

# Clinical Heme Panel Optimisation

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## 1 The process

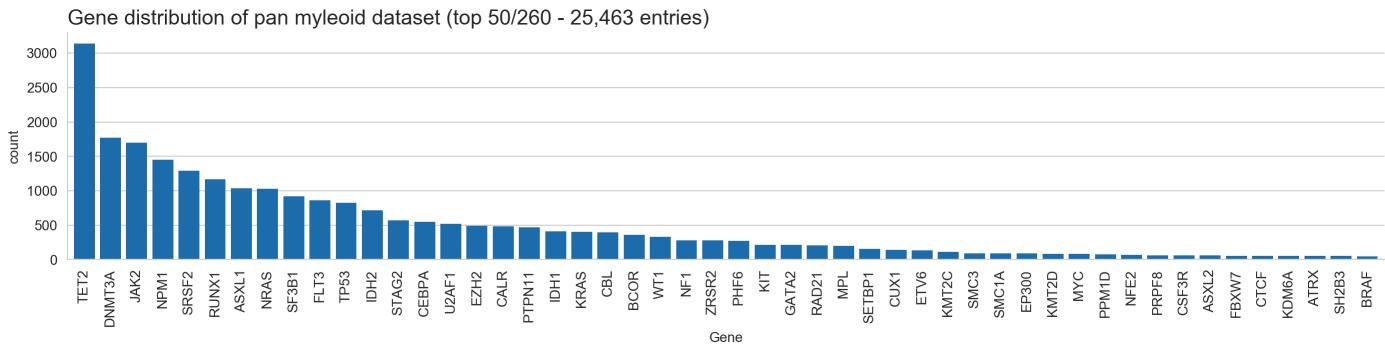
### 1.1 The pan-myeloid dataset

Brief overview of the pan-myeloid dataset:

- 25,463 mutations
- 8,966 patients
- only oncogenic mutations
- 3 different diseases:

disease	count	frequency	patient
AML	13,656	54%	4,348
MDS	8,179	32%	2,586
MPN	3,628	14%	2,063

- 260 genes:



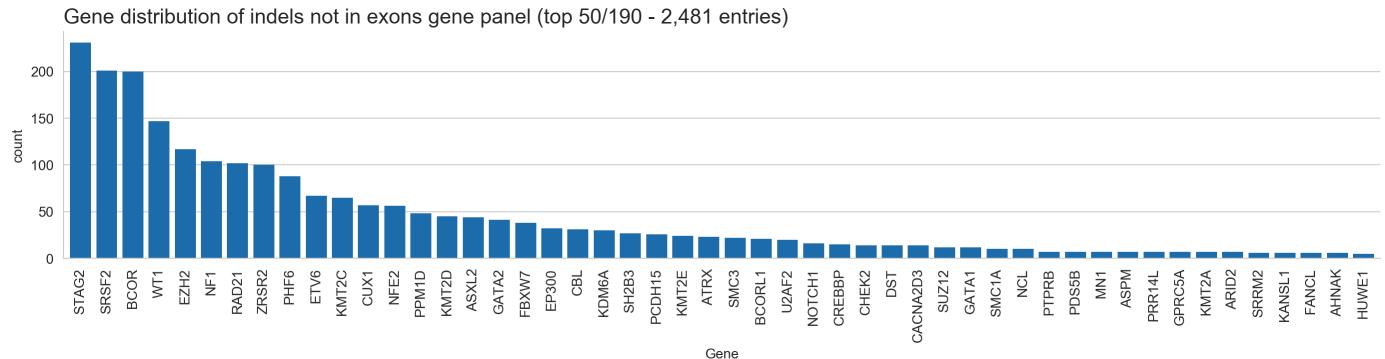
### 1.2 Methodology

- Filter the indels between reported and unreported following the selected exons list. An indel is considered as reported if its beginning or end is within one of the exons of the selected exons list.
- Filter the substitutions between reported and unreported following the selected hotspots list. A substitution is considered as reported if it hits one of the three nucleotides of an amino acid present in the selected hotspots list.

## 2 Unreported indels

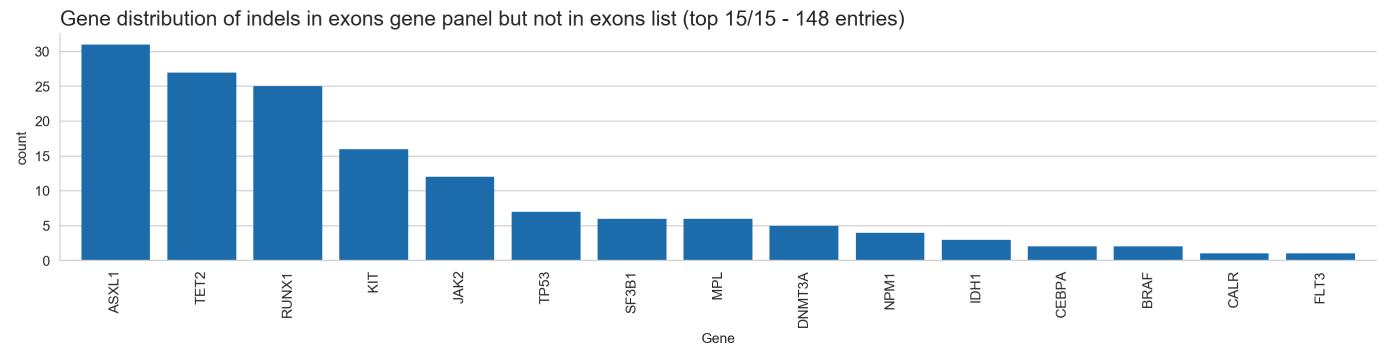
### 2.1 Genes not included in the selected exons list

$\approx 31\%$  of the indels are not in the selected exons gene panel. The following plot shows the distribution of the main unreported genes:



### 2.2 Indels identified in genes with partial exon reporting

Only  $\approx 2\%$  of the indels are in the selected exons gene panel, but not in the selected exons list. The following plot shows the distribution of the genes having some unreported exons:



Here is a table of the unreported exons for the genes above (NA  $\leftrightarrow$  no corresponding exon found in transcript list):

gene	missed exons
ASXL1	11 (n = 30), NA (n = 1)
TET2	NA (n = 27)
RUNX1	NA (n = 25)
KIT	8 (n = 8), 2 (n = 2), 9 (n = 1), 10 (n = 1), 5 (n = 1), 11 (n = 1), 21 (n = 1), NA (n = 1)
JAK2	12 (n = 5), 11 (n = 5), 19 (n = 1), NA (n = 1)
TP53	NA (n = 7)
SF3B1	16 (n = 5), NA (n = 1)
MPL	12 (n = 3), 3 (n = 1), 11 (n = 1), NA (n = 1)
DNMT3A	NA (n = 5)
NPM1	10 (n = 2), NA (n = 2)
IDH1	6 (n = 1), 3 (n = 1), NA (n = 1)
CEBPA	NA (n = 2)
BRAF	14 (n = 1), 3 (n = 1)
CALR	7 (n = 1)
FLT3	3 (n = 1)

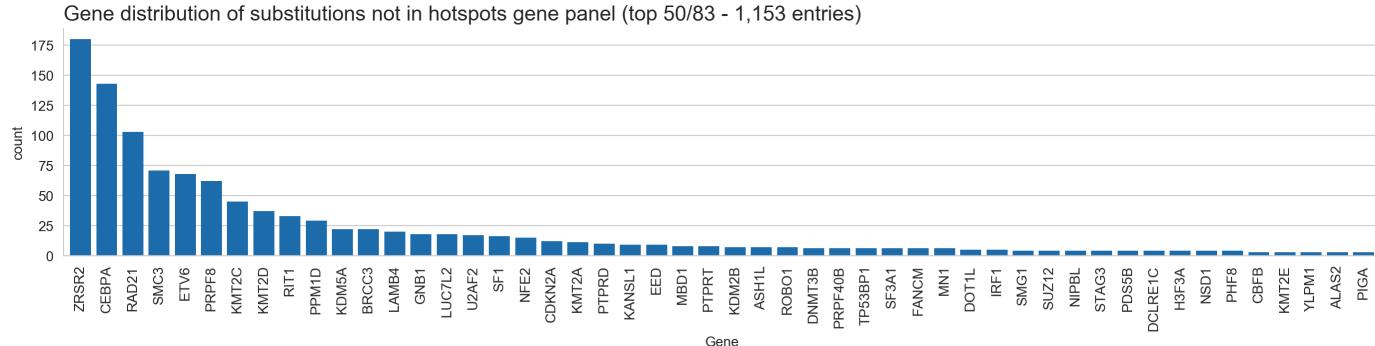
### 2.3 Summary table

indel status	count	frequency
in exons list	5, 267	66, 70%
not in exons gene panel	2, 481	31, 42%
in exons gene panel but not in exons list	148	1, 87%

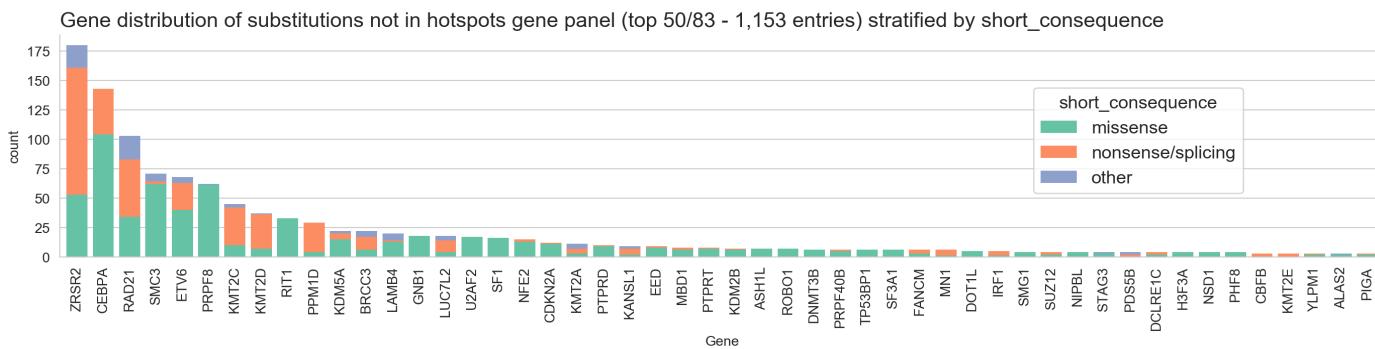
### 3 Unreported substitutions

#### 3.1 Genes not included in the selected hotspots list

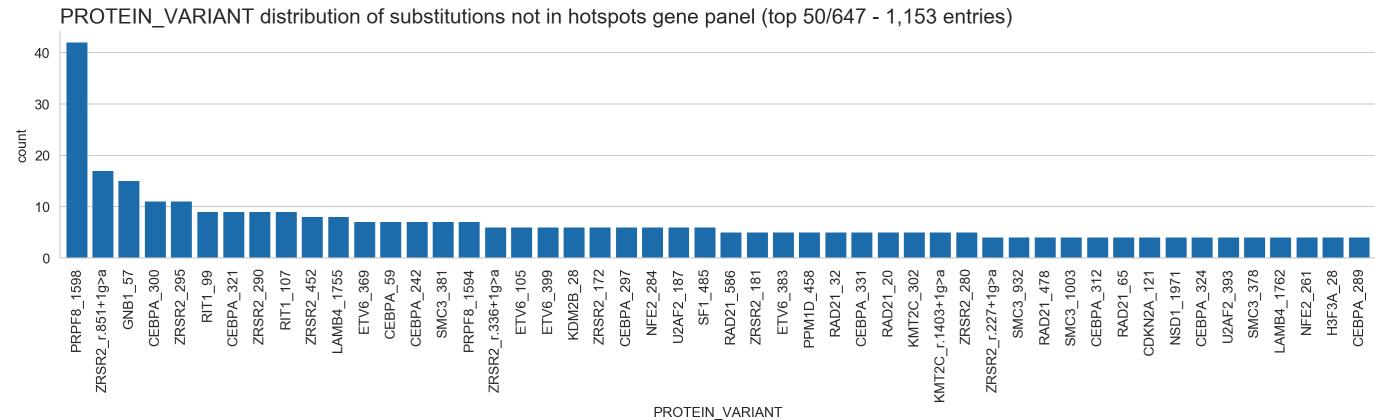
$\approx 7\%$  of the substitutions are not in the selected hotspots gene panel. The following plot shows the distribution of the main unreported genes:



Here is the same plot stratified by mutation consequence:

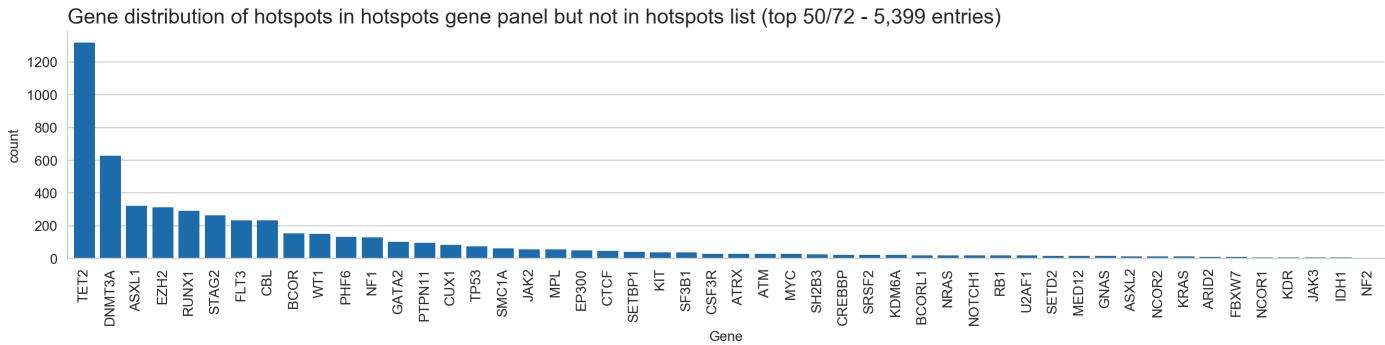


Here is a figure of the 50 most recurrent unreported substitutions which are not in the selected hotspots gene panel:

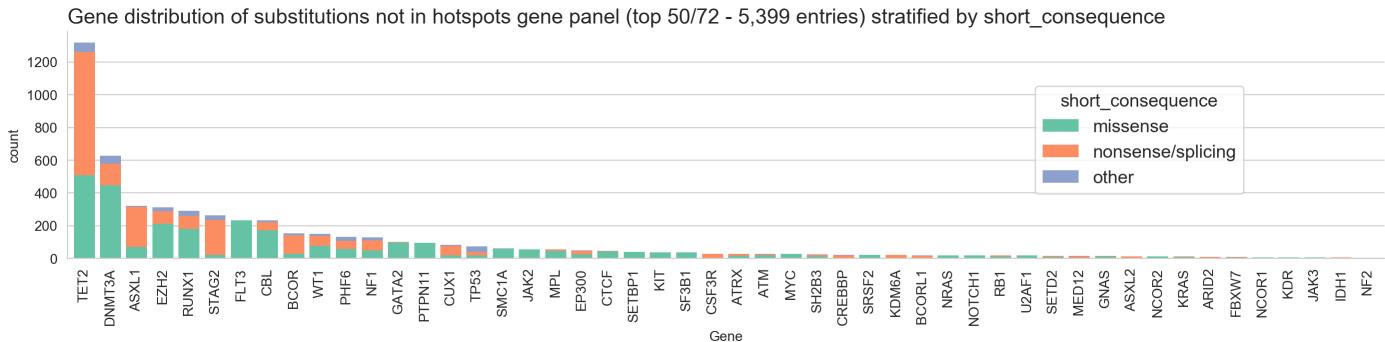


#### 3.2 Substitutions identified in genes with partial hotspot reporting

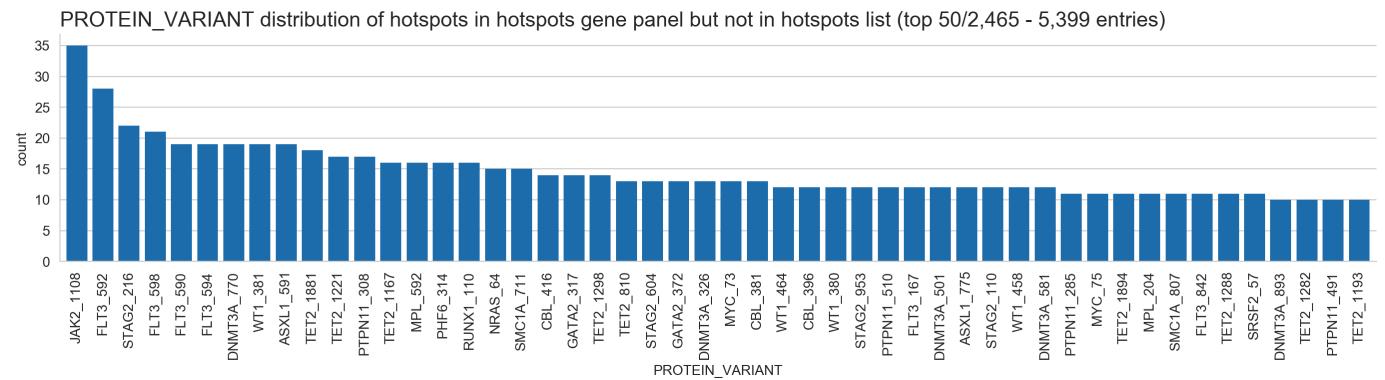
$\approx 31\%$  of the substitutions are in the selected hotspots gene panel, but not in the selected hotspots list. The following plot shows the distribution of the main genes having some unreported substitutions:



