

SOMATIC VARIANT ANNOTATION

**Sample Name** SCLC\_hyperlinks  
**Date** Thu, Apr 18, 2019  
**Time Processed** 11:42:17 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:** MTHFR A222V Supports Better Outcome for patients with Pancreatic Cancer

* **CIViC EID(s):** [EID1756](https://civicdb.org/links?idtype=evidence&id=EID1756)
* **PubMed ID(s):** [27819322](https://www.ncbi.nlm.nih.gov/pubmed/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:** FCGR2A H167R Does Not Support N/A for patients with Breast Cancer

* **CIViC EID(s):** [EID1088](https://civicdb.org/links?idtype=evidence&id=EID1088), [EID1084](https://civicdb.org/links?idtype=evidence&id=EID1084)
* **PubMed ID(s):** [24989892](https://www.ncbi.nlm.nih.gov/pubmed/24989892), [22906996](https://www.ncbi.nlm.nih.gov/pubmed/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:

# 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:** TP53 P72R Does Not Support Positive Predisposition For Cancer for patients with Breast Cancer

* **CIViC EID(s):** [EID1302](https://civicdb.org/links?idtype=evidence&id=EID1302)
* **PubMed ID(s):** [17909070](https://www.ncbi.nlm.nih.gov/pubmed/17909070)

**Description:** TP53 P72R Does Not Support Poor Outcome for patients with Cervical Cancer

* **CIViC EID(s):** [EID1304](https://civicdb.org/links?idtype=evidence&id=EID1304)
* **PubMed ID(s):** [11535556](https://www.ncbi.nlm.nih.gov/pubmed/11535556)

**Description:** TP53 P72R Does Not Support Poor Outcome for patients with Lung Carcinoma

* **CIViC EID(s):** [EID1303](https://civicdb.org/links?idtype=evidence&id=EID1303)
* **PubMed ID(s):** [12840112](https://www.ncbi.nlm.nih.gov/pubmed/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:** ERCC2 K751Q Supports Sensitivity/Response to combination of Carboplatin and Paclitaxel for patients with Non-small Cell Lung Carcinoma

* **CIViC EID(s):** [EID677](https://civicdb.org/links?idtype=evidence&id=EID677)
* **PubMed ID(s):** [19470925](https://www.ncbi.nlm.nih.gov/pubmed/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.