CANCER GENOMICS CONSORTIUM

Educating for Best Practices in Clinical Cancer Genomics

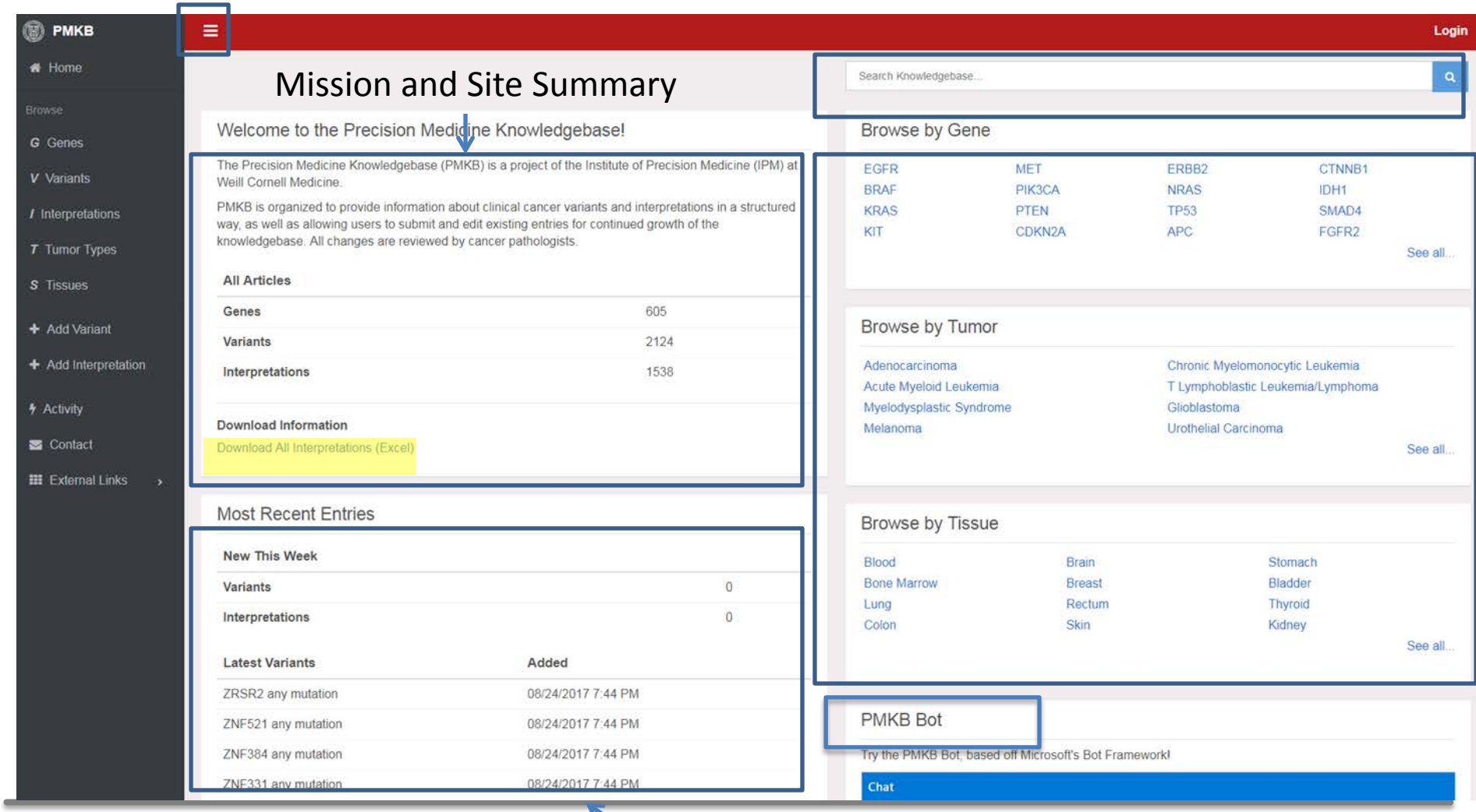
Precision Medicine Knowledgebase (PMKB)

https://pmkb.weill.cornell.edu/

https://academic.oup.com/jamia/article/24/3/513/2418181/The-cancer-precision-medicine-knowledge-base-for

Home Page

Click on this to expand/collapse side menu



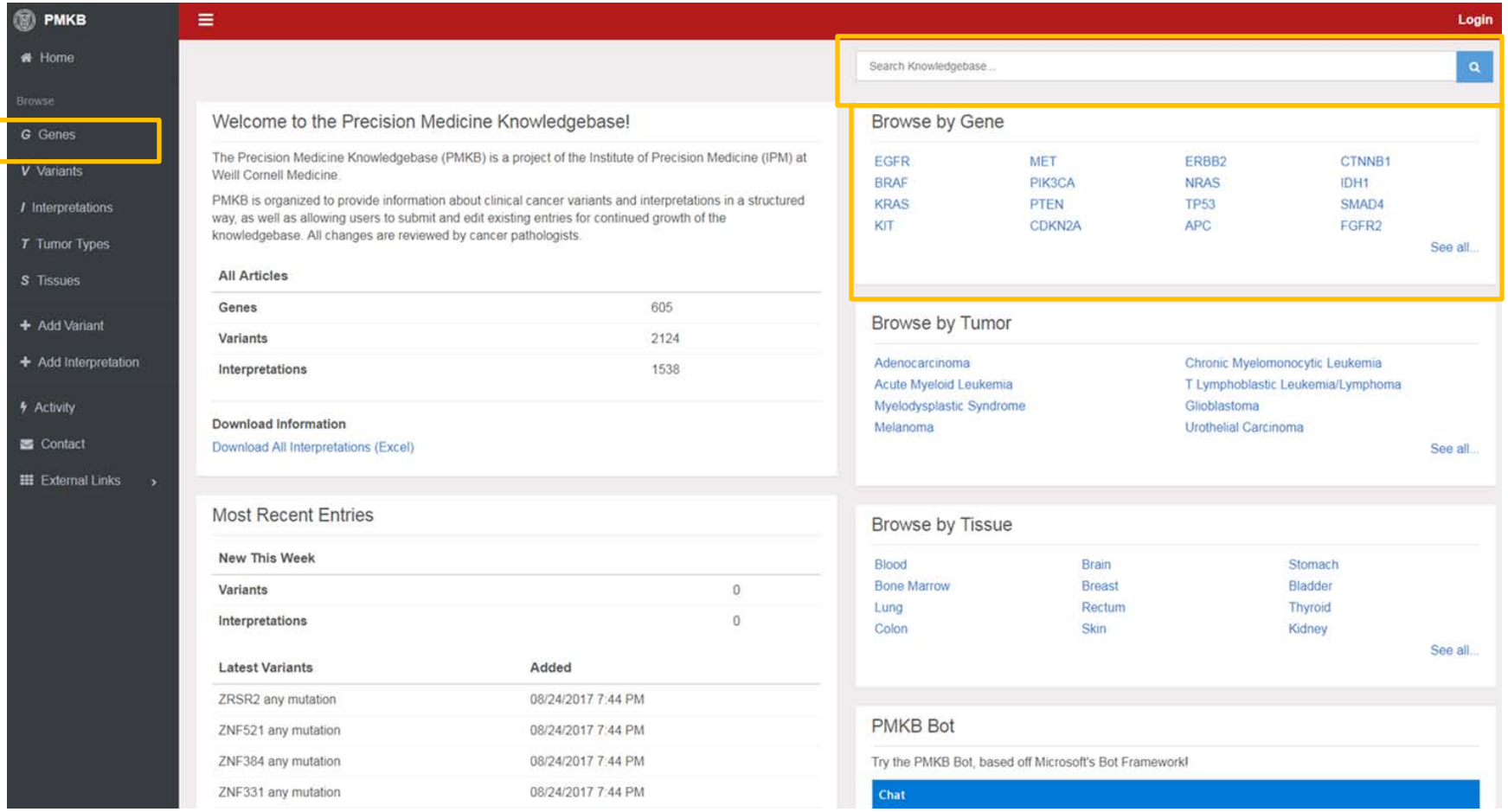
Mission and Site Summary

Recent Additions to PMKB

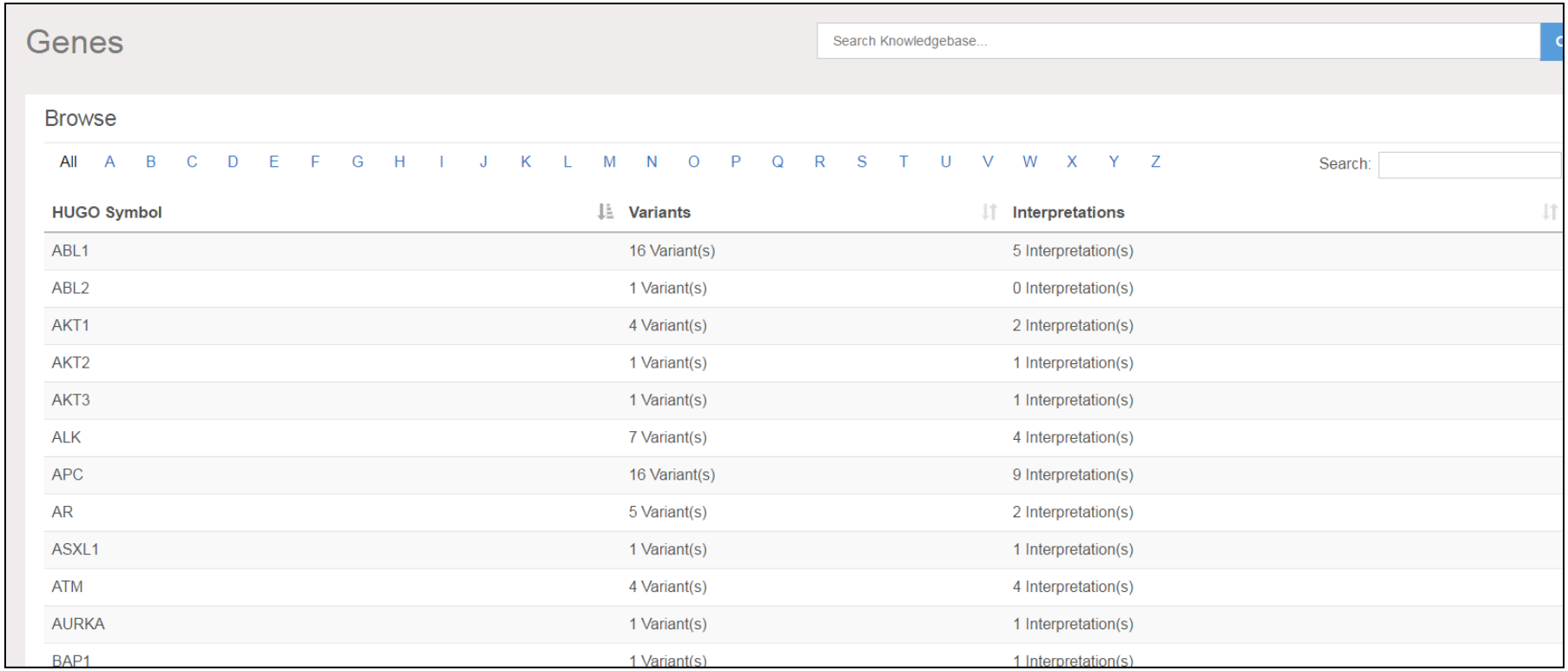
CANCER GENOMICS CONSORTIUM

Educating for best practices in clinical cancer genomics

Search for Genes



Browse Genes



Gene Page

PMKB

ABL1

Search Knowledgebase...

