4302G>A c.4302G>A(p.(Thr1434=))	Passed - NGS

Variant Summary

No data available.

Small Variants

Variant Details

VARIANT	GENE	ASSOCIATION TYPE	CONSEQUENCE	VALIDATION

7:140453136	BRAF	Prognostic	Non Coding Transcript Exon Variant XR_242190.1 Exon 15 n.1807T>A	Passed - NGS

Variant Summary

No data available.

Appendix

Annotation Data Sources

VEP(91), ClinVar(20190204), COSMIC(84), dbSNP(151), gnomAD(2.1), gnomAD_exome(2.1), MITOMAP(20190225), 1000 Genomes Project (Phase 3 v5a), TOPMed(freeze_5), ClinGen(20160414), DGV(20160515), MITOMAP_SV(20190225), OMIM(20190225), ExAC(0.3.1), phyloP (hg19)