Here is the same plot stratified by mutation consequence:



Here is a figure of the 50 most recurrent unreported substitutions<sup>1</sup>:



### 3.3 Summary table

substitution status	count	frequency
in hotspots list	11,015	62, 70%
not in hotspots gene panel	1,153	6, 56%
in hotspots gene panel but not in hotspots list	5,399	30, 73%

<sup>1</sup> see supplementary material 1 for the complete table

## 4 Summary by patient

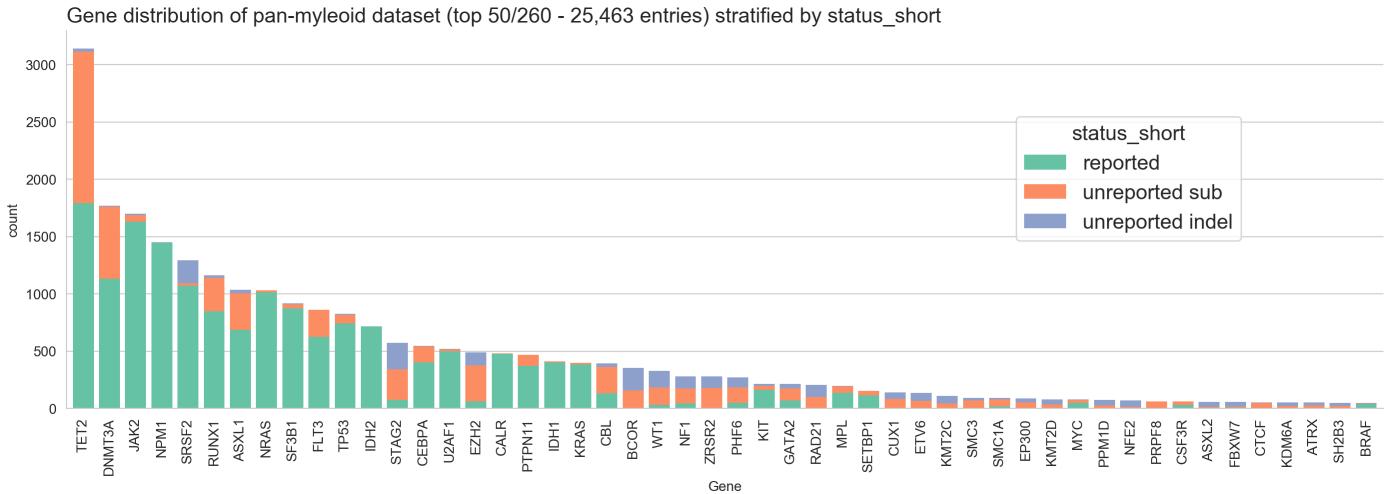
At the patient level, we have:

- a mean of [31%] oncogenic mutations unreported by patient
- for [8%] (711/8,966) of the patients, **not a single oncogenic mutation reported**

## 5 Summary by gene

### 5.1 Overview

The following plot shows the proportion of unreported mutation for the most recurrent mutated genes<sup>2</sup>:



In the next part we propose an analysis gene by gene for the 33 most recurrent mutated genes (see lookup table under) gathering:

- The OS (overall survival) plot between:
  - wildtype**: patients without any mutation in the studied gene
  - gene name - reported**: patients with at least one reported mutation in the studied gene
  - gene name - unreported**: patients with mutations in the studied gene but not a single reported mutation

Warning, the number indicated in each category represents the number of patient used to plot the curve (modulo the missing values).

- A pivot table showing the count of mutation reported (✓) and unreported (✗) stratified by disease (AML, MDS or MPN) and mutation consequence (inframe, missense, nonsense/splicing, truncating or other).
- 4 lollipop plots of the gene, two for indels (reported vs unreported) and two for substitutions (reported vs unreported). Warning, these lollipop plots don't take into account the splicing mutations. The color of the mutation circle represents its consequence, the size of the mutation circle represents its occurrence in the dataset, see legend:

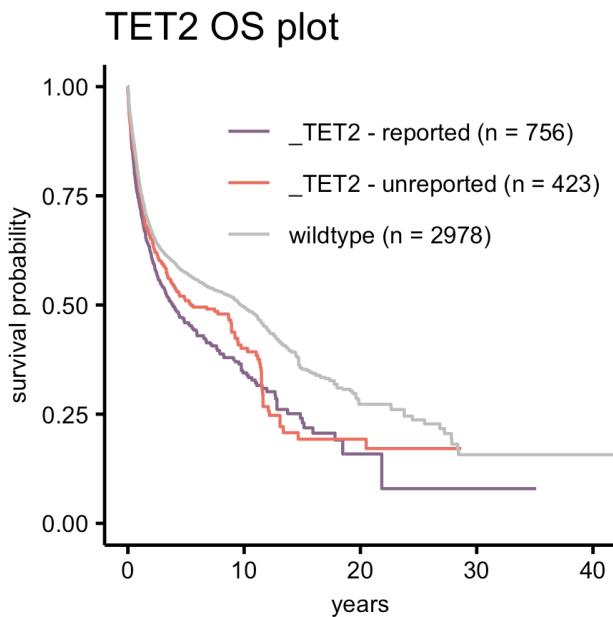


Lookup table gene ↔ page:

gene	page	gene	page	gene	page	gene	page	gene	page	gene	page
TET2	7	ASXL1	13	STAG2	19	IDH1	25	ZRSR2	31	SETBP1	37
DNMT3A	8	NRAS	14	CEBPA	20	KRAS	26	PHF6	32	CUX1	38
JAK2	9	SF3B1	15	U2AF1	21	CBL	27	KIT	33	ETV6	39
NPM1	10	FLT3	16	EZH2	22	BCOR	28	GATA2	34		
SRSF2	11	TP53	17	CALR	23	WT1	29	RAD21	35		
RUNX1	12	IDH2	18	PTPN11	24	NF1	30	MPL	36		

<sup>2</sup> see supplementary material 2 for the complete table

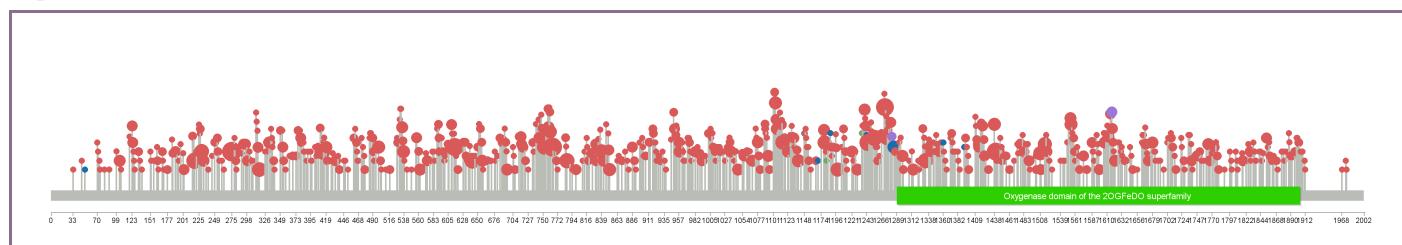
## 5.2 TET2



disease	status consequence	x indel	x sub	✓
AML	inframe	0	0	6
	missense	0	189	86
	nonsense/splicing	4	266	92
	other	10	31	4
	truncating	0	0	466
MDS	inframe	0	0	6
	missense	0	245	110
	nonsense/splicing	5	422	114
	other	2	15	3
	truncating	2	0	714
MPN	inframe	0	0	1
	missense	0	73	35
	nonsense/splicing	0	66	17
	other	3	13	0
	truncating	1	0	139
<b>TOTAL</b>		27	1320	1793
		0.01%	0.42%	0.57%

### Indels

#### reported

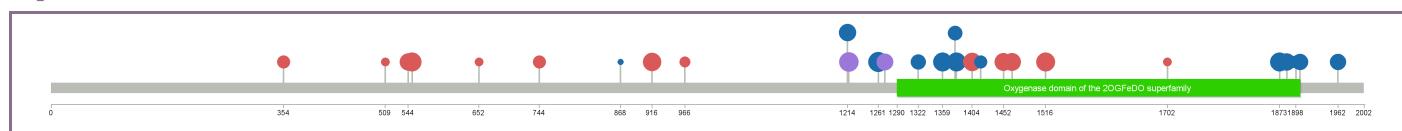


#### unreported

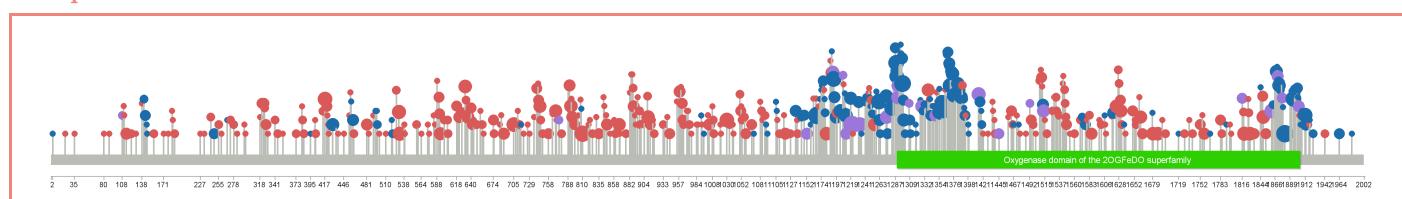
No unreported indel found.

### Substitutions

#### reported

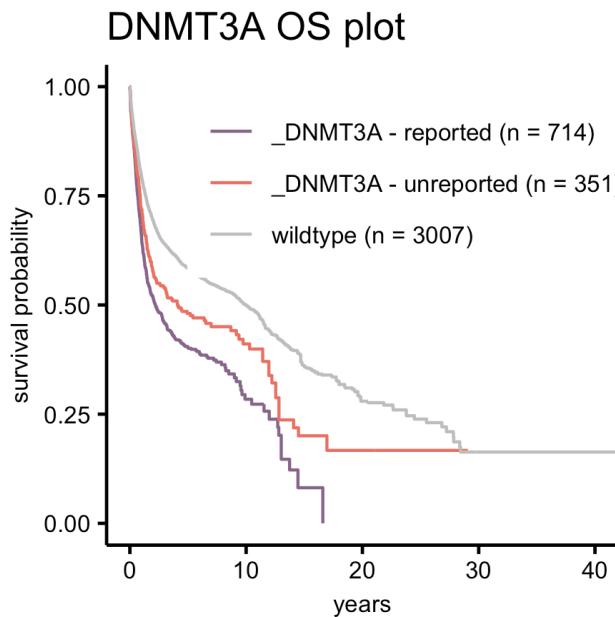


#### unreported



\* likely germline infiltration

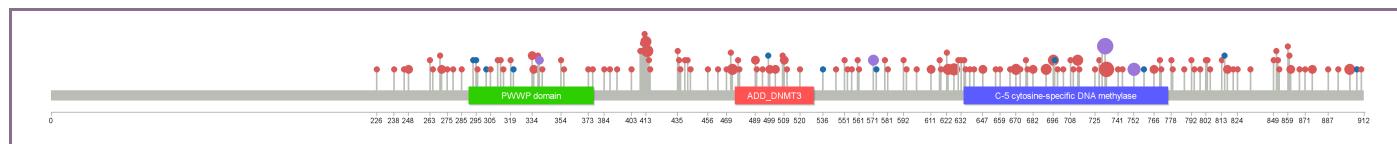
### 5.3 DNMT3A



disease	status consequence	x indel	x sub	✓
AML	inframe	1	0	14
	missense	0	297	733
	nonsense/splicing	1	70	6
	other	1	27	2
	truncating	0	0	119
MDS	inframe	0	0	11
	missense	0	105	126
	nonsense/splicing	0	48	4
	other	0	12	1
	truncating	0	0	51
MPN	inframe	0	0	1
	missense	0	44	45
	nonsense/splicing	0	16	3
	other	2	10	0
	truncating	0	0	18
<b>TOTAL</b>		5	629	1134
		0.00%	0.36%	0.64%

#### Indels

##### reported

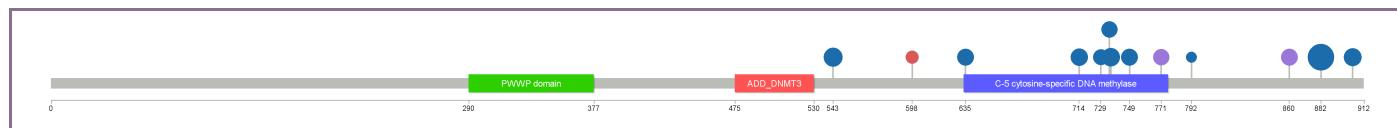


##### unreported

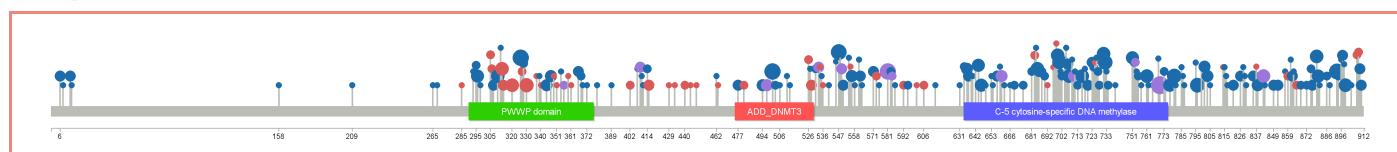
No unreported indel found.

#### Substitutions

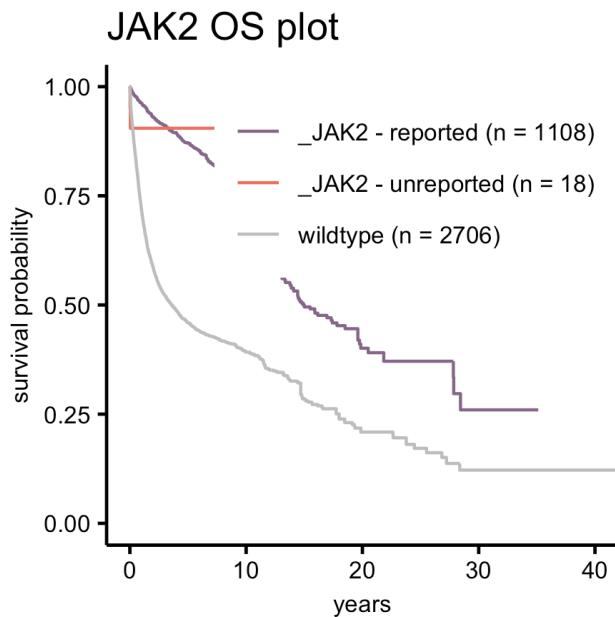
##### reported



##### unreported



## 5.4 JAK2



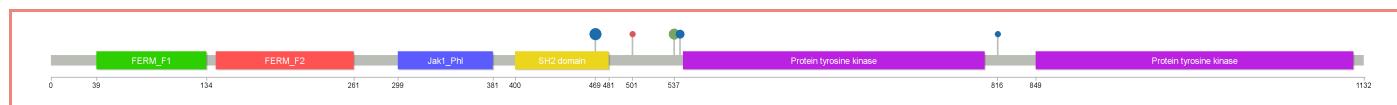
disease	status consequence	× indel	× sub	✓
<b>AML</b>	missense	0	8	134
	truncating	1	0	0
<b>MDS</b>	inframe	1	0	0
	missense	0	20	109
<b>MPN</b>	inframe	6	0	0
	missense	0	28	1388
	other	4	1	0
<b>TOTAL</b>		12	57	1631
		0.01%	0.03%	0.96%

### Indels

#### reported

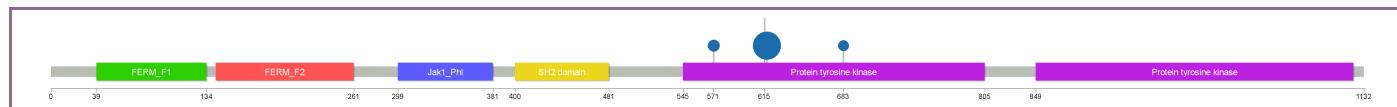
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported



### Substitutions

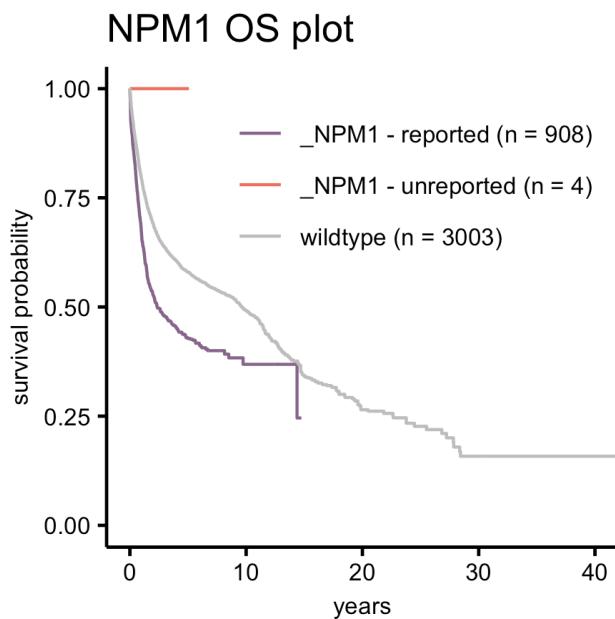
#### reported



#### unreported



## 5.5 NPM1



disease	status consequence	× indel	✓
AML	other	1	0
	truncating	1	1385
MDS	truncating	0	63
MPN	other	2	0
<b>TOTAL</b>		4	1448
		0.00%	1.00%

### Indels

#### reported



#### unreported



### Substitutions

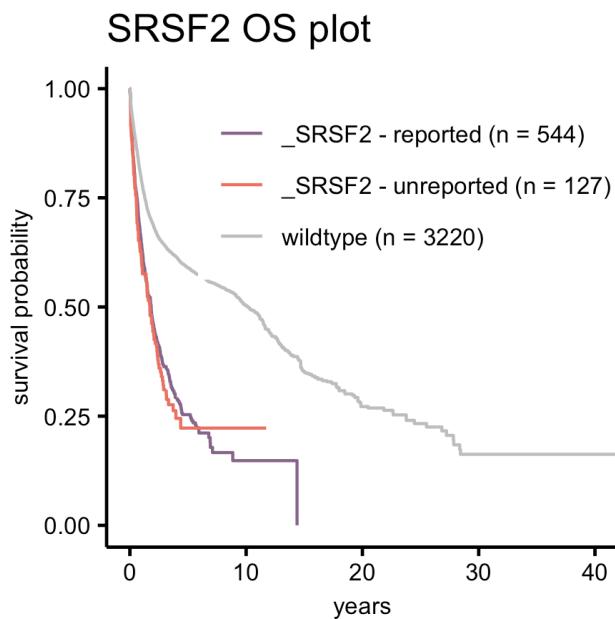
#### reported

This gene was not reported for hostpots (or no reported mutation was found in the dataset).

#### unreported

No unreported substitution found.

## 5.6 SRSF2



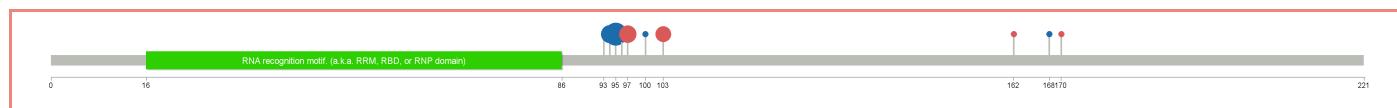
disease	status consequence	× indel	× sub	✓
AML	inframe	100	0	0
	missense	0	9	494
	other	1	0	0
	truncating	16	0	0
MDS	inframe	67	0	0
	missense	0	13	540
	truncating	15	0	0
MPN	inframe	2	0	0
	missense	0	1	36
<b>TOTAL</b>		201	23	1070
		0.16%	0.02%	0.83%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported



### Substitutions

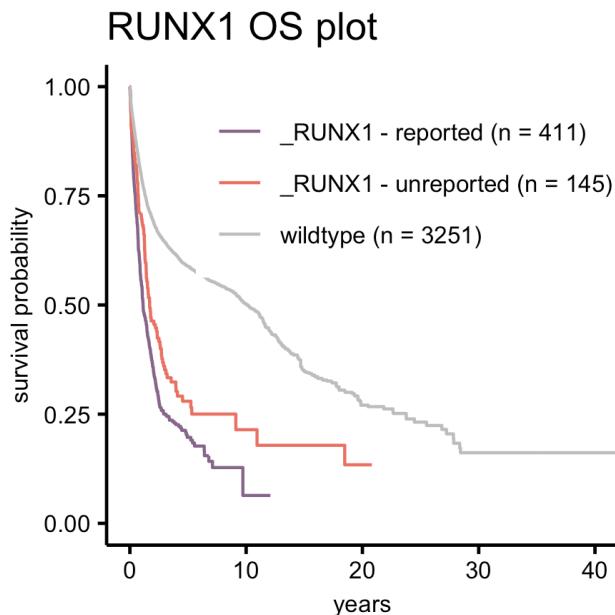
#### reported



#### unreported



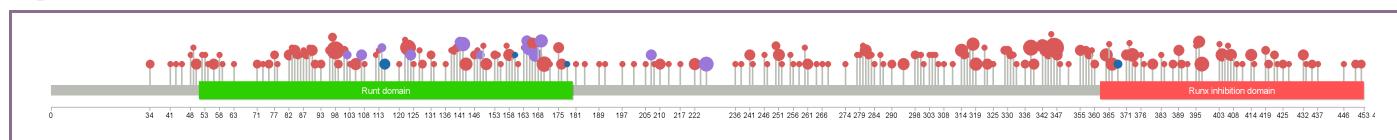
## 5.7 RUNX1



disease	status consequence	× indel	× sub	✓
<b>AML</b>	inframe	0	0	20
	missense	0	108	170
	nonsense/splicing	4	46	60
	other	12	23	6
	truncating	2	0	296
<b>MDS</b>	inframe	0	0	5
	missense	0	65	72
	nonsense/splicing	6	36	34
	other	1	5	0
	truncating	0	0	180
<b>MPN</b>	missense	0	7	2
	nonsense/splicing	0	0	2
	other	0	2	0
	truncating	0	0	1
<b>TOTAL</b>		25	292	848
		0.02%	0.25%	0.73%

### Indels

#### reported



#### unreported

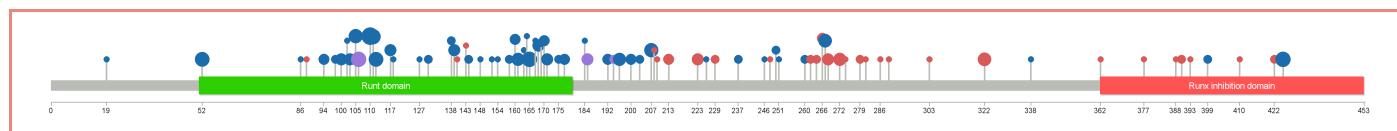
No unreported indel found.

### Substitutions

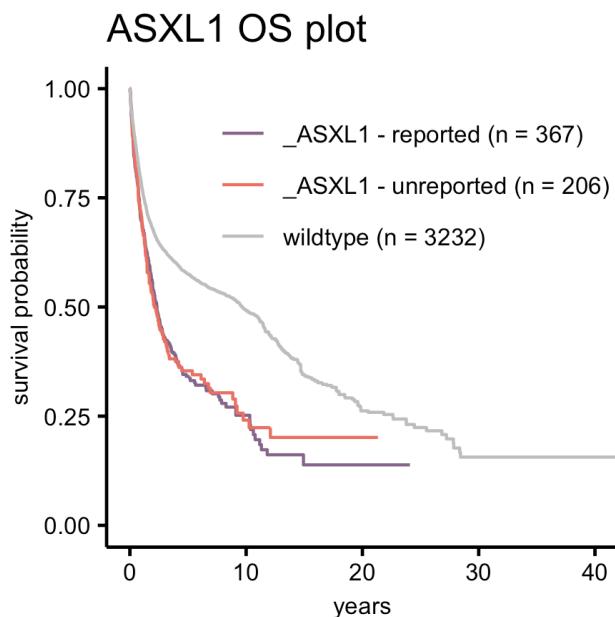
#### reported



#### unreported



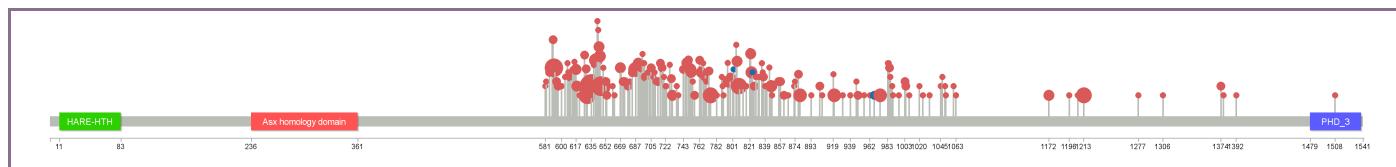
## 5.8 ASXL1



disease	status consequence	x indel	x sub	✓
AML	inframe	0	0	2
	missense	0	42	0
	nonsense/splicing	0	86	36
	other	1	0	0
	truncating	9	0	191
MDS	inframe	0	0	2
	missense	0	16	0
	nonsense/splicing	0	121	47
	other	0	1	0
	truncating	15	0	325
MPN	missense	0	14	0
	nonsense/splicing	0	37	9
	other	1	4	0
	truncating	5	0	73
<b>TOTAL</b>		31	321	685
		0.03%	0.31%	0.66%

### Indels

#### reported

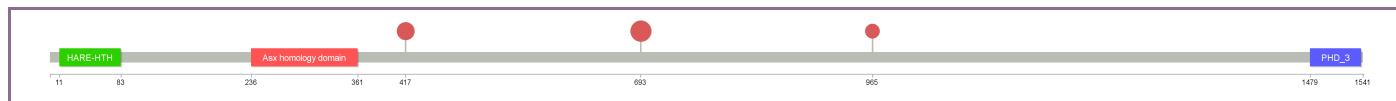


#### unreported

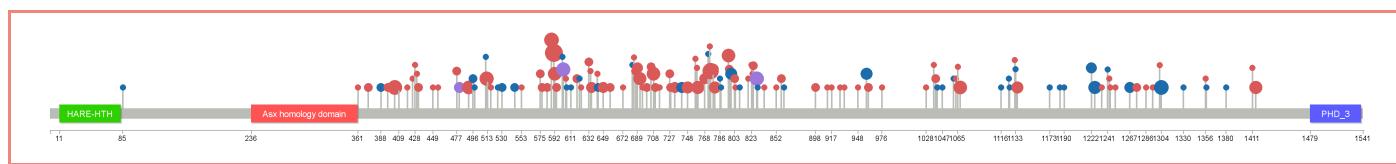


### Substitutions

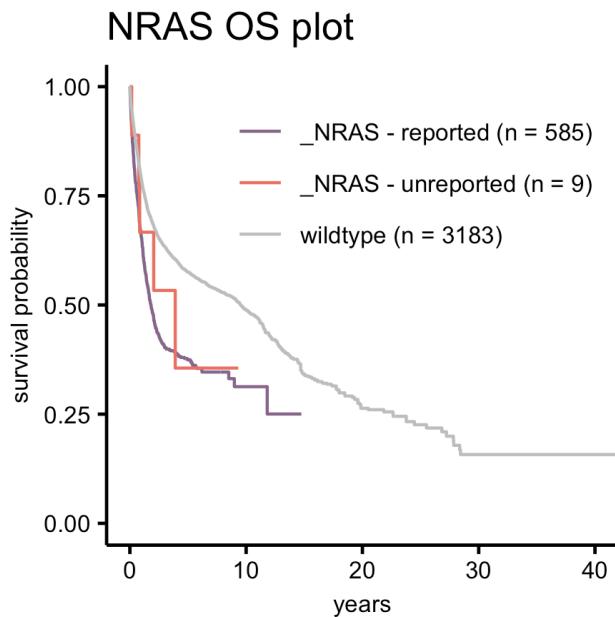
#### reported



#### unreported



## 5.9 NRAS



disease	status consequence	× sub	✓
AML	missense	6	823
MDS	missense	13	181
MPN	missense	0	9
<b>TOTAL</b>		19	1013
		0.02%	0.98%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

No unreported indel found.

### Substitutions

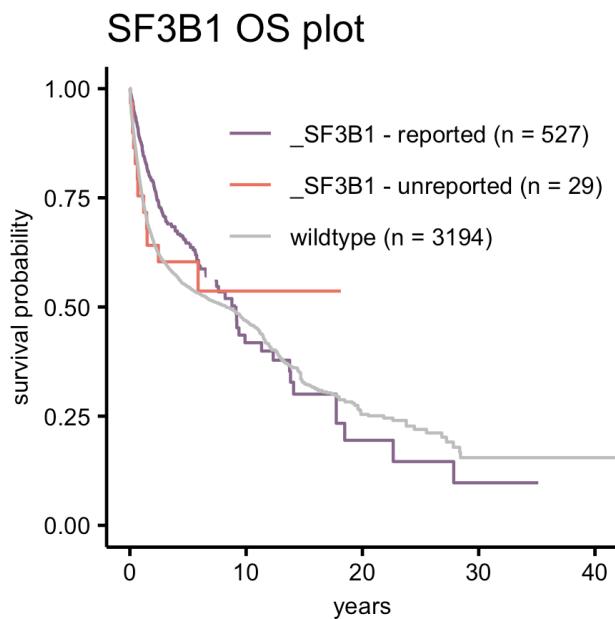
#### reported



#### unreported



## 5.10 SF3B1



disease	status consequence	× indel	× sub	✓
AML	missense	0	15	132
	other	0	1	0
MDS	missense	0	12	704
	other	4	0	0
MPN	inframe	1	0	0
	missense	0	9	37
	nonsense/splicing	0	1	0
	other	1	0	0
<b>TOTAL</b>		6	38	873
		0.01%	0.04%	0.95%

### Indels

#### reported

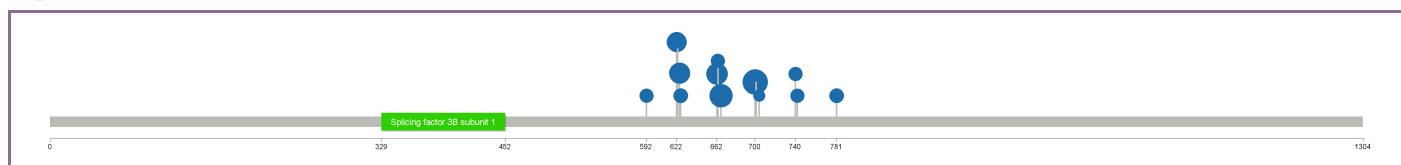
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

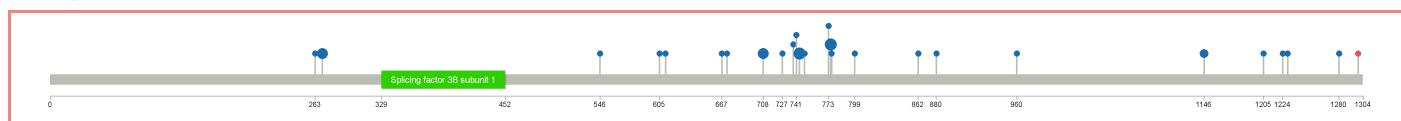


### Substitutions

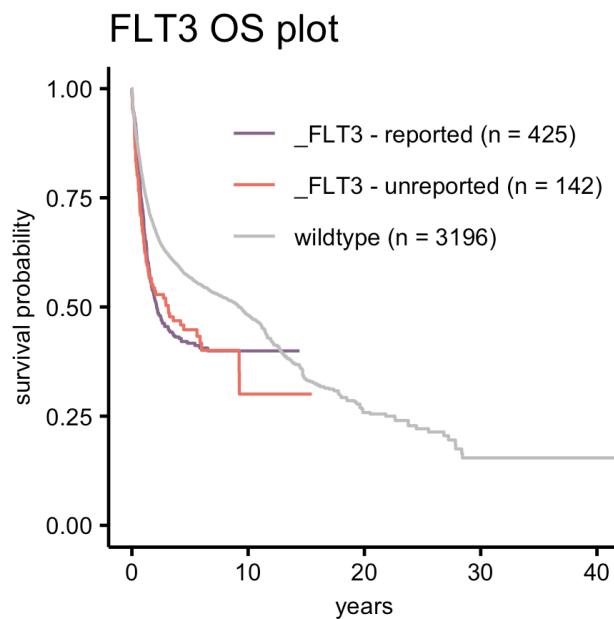
#### reported



#### unreported



## 5.11 FLT3



disease	status consequence	× indel	× sub	✓
AML	inframe	0	0	141
	missense	0	203	425
	nonsense/splicing	0	1	11
	other	0	0	26
MDS	inframe	0	0	9
	missense	0	20	16
MPN	missense	0	9	0
	truncating	1	0	0
<b>TOTAL</b>		1	233	628
		0.00%	0.27%	0.73%