Interpretations

By Variant(s)

- ABL1 any mutation
- ABL1 D276G
- ABL1 E255K
- ABL1 E255V
- ABL1 G250E
- ABL1 H396P
- ABL1 L387M
- ABL1 M351T
- ABL1 Q252H
- ABL1 T315I
- ABL1 V299L
- ABL1 Y253F
- ABL1 Y253H
- ABL1 F359V
- ABL1 M244V
- ABL1 E355G

By Tumor

- Acute Myeloid Leukemia
  - ABL1 F359V
- B Lymphoblastic Leukemia/Lymphoma
  - ABL1 any mutation, ABL1 T315I
- Chronic Myeloid Leukemia
  - ABL1 any mutation, ABL1 T315I
  - ABL1 F359V
  - ABL1 E355G
  - ABL1 M244V
  - ABL1 F359V
- Other Acute Leukemia
  - ABL1 F359V
  - ABL1 E355G
  - ABL1 M244V

Variants

+ Add New Variant

Show 10 entries Search:

Gene	Type	Description	COSMIC ID	DNA Change (Coding Nucleotide)	Exon
ABL1	any	ABL1 any mutation			
ABL1	missense	ABL1 D276G	COSM12602	827A>G	5
ABL1	missense	ABL1 E255K	COSM12573	763G>A	4
ABL1	missense	ABL1 E255V	COSM12574	764A>T	4
ABL1	missense	ABL1 G250E	COSM12577	749G>A	4
ABL1	missense	ABL1 H396P	COSM12564	1187A>C	7
ABL1	missense	ABL1 L387M	COSM131574	1159T>A	7
ABL1	missense	ABL1 M351T	COSM12578	1052T>C	6
ABL1	missense	ABL1 Q252H	COSM12609	756G>C	4
ABL1	missense	ABL1 T315I	COSM12560	944C>T	6

Showing 1 to 10 of 16 entries First Previous 1 2 Next Last

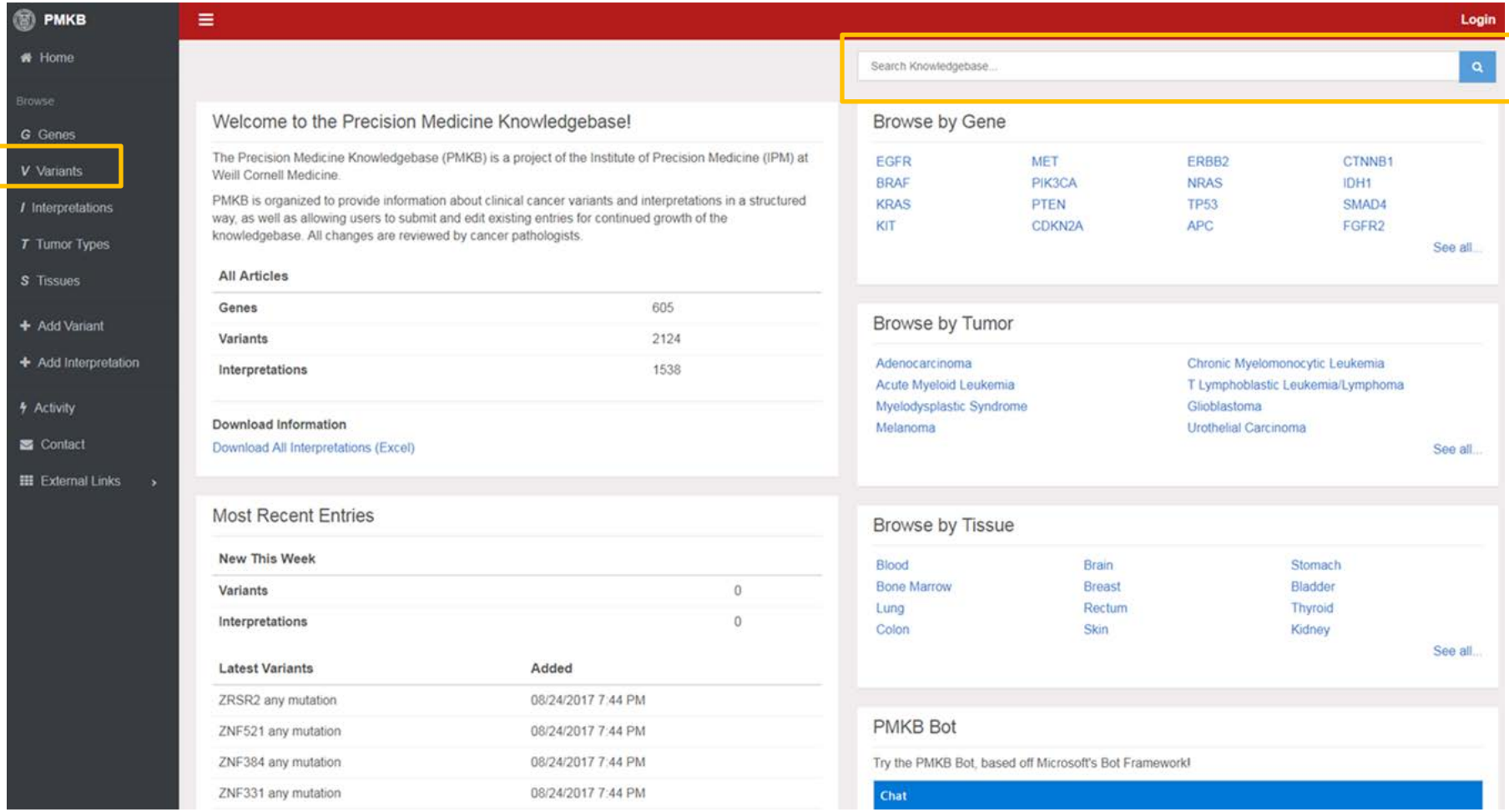
Interpretations

+ Add New Interpretation

Show 10 entries Search:

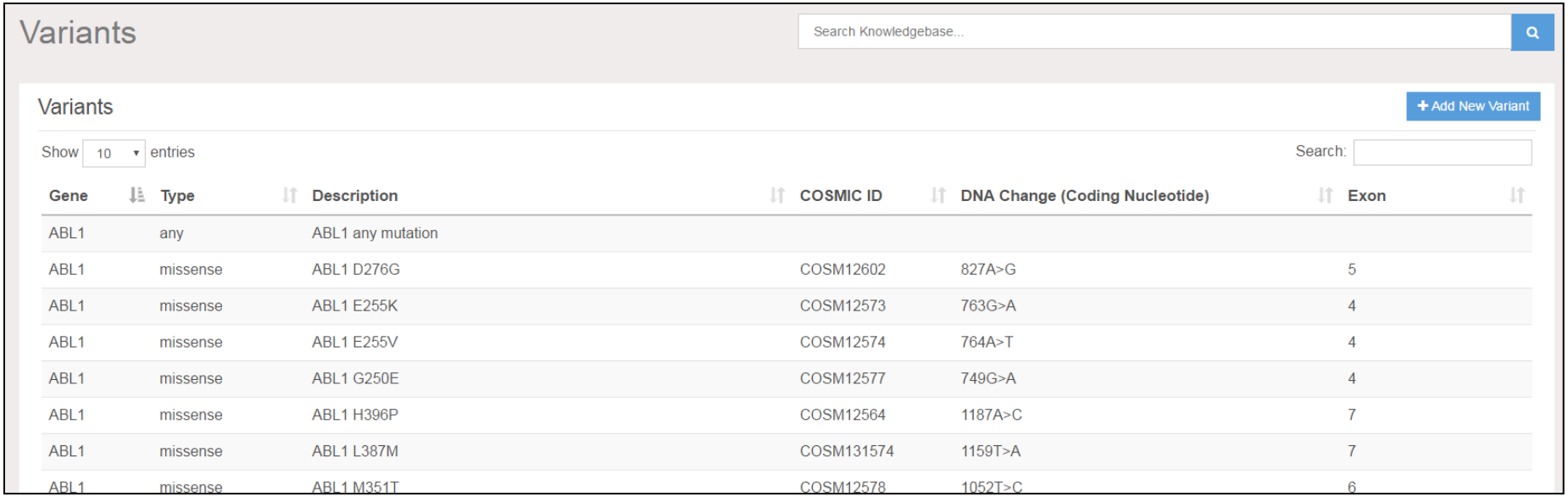
Tumor	Tissue	Variants	Interpretation
Chronic Myeloid Leukemia Acute Myeloid Leukemia Other Acute Leukemia	Blood Bone Marrow	ABL1 F359V	The p.F359V mutation in the context of the BCR-ABL1 translocation may associated with resistance to ABL kinase inhibitors.
Chronic Myeloid Leukemia B Lymphoblastic	Blood Bone Marrow	ABL1 any mutation	ABL1 kinase domain mutations in Philadelphia chromosome positive acute lymphoblastic leukemia and chronic myelogenous leukemia are associated with resistance to some types of tryosine kinase inhibitor therapy. The various mutations span several hundred amino acids (M237 thru E507) and vary in their

Search for Variants



- Either search by variants through the gene page or on their own using either of the boxed regions above. Includes some copy number variants.

