

SOMATIC VARIANT ANNOTATION

**Sample Name** SCLC\_9  
**Date** Fri, Jul 19, 2019  
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# Clinical Variant #1

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

### External Databases:

**ClinVar Allele ID:** [3520](https://www.ncbi.nlm.nih.gov/clinvar/variation/3520/)  
**dbSNP ID:** [rs1801133](https://www.ncbi.nlm.nih.gov/snp/rs1801133/)  
**COSMIC ID:** [146404](https://cancer.sanger.ac.uk/cosmic/mutation/overview?id=146404)

### CIViC Variant Description:

N/A

### Associated CIViC Assertions:

N/A

### Associated CIViC Evidence Items:

**Description:** MTHFR A222V Supports Better Outcome for patients with Pancreatic Cancer  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID1756](https://civicdb.org/links?idtype=evidence&id=EID1756) [Wu et al., 2016, Sci Rep](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

### External Databases:

**ClinVar Allele ID:** [14823](https://www.ncbi.nlm.nih.gov/clinvar/variation/14823/)  
**dbSNP ID:** [rs1801274](https://www.ncbi.nlm.nih.gov/snp/rs1801274/)

### CIViC Variant Description:

N/A

### Associated CIViC Assertions:

N/A

### Associated CIViC Evidence Items:

**Description:** FCGR2A H167R Does Not Support N/A for patients with Breast Cancer  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID1088](https://civicdb.org/links?idtype=evidence&id=EID1088) [Norton et al., 2014, Cancer Immunol Res](https://www.ncbi.nlm.nih.gov/pubmed/24989892)  
 [EID1084](https://civicdb.org/links?idtype=evidence&id=EID1084) [Kim et al., 2012, Oncology](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

### External Databases:

**ClinVar Allele ID:** [134447](https://www.ncbi.nlm.nih.gov/clinvar/variation/134447/)  
**dbSNP ID:** [rs1933437](https://www.ncbi.nlm.nih.gov/snp/rs1933437/)

### CIViC 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 CIViC Assertions:

N/A

### Associated CIViC Evidence Items:

N/A

# Clinical Variant #4

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

### External Databases:

**ClinVar Allele ID:** [12351](https://www.ncbi.nlm.nih.gov/clinvar/variation/12351/)  
**dbSNP ID:** [rs1042522](https://www.ncbi.nlm.nih.gov/snp/rs1042522/)  
**COSMIC ID:** [250061](https://cancer.sanger.ac.uk/cosmic/mutation/overview?id=250061)

### CIViC 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 CIViC Assertions:

N/A

### Associated CIViC Evidence Items:

**Description:** TP53 P72R Does Not Support Positive Predisposition For Cancer for patients with Breast Cancer  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID1302](https://civicdb.org/links?idtype=evidence&id=EID1302) [Schmidt et al., 2007, Cancer Res.](https://www.ncbi.nlm.nih.gov/pubmed/17909070)

**Description:** TP53 P72R Does Not Support Poor Outcome for patients with Cervical Cancer  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID1304](https://civicdb.org/links?idtype=evidence&id=EID1304) [Klug et al., 2001, Cancer Epidemiol. Biomarkers Prev.](https://www.ncbi.nlm.nih.gov/pubmed/11535556)

**Description:** TP53 P72R Does Not Support Poor Outcome for patients with Lung Carcinoma  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID1303](https://civicdb.org/links?idtype=evidence&id=EID1303) [Matakidou et al., 2003, Mutagenesis](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

### External Databases:

**ClinVar Allele ID:** [134105](https://www.ncbi.nlm.nih.gov/clinvar/variation/134105/)  
**dbSNP ID:** [rs13181](https://www.ncbi.nlm.nih.gov/snp/rs13181/)

### CIViC Variant Description:

N/A

### Associated CIViC Assertions:

N/A

### Associated CIViC Evidence Items:

**Description:** ERCC2 K751Q Supports Sensitivity/Response to combination of Carboplatin and Paclitaxel for patients with Lung Non-small Cell Carcinoma  
  
 **CIViC ID(s)** **Citation(s)**  
 [EID677](https://civicdb.org/links?idtype=evidence&id=EID677) [Gandara et al., 2009, J. Clin. Oncol.](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.