

Pathology Mutations

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Lecture 1

Disease	Mutation
Thrombotic Thrombocytopenic Purpura (TTP)	ADAMTS13

Lecture 2

Disease	Mutation
Hemophagocytic lymphohistocytosis (Infants and young children)	PRF1
Hemophagocytic lymphohistocytosis (Adolescents and adults)	SLAM (it's not a gene)

Lecture 3

Disease	Mutation	CD
Nodular sclerosis (MC)		CD15, CD3
Mixed cellularity		CD15, CD3
Lymphocyte-rich		CD45+ and CD20+, negative for CD30 and CD15
Nodular lymphocyte-predominant		CD20+, negative for CD30 and CD15
Diffuse Large B-cell Lymphoma	Bcl6 promoter gene (60%), t(14;18) (Bcl2-IgH fusion) (30%), MYC gene.	CD20

Disease	Mutation	CD
Follicular Lymphoma	t(14;18) (Bcl2-IgH), histone-modifying proteins (epigenetic).	CD20, CD10, Bcl2, Bcl6.
Burkitt Lymphoma	t(8;14) (MYC-IgH)	CD20, CD10, Bcl6
Mantle Cell Lymphoma	t(11;14): ↑ expression of cyclinD1	CD5, CD20
Small Lymphocytic Lymphoma		CD20, Bcl2 and CD5.
Peripheral T-cell Lymphoma		CD2, CD3, CD5, CD7, but TdT-

Disease	Mutation	CD
Cutaneous T-cell Lymphoma		CD4+ T-cell.
Adult T-cell Lymphoma/Leukemia		CD4,CD25

Lecture 4

Disease	Mutation	CD
B-ALL	PAX5	CD10+, CD34+ and TdT+
Childhood B-ALL	t(12;21), ETV6 and TUNX1 genes	
Adult B-ALL	t(9;22), ABL-BCR genes (Philadelphia chromosome)	
T-ALL	NOTCH1, PTEN, CDKN2A	CD7+, TdT+
Acute Myeloid Leukemia (AML)	tyrosine kinase pathways (RAS),IDH	CD34, MPO, CD13, CD33

Disease	Mutation	CD
Acute Promyelocytic Leukemia	Due to t(15:17): PML-RARA fusion	negative for CD34
Chronic Lymphoblastic Leukemia (CLL)		CD20, Bcl2 and CD5.
Plasma Cell Myeloma (Multiple Myeloma)	t(11;14) IgH-cyclinD1 and cyclinD3,MYC	
Hairy Cell Leukemia	serine/threonine kinase BRAF gene.	

Lecture 5

Disease	Mutation
Myelodysplastic Syndrome	Chromosomal abnormality in 50% of cases: monosomy 5, monosomy 7, deletions of 5q, 7q, 20q, trisomy 8. Mutations in epigenetic factor. RNA splicing factors Transcription factors. P53
Chronic Myeloid Leukemia	t(9;22) Philadelphia chromosome
Primary Myelofibrosis	50% have mutation in JAK2, 5% in MPL gene

Disease	Mutation
Essential Thrombocythemia	JAK2
Langerhans Cells Histiocytosis	BRAF serine-threonine kinase.