### Indels

#### reported

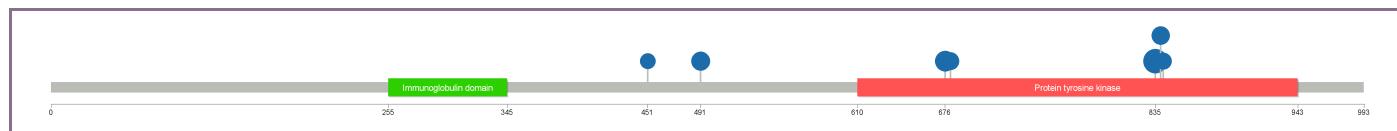


#### unreported

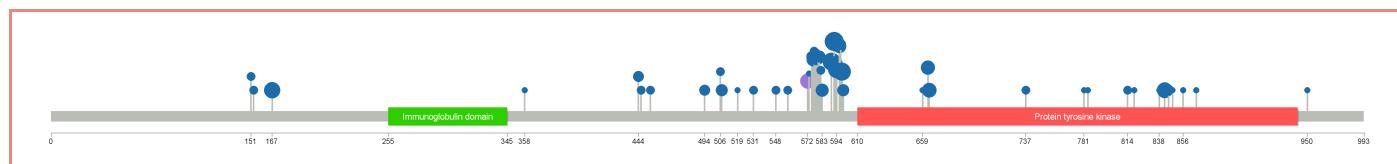


### Substitutions

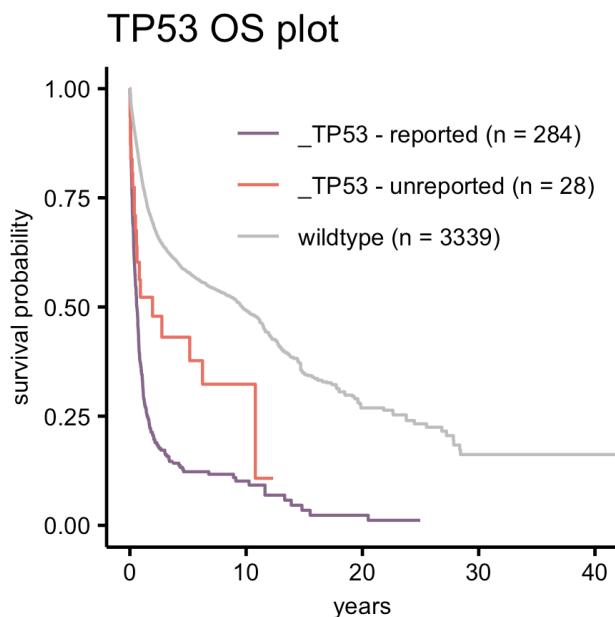
#### reported



#### unreported



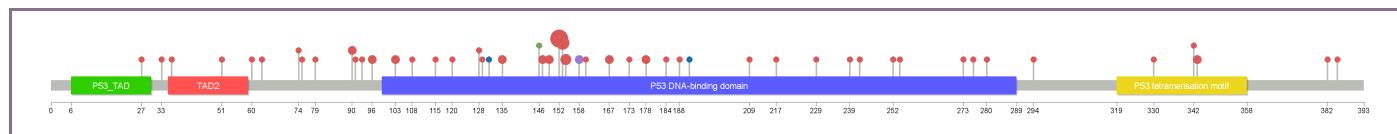
## 5.12 TP53



disease	status consequence	x indel	x sub	✓
AML	inframe	0	0	1
	missense	0	5	353
	nonsense/splicing	1	12	33
	other	2	23	7
	truncating	0	0	43
MDS	inframe	0	0	2
	missense	0	10	227
	nonsense/splicing	3	10	10
	other	0	5	2
	truncating	0	0	40
MPN	missense	0	4	23
	nonsense/splicing	0	0	2
	other	1	5	1
	truncating	0	0	1
TOTAL		7	74	745
		0.01%	0.09%	0.90%

### Indels

#### reported

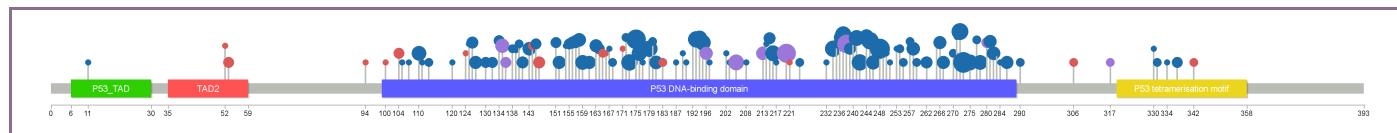


#### unreported

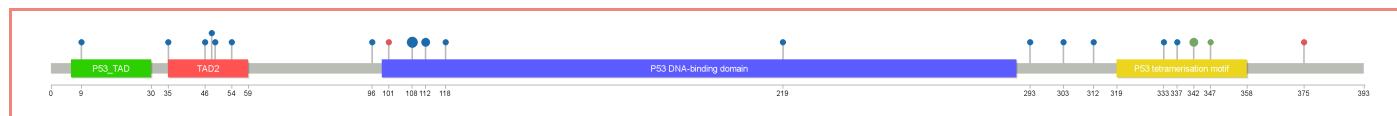
No unreported indel found.

### Substitutions

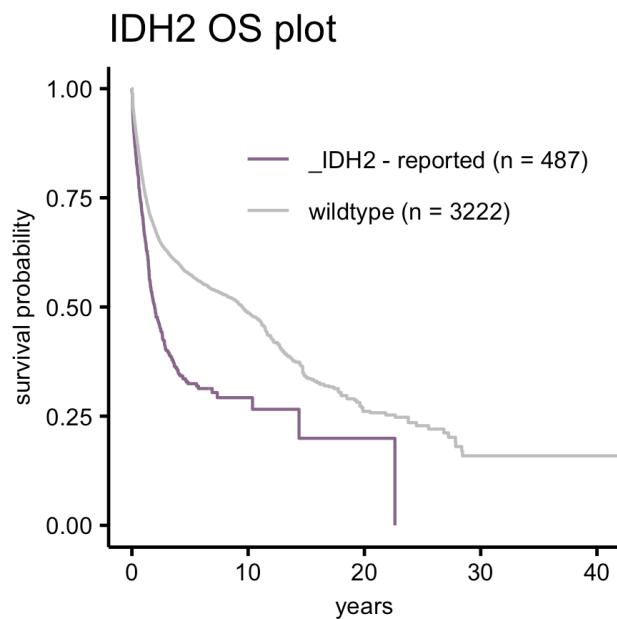
#### reported



#### unreported



## 5.13 IDH2



disease	status consequence	× sub	✓
AML	missense	2	578
MDS	missense	0	123
MPN	missense	0	16
<b>TOTAL</b>		2	717
		0.00%	1.00%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

No unreported indel found.

### Substitutions

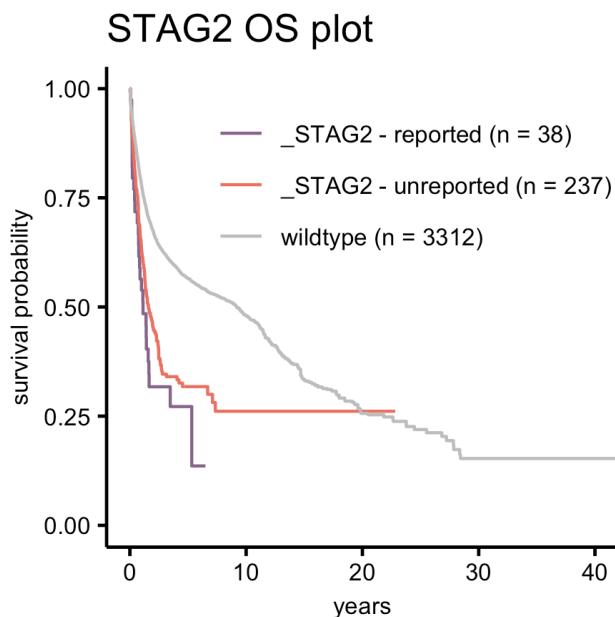
#### reported



#### unreported



## 5.14 STAG2



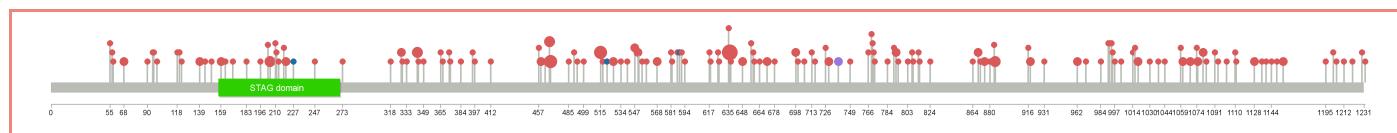
disease	status consequence	× indel	× sub	✓
AML	inframe	3	0	0
	missense	0	16	0
	nonsense/splicing	2	104	39
	other	7	19	0
	truncating	105	0	0
MDS	inframe	1	0	0
	missense	0	3	0
	nonsense/splicing	3	107	35
	other	6	10	0
MPN	truncating	102	0	0
	missense	0	4	0
	nonsense/splicing	0	1	2
	other	1	1	0
TOTAL	truncating	1	0	0
		231	265	76
		0.40%	0.46%	0.13%

### Indels

#### reported

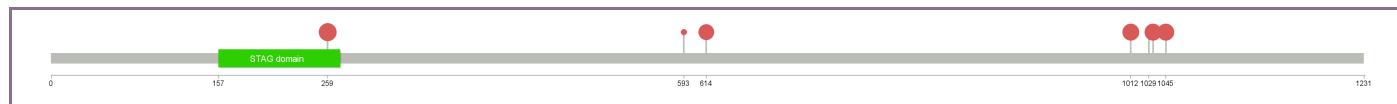
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

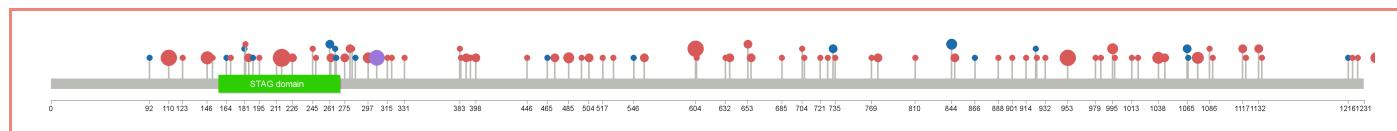


### Substitutions

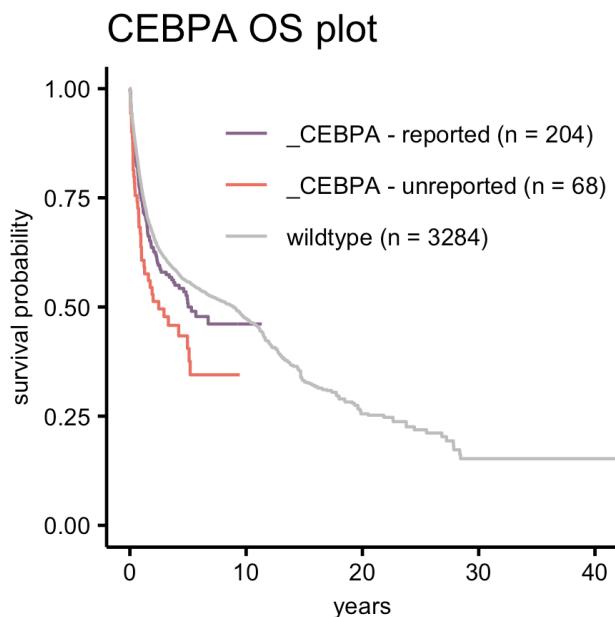
#### reported



#### unreported



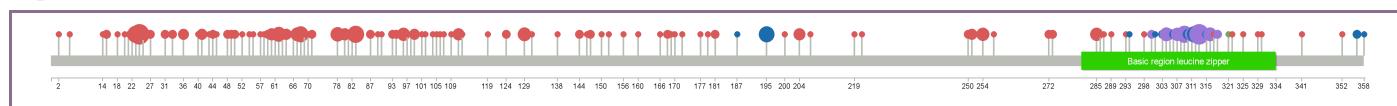
## 5.15 CEBPA



disease	status consequence	× indel	× sub	✓
AML	inframe	0	0	98
	missense	0	88	0
	nonsense/splicing	0	33	0
	other	2	0	17
	truncating	0	0	239
MDS	inframe	0	0	11
	missense	0	14	0
	nonsense/splicing	0	6	0
	truncating	0	0	36
MPN	missense	0	2	0
<b>TOTAL</b>		2	143	401
		0.00%	0.26%	0.73%

### Indels

#### reported



#### unreported

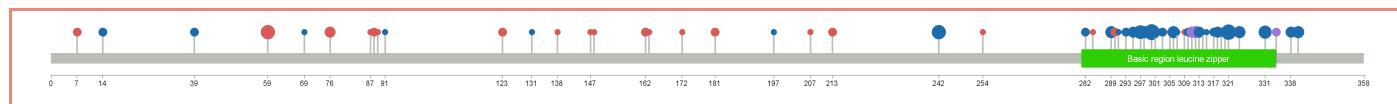
No unreported indel found.

### Substitutions

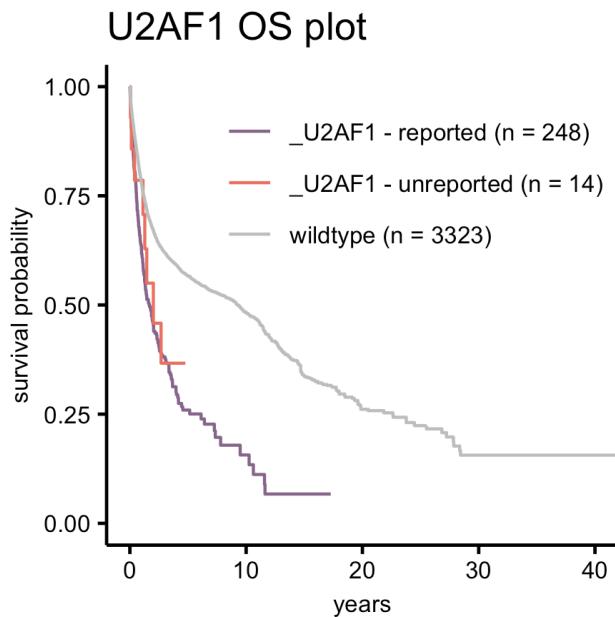
#### reported

This gene was not reported for hostpots (or no reported mutation was found in the dataset).

