## PACIENTE

| Número.de.Protocolo | Nombre | Fecha.de.recepción.de.la.muestra | Tipo.de.muestra |
| --- | --- | --- | --- |
| 36919 | --- | 01/07/2019 | --- |

## ANÁLISIS

### Tipo de estudio

Secuenciación masiva de ARN total obtenido de tejido tumoral congelado.

### Metodología

Las muestras se prepararon de acuerdo a las guías de preparación del kit TruSeq Stranded Total RNA LT. Las bibliotecas fueron secuenciadas con el secuenciador de la plataforma NovaSeq 6000.

### Softwares utilizados

Para el análisis de los datos de secuenciación se utilizaron los siguientes programas:
1. FastQC v0.11.7 (http://www.bioinformatics.babraham.ac.uk/projects/fastqc/)
2. TrimGalore v0.6.10
3. STAR v2.7.11a
4. ARRIBA v2.4.0
Para realizar el mapeo de las lecturas se utilizó como referencia el hg38 de Ensembl.

## Quality Metrics

| Metrica | Valor | Fuente |
| --- | --- | --- |
| Q\_Mean\_R1 | 36 | QC Secuenciación |
| %\_>=Q30\_R1 | 99 | QC Secuenciación |
| Q\_Mean\_R2 | 36 | QC Secuenciación |
| %\_>=Q30\_R2 | 98 | QC Secuenciación |
| Number of input reads | 58,792,011 | QC Alineamiento |
| Average input read length | 202 | QC Alineamiento |
| Uniquely mapped reads number | 54,730,506 | QC Alineamiento |
| Uniquely mapped reads % | 93 | QC Alineamiento |
| Average mapped length | 202 | QC Alineamiento |

### RESULTADOS

## Fusion 1: LINC03051 - LSAMP

Transcript 1: ENST00000484092

Transcript 2: ENST00000474851

Peptide sequence: .

### Drugs for Gene 1

| gene\_name | drug\_name |
| --- | --- |
| LINC03051 | No drugs approved for this gene |

### Drugs for Gene 2

| gene\_name | drug\_name |
| --- | --- |
| LSAMP | No drugs approved for this gene |

## Fusion 2: KIAA1549 - BRAF

Transcript 1: ENST00000422774

Transcript 2: ENST00000496384

Peptide sequence: PASTAGVGPGVPPGLPANSTPSQEERRATQWGSFYSPAQTANNPCS|DLIRDQGFRGDGGSTT

### Drugs for Gene 1

| gene\_name | drug\_name |
| --- | --- |
| KIAA1549 | No drugs approved for this gene |