Browse Variants

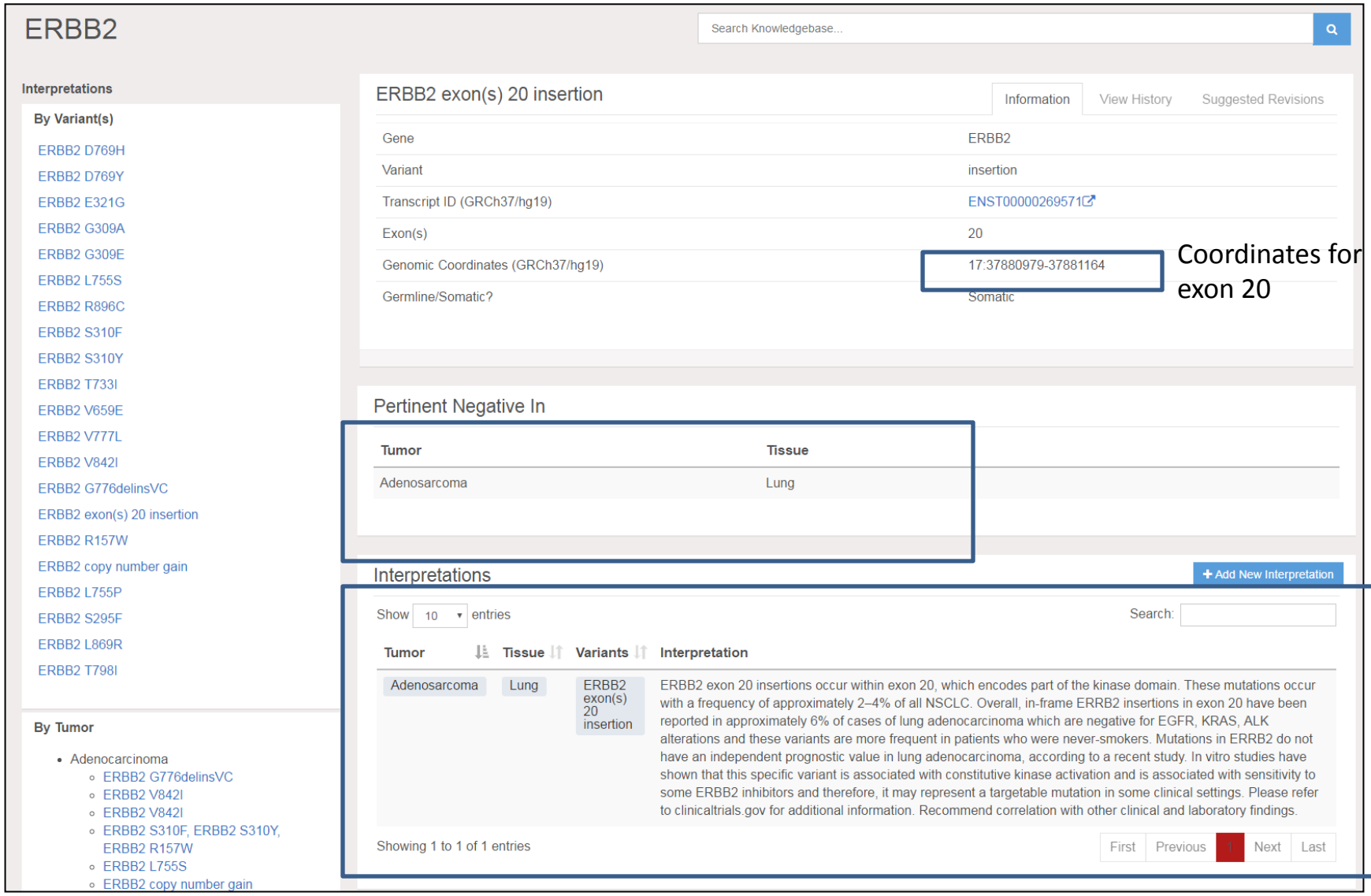


Gene	Type	Description	COSMIC ID	DNA Change (Coding Nucleotide)	Exon
ABL1	any	ABL1 any mutation			
ABL1	missense	ABL1 D276G	COSM12602	827A>G	5
ABL1	missense	ABL1 E255K	COSM12573	763G>A	4
ABL1	missense	ABL1 E255V	COSM12574	764A>T	4
ABL1	missense	ABL1 G250E	COSM12577	749G>A	4
ABL1	missense	ABL1 H396P	COSM12564	1187A>C	7
ABL1	missense	ABL1 L387M	COSM131574	1159T>A	7
ABL1	missense	ABL1 M351T	COSM12578	1052T>C	6

- 513 Variants described in PMKB

Variant Page

PMKB



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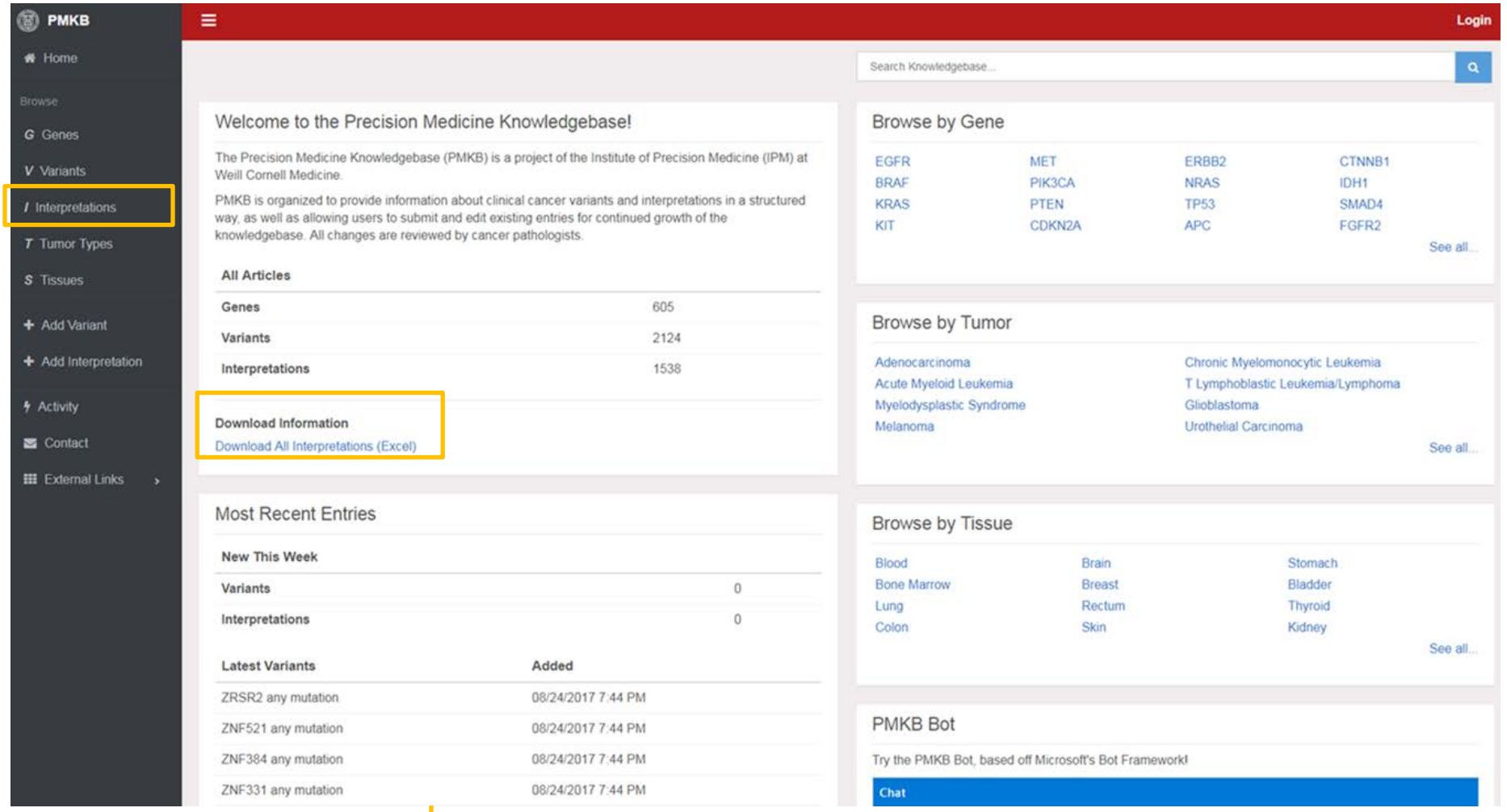
Interpretations



Scroll down to view latest interpretations

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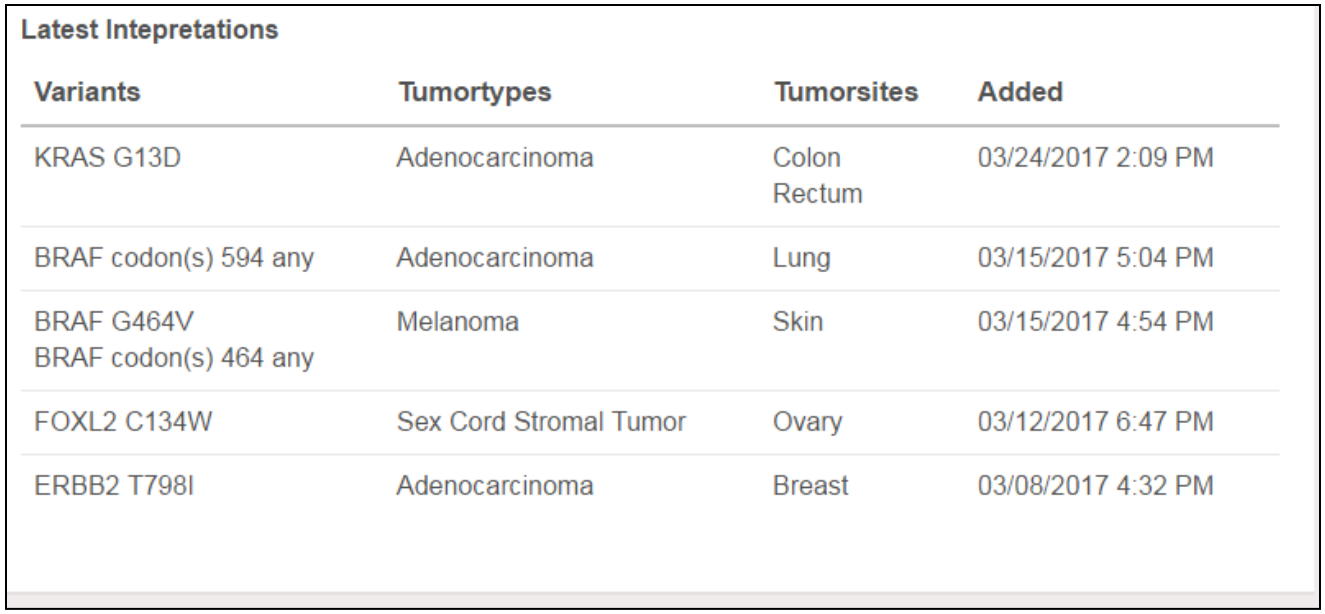
View Latest Interpretations



Latest Intepretations

Variants	Tumortypes	Tumorsites	Added
KRAS G13D	Adenocarcinoma	Colon Rectum	03/24/2017 2:09 PM
BRAF codon(s) 594 any	Adenocarcinoma	Lung	03/15/2017 5:04 PM
BRAF G464V BRAF codon(s) 464 any	Melanoma	Skin	03/15/2017 4:54 PM
FOXL2 C134W	Sex Cord Stromal Tumor	Ovary	03/12/2017 6:47 PM
ERBB2 T798I	Adenocarcinoma	Breast	03/08/2017 4:32 PM

