Gene-Specific Template

(*General Instructions – The main focus of these pages is providing a quick reference for cancer associations and linking to internal and external resources providing genetics-related information for the gene of interest. Use* [*HUGO-approved gene names and symbols*](https://www.genenames.org/) *(italicized when appropriate) and* [*HGVS-based nomenclature for variants*](https://varnomen.hgvs.org/) *if applicable. Please complete the table and do not delete it. The use of bullet points rather than large paragraphs is encouraged. Additional instructions below in italicized blue text or examples should not be included in the final page content. Please also see* [*Author Instructions*](https://ccga.io/index.php/Author_Instructions) *and* [*FAQs*](https://ccga.io/index.php/Frequently_Asked_Questions_%28FAQs%29) *as well as contact your* [*Associate Editor*](https://ccga.io/index.php/Leadership) *or* *Technical Support**.*)

**Primary Author(s)\***

Put your text here (*Name and affiliation; EXAMPLE: Jane Smith, PhD, Institute of Genomics*)

**Gene Characteristics**

|  |  |
| --- | --- |
| **Synonyms** | EXAMPLE: Tumor protein p53, 'LFS1, p53, BCC7, TRP53 |
| **Cytoband** | EXAMPLE: 17p13.1 |
| **Genomic Coordinates** | EXAMPLE: chr17:7,668,402-7,687,538 [GRCh38/hg38]EXAMPLE: chr17:7,571,720-7,590,868 [GRCh37/hg19] |
| **Gene/Protein Native Function** | EXAMPLES: Growth factor, DNA repair, Apoptosis |

**Gene in Cancer Overview**

* Put your text here
* Put your text here
* Put your text here

(*Instruction: Use a list of three bullets, targeting one to three sentences each for gene function, germline associations, and somatic associations. Include gene/protein function in cancer such as tumor suppressor, oncogene or other. The information provided can include high level clinically significant details such as if mutations in this gene confirm or exclude certain diseases. Of note, this gene page is not meant to recapitulate the very detailed content on related linked disease entity pages.*)

EXAMPLE:

* *TP53* is a well-described tumor suppressor gene that functions in apoptosis, genomic stability and inhibition of angiogenesis (RefSeq, Dec 2016).
* Germline loss-of-function alterations, including deletions, are associated with Li-Fraumeni cancer predisposition syndrome (LFS). Individuals with LFS are at an increased risk of developing cancer, particularly sarcomas (PMID: 20586629, 27621308, 25896519).
* Somatic *TP53* alterations are reported at a high frequency across a wide range of tumors, arising in ~30% of all cancer patients (PMID: 25400752, 27239089, cbioportal.org accessed 6/2/21).

**Internal Pages**

Put your link placeholder here (links will be converted using the link icon at top of page in the CCGA site)

**External Links**

Put your text here - Include as applicable links to the following using the example format provided:

1) Atlas of Genetics and Cytogenetics in Oncology and Haematology

2) COSMIC

3) CIViC

4) St. Jude ProteinPaint

5) Precision Medicine Knowledgebase (Weill Cornell)

6) Cancer Index

7) OncoKB

8) NCBI Gene

9) My Cancer Genome

10) UniProt

11) Pfam

12) GeneCards

13) OMIM

14) LOVD(3) - Leiden Open Variation Database

15) TICdb - database of Translocation breakpoints In Cancer

16) GeneReviews

17) ClinGen

18) Any gene-specific databases.

EXAMPLES (these may be filled in already)

'''[http://atlasgeneticsoncology.org/Genes/P53ID88.html ''TP53'' by Atlas of Genetics and Cytogenetics in Oncology and Haematology]''' - detailed gene information

'''[https://cancer.sanger.ac.uk/cosmic/gene/analysis?ln=TP53 ''TP53'' by COSMIC]''' - sequence information, expression, catalogue of mutations

'''[https://civicdb.org/links/entrez\_name/TP53 by CIViC]''' - general knowledge and evidence-based variant specific information

'''[http://p53.iarc.fr/ ''TP53'' by IARC]''' - ''TP53'' database with reference sequences and mutational landscape

'''[https://pecan.stjude.cloud/proteinpaint/tp53 ''TP53'' by St. Jude ProteinPaint]''' mutational landscape and matched expression data.

'''[https://pmkb.weill.cornell.edu/search?utf8=%E2%9C%93&search=tp53 ''TP53'' by Precision Medicine Knowledgebase (Weill Cornell)]''' - manually vetted interpretations of variants and CNVs

'''[http://www.cancerindex.org/geneweb/TP53.htm ''TP53'' by Cancer Index]''' - gene, pathway, publication information matched to cancer type

'''[http://oncokb.org/#/gene/TP53 ''TP53'' by OncoKB]''' - mutational landscape, mutation effect, variant classification

'''[https://www.ncbi.nlm.nih.gov/gene/2122 ''MECOM'' by NCBI Gene]''' - brief gene overview

'''[https://www.mycancergenome.org/content/gene/tp53/ ''TP53'' by My Cancer Genome]''' - brief gene overview

'''[http://www.uniprot.org/uniprot/P04637 ''TP53'' by UniProt]''' - protein and molecular structure and function

'''[https://pfam.xfam.org/family/p53 ''TP53'' by Pfam]''' - gene and protein structure and function information

'''[http://www.genecards.org/cgi-bin/carddisp.pl?gene=tp53 ''TP53'' by GeneCards]''' - general gene information and summaries

'''[http://www.omim.org/entry/165215 ''MECOM'' by OMIM]''' - compendium of human genes and genetic phenotypes

'''[https://databases.lovd.nl/shared/genes/MECOM ''MECOM'' by LOVD(3)]''' - Leiden Open Variation Database

'''[http://www.unav.es/genetica/TICdb/results.php?hgnc=MECOM ''MECOM'' by TICdb]''' - database of Translocation breakpoints In Cancer

'''[https://search.clinicalgenome.org/kb/genes/HGNC:11998]''' - germline gene significance with expert curation

'''[https://www.ncbi.nlm.nih.gov/books/NBK1311/ GeneReviews]''' - information on Li Fraumeni Syndrome

**Additional Information**

Put your text here

**References**

(*Instruction: Add PMIDs into the text above where references are appropriate - PMIDs will be used to insert references on the CCGA site* *and the reference list automatically generated*)

(*Instruction: If a PMID is not available, such as for a book, please include the entire reference in this section*)

BOOK EXAMPLE: Arber DA, et al., (2017). Acute myeloid leukaemia with recurrent genetic abnormalities, in World Health Organization Classification of Tumours of Haematopoietic and Lymphoid Tissues, Revised 4th edition. Swerdlow SH, Campo E, Harris NL, Jaffe ES, Pileri SA, Stein H, Thiele J, Arber DA, Hasserjian RP, Le Beau MM, Orazi A, and Siebert R, Editors. IARC Press: Lyon, France, p130-149.

INTERNET RESOURCE EXAMPLE:

Author name(s). Date (if possible). page title, website title, web address, and date accessed.

Cingam, S. R. and Koshy, N.V. (2017). Cancer, Leukemia, Promyelocytic, Acute (APL, APML). https://www.ncbi.nlm.nih.gov/books/NBK459352/ Accessed August 3, 2018.

**Notes**

\*Primary authors will typically be those that initially create and complete the content of a page. If a subsequent user modifies the content and feels the effort put forth is of high enough significance to warrant listing in the authorship section, please contact the CCGA coordinators (contact information provided on the homepage). Additional global feedback or concerns are also welcome.