Disease or Entity-Specific Template with Instructions

*(General Instructions – The main focus of these pages is the clinically significant genetic alterations in each disease type. Use* [*HUGO-approved gene names and symbols*](https://www.genenames.org/) *(italicized when appropriate),* [*HGVS-based nomenclature for variants*](https://varnomen.hgvs.org/)*, as well as generic names of drugs and testing platforms or assays if applicable. Please complete tables whenever possible and do not delete them (add N/A if not applicable in the table and delete the examples). Please do not delete or alter the section headings. The use of bullet points alongside short blocks of text rather than only large paragraphs is encouraged. Additional instructions below in italicized blue text 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_(FAQs)) *as well as contact your* [*Associate Editor*](https://ccga.io/index.php/Leadership) *or* [*Technical Support*](mailto:CCGA@cancergenomics.org)*.)*

**Primary Author(s)\***

Put your text here (EXAMPLE*:* Jane Smith, PhD)

**WHO Classification of Disease**

(Will be autogenerated; Book will include name of specific book and have a link to the online WHO site)

|  |  |
| --- | --- |
| Book |  |
| Category |  |
| Family |  |
| Type |  |
| Subtype(s) |  |

**Definition/Description of Disease**

Put your text here (*Instructions: Brief description of approximately one paragraph - include disease context relative to other WHO classification categories, diagnostic criteria if applicable, and differential diagnosis if applicable. Other classifications can be referenced for comparison.*)

**Synonyms/Terminology**

Put your text here (*Instructions: Include currently used terms and major historical ones, adding “(historical)” after the latter.*)

**Epidemiology/Prevalence**

Put your text here

**Clinical Features**

Put your text here and fill in the table (*Instruction: Can include references in the table. Do not delete table.*)

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| **Signs and Symptoms** | EXAMPLE: Asymptomatic (incidental finding on complete blood counts)  EXAMPLE: B-symptoms (weight loss, fever, night sweats)  EXAMPLE: Lymphadenopathy (uncommon) |
| **Laboratory Findings** | EXAMPLE: Cytopenias  EXAMPLE: Lymphocytosis (low level) |

**Sites of Involvement**

Put your text here (*Instruction: Indicate physical sites; EXAMPLE: nodal, extranodal, bone marrow*)

**Morphologic Features**

Put your text here (*Instructions: Brief description typically of approximately one paragraph)*

**Immunophenotype**

Put your text here and fill in the table (*Instruction: Can include references in the table. Do not delete table.*)

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| **Positive (universal)** | EXAMPLE: CD1 |
| **Positive (subset)** |  |
| **Negative (universal)** |  |
| **Negative (subset)** |  |

**Chromosomal Rearrangements (Gene Fusions)**

Put your text here and fill in the table (*Instruction: Can include references in the table. Do not delete table.*)

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| **Chromosomal Rearrangement** | **Genes in Fusion**  **(5’ or 3’ Segments)** | **Pathogenic Derivative** | **Prevalence** | **Diagnostic Significance (Yes, No or Unknown)** | **Prognostic Significance (Yes, No or Unknown)** | **Therapeutic Significance (Yes, No or Unknown)** | **Notes** |
| EXAMPLE: t(9;22)(q34;q11.2) | EXAMPLE: 3'ABL1 / 5'BCR | EXAMPLE: der(22) | EXAMPLE: 20% (COSMIC)  EXAMPLE: 30% (add reference) | EXAMPLE: Yes | EXAMPLE: No | EXAMPLE: Yes | EXAMPLE:  The t(9;22) is diagnostic of CML in the appropriate morphology and clinical context (add reference). This fusion is responsive to targeted therapy such as Imatinib (Gleevec) (add reference). |
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**Individual Region Genomic Gain/Loss/LOH**

Put your text here and fill in the table (*Instructions: Includes aberrations not involving gene fusions. Can include references in the table. Can refer to CGC workgroup tables as linked on the homepage if applicable. Do not delete table.*)

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| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr #** | **Gain/Loss/Amp/LOH** | **Minimal Region Genomic Coordinates [Genome Build]** | **Minimal Region Cytoband** | **Diagnostic Significance (Yes, No or Unknown)** | **Prognostic Significance**  **(Yes, No or Unknown)** | **Therapeutic Significance**  **(Yes, No or Unknown)** | **Notes** |
| EXAMPLE:  7 | EXAMPLE: Loss | EXAMPLE:  chr7:1-159,335,973 [hg38] | EXAMPLE:  chr7 | EXAMPLE: Yes | EXAMPLE: Yes | EXAMPLE: No | EXAMPLE:  Presence of monosomy 7 (or 7q deletion) is sufficient for a diagnosis of AML with MDS-related changes when there is ≥20% blasts and no prior therapy (add reference). Monosomy 7/7q deletion is associated with a poor prognosis in AML (add reference). |
| EXAMPLE:  8 | EXAMPLE: Gain | EXAMPLE:  chr8:1-145,138,636 [hg38] | EXAMPLE:  chr8 | EXAMPLE: No | EXAMPLE: No | EXAMPLE: No | EXAMPLE:  Common recurrent secondary finding for t(8;21) (add reference). |
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**Characteristic Chromosomal Patterns**