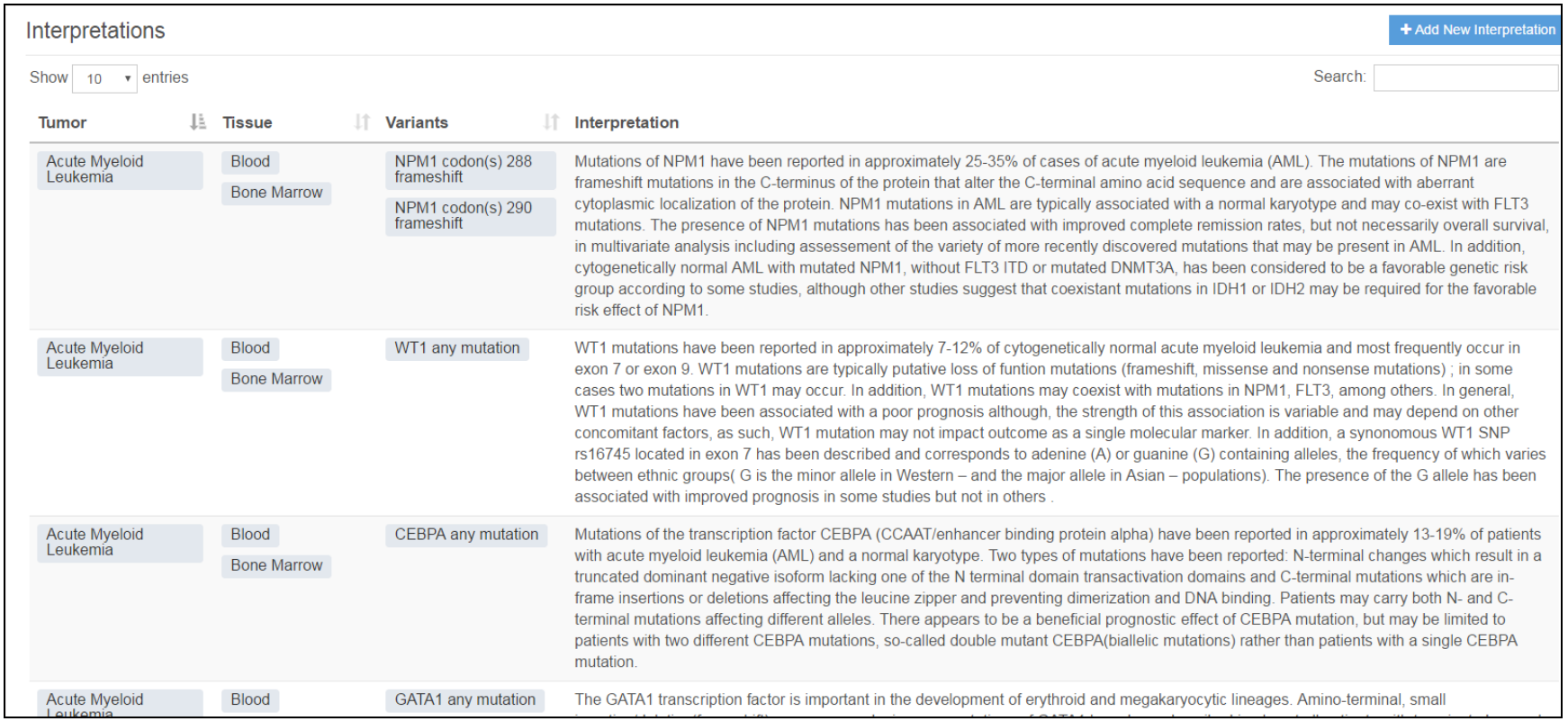
- At bottom of home page

Excel of All Interpretations Available for Download

Gene	Tumor Type(s)	Tissue Type(s)	Variant(s)	Tier	Interpretations	Citations	
CSF3R	Myeloproliferative Neoplasm, Chronic Neutrophilic Leukemia, Atypical Chronic Myeloid Leukemia	Blood, Bone Marrow	CSF3R T618I, CSF3R any nonsense, CSF3R any frameshift	1	The activating missense membrane-proximal mutation in CSF3R (p.T618I) has been reported to occur in approximately 83% of cases of chronic neutrophilic leukemia; some reports indicate this mutation may be present in cases of atypical chronic myeloid leukemia as well. The CS3R T618I mutation has been associated with response to JAK2 inhibitors but not dasatinib. A germline activating CSF3R mutation (p. T617N) has been described in autosomal dominant hereditary neutrophilia associated with splenomegaly and increased circulating CD34-positive myeloid progenitors. Nonsense and/or frameshift somatic mutations truncating the cytoplasmic domain of CSF3R have been described in approximately 40% of patients with severe congenital neutropenia and in the context of mutations in other genes may be associated with progression to acute myeloid leukemia. These activating truncating mutations have also been found in patients with chronic neutrophilic leukemia or atypical chronic myeloid leukemia. Some of these cytoplasmic truncating mutations have been associated with responses to dasatinib but not JAK2 inhibitors.	Pardanani A, et al. CSF3R T618I is a highly prevalent and specific mutation in chronic neutrophilic leukemia. Leukemia 2013;27(9):1870-3	Maxson JE, et al. Oncogenic CSF3R mutations in chronic neutrophilic leukemia and atypical CML. N Engl J Med 2013;368(19):1781-90
MPL	Myeloproliferative Neoplasm, Essential Thrombocythemia, Primary Myelofibrosis	Blood, Bone Marrow	MPL codon(s) 515 missense, MPL W515L, MPL W515K	1	Somatic activating mutation in MPL (W515L, W515K) has been reported in approximately 1%-10% of cases of JAK2 Val617Phe-negative myelofibrosis, essential thrombocythemia, a subset of cases of acute megakaryoblastic leukemia and has been associated with sensitivity to JAK inhibitors. The W515 mutations are typically not observed in polycythemia vera or other myeloid disorders (chronic myelomonocytic leukemia, myelodysplastic syndrome). A Ser505Asn activating mutation has also been described in familial essential thrombocythemia.	Pikman Y, et al. MPLW515L is a novel somatic activating mutation in myelofibrosis with myeloid metaplasia. PLoS Med 2006;3(7):e270	Ding J, et al. Familial essential thrombocythemia associated with a dominant-positive activating mutation of the c-MPL gene, which encodes for the receptor for thrombopoietin. Blood 2004;103(11):4198-200
JAK1	T Lymphoblastic Leukemia/Lymphoma, Acute Myeloid Leukemia	Blood, Bone Marrow	JAK1 any mutation	1	Activating mutations in JAK1 SH2, pseudokinase and kinase domains have been reported in approximately 5-20% of cases of T-Cell Acute Lymphoblastic Leukemia and less than 5% of Acute Myeloid Leukemia. Some, but not all, of these mutations have been shown to be inhibitable by ATP-competitive JAK inhibitors or Type I interferon.	Flex E, et al. Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. J Exp Med 2008;205(4):751-8	Aranaz P, et al. A new potential oncogenic mutation in the FERM domain of JAK2 in BCR/ABL1-negative and V617F-negative chronic myeloproliferative neoplasms revealed by a comprehensive screening of 17 tyrosine kinase coding genes. Cancer Genet Cytogenet 2010;199(1):1-8
NRAS	Acute Myeloid Leukemia, Chronic Myelomonocytic Leukemia, Myelodysplastic Syndrome, B Lymphoblastic Leukemia/Lymphoma, MDS/MPN	Blood, Bone Marrow	NRAS any mutation	1	Mutations in codons 12, 13, and 61 of NRAS have been reported in 7-12% of acute myeloid leukemia and may not be responsive to some MEK inhibitors according to some studies. Mutations in these codons of NRAS have also been reported in 10-30% of chronic myelomonocytic leukemia and 4-10% of myelodysplastic syndromes and may be associated with a worse prognosis according to some but not all studies. In addition, NRAS mutations have been described in approximately 15% of cases of B-ALL and, interestingly, some cases of ALL may show more than one abnormality in this pathway.	Jain N, et al. Phase II study of the oral MEK inhibitor selumetinib in advanced acute myelogenous leukemia: a University of Chicago phase II consortium trial. Clin Cancer Res 2014;20(2):490-8	Murphy DM, et al. NRAS mutations with low allele burden have independent prognostic significance for patients with lower risk myelodysplastic syndromes. Leukemia 2013;27(10):2077-81
NOTCH2	Marginal Zone B Cell Lymphoma, Diffuse Large B Cell Lymphoma	Blood, Bone Marrow	NOTCH2 any mutation, NOTCH2 I2304fs, NOTCH2 exon(s) 34 frameshift	1	NOTCH2 gain of function mutations have been reported in approximately 25% of splenic marginal zone lymphomas and are thought to be rare in non-splenic marginal zone lymphomas. These mutations are typically located near the C-terminal PEST domain and lead to protein truncation or, more rarely, are nonsynonymous substitution mutations affected the extracellular heterodimerization domain. NOTCH2 mutations may be associated with a worse prognosis among cases of splenic marginal zone lymphoma. In addition, NOTCH2 PEST domain mutations have been reported in approximately 8% of diffuse large B cell lymphomas and in vitro systems have demonstrated these PEST domain mutant NOTCH2 receptors have increased activity compared to wild type NOTCH2. In addion, copy number gain has been reported in a subset of DLBCL cases with NOTCH2 mutations.	Lee SY, et al. Gain-of-function mutations and copy number increases of Notch2 in diffuse large B-cell lymphoma. Cancer Sci 2009;100(5):920-6	Kiel MJ, et al. Whole-genome sequencing identifies recurrent somatic NOTCH2 mutations in splenic marginal zone lymphoma. J Exp Med 2012;209(9):1553-65
RIT1	Acute Myeloid Leukemia, Myelodysplastic Syndrome, Chronic Myelomonocytic Leukemia	Blood, Bone Marrow	RIT1 any mutation	1	Ras-like-without-CAAX-1 (RIT1) gene is a member of the RAS gene family. Recurrent somatic mutations of RIT1 have been reported in approximately 7% of cases of chronic myelomonocytic leukemia, and less than 5% of cases of myelodysplastic syndrome and less than 5% of acute myeloid leukemia. The mutations typically occur in the Switch II effector domain, and the affected residues are close to codon Q79, which is analogous to amino acid Q61 of NRAS or KRAS where mutations frequently occur in cancer. Moreover, the experimental Q79L mutation in RIT1 has been reported to confer constitutive activation of the protein. RIT1 mutations are typically mutually exclusive of mutations in other RAS family members. In addition, RIT1 maps to the minimal common amplified region (1q21–22) in 1q gains frequently found in other cancers. 1q amplification involving the RIT1 locus has been reported in 4-18% of cases of myelofibrosis as well as less than 5% of chronic myelomonocytic leukemia, less than 5% of myelodysplastic syndromes and less than 5% of acute myeloid luekemia. In rare cases mutations and amplifcations of RIT1 may coexist. In general, RIT1 has been reported to increased phosphorylation of AKT and activate proliferation through the mitogen activated protein kinase pathway.	Gómez-Seguí I, et al. Novel recurrent mutations in the RAS-like GTP-binding gene RIT1 in myeloid malignancies. Leukemia 2013;27(9):1943-6	
DNMT3A	Myeloproliferative Neoplasm, Acute Myeloid Leukemia, T Lymphoblastic Leukemia/Lymphoma, Myelodysplastic Syndrome, Chronic Myelomonocytic Leukemia	Blood, Bone Marrow	DNMT3A any mutation	1	DNMT3A is a DNA methyltransferase. Recurrent, somatic, heterozygous mutations in DNMT3A have been reported in approximately 18-25% of cases of acute myeloid leukemia, 8% of cases of myelodysplastic syndrome, up to 15% of myeloproliferative neoplasms and less than 5% of cases of chronic myelomonocytic leukemia. Mutations in DNMT3A have been reported to occur together with mutations in other genes including JAK2, FLT3, IDH1/IDH2, ASXL1, TET2 and NPM1. DNMT3A mutations have been associated with reduced enzymatic activity or altered histone binding, as well as reduced DNA methylation in various genomic regions and altered gene expression in some models. DNMT3A mutations may be associated with adverse prognosis . In addition to myeloid disorders, mutations in DNMT3A have also been reported in approximately 15% of cases of adult, early T cell precursor acute lymphoblastic leukemia.	Larsson CA, et al. The changing mutational landscape of acute myeloid leukemia and myelodysplastic syndrome. Mol Cancer Res 2013;11(8):815-27	Ley TJ, et al. DNMT3A mutations in acute myeloid leukemia. N Engl J Med 2010;363(25):2424-33

- Columns include Genes, Tumor types, Variants, Tier, Interpretations, and Citations
- Each row represents an interpretation

Browse Interpretations



Interpretations

+ Add New Interpretation

Show 10 entries

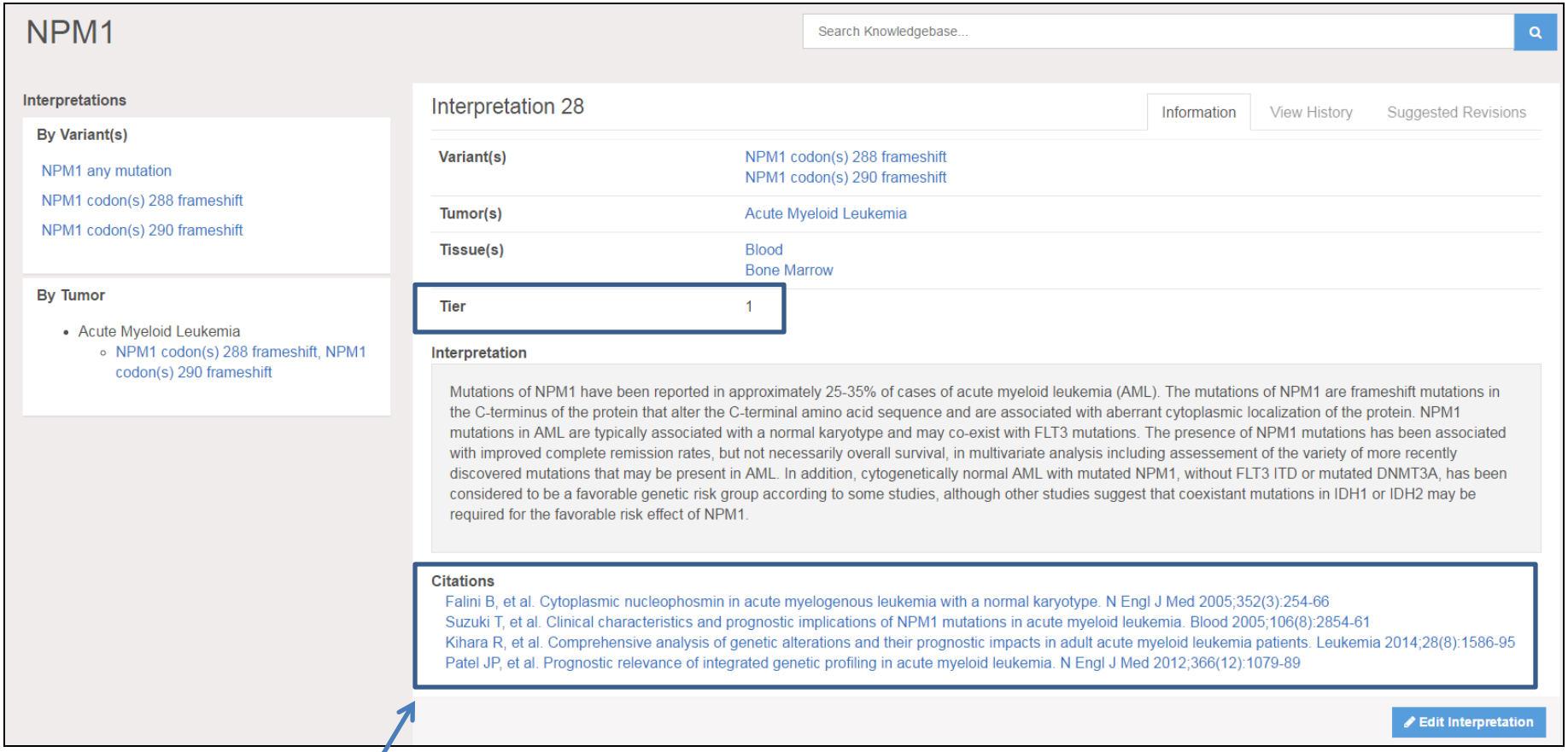
Search:

Tumor	Tissue	Variants	Interpretation
Acute Myeloid Leukemia	Blood Bone Marrow	NPM1 codon(s) 288 frameshift NPM1 codon(s) 290 frameshift	Mutations of NPM1 have been reported in approximately 25-35% of cases of acute myeloid leukemia (AML). The mutations of NPM1 are frameshift mutations in the C-terminus of the protein that alter the C-terminal amino acid sequence and are associated with aberrant cytoplasmic localization of the protein. NPM1 mutations in AML are typically associated with a normal karyotype and may co-exist with FLT3 mutations. The presence of NPM1 mutations has been associated with improved complete remission rates, but not necessarily overall survival, in multivariate analysis including assessement of the variety of more recently discovered mutations that may be present in AML. In addition, cytogenetically normal AML with mutated NPM1, without FLT3 ITD or mutated DNMT3A, has been considered to be a favorable genetic risk group according to some studies, although other studies suggest that coexistant mutations in IDH1 or IDH2 may be required for the favorable risk effect of NPM1.
Acute Myeloid Leukemia	Blood Bone Marrow	WT1 any mutation	WT1 mutations have been reported in approximately 7-12% of cytogenetically normal acute myeloid leukemia and most frequently occur in exon 7 or exon 9. WT1 mutations are typically putative loss of funtion mutations (frameshift, missense and nonsense mutations) ; in some cases two mutations in WT1 may occur. In addition, WT1 mutations may coexist with mutations in NPM1, FLT3, among others. In general, WT1 mutations have been associated with a poor prognosis although, the strength of this association is variable and may depend on other concomitant factors, as such, WT1 mutation may not impact outcome as a single molecular marker. In addition, a synonomous WT1 SNP rs16745 located in exon 7 has been described and corresponds to adenine (A) or guanine (G) containing alleles, the frequency of which varies between ethnic groups(G is the minor allele in Western – and the major allele in Asian – populations). The presence of the G allele has been associated with improved prognosis in some studies but not in others .
Acute Myeloid Leukemia	Blood Bone Marrow	CEBPA any mutation	Mutations of the transcription factor CEBPA (CCAAT/enhancer binding protein alpha) have been reported in approximately 13-19% of patients with acute myeloid leukemia (AML) and a normal karyotype. Two types of mutations have been reported: N-terminal changes which result in a truncated dominant negative isoform lacking one of the N terminal domain transactivation domains and C-terminal mutations which are in-frame insertions or deletions affecting the leucine zipper and preventing dimerization and DNA binding. Patients may carry both N- and C-terminal mutations affecting different alleles. There appears to be a beneficial prognostic effect of CEBPA mutation, but may be limited to patients with two different CEBPA mutations, so-called double mutant CEBPA(biallelic mutations) rather than patients with a single CEBPA mutation.
Acute Myeloid Leukemia	Blood	GATA1 any mutation	The GATA1 transcription factor is important in the development of erythroid and megakaryocytic lineages. Amino-terminal, small

- Each interpretation is within disease context and may be specific to a variant(s).
- Click on interpretation to see interpretation page w/citations and more information.

PMKB

Interpretation Page



Links to PubMed

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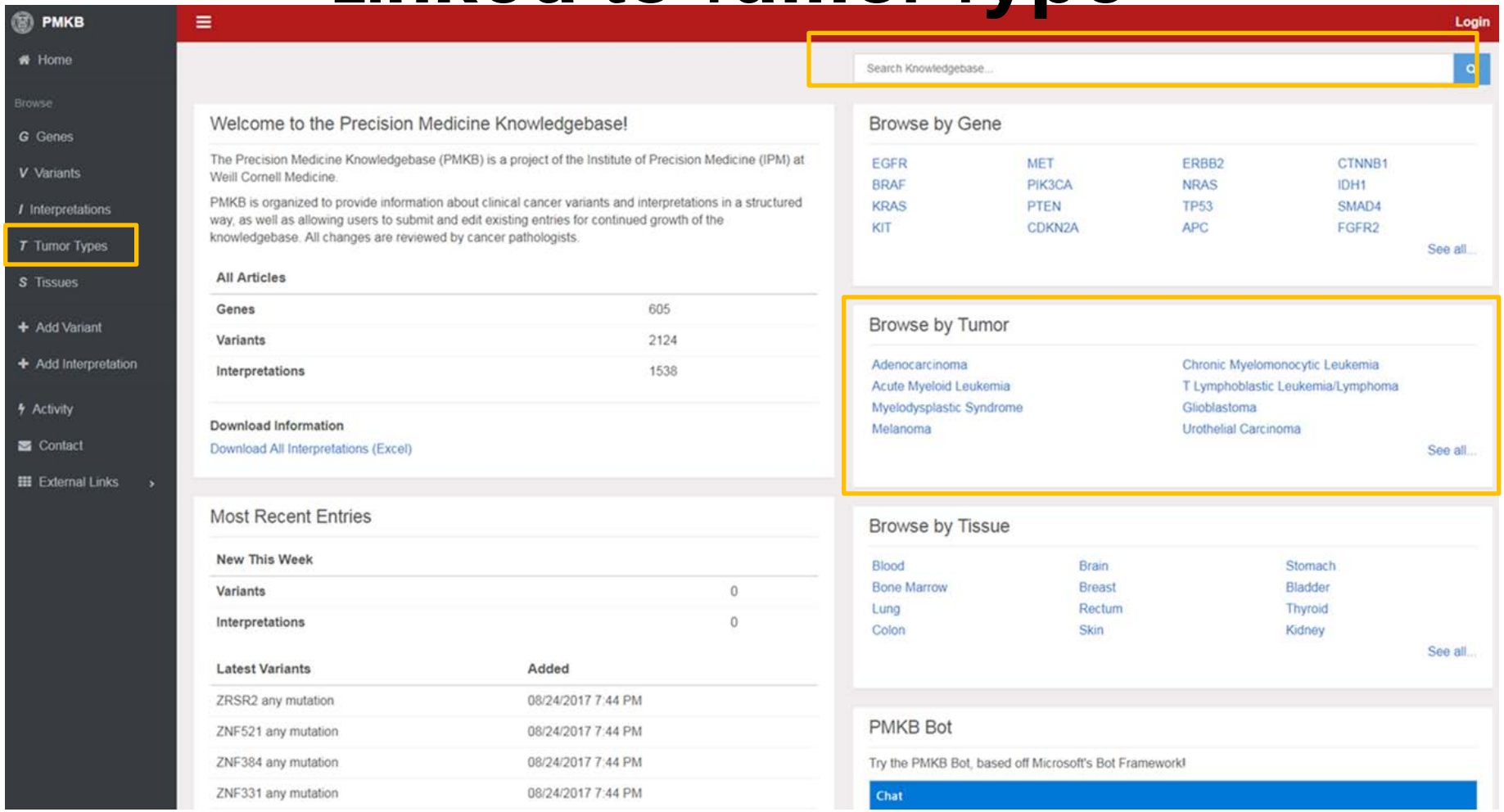
Tier Definitions

- **Tier 1** - Clinical utility has been demonstrated - Actionable / Clinically Relevant variants. Variants in genes with approved therapeutic implications in specified tumors.

- **Tier 2** - Clinical utility/actionability has diagnostic, prognostic or therapeutic implications. Variants with potential diagnostic/classification, prognostic implications. Variants with approved therapeutic implications in a different tumor type. Novel variants in genes that have approved therapeutic implications. Variants associated with Clinical trials.

- **Tier 3** - Variants of Unknown Significance.

Search for Variants Linked to Tumor Type



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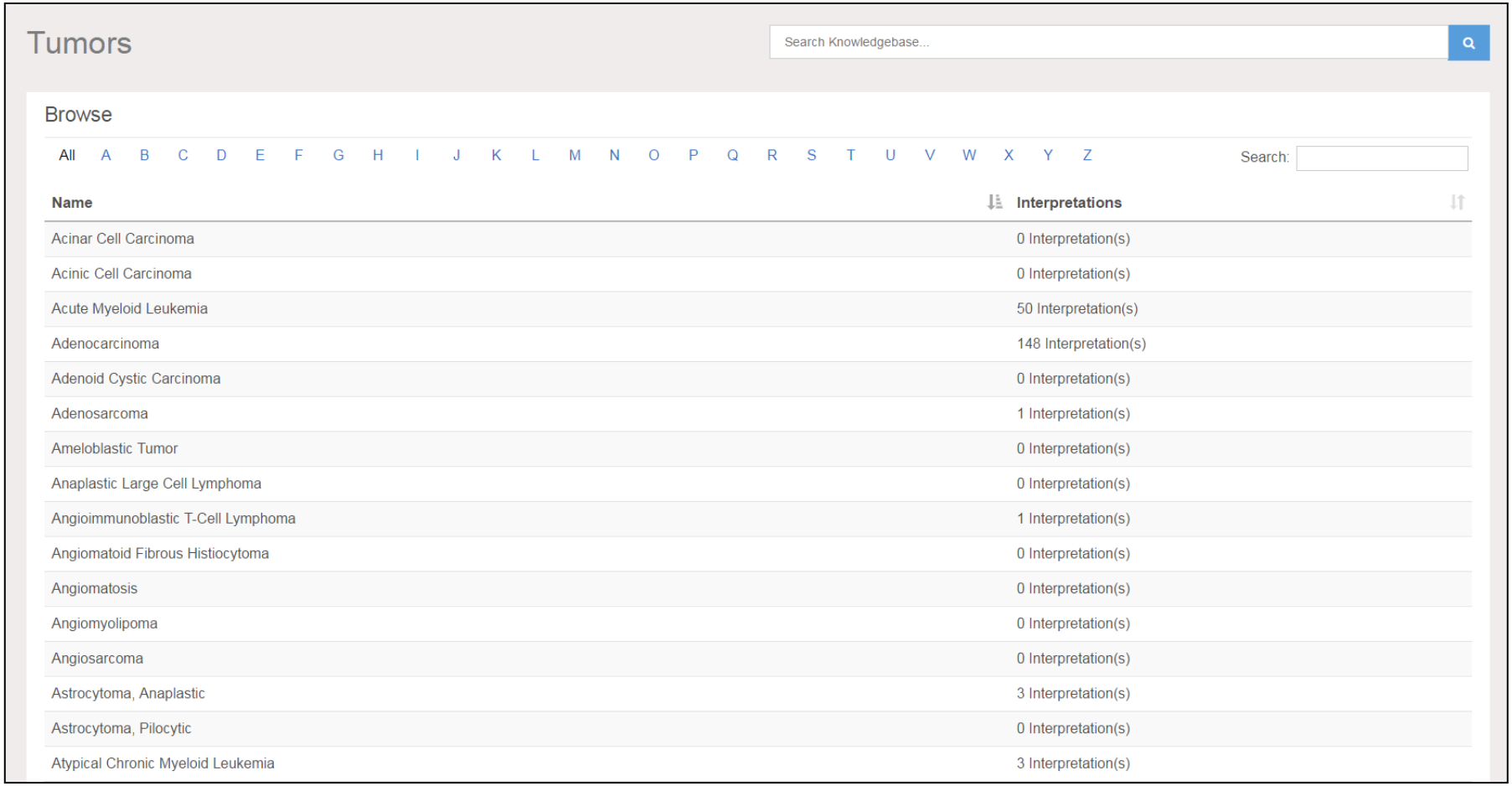
PMKB

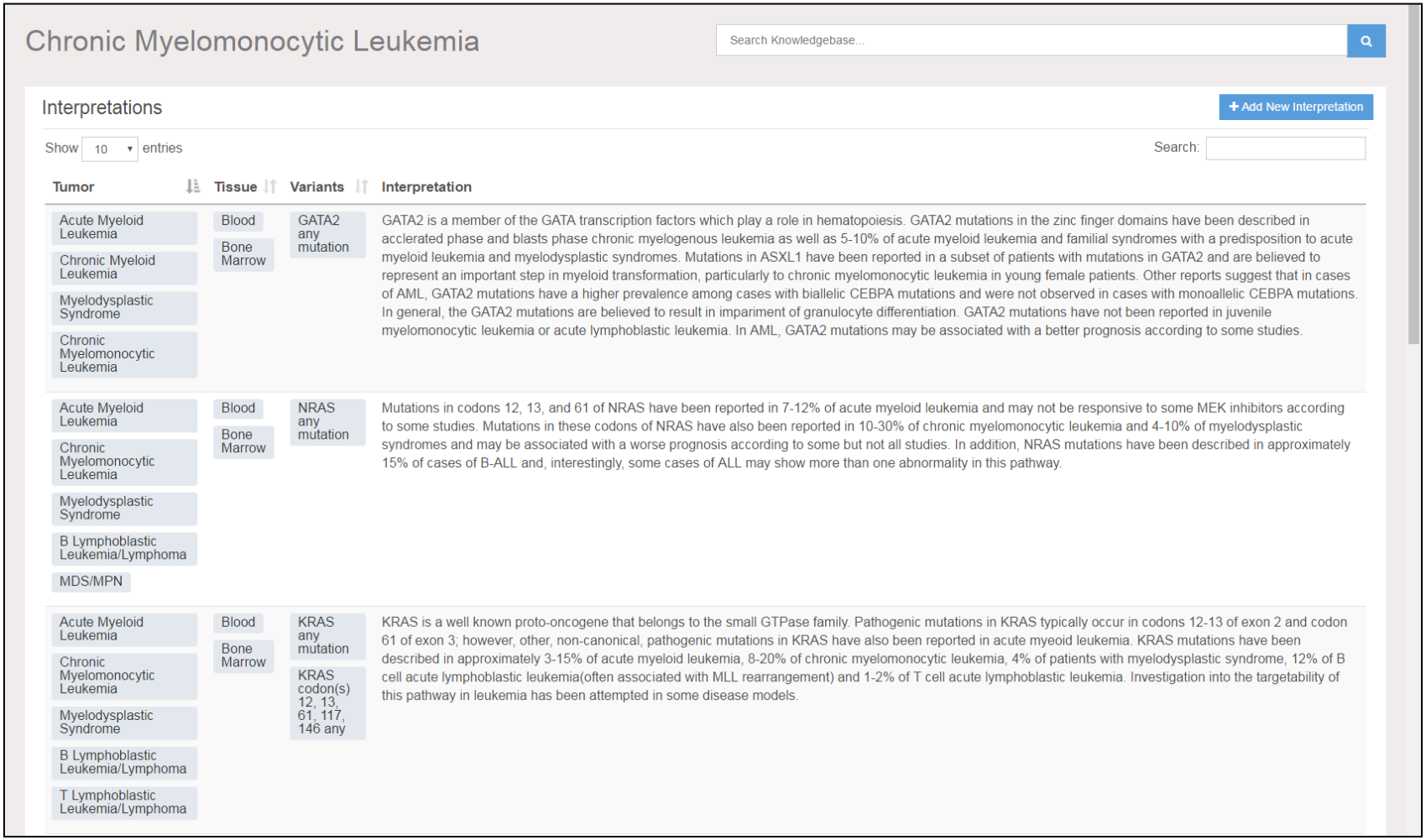
Browse Tumor Types



- Click on tumor type to see all associated interpretations.
- 143 tumor types listed – not all have interpretations.

Tumor-Specific Page of Interpretations

PMKB



Chronic Myelomonocytic Leukemia

Search Knowledgebase...

Interpretations

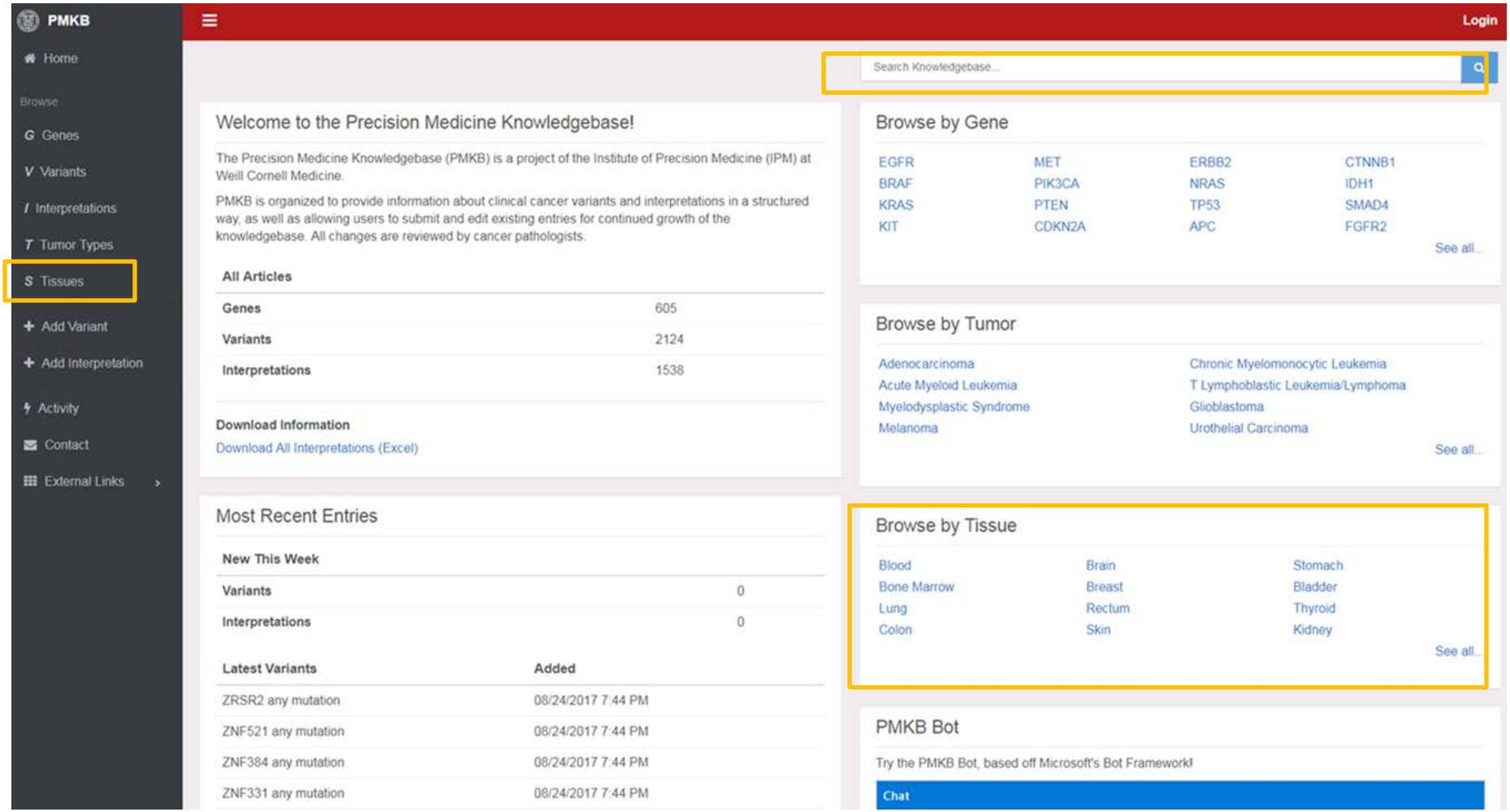
+ Add New Interpretation

Show 10 entries

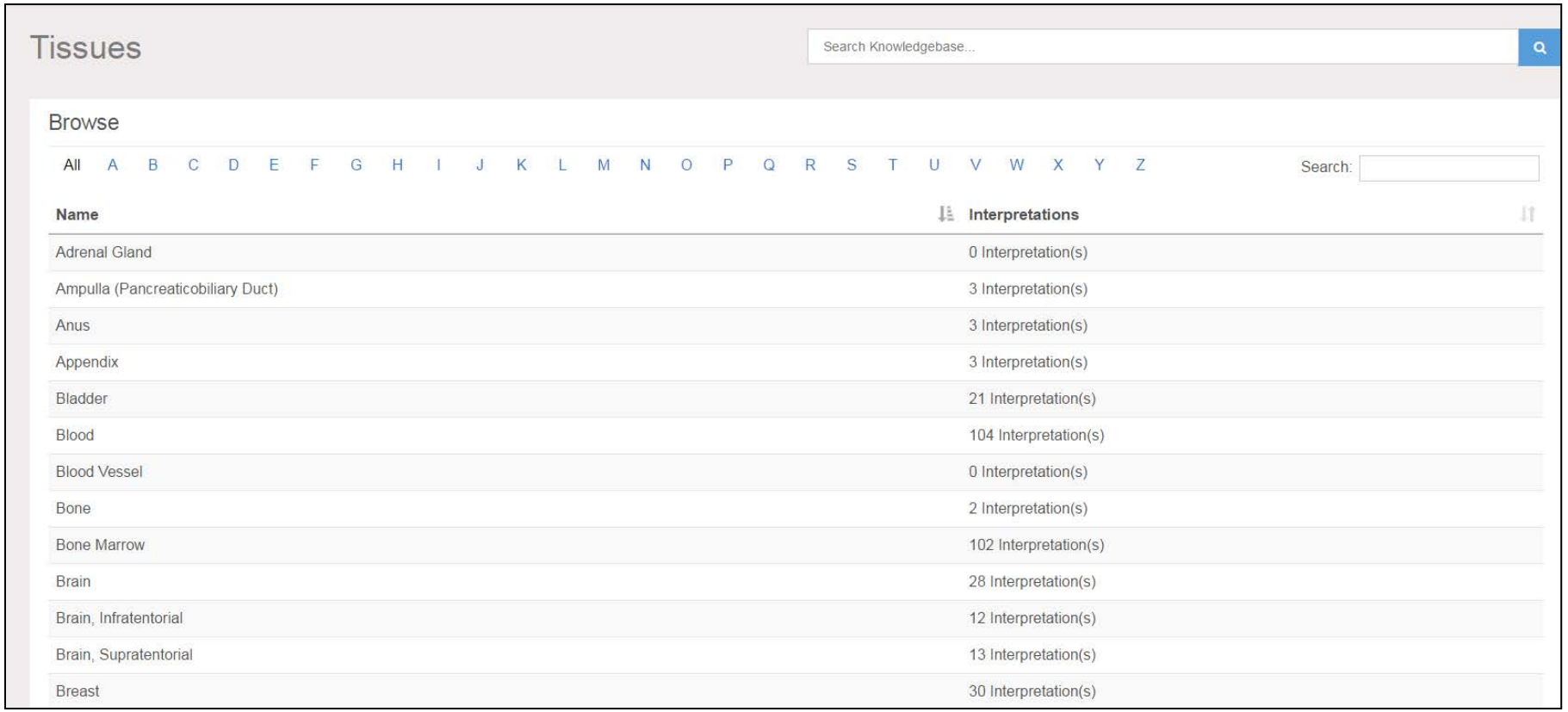
Search:

Tumor	Tissue	Variants	Interpretation
Acute Myeloid Leukemia Chronic Myeloid Leukemia Myelodysplastic Syndrome Chronic Myelomonocytic Leukemia	Blood Bone Marrow	GATA2 any mutation	GATA2 is a member of the GATA transcription factors which play a role in hematopoiesis. GATA2 mutations in the zinc finger domains have been described in acclerated phase and blasts phase chronic myelogenous leukemia as well as 5-10% of acute myeloid leukemia and familial syndromes with a predisposition to acute myeloid leukemia and myelodysplastic syndromes. Mutations in ASXL1 have been reported in a subset of patients with mutations in GATA2 and are believed to represent an important step in myeloid transformation, particularly to chronic myelomonocytic leukemia in young female patients. Other reports suggest that in cases of AML, GATA2 mutations have a higher prevalence among cases with biallelic CEBPA mutations and were not observed in cases with monoallelic CEBPA mutations. In general, the GATA2 mutations are believed to result in impariment of granulocyte differentiation. GATA2 mutations have not been reported in juvenile myelomonocytic leukemia or acute lymphoblastic leukemia. In AML, GATA2 mutations may be associated with a better prognosis according to some studies.
Acute Myeloid Leukemia Chronic Myelomonocytic Leukemia Myelodysplastic Syndrome B Lymphoblastic Leukemia/Lymphoma MDS/MPN	Blood Bone Marrow	NRAS any mutation	Mutations in codons 12, 13, and 61 of NRAS have been reported in 7-12% of acute myeloid leukemia and may not be responsive to some MEK inhibitors according to some studies. Mutations in these codons of NRAS have also been reported in 10-30% of chronic myelomonocytic leukemia and 4-10% of myelodysplastic syndromes and may be associated with a worse prognosis according to some but not all studies. In addition, NRAS mutations have been described in approximately 15% of cases of B-ALL and, interestingly, some cases of ALL may show more than one abnormality in this pathway.
Acute Myeloid Leukemia Chronic Myelomonocytic Leukemia Myelodysplastic Syndrome B Lymphoblastic Leukemia/Lymphoma T Lymphoblastic Leukemia/Lymphoma	Blood Bone Marrow	KRAS any mutation KRAS codon(s) 12, 13, 61, 117, 146 any	KRAS is a well known proto-oncogene that belongs to the small GTPase family. Pathogenic mutations in KRAS typically occur in codons 12-13 of exon 2 and codon 61 of exon 3; however, other, non-canonical, pathogenic mutations in KRAS have also been reported in acute myeoid leukemia. KRAS mutations have been described in approximately 3-15% of acute myeloid leukemia, 8-20% of chronic myelomonocytic leukemia, 4% of patients with myelodysplastic syndrome, 12% of B cell acute lymphoblastic leukemia(often associated with MLL rearrangement) and 1-2% of T cell acute lymphoblastic leukemia. Investigation into the targetability of this pathway in leukemia has been attempted in some disease models.

Search for Tissue type



Browse Tissue Types



Tissues

Search Knowledgebase...