#### unreported



## 5.16 U2AF1



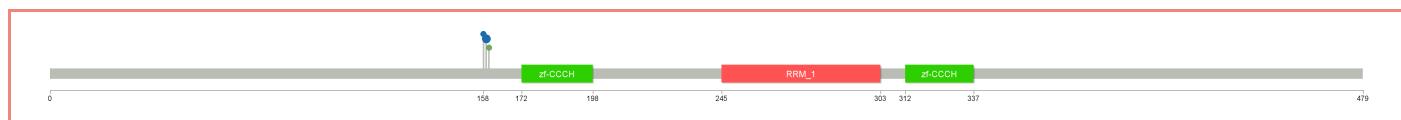
disease	status consequence	× indel	× sub	✓
<b>AML</b>	inframe	1	0	0
	missense	0	14	216
<b>MDS</b>	inframe	1	0	0
	missense	0	4	252
<b>MPN</b>	other	1	0	0
	inframe	1	0	0
	missense	0	0	30
<b>TOTAL</b>	other	1	0	0
		5	18	498
		0.01%	0.03%	0.96%

### Indels

#### reported

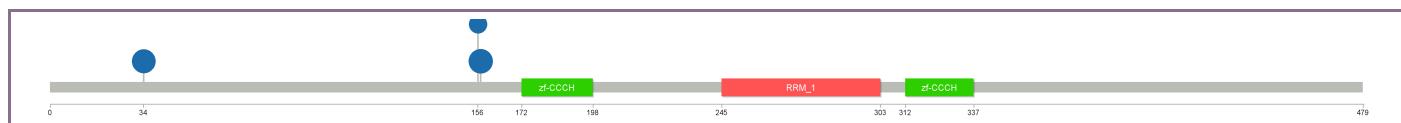
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

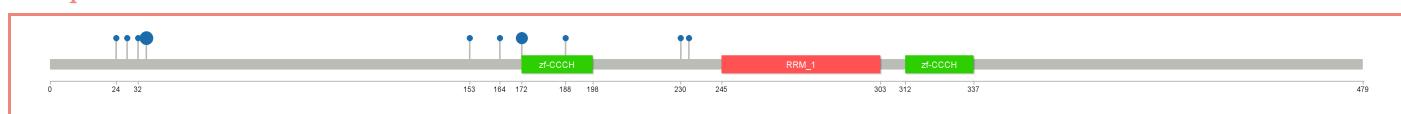


### Substitutions

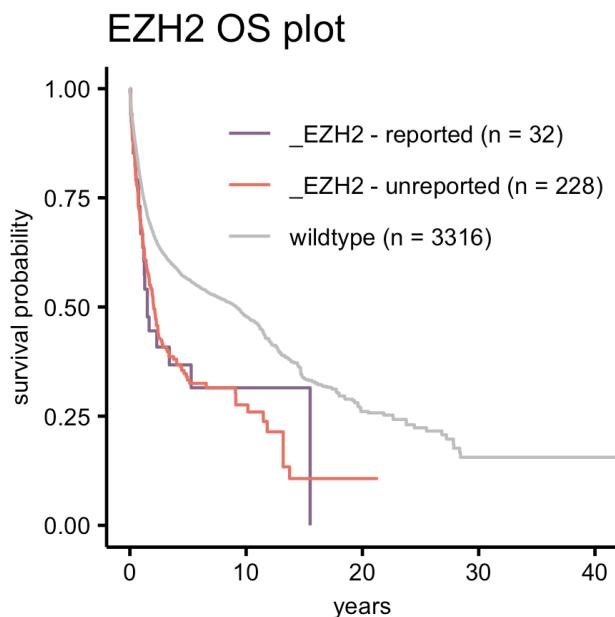
#### reported



#### unreported



## 5.17 EZH2



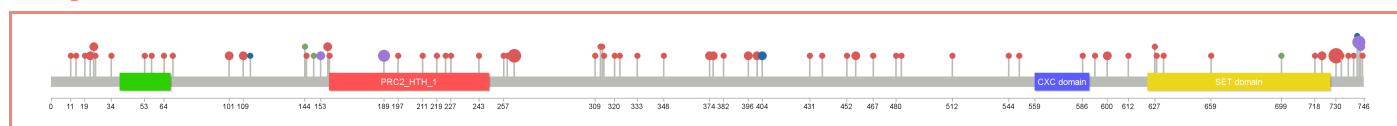
disease	status consequence	× indel	× sub	✓
AML	inframe	5	0	0
	missense	0	91	23
	nonsense/splicing	1	25	0
	other	4	12	0
	truncating	47	0	0
MDS	inframe	1	0	0
	missense	0	91	27
	nonsense/splicing	3	47	3
	other	2	5	0
	truncating	47	0	0
MPN	inframe	3	0	0
	missense	0	31	8
	nonsense/splicing	0	3	0
	other	2	9	0
	truncating	2	0	0
<b>TOTAL</b>		117	314	61
		0.24%	0.64%	0.12%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

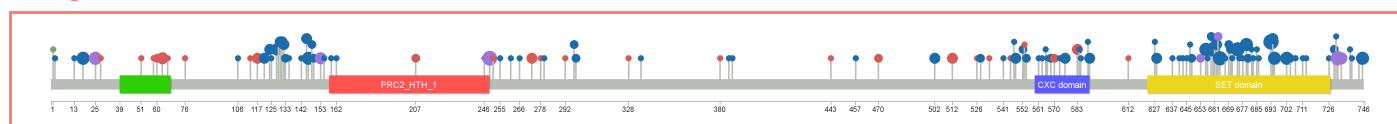


### Substitutions

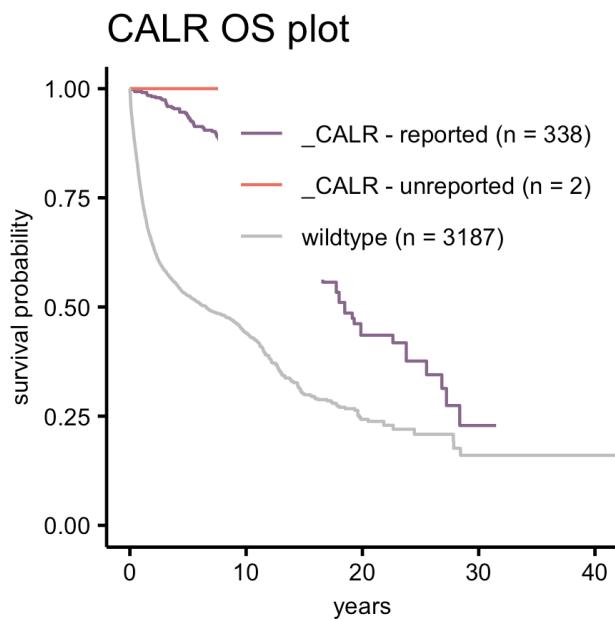
#### reported



#### unreported



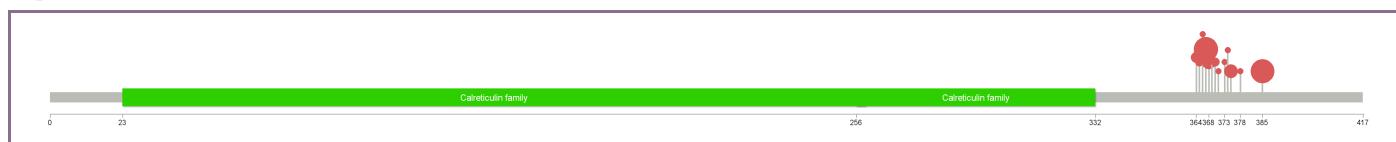
## 5.18 CALR



disease	status consequence	× indel	× sub	✓
AML	truncating	0	0	28
MDS	truncating	0	0	4
MPN	nonsense/splicing	0	2	0
	truncating	1	0	446
<b>TOTAL</b>		1	2	478
		0.00%	0.00%	0.99%

### Indels

#### reported



#### unreported



### Substitutions

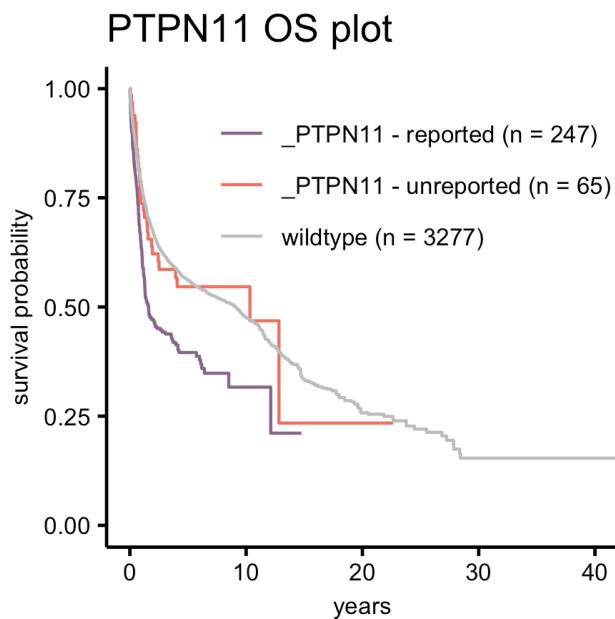
#### reported

This gene was not reported for hotspots (or no reported mutation was found in the dataset).

#### unreported



## 5.19 PTPN11



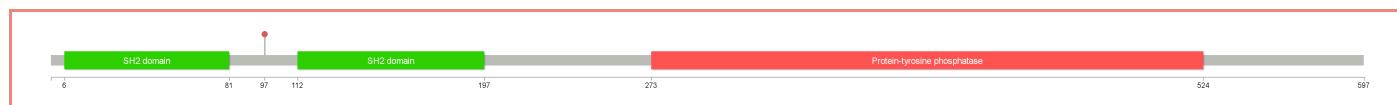
disease	status consequence	× indel	× sub	✓
AML	missense	0	67	329
MDS	missense	0	20	38
MPN	missense	0	9	4
	other	1	0	0
	truncating	1	0	0
<b>TOTAL</b>		2	96	371
		0.00%	0.20%	0.79%

### Indels

#### reported

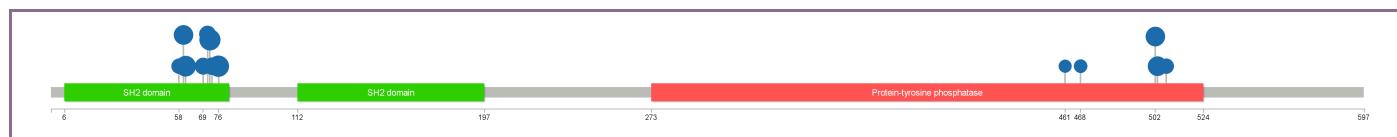
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

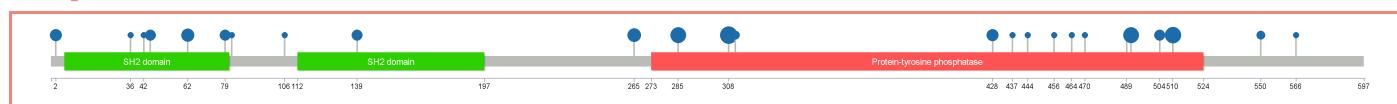


### Substitutions

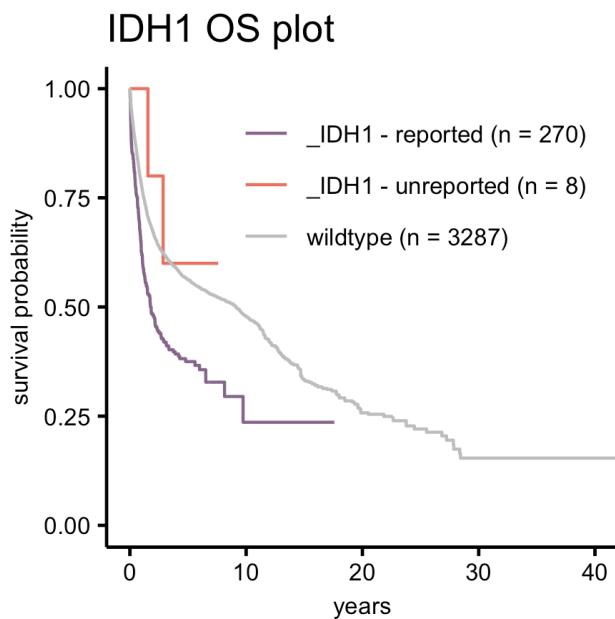
#### reported



#### unreported



## 5.20 IDH1



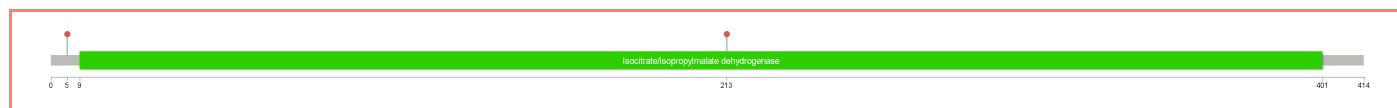
disease	status consequence	× indel	× sub	✓
AML	missense	0	1	337
MDS	missense	0	3	60
	nonsense/splicing	0	1	0
	truncating	1	0	0
MPN	missense	0	1	7
	other	1	0	0
	truncating	1	0	0
<b>TOTAL</b>		3	6	404
		0.01%	0.01%	0.98%

### Indels

#### reported

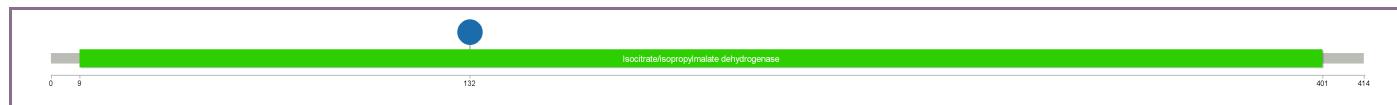
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

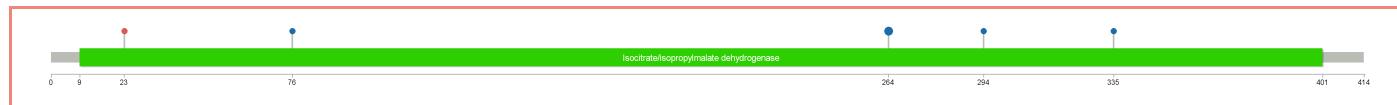


### Substitutions

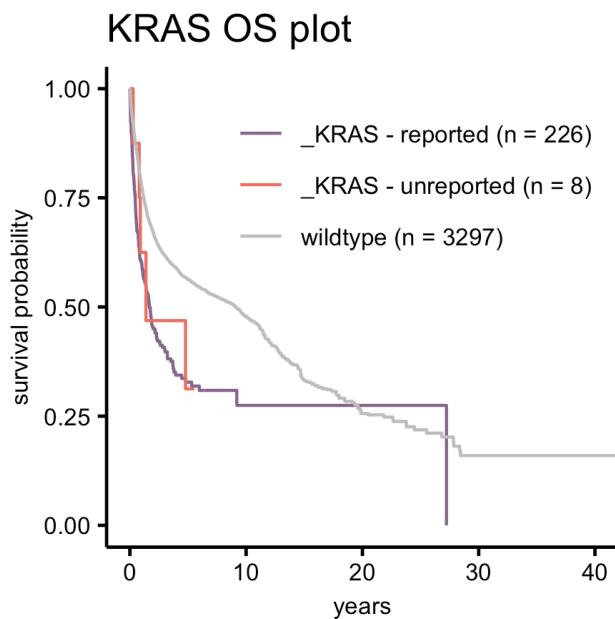
#### reported



#### unreported



## 5.21 KRAS



disease	status consequence	× indel	× sub	✓
AML	inframe	2	0	0
	missense	0	1	258
	nonsense/splicing	0	1	1
MDS	missense	0	8	116
MPN	missense	0	2	10
<b>TOTAL</b>		2	12	385
		0.01%	0.03%	0.96%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported



### Substitutions

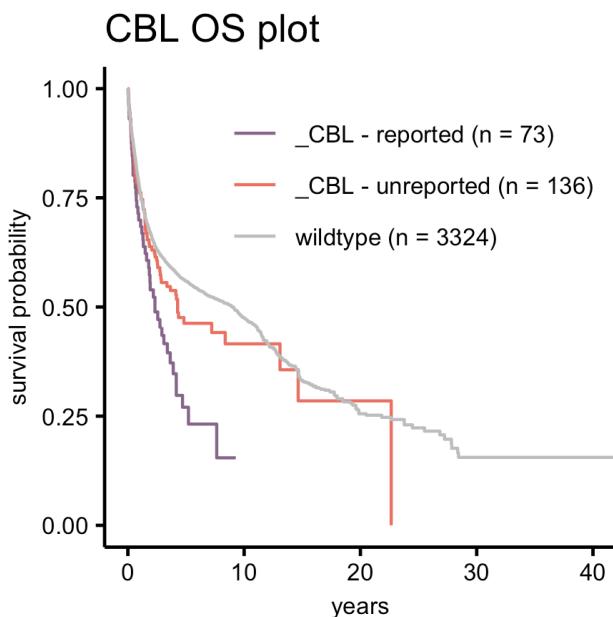
#### reported



#### unreported



## 5.22 CBL



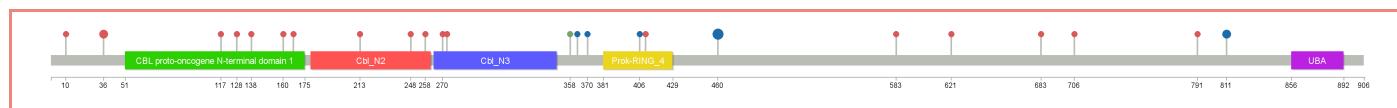
disease	status	indel	sub	✓
	consequence			
AML	inframe	1	0	0
	missense	0	65	54
	nonsense/splicing	0	10	0
	other	3	6	0
	truncating	2	0	0
		2	0	0
MDS	inframe	2	0	0
	missense	0	84	69
	nonsense/splicing	0	35	0
	other	1	3	0
	truncating	14	0	0
		2	0	0
MPN	inframe	5	0	0
	missense	0	24	8
	nonsense/splicing	0	4	0
	other	0	1	0
	truncating	3	0	0
		2	0	0
<b>TOTAL</b>		31	232	131
		0.08%	0.59%	0.33%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

