Disease or Entity-Specific Template

*(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. 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 (*Name and affiliation; example:* Jane Smith, PhD, Institute of Genomics)

**Cancer Category/Type**

Put your text here

**Cancer Sub-Classification/Subtype**

Put your text here

**Definition/Description of Disease**

Put your text here (*Instructions: Brief description of approximately one paragraph - include disease context relative to other WHO classification categories referring to the specific WHO book pages, diagnostic criteria if applicable, and differential diagnosis if applicable*)

**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*)

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| --- | --- |
| **Signs and Symptoms** | EXAMPLE Asymptomatic (incidental finding on complete blood counts)  EXAMPLE B-symptoms (weight loss, fever, night sweats)  EXAMPLE Fatigue  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

**Immunophenotype**

Put your text here and fill in the table (*Instruction: Can include references in the table*)

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| --- | --- |
| **Positive (universal)** | EXAMPLE CD1 |
| **Positive (subset)** | EXAMPLE CD2 |
| **Negative (universal)** | EXAMPLE CD3 |
| **Negative (subset)** | EXAMPLE CD4 |

**Chromosomal Rearrangements (Gene Fusions)**

Put your text here and fill in the 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) | Yes | No | 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.*)

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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 | Yes | Yes | 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 | No | No | 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)*

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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 | Yes | No | 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.)*

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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: 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.)*

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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 |
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**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 your text placeholder here (use "Link" icon at top of page) (*Instructions: For example, link to related gene pages within the site. Note: links will be converted using the link icon at top of page in the CCGA site.*)

**References**

(use "Cite" icon at top of page)

(*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.

**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.