Put your text here *(EXAMPLE PATTERNS: hyperdiploid; gain of odd number chromosomes including typically chromosome 1, 3, 5, 7, 11, and 17; co-deletion of 1p and 19q; complex karyotypes without characteristic genetic findings; chromothripsis. Do not delete table.)*

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| **Chromosomal Pattern** | **Diagnostic Significance (Yes, No or Unknown)** | **Prognostic Significance**  **(Yes, No or Unknown)** | **Therapeutic Significance**  **(Yes, No or Unknown)** | **Notes** |
| EXAMPLE:  Co-deletion of 1p and 18q | EXAMPLE: Yes | EXAMPLE: No | EXAMPLE: No | EXAMPLE:  See chromosomal rearrangements table as this pattern is due to an unbalanced derivative translocation associated with oligodendroglioma (add reference). |
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**Gene Mutations (SNV/INDEL)**

Put your text here and fill in the table (*Instructions: This table is not meant to be an exhaustive list; please include only genes/alterations that are recurrent and common as well either disease defining and/or clinically significant. Can include references in the table. For clinical significance, denote associations with FDA-approved therapy (not an extensive list of applicable drugs) and NCCN or other national guidelines if applicable; Can also refer to CGC workgroup tables as linked on the homepage if applicable as well as any high impact papers or reviews of gene mutations in this entity. Do not delete table.)*

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| **Gene; Genetic Alteration** | **Presumed Mechanism (Tumor Suppressor Gene (TSG)/Oncogene/Other)** | **Prevalence (COSMIC/ TCGA/Other)** | **Concomitant Mutations** | **Mutually Exclusive Mutations** | **Diagnostic Significance (Yes, No or Unknown)** | **Prognostic Significance**  **(Yes, No or Unknown)** | **Therapeutic Significance**  **(Yes, No or Unknown)** | **Notes** |
| EXAMPLE: *TP53*; Variable LOF mutations  EXAMPLE:  *EGFR*; Exon 20 mutations  EXAMPLE: *BRAF*; Activating mutations | EXAMPLE: TSG | EXAMPLE: 20% (COSMIC)  EXAMPLE: 30% (add Reference) | EXAMPLE: *IDH1* R123H | EXAMPLE: *EGFR* amplification | EXAMPLE: Yes | EXAMPLE: No | EXAMPLE: No | EXAMPLE: Excludes hairy cell leukemia (HCL) (add reference). |
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Note: A more extensive list of mutations can be found in cBioportal (<https://www.cbioportal.org/>), COSMIC (<https://cancer.sanger.ac.uk/cosmic>), ICGC (<https://dcc.icgc.org/>) and/or other databases. When applicable, gene-specific pages within the CCGA site directly link to pertinent external content.

**Epigenomic Alterations**

Put your text here

**Genes and Main Pathways Involved**

Put your text here and fill in the table *(Instructions: Can include references in the table. Do not delete table.)*

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| **Gene; Genetic Alteration** | **Pathway** | **Pathophysiologic Outcome** |
| EXAMPLE: *BRAF* and *MAP2K1*; Activating mutations | EXAMPLE: MAPK signaling | EXAMPLE: Increased cell growth and proliferation |
| EXAMPLE: *CDKN2A*; Inactivating mutations | EXAMPLE: Cell cycle regulation | EXAMPLE: Unregulated cell division |
| EXAMPLE: *KMT2C* and *ARID1A*; Inactivating mutations | EXAMPLE: Histone modification, chromatin remodeling | EXAMPLE: Abnormal gene expression program |
|  |  |  |

**Genetic Diagnostic Testing Methods**

Put your text here

**Familial Forms**

Put your text here (*Instructions: Include associated hereditary conditions/syndromes that cause this entity or are caused by this entity.*)

**Additional Information**

Put your text here

**Links**

Put a link here or anywhere appropriate in this page (*Instructions: Links can be added to internal pages by the page name or to an external internet address by including the http://www. portion; links will be created using the link icon at the top of the page in the CCGA site.*)

**References**

(use "Cite" icon at top of page)

(*Instructions: 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 will be automatically generated. If a PMID is not available, such as for a book, please include the entire reference in this section.*)

**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.