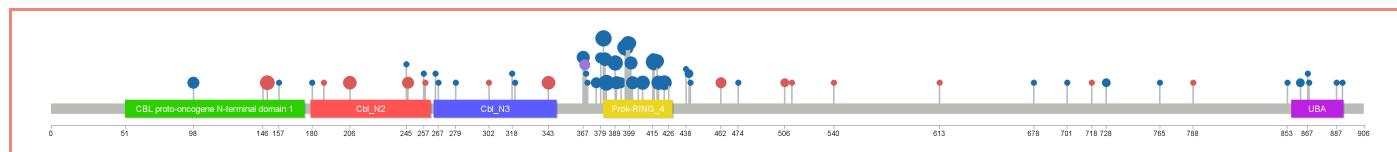


### Substitutions

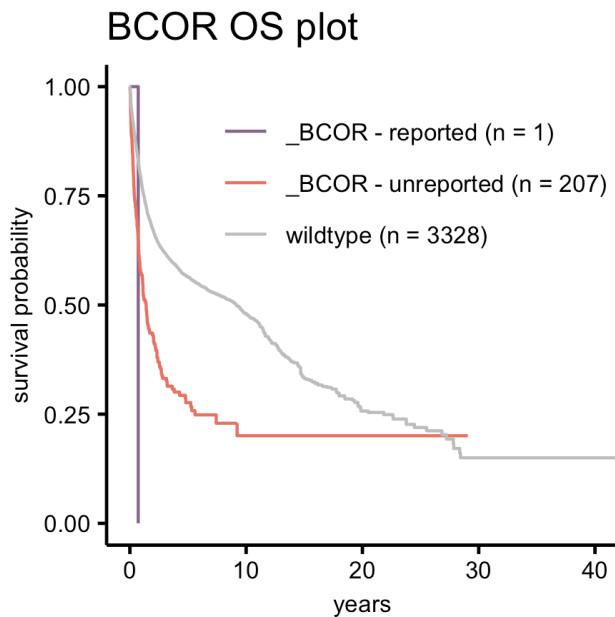
#### reported



#### unreported



## 5.23 BCOR



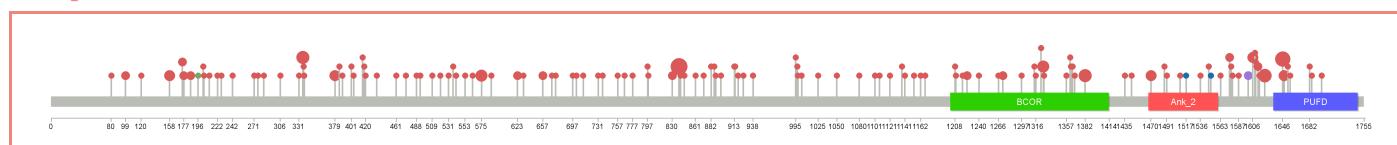
disease	status consequence	x indel	x sub	✓
AML	inframe	2	0	0
	missense	0	14	1
	nonsense/splicing	0	76	0
	other	0	8	0
	truncating	136	0	0
MDS	inframe	1	0	0
	missense	0	8	1
	nonsense/splicing	0	37	0
	other	2	6	0
	truncating	54	0	0
MPN	missense	0	6	0
	truncating	5	0	0
<b>TOTAL</b>		200	155	2
		0.56%	0.43%	0.01%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

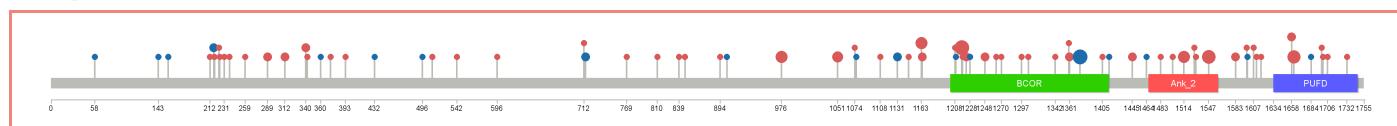


### Substitutions

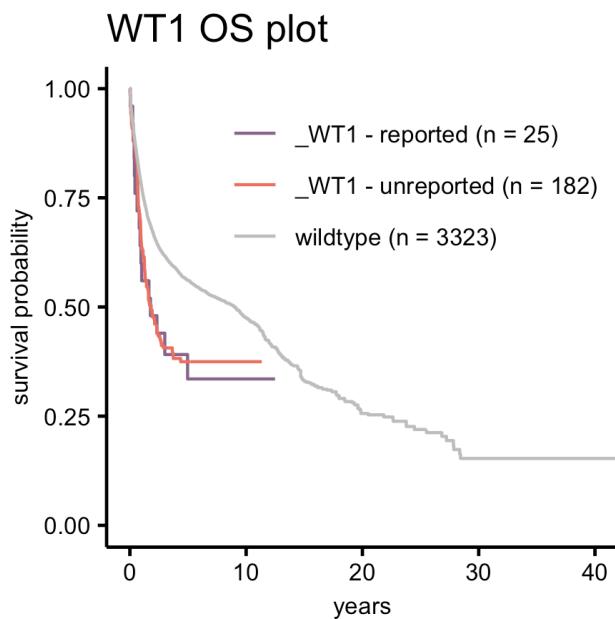
#### reported



#### unreported



## 5.24 WT1



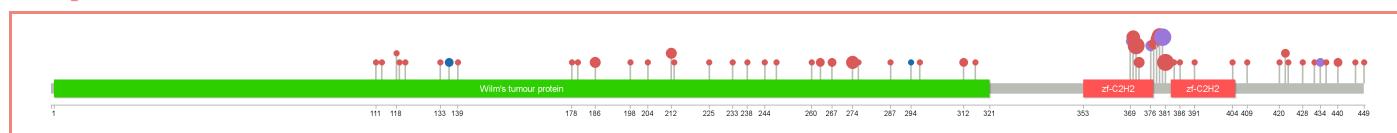
disease	status consequence	× indel	× sub	✓
AML	inframe	6	0	0
	missense	0	67	30
	nonsense/splicing	2	58	0
	other	4	13	0
MDS	truncating	121	0	0
	missense	0	10	1
	nonsense/splicing	0	4	0
TOTAL	truncating	14	0	0
		147	152	31
		0.45%	0.46%	0.09%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

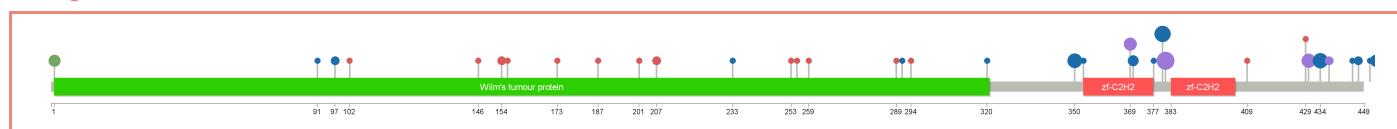


### Substitutions

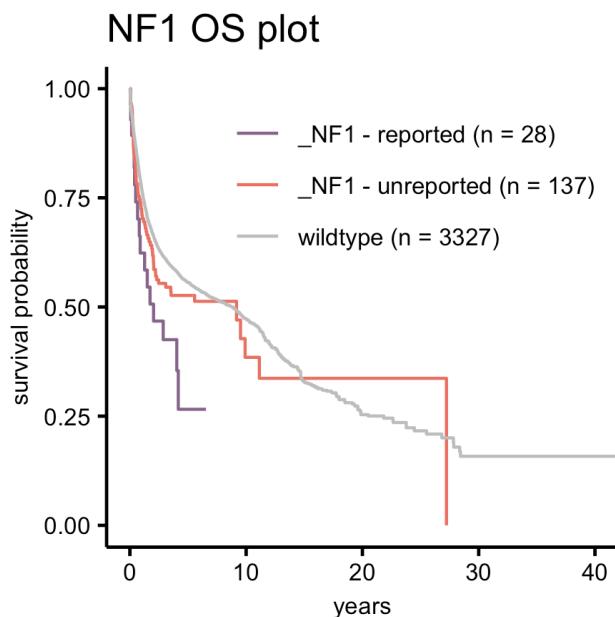
#### reported



#### unreported



## 5.25 NF1



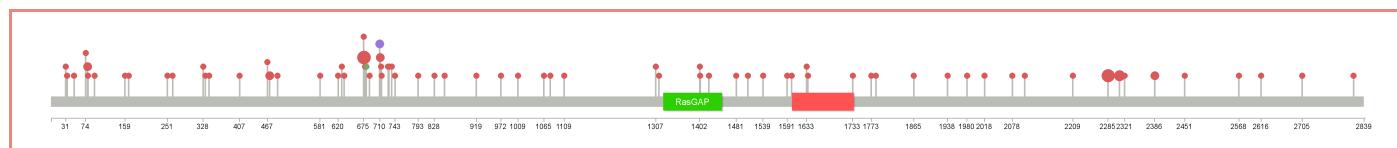
disease	status consequence	× indel	× sub	✓
AML	inframe	1	0	0
	missense	0	24	7
	nonsense/splicing	1	42	19
	other	3	12	0
	truncating	59	0	0
MDS	missense	0	10	12
	nonsense/splicing	1	17	7
	other	1	3	0
	truncating	25	0	0
MPN	missense	0	16	0
	nonsense/splicing	1	1	1
	other	9	5	0
	truncating	3	0	0
<b>TOTAL</b>		104	130	46
		0.37%	0.46%	0.16%

### Indels

#### reported

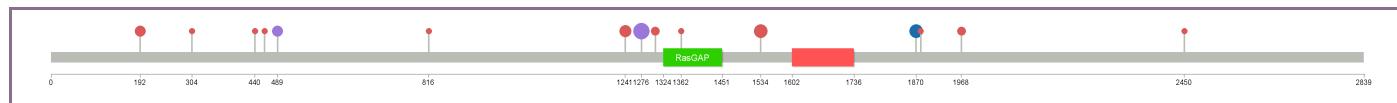
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

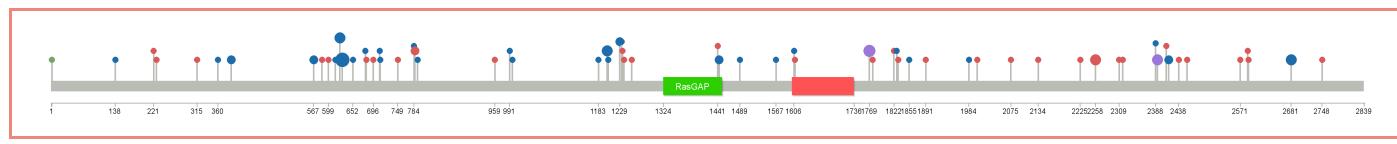


### Substitutions

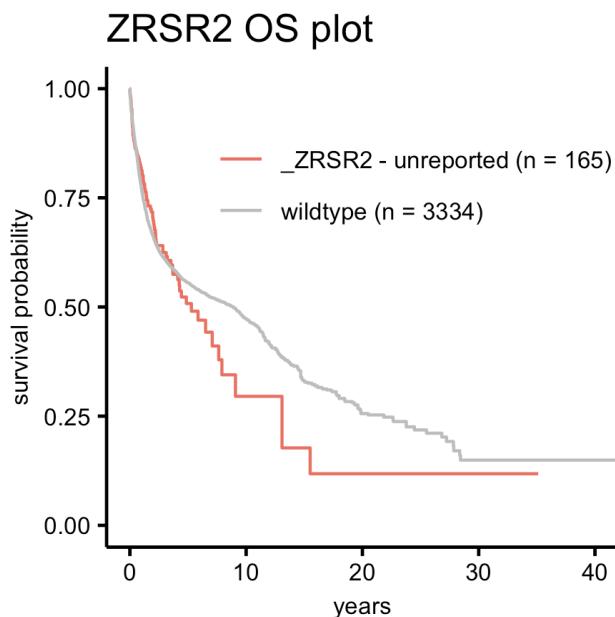
#### reported



#### unreported



## 5.26 ZRSR2



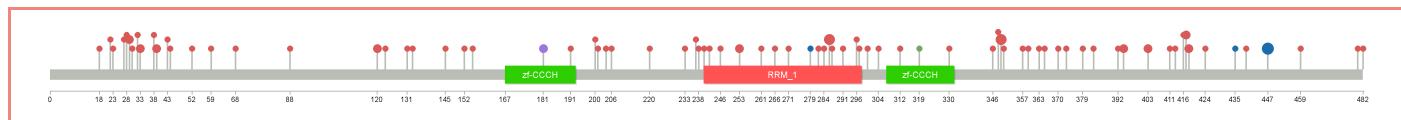
disease	status consequence	× indel	× sub
AML	inframe	1	0
	missense	0	9
	nonsense/splicing	0	22
	other	0	2
	truncating	24	0
MDS	inframe	1	0
	missense	0	43
	nonsense/splicing	2	80
	other	1	15
	truncating	57	0
MPN	inframe	5	0
	missense	0	1
	nonsense/splicing	0	6
	other	0	2
	truncating	9	0
<b>TOTAL</b>		100	180
		0.36%	0.64%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

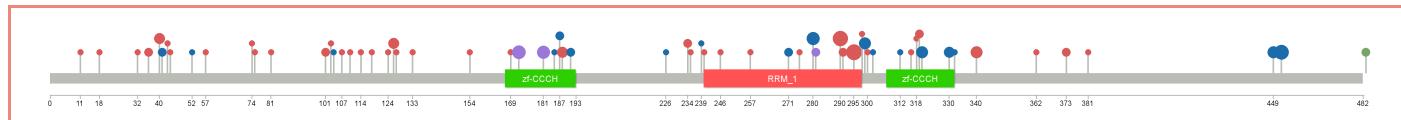


### Substitutions

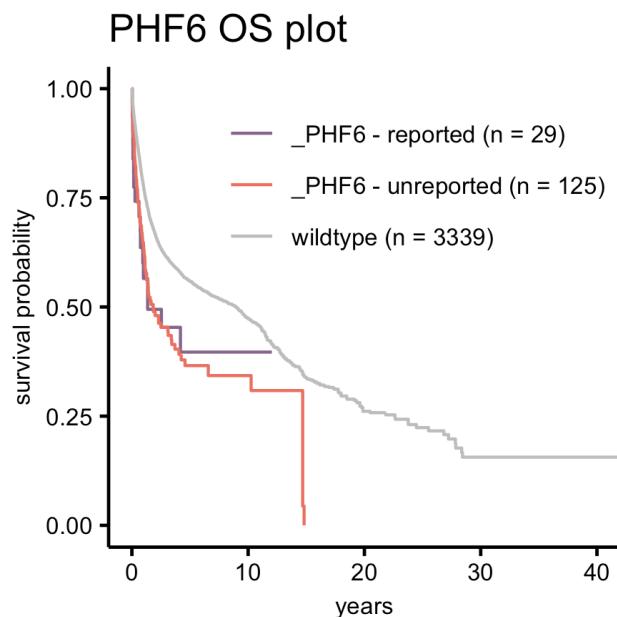
#### reported

This gene was not reported for hotspots (or no reported mutation was found in the dataset).

#### unreported



## 5.27 PHF6



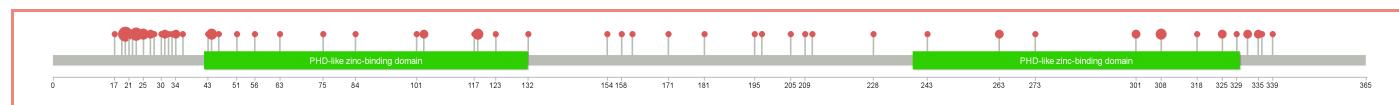
disease	status consequence	× indel	× sub	✓
AML	missense	0	35	12
	nonsense/splicing	1	26	19
	other	2	20	0
	truncating	47	0	0
MDS	missense	0	22	6
	nonsense/splicing	0	21	13
	other	2	5	0
	truncating	27	0	0
MPN	missense	0	3	0
	nonsense/splicing	0	1	0
	other	1	0	0
	truncating	8	0	0
<b>TOTAL</b>		88	133	50
		0.32%	0.49%	0.18%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

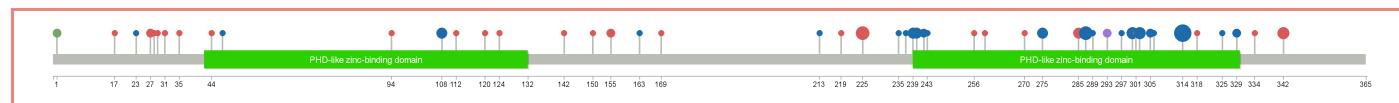


### Substitutions

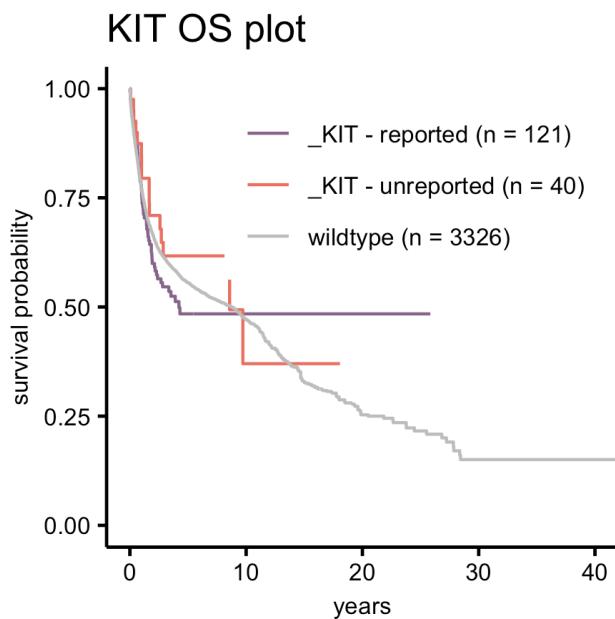
#### reported



#### unreported



## 5.28 KIT



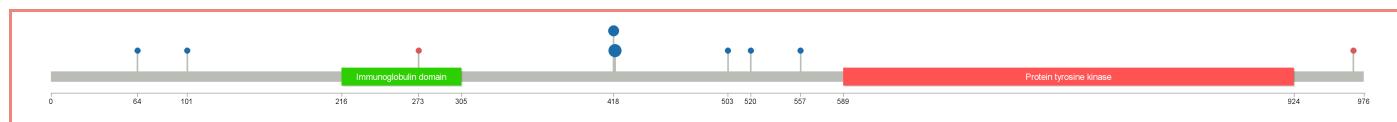
disease	status consequence	× indel	× sub	✓
AML	inframe	13	0	0
	missense	0	18	141
	truncating	1	0	0
MDS	missense	0	12	17
	other	1	0	0
MPN	missense	0	8	3
	truncating	1	0	0
TOTAL		16	38	161
		0.07%	0.18%	0.75%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