### Drugs for Gene 2

| drug\_name | interaction\_type | interaction\_score | approved | anti\_neoplastic |
| --- | --- | --- | --- | --- |
| VEMURAFENIB | activator | 1.657092009619794 | TRUE | TRUE |
| VEMURAFENIB | inhibitor | 1.657092009619794 | TRUE | TRUE |
| DASATINIB ANHYDROUS | inhibitor | 0.082115757 | TRUE | TRUE |
| TEMSIROLIMUS | inhibitor | 0.063440602 | TRUE | TRUE |
| SORAFENIB | inhibitor | 0.091636424 | TRUE | TRUE |
| ENCORAFENIB | inhibitor | 1.282909942931453 | TRUE | TRUE |
| DABRAFENIB | inhibitor | 2.341819737097098 | TRUE | TRUE |
| REGORAFENIB | inhibitor | 0.074975256 | TRUE | TRUE |
| BOSUTINIB | NULL | 0.023905154 | TRUE | TRUE |
| SORAFENIB | NULL | 0.091636424 | TRUE | TRUE |
| VENETOCLAX | NULL | 0.026181836 | TRUE | TRUE |
| ENZALUTAMIDE | NULL | 0.042293734 | TRUE | TRUE |
| LEUCOVORIN CALCIUM | NULL | 0.037149902 | TRUE | TRUE |
| GEFITINIB | NULL | 0.024256701 | TRUE | TRUE |
| ERLOTINIB | NULL | 0.032616354 | TRUE | TRUE |
| NERATINIB | NULL | 0.05890913 | TRUE | TRUE |
| CAPECITABINE | NULL | 0.015272737 | TRUE | TRUE |
| GEMCITABINE | NULL | 0.005912027 | TRUE | TRUE |
| RIFAMPIN | NULL | 0.019636377 | TRUE | FALSE |
| COBIMETINIB | NULL | 0.4734548598913698 | TRUE | TRUE |
| RUXOLITINIB | NULL | 0.032342267 | TRUE | TRUE |
| OSIMERTINIB | NULL | 0.07330914 | TRUE | TRUE |
| AFATINIB | NULL | 0.043406727 | TRUE | TRUE |
| LENVATINIB | NULL | 0.030545475 | TRUE | TRUE |
| ROMIDEPSIN | NULL | 0.019636377 | TRUE | TRUE |
| TALAZOPARIB | NULL | 0.02036365 | TRUE | TRUE |
| IRINOTECAN HYDROCHLORIDE | NULL | 0.043636393 | TRUE | TRUE |
| VALPROIC ACID | NULL | 0.009645939 | TRUE | TRUE |
| HYDROXYCHLOROQUINE | NULL | 0.030545475 | TRUE | FALSE |
| AXITINIB | NULL | 0.024991752 | TRUE | TRUE |
| DABRAFENIB | NULL | 2.341819737097098 | TRUE | TRUE |
| ALECTINIB | NULL | 0.082472782 | TRUE | TRUE |
| OLAPARIB | NULL | 0.012495876 | TRUE | TRUE |
| VORINOSTAT | NULL | 0.014995051 | TRUE | TRUE |
| IMATINIB | NULL | 0.007330914 | TRUE | TRUE |
| VEMURAFENIB | NULL | 1.657092009619794 | TRUE | TRUE |
| FUTIBATINIB | NULL | 0.049983504 | TRUE | TRUE |
| LORLATINIB | NULL | 0.042293734 | TRUE | TRUE |
| TEMSIROLIMUS | NULL | 0.063440602 | TRUE | TRUE |
| DASATINIB ANHYDROUS | NULL | 0.082115757 | TRUE | TRUE |
| OXALIPLATIN | NULL | 0.040230625 | TRUE | TRUE |
| GILTERITINIB | NULL | 0.028937818 | TRUE | TRUE |
| BINIMETINIB | NULL | 0.4948366922735607 | TRUE | TRUE |
| DOXORUBICIN HYDROCHLORIDE | NULL | 0.003665457 | TRUE | TRUE |
| SELUMETINIB | NULL | 0.2332563532602643 | TRUE | TRUE |
| PONATINIB | NULL | 0.021992742 | TRUE | TRUE |
| DOXORUBICIN LIPOSOME | NULL | 0.1374546367426557 | TRUE | FALSE |
| RABEPRAZOLE | NULL | 0.021146867 | TRUE | TRUE |
| ALPELISIB | NULL | 0.070690956 | TRUE | TRUE |
| CHLOROQUINE | NULL | 0.034363659 | TRUE | TRUE |
| SAPANISERTIB | NULL | 0.024991752 | TRUE | TRUE |
| PALBOCICLIB | NULL | 0.03366236 | TRUE | TRUE |
| TIVOZANIB | NULL | 0.06109095 | TRUE | TRUE |
| RIBOCICLIB | NULL | 0.030545475 | TRUE | TRUE |
| TRAMETINIB DIMETHYL SULFOXIDE | NULL | 1.336059069138614 | TRUE | TRUE |
| ENCORAFENIB | NULL | 1.282909942931453 | TRUE | TRUE |
| PANOBINOSTAT | NULL | 0.024991752 | TRUE | TRUE |
| TORIPALIMAB-TPZI | NULL | 0.1099637093941246 | TRUE | FALSE |
| ABEMACICLIB | NULL | 0.028937818 | TRUE | TRUE |
| VINBLASTINE | NULL | 0.016171134 | TRUE | TRUE |
| IDELALISIB | NULL | 0.06109095 | TRUE | TRUE |
| CRIZOTINIB | NULL | 0.010573434 | TRUE | TRUE |
| CELECOXIB | NULL | 0.008206247 | TRUE | TRUE |
| FLUOROURACIL | NULL | 0.013860972 | TRUE | TRUE |
| LAPATINIB | NULL | 0.026604123 | TRUE | TRUE |
| DENOSUMAB | NULL | 0.1832728489902076 | TRUE | TRUE |
| RUCAPARIB | NULL | 0.024991752 | TRUE | TRUE |
| REGORAFENIB | NULL | 0.074975256 | TRUE | TRUE |

Los datos de secuenciación quedarán disponibles para un posible pedido de re-análisis y búsqueda de fusiones en otros genes.

### INFORME

## Interpretación Clínica

## Limitaciones de la técnica

El análisis de fusiones mediante secuenciación de ARN no detecta inserciones o deleciones grandes, ni cambios en el número de copias (CNVs).
Un resultado negativo no descarta la presencia de fusiones o rearreglos que estén por debajo del límite de detección de la prueba.
La presencia de polimorfismos atípicos puede llevar a resultados que sean falsos positivos o negativos.

## Información adicional

Ensayos clínicos disponibles al día de la fecha pueden ser encontrados en los siguientes sitios:
1. ClinicalTrials.gov (www.clinicaltrials.gov)
2. Mayo Clinic (www.mayo.edu/research/clinical-trials)
3. National Cancer Institute (www.cancer.gov/clinicaltrials)

## Referencias