

SOMATIC VARIANT ANNOTATION

**Sample Name** SCLC\_9  
**Date** Thu, Mar 21, 2019  
**Time Processed** 11:06:36 AM

# Clinical Variant #1

**Gene Name** MTHFR  
**Protein Change** A222V  
**Coordinates** chr1:g.11856378G>A  
**ENST ID** ENST00000376592.1

### Variant Description:

N/A

### Associated Assertions:

N/A

### Associated Evidence Items:

|  |  |  |
| --- | --- | --- |
| **Description** | **CIViC EID** | **PubMedID** |
| MTHFR A222V Supports Better Outcome for patients with Pancreatic Cancer | EID1756 | 27819322 |

# Clinical Variant #2

**Gene Name** FCGR2A  
**Protein Change** H167R  
**Coordinates** chr1:g.161479745A>G  
**ENST ID** ENST00000271450.6

### Variant Description:

N/A

### Associated Assertions:

N/A

### Associated Evidence Items:

|  |  |  |
| --- | --- | --- |
| **Description** | **CIViC EID** | **PubMedID** |
| FCGR2A H167R Does Not Support N/A for patients with Breast Cancer | EID1088, EID1084 | 24989892, 22906996 |

# Clinical Variant #3

**Gene Name** FLT3  
**Protein Change** T227M  
**Coordinates** chr13:g.28624294G>A  
**ENST ID** ENST00000241453.7

### Variant Description:

FLT3 T227M (rs1933437) is a common polymorphism with a GMAF around .60 based on the Exome Aggregation Consortium (ExAC) data. Its role in cancer predisposition is still unknown, however it may be associated with the development of leukopenia in patients treated with sunitinib.

### Associated Assertions:

N/A

### Associated Evidence Items:

|  |  |  |
| --- | --- | --- |
| **Description** | **CIViC EID** | **PubMedID** |

# Clinical Variant #4

**Gene Name** TP53  
**Protein Change** P72R  
**Coordinates** chr17:g.7579472G>C  
**ENST ID** ENST00000269305.4

### Variant Description:

This polymorphism is relatively widely studied across cancer types, but meta-analyses in breast, lung and cervical cancer cohorts have so far been inconclusive as to the significance of a patient's genotype at this locus as it relates to cancer susceptibility and prognosis.

### Associated Assertions:

N/A

### Associated Evidence Items:

|  |  |  |
| --- | --- | --- |
| **Description** | **CIViC EID** | **PubMedID** |
| TP53 P72R Does Not Support Positive Predisposition For Cancer for patients with Breast Cancer | EID1302 | 17909070 |
| TP53 P72R Does Not Support Poor Outcome for patients with Cervical Cancer | EID1304 | 11535556 |
| TP53 P72R Does Not Support Poor Outcome for patients with Lung Carcinoma | EID1303 | 12840112 |

# Clinical Variant #5

**Gene Name** ERCC2  
**Protein Change** K751Q  
**Coordinates** chr19:g.45854919T>G  
**ENST ID** ENST00000391945.4

### Variant Description:

N/A

### Associated Assertions:

N/A

### Associated Evidence Items:

|  |  |  |
| --- | --- | --- |
| **Description** | **CIViC EID** | **PubMedID** |
| ERCC2 K751Q Supports Sensitivity/Response to combination of Carboplatin and Paclitaxel for patients with Non-small Cell Lung Carcinoma | EID677 | 19470925 |

# Processing information

**Variants Processed:** 62  
**Clinical Annotations:** 5

OpenCAP is intended for research use only and clinical applications of subsequent panels designed using the SOP would require further panel validation.