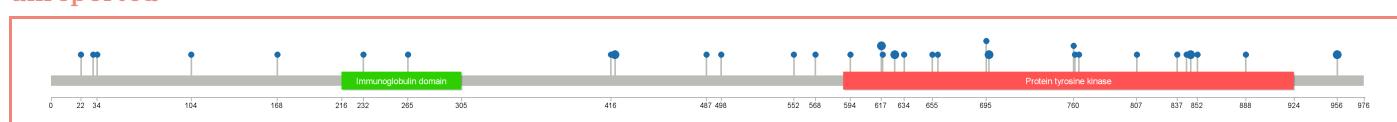


### Substitutions

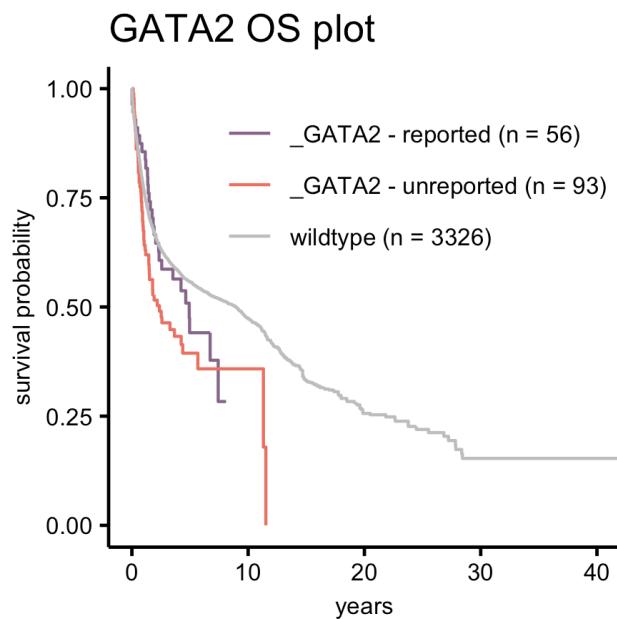
#### reported



#### unreported



## 5.29 GATA2



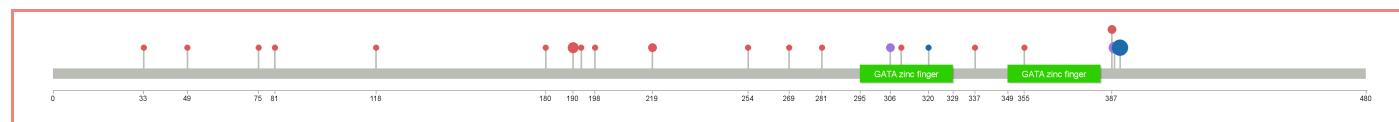
disease	status consequence	× indel	× sub	✓
AML	inframe	7	0	0
	missense	0	78	66
	other	2	0	0
	truncating	15	0	0
MDS	inframe	8	0	0
	missense	0	18	6
	nonsense/splicing	0	1	0
	truncating	6	0	0
MPN	inframe	2	0	0
	missense	0	4	1
	truncating	1	0	0
<b>TOTAL</b>		41	101	73
		0.19%	0.47%	0.34%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

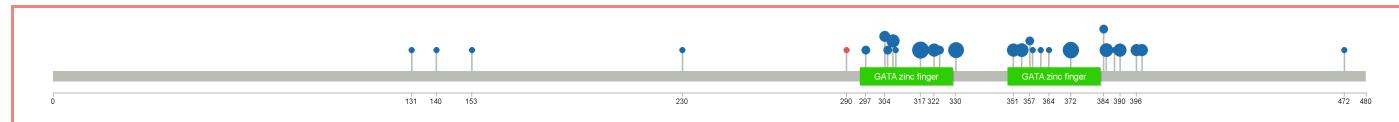


### Substitutions

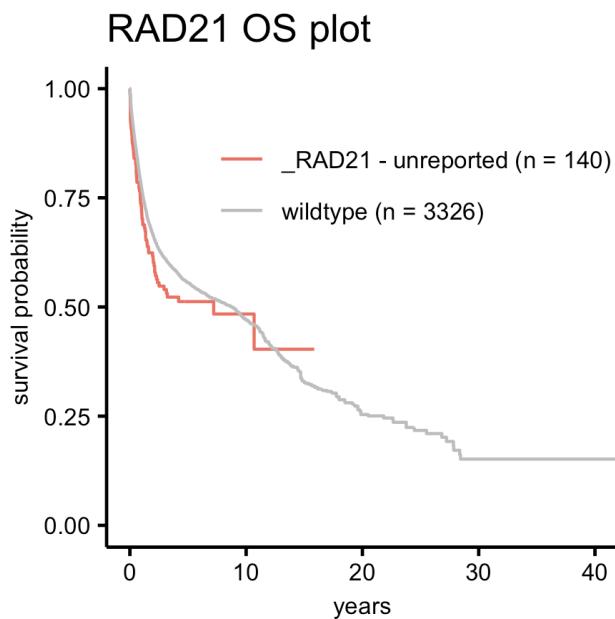
#### reported



#### unreported



### 5.30 RAD21



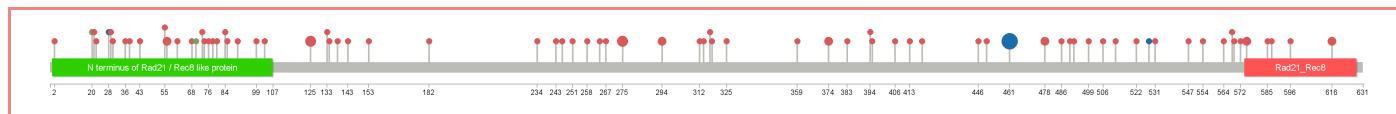
disease	status consequence	× indel	× sub
AML	inframe	8	0
	missense	0	26
	nonsense/splicing	0	40
	other	2	15
	truncating	68	0
MDS	inframe	6	0
	missense	0	8
	nonsense/splicing	0	9
	other	1	4
	truncating	12	0
MPN	other	2	1
	truncating	3	0
<b>TOTAL</b>		102	103
0.50%		0.50%	0.50%

#### Indels

##### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

##### unreported

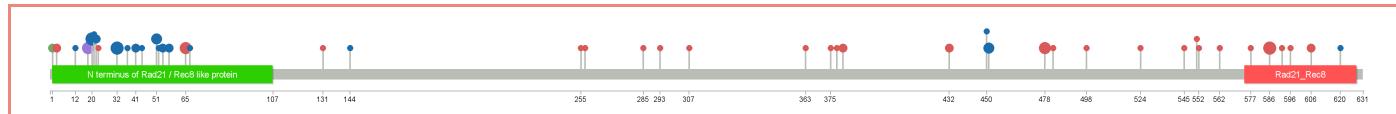


#### Substitutions

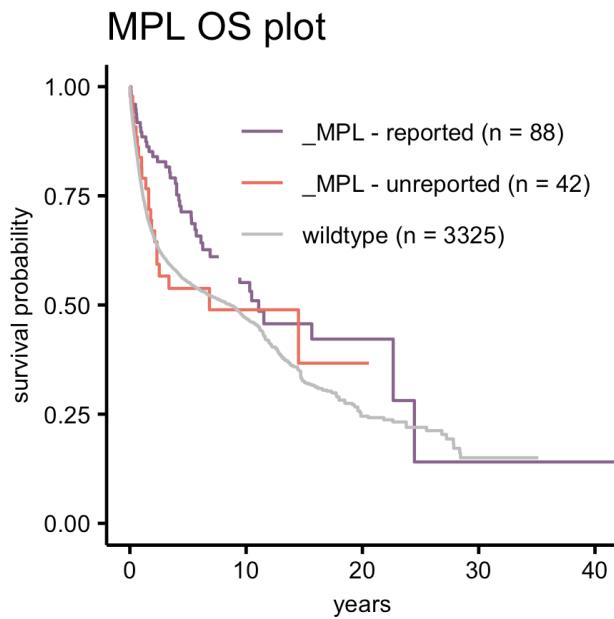
##### reported

This gene was not reported for hostpots (or no reported mutation was found in the dataset).

##### unreported



### 5.31 MPL



disease	status consequence	× indel	× sub	✓
AML	missense	0	10	20
	nonsense/splicing	0	1	2
	other	1	0	0
MDS	missense	0	15	29
	nonsense/splicing	0	8	1
	truncating	4	0	0
MPN	missense	0	22	75
	nonsense/splicing	0	0	11
	truncating	1	0	0
<b>TOTAL</b>		6	56	138
		0.03%	0.28%	0.69%

#### Indels

##### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

##### unreported

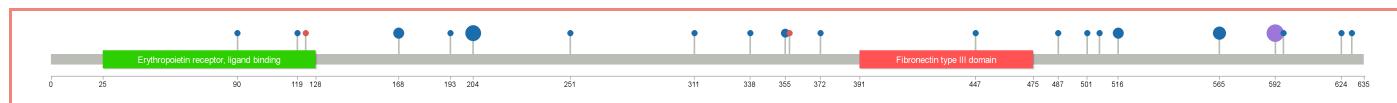


#### Substitutions

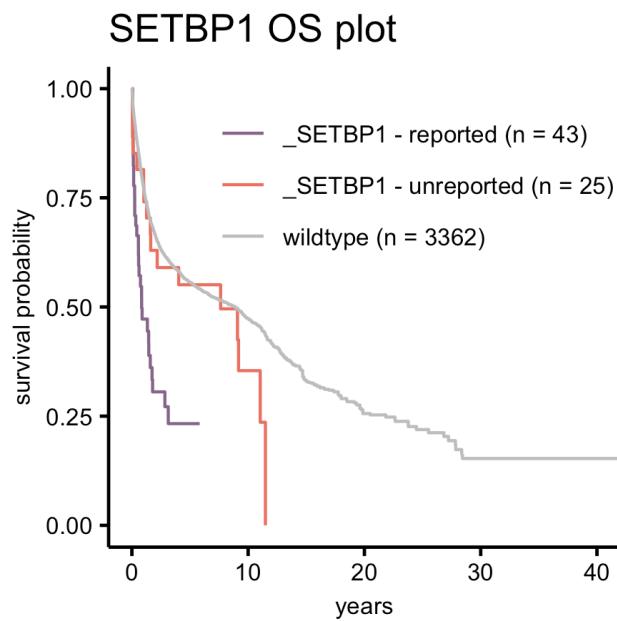
##### reported



##### unreported



## 5.32 SETBP1



disease	status consequence	× sub	✓
AML	missense	20	58
MDS	missense	6	59
MPN	missense	13	0
<b>TOTAL</b>		39	117
		0.25%	0.75%

### Indels

#### reported

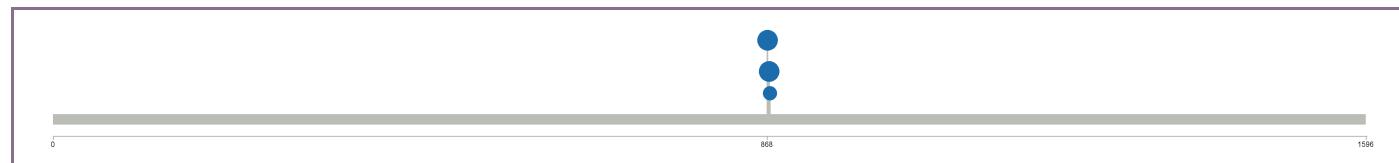
This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

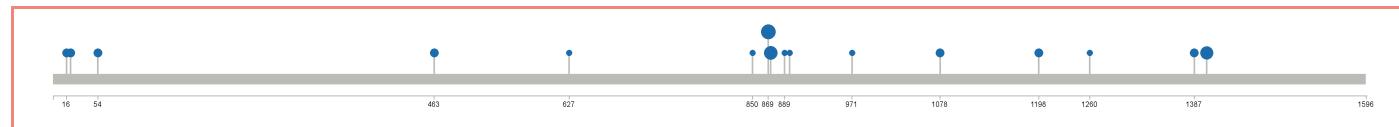
No unreported indel found.

### Substitutions

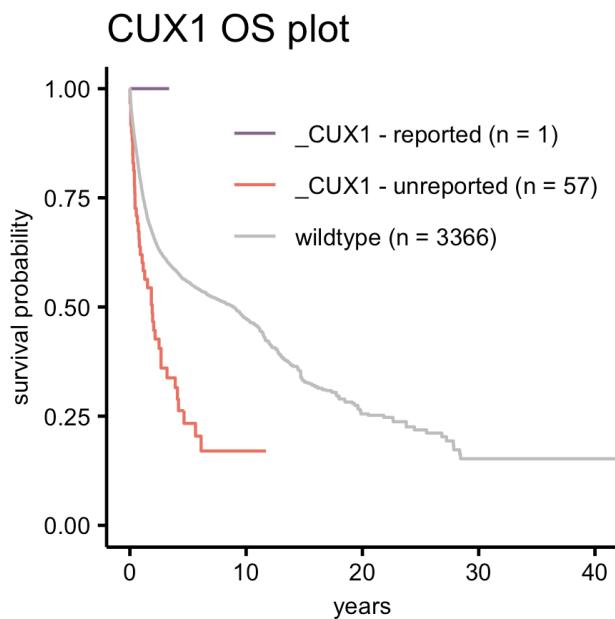
#### reported



#### unreported



### 5.33 CUX1



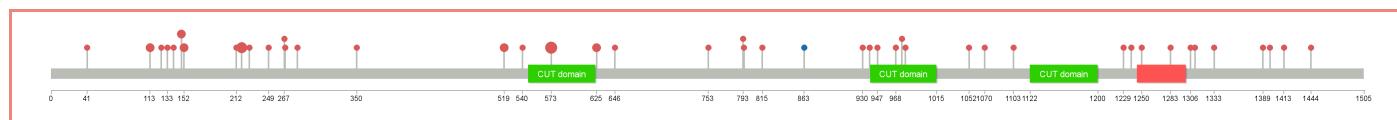
disease	status consequence	× indel	× sub	✓
AML	missense	0	13	0
	nonsense/splicing	0	12	1
	other	1	3	0
MDS	truncating	23	0	0
	inframe	1	0	0
	missense	0	1	0
MPN	nonsense/splicing	1	46	0
	other	0	2	0
	truncating	28	0	0
TOTAL	missense	0	4	0
	nonsense/splicing	0	1	0
	truncating	3	0	0
TOTAL		57	82	1
		0.41%	0.59%	0.01%

#### Indels

##### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

##### unreported

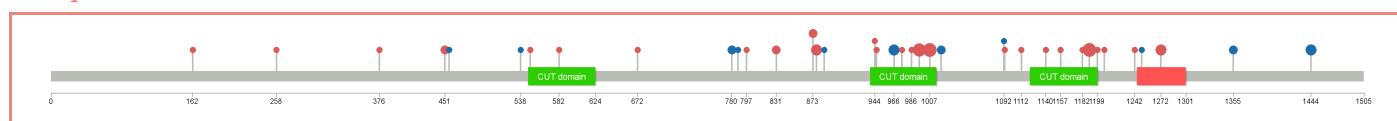


#### Substitutions

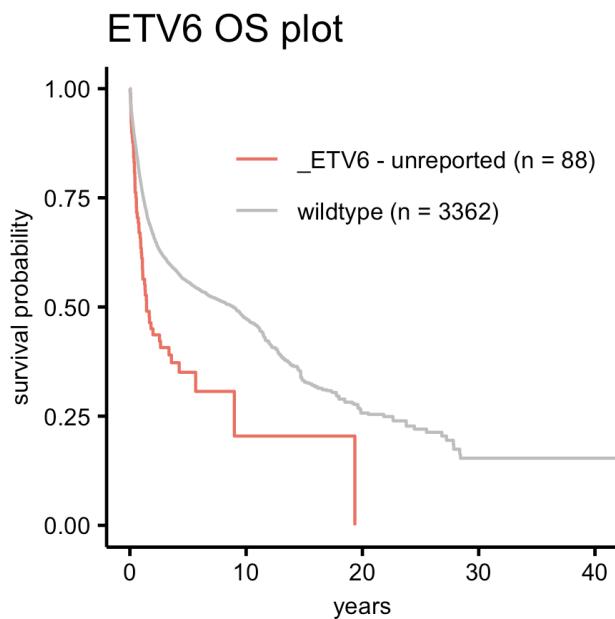
##### reported



##### unreported



## 5.34 ETV6



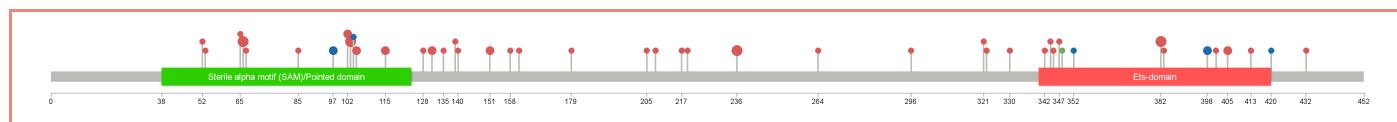
disease	status consequence	× indel	× sub
AML	inframe	5	0
	missense	0	21
	nonsense/splicing	0	13
	other	4	3
	truncating	29	0
MDS	inframe	2	0
	missense	0	19
	nonsense/splicing	1	10
	other	2	2
	truncating	23	0
MPN	truncating	1	0
<b>TOTAL</b>		67	68
		0.50%	0.50%

### Indels

#### reported

This gene was not reported for exons (or no reported mutation was found in the dataset).