Browse

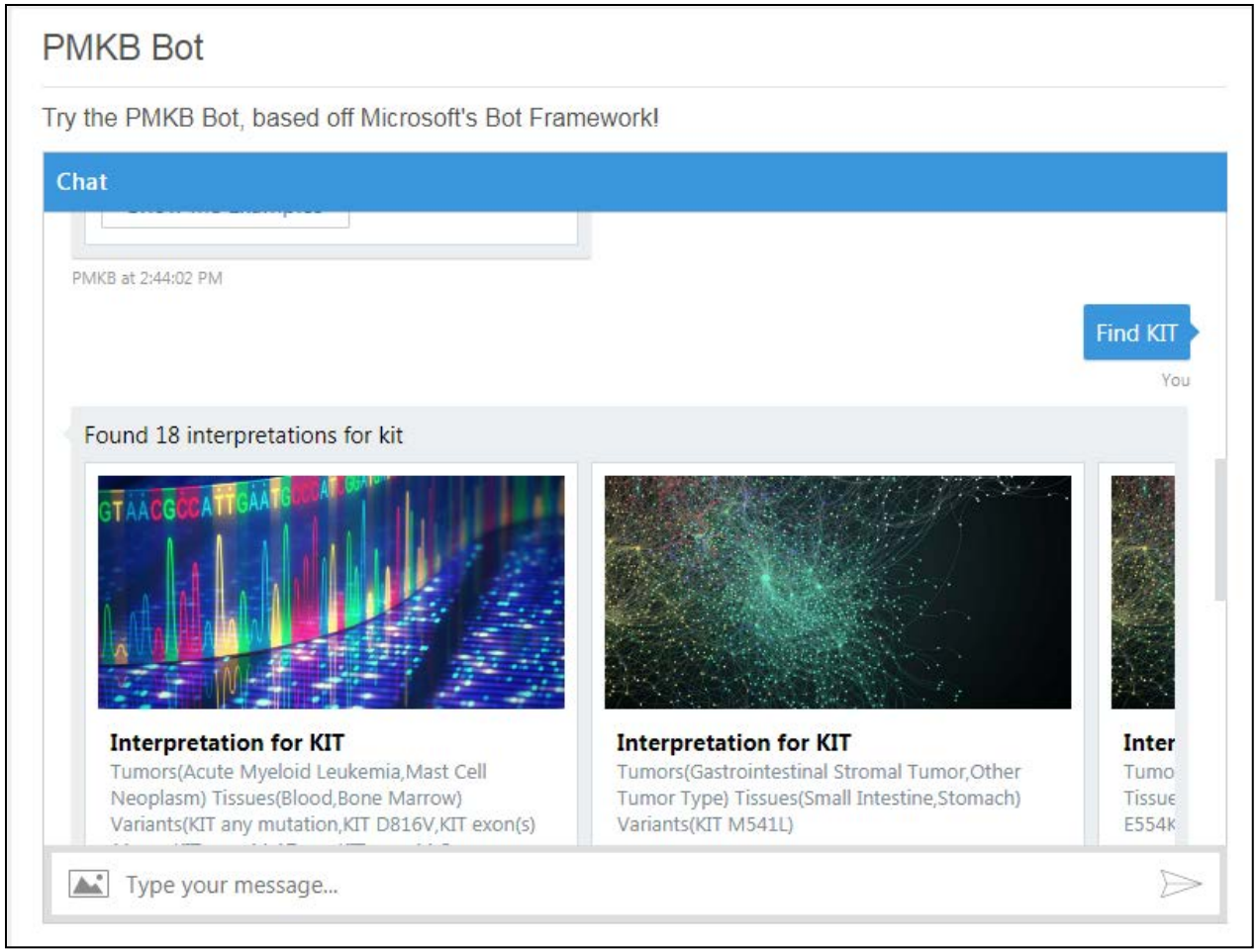
All A B C D E F G H I J K L M N O P Q R S T U V W X Y Z Search:

Name	Interpretations
Adrenal Gland	0 Interpretation(s)
Ampulla (Pancreaticobiliary Duct)	3 Interpretation(s)
Anus	3 Interpretation(s)
Appendix	3 Interpretation(s)
Bladder	21 Interpretation(s)
Blood	104 Interpretation(s)
Blood Vessel	0 Interpretation(s)
Bone	2 Interpretation(s)
Bone Marrow	102 Interpretation(s)
Brain	28 Interpretation(s)
Brain, Infratentorial	12 Interpretation(s)
Brain, Supratentorial	13 Interpretation(s)
Breast	30 Interpretation(s)

- 68 tissue types listed – not all have interpretations.

PMKB Bot

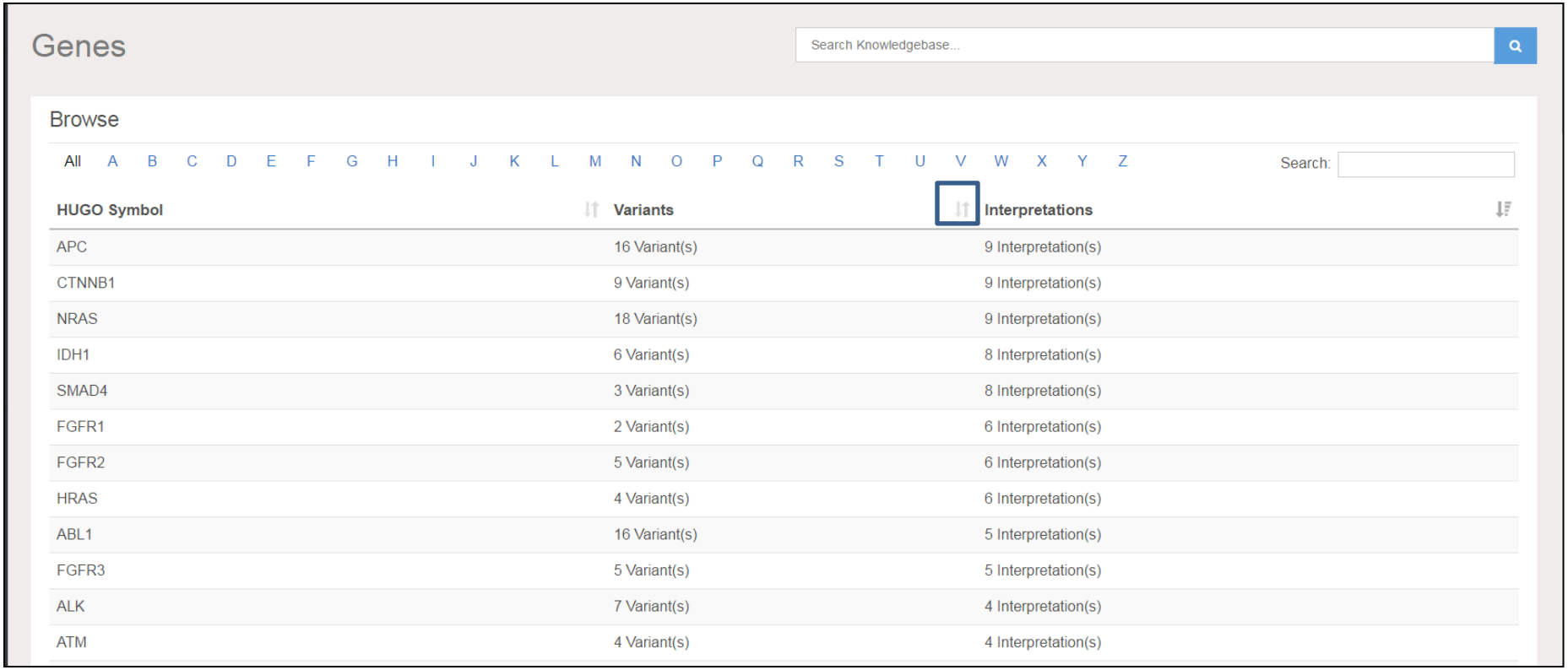
- Type in a gene name, disease, tissue, or variant
- PMKB Bot will output interpretations that fit your criteria.
- Click anywhere in the interpretation or the image above the interpretation to see the gene page housing that interpretation.



Scenario #1

- You are a laboratory professional putting together a Pan Cancer gene list.
  - Browse Genes and add all
  - OR add only genes with interpretations

Browse Genes and sort genes by the number of associated interpretations.



Genes

Search Knowledgebase...

Browse

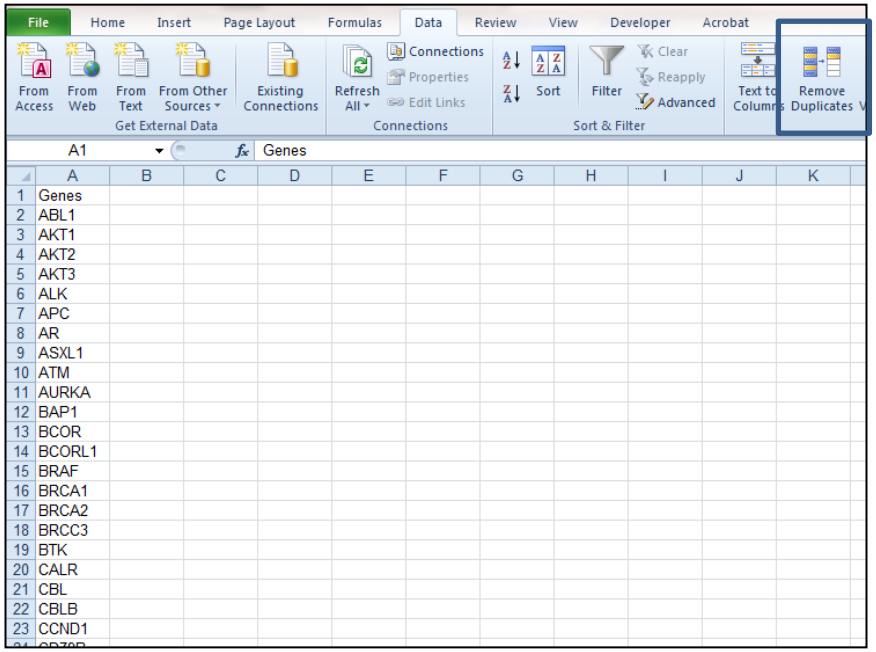
All A B C D E F G H I J K L M N O P Q R S T U V W X Y Z

Search:

HUGO Symbol	Variants	Interpretations
APC	16 Variant(s)	9 Interpretation(s)
CTNNB1	9 Variant(s)	9 Interpretation(s)
NRAS	18 Variant(s)	9 Interpretation(s)
IDH1	6 Variant(s)	8 Interpretation(s)
SMAD4	3 Variant(s)	8 Interpretation(s)
FGFR1	2 Variant(s)	6 Interpretation(s)
FGFR2	5 Variant(s)	6 Interpretation(s)
HRAS	4 Variant(s)	6 Interpretation(s)
ABL1	16 Variant(s)	5 Interpretation(s)
FGFR3	5 Variant(s)	5 Interpretation(s)
ALK	7 Variant(s)	4 Interpretation(s)
ATM	4 Variant(s)	4 Interpretation(s)

Another way to compile the same list of genes with interpretations is to download the Excel file of all interpretations, sort by gene name, and expect duplicates for genes with >1 interpretation. I like to copy the gene column over to another worksheet/tab.

- One you have the gene list in Excel (or similar), go to Data/Remove Duplicates to have a full list of candidate genes.
- I like to alphabetize before integrating with other lists/resources.



Scenario #2

- You are putting together a list for a potential AML NGS panel. Using PMKB, what genes might you consider including?
  - Search for AML Tumor Type (see slides 14-16)
  - Filter spreadsheet for rows/interpretations related to AML
    - Copy Information over to new unprotected worksheet
    - Filter Tumor Type(s) column by “Contains” criteria for “Acute Myeloid Leukemia”

Scenario #3

- Your laboratory is interpreting a small NGS panel for a patient with lung adenosarcoma. How can PMKB aid in interpretation?

- Abnormal genes
  - EGFR L858R
  - RET V804M

- Normal genes
  - ALK
  - ERBB2
  - KRAS
  - PIK3CA
  - BRAF

PMKB Relevant Interpretations

- EGFR: positive for L858R mutation https://pmkb.weill.cornell.edu/variants/115
- ERBB2: pertinent negative! https://pmkb.weill.cornell.edu/genes/95/variants/366
- KRAS: pertinent negative! https://pmkb.weill.cornell.edu/genes/50/variants/368
- PIK3CA: pertinent negative! https://pmkb.weill.cornell.edu/genes/19/variants/367

Future Features: Coming Soon!

- Shared API
- Opportunities for Crowd-sourcing: Addition of variants
  - Addition of interpretation

Contacts

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