#### unreported

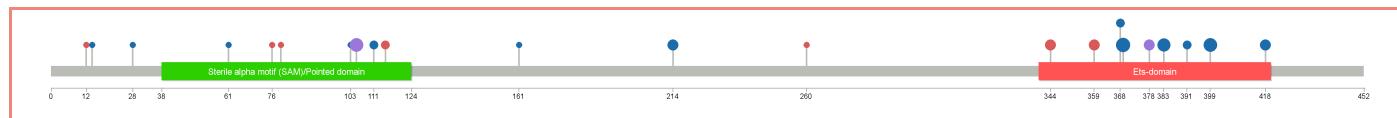


### Substitutions

#### reported

This gene was not reported for hotspots (or no reported mutation was found in the dataset).

#### unreported



## 6 Supplementary material

### 6.1 Unreported substitutions table (with count $\geq 5$ )

Table of the substitutions identified in genes with partial hotspot reporting.

protein variant	count
JAK2_1108	35
FLT3_592	28
STAG2_216	22
FLT3_598	21
FLT3_590	19
FLT3_594	19
DNMT3A_770	19
WT1_381	19
ASXL1_591	19
TET2_1881	18
TET2_1221	17
PTPN11_308	17
TET2_1167	16
MPL_592	16
PHF6_314	16
RUNX1_110	16
NRAS_64	15
SMC1A_711	15
CBL_416	14
GATA2_317	14
TET2_1298	14
TET2_810	13
STAG2_604	13
GATA2_372	13
DNMT3A_326	13
MYC_73	13
CBL_381	13
WT1_464	12
CBL_396	12
WT1_380	12
STAG2_953	12
PTPN11_510	12
FLT3_167	12
DNMT3A_501	12
ASXL1_775	12
STAG2_110	12
WT1_458	12
DNMT3A_581	12
PTPN11_285	11
MYC_75	11
TET2_1894	11
MPL_204	11
SMC1A_807	11
FLT3_842	11
TET2_1288	11
SRSF2_57	11
DNMT3A_893	10
TET2_1282	10
PTPN11_491	10

Continued on next page

protein variant	count
TET2_1193	10
TET2_1262	10
GATA2_330	10
CBL_383	10
STAG2_305	10
TET2_r.4814+5g>a	10
DNMT3A_547	10
CBL_389	9
TP53_r.973-1g>a	9
WT1_434	9
RUNX1_106	9
FLT3_572	9
RUNX1_111	9
RUNX1_165	9
EZH2_693	9
RUNX1_425	9
TET2_1355	9
FLT3_664	8
RB1_786	8
WT1_465	8
FLT3_579	8
TET2_1912	8
DNMT3A_796	8
CTCF_339	8
EZH2_249	8
TET2_1289	8
DNMT3A_732	8
RUNX1_112	8
TET2_1861	8
DNMT3A_r.2005+1g>a	8
BCOR_1217	8
EZH2_r.1029+5g>a	8
RUNX1_52	8
TET2_r.4663+5g>a	8
ASXL1_1304	8
SETBP1_869	8
CBL_423	8
TET2_1233	8
TET2_417	8
TET2_1361	8
ASXL1_588	8
TET2_1030	8
ASXL1_404	8
CBL_149	8
BCOR_1375	8
WT1_350	8
DNMT3A_780	8
TET2_705	8
EP300_2266	8
WT1_430	7
ASXL1_829	7
DNMT3A_330	7
TET2_1819	7
TET2_1211	7

Continued on next page

protein variant	count
TET2_r.4904+1g>a	7
ASXL1_512	7
TET2_1414	7
CBL_398	7
SETD2_629	7
TET2_1358	7
FLT3_663	7
TET2_1370	7
RUNX1_105	7
EZH2_674	7
TET2_1376	7
TET2_1191	7
NF1_629	7
CBL_408	7
CBL_419	7
ASXL1_602	7
EZH2_664	7
PHF6_r.932-2a>g	7
EZH2_731	7
TET2_531	7
RUNX1_207	7
SMC1A_1131	7
TET2_530	7
FLT3_593	7
EZH2_660	7
GATA2_354	6
TET2_1680	6
DNMT3A_310	6
SETBP1_872	6
DNMT3A_699	6
RUNX1_322	6
DNMT3A_707	6
DNMT3A_899	6
TET2_r.4905-1g>a	6
TET2_1687	6
TET2_1273	6
CBL_382	6
DNMT3A_716	6
CBL_343	6
TP53_r.749+1g>a	6
SRSF2_96	6
FLT3_596	6
TET2_1382	6
TET2_910	6
CUX1_1007	6
CBL_418	6
TET2_792	6
RUNX1_267	6
CREBBP_1745	6
ASXL1_491	6
ASXL1_708	6
TET2_r.4269+1g>a	6
GATA2_385	6
DNMT3A_731	6

Continued on next page

protein variant	count
PHF6_225	6
TET2_1848	6
TET2_1182	6
DNMT3A_313	6
TET2_1291	6
TET2_1203	6
U2AF1_35	6
TET2_631	6
BCOR_1547	6
DNMT3A_842	6
CUX1_1190	6
TET2_1387	6
DNMT3A_879	6
DNMT3A_761	6
DNMT3A_320	6
TET2_1828	6
PHF6_287	6
CTCF_371	6
CBL_206	6
DNMT3A_r.1767+1g>a	6
DNMT3A_685	6
TET2_1374	6
PTPN11_265	6
ASXL1_592	6
BCOR_r.4657+1g>a	6
ASXL1_796	6
TET2_1872	6
ASXL1_1068	6
TET2_1274	6
DNMT3A_700	6
TET2_1624	6
SH2B3_566	5
TET2_1384	5
TET2_1377	5
CUX1_995	5
CBL_400	5
TET2_1300	5
EZH2_695	5
TET2_593	5
EZH2_679	5
ASXL1_1226	5
FLT3_576	5
EZH2_146	5
CBL_367	5
GATA2_351	5
RUNX1_r.796+1g>a	5
JAK2_797	5
RUNX1_r.953+2u>c	5
RUNX1_196	5
TET2_769	5
CSF3R_776	5
RUNX1_161	5
DNMT3A_751	5
TET2_429	5

Continued on next page

protein variant	count
TET2_1661	5
TET2_745	5
EZH2_702	5
ASXL1_760	5
TET2_1868	5
EZH2_745	5
ASXL1_692	5
ASXL1_1415	5
ASXL2_635	5
TET2_1197	5
EZH2_656	5
SETBP1_1402	5
DNMT3A_880	5
EZH2_18	5
TET2_1219	5
DNMT3A_296	5
EP300_638	5
TET2_635	5
ASXL2_614	5
STAG2_146	5
RUNX1_272	5
CBL_390	5
CBL_401	5
PTPN11_62	5
TET2_1327	5
TET2_r.4814+1g>u	5
BCOR_1661	5
DNMT3A_777	5
MPL_565	5
FLT3_583	5
DNMT3A_644	5
CTCF_342	5
TET2_1337	5
CBL_399	5
TET2_1255	5
GATA2_390	5
GATA2_322	5
TET2_1631	5
CSF3R_770	5
WT1_369	5
GATA2_307	5
TET2_r.4454+1g>u	5
TET2_1513	5
SH2B3_395	5
EZH2_25	5
DNMT3A_646	5
TET2_866	5

## 6.2 Gene summary table

gene	count	freq	unreported	reported	unreported	sub	unreported	indel	%	unreported
<b>TET2</b>	3140	12.33%	1347	1793		1320		27	42.9%	
<b>DNMT3A</b>	1768	6.94%	634	1134		629		5	35.9%	
<b>JAK2</b>	1700	6.68%	69	1631		57		12	4.1%	
<b>NPM1</b>	1452	5.70%	4	1448		0		4	0.3%	
<b>SRSF2</b>	1294	5.08%	224	1070		23		201	17.3%	
<b>RUNX1</b>	1165	4.58%	317	848		292		25	27.2%	
<b>ASXL1</b>	1037	4.07%	352	685		321		31	33.9%	
<b>NRAS</b>	1032	4.05%	19	1013		19		0	1.8%	
<b>SF3B1</b>	917	3.60%	44	873		38		6	4.8%	
<b>FLT3</b>	862	3.39%	234	628		233		1	27.1%	
<b>TP53</b>	826	3.24%	81	745		74		7	9.8%	
<b>IDH2</b>	719	2.82%	2	717		2		0	0.3%	
<b>STAG2</b>	572	2.25%	496	76		265		231	86.7%	
<b>CEBPA</b>	546	2.14%	145	401		143		2	26.6%	
<b>U2AF1</b>	521	2.05%	23	498		18		5	4.4%	
<b>EZH2</b>	492	1.93%	431	61		314		117	87.6%	
<b>CALR</b>	481	1.89%	3	478		2		1	0.6%	
<b>PTPN11</b>	469	1.84%	98	371		96		2	20.9%	
<b>IDH1</b>	413	1.62%	9	404		6		3	2.2%	
<b>KRAS</b>	399	1.57%	14	385		12		2	3.5%	
<b>CBL</b>	394	1.55%	263	131		232		31	66.8%	
<b>BCOR</b>	357	1.40%	355	2		155		200	99.4%	
<b>WT1</b>	330	1.30%	299	31		152		147	90.6%	
<b>NF1</b>	280	1.10%	234	46		130		104	83.6%	
<b>ZRSR2</b>	280	1.10%	280	0		180		100	100.0%	
<b>PHF6</b>	271	1.06%	221	50		133		88	81.5%	
<b>KIT</b>	215	0.84%	54	161		38		16	25.1%	
<b>GATA2</b>	215	0.84%	142	73		101		41	66.0%	
<b>RAD21</b>	205	0.81%	205	0		103		102	100.0%	
<b>MPL</b>	200	0.79%	62	138		56		6	31.0%	
<b>SETBP1</b>	156	0.61%	39	117		39		0	25.0%	
<b>CUX1</b>	140	0.55%	139	1		82		57	99.3%	
<b>ETV6</b>	135	0.53%	135	0		68		67	100.0%	
<b>KMT2C</b>	110	0.43%	110	0		45		65	100.0%	
<b>SMC3</b>	93	0.37%	93	0		71		22	100.0%	
<b>SMC1A</b>	91	0.36%	71	20		61		10	78.0%	
<b>EP300</b>	87	0.34%	81	6		49		32	93.1%	
<b>KMT2D</b>	82	0.32%	82	0		37		45	100.0%	
<b>MYC</b>	80	0.31%	32	48		27		5	40.0%	
<b>PPM1D</b>	77	0.30%	77	0		29		48	100.0%	
<b>NFE2</b>	71	0.28%	71	0		15		56	100.0%	
<b>PRPF8</b>	63	0.25%	63	0		62		1	100.0%	
<b>CSF3R</b>	63	0.25%	31	32		29		2	49.2%	
<b>ASXL2</b>	58	0.23%	58	0		14		44	100.0%	
<b>FBXW7</b>	56	0.22%	47	9		9		38	83.9%	
<b>CTCF</b>	54	0.21%	52	2		48		4	96.3%	
<b>KDM6A</b>	53	0.21%	52	1		22		30	98.1%	
<b>ATRX</b>	52	0.20%	51	1		28		23	98.1%	
<b>SH2B3</b>	51	0.20%	51	0		24		27	100.0%	
<b>BRAF</b>	48	0.19%	6	42		4		2	12.5%	
<b>GNAS</b>	47	0.18%	19	28		15		4	40.4%	

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gene	count	freq	unreported	reported	unreported sub	unreported	indel	% unreported
<b>NOTCH1</b>	41	0.16%	35	6	19		16	85.4%
<b>BCORL1</b>	40	0.16%	40	0	19		21	100.0%
<b>CREBBP</b>	38	0.15%	38	0	23		15	100.0%
<b>U2AF2</b>	37	0.15%	37	0	17		20	100.0%
<b>ATM</b>	36	0.14%	32	4	27		5	88.9%
<b>RB1</b>	36	0.14%	20	16	18		2	55.6%
<b>RIT1</b>	33	0.13%	33	0	33		0	100.0%
<b>ETNK1</b>	30	0.12%	1	29	1		0	3.3%
<b>KMT2E</b>	27	0.11%	27	0	3		24	100.0%
<b>PCDH15</b>	26	0.10%	26	0	0		26	100.0%
<b>BRCC3</b>	26	0.10%	26	0	22		4	100.0%
<b>KDM5A</b>	23	0.09%	23	0	22		1	100.0%
<b>LAMB4</b>	21	0.08%	21	0	20		1	100.0%
<b>SF1</b>	21	0.08%	21	0	16		5	100.0%
<b>SETD2</b>	20	0.08%	20	0	17		3	100.0%
<b>LUC7L2</b>	19	0.07%	19	0	18		1	100.0%
<b>MED12</b>	19	0.07%	15	4	15		0	78.9%
<b>CHEK2</b>	19	0.07%	15	4	1		14	78.9%
<b>KMT2A</b>	18	0.07%	18	0	11		7	100.0%
<b>GNB1</b>	18	0.07%	18	0	18		0	100.0%
<b>ARID2</b>	18	0.07%	18	0	11		7	100.0%
<b>GATA1</b>	16	0.06%	16	0	4		12	100.0%
<b>SUZ12</b>	16	0.06%	16	0	4		12	100.0%
<b>NCOR2</b>	15	0.06%	15	0	13		2	100.0%
<b>KANSL1</b>	15	0.06%	15	0	9		6	100.0%
<b>JAK3</b>	15	0.06%	7	8	6		1	46.7%
<b>DST</b>	14	0.05%	14	0	0		14	100.0%
<b>CACNA2D3</b>	14	0.05%	14	0	0		14	100.0%
<b>CDKN2A</b>	13	0.05%	13	0	12		1	100.0%
<b>MN1</b>	13	0.05%	13	0	6		7	100.0%
<b>PTPRD</b>	12	0.05%	12	0	10		2	100.0%
<b>PDS5B</b>	11	0.04%	11	0	4		7	100.0%
<b>EED</b>	11	0.04%	11	0	9		2	100.0%
<b>NCL</b>	10	0.04%	10	0	0		10	100.0%
<b>ROBO1</b>	10	0.04%	10	0	7		3	100.0%
<b>KDM2B</b>	10	0.04%	10	0	7		3	100.0%
<b>IRF1</b>	9	0.04%	9	0	5		4	100.0%
<b>NCOR1</b>	9	0.04%	9	0	8		1	100.0%
<b>FANCA</b>	9	0.04%	9	0	4		5	100.0%
<b>FANCM</b>	8	0.03%	8	0	6		2	100.0%
<b>ARID1A</b>	8	0.03%	7	1	3		4	87.5%
<b>DCLRE1C</b>	8	0.03%	8	0	4		4	100.0%
<b>ARID1B</b>	8	0.03%	8	0	4		4	100.0%
<b>MBD1</b>	8	0.03%	8	0	8		0	100.0%
<b>PTPRT</b>	8	0.03%	8	0	8		0	100.0%
<b>NSD1</b>	8	0.03%	8	0	4		4	100.0%
<b>KDR</b>	8	0.03%	8	0	8		0	100.0%
<b>PTEN</b>	7	0.03%	6	1	4		2	85.7%
<b>FANCL</b>	7	0.03%	7	0	1		6	100.0%
<b>GPRC5A</b>	7	0.03%	7	0	0		7	100.0%
<b>CBFB</b>	7	0.03%	7	0	3		4	100.0%
<b>ASPM</b>	7	0.03%	7	0	0		7	100.0%
<b>DNMT3B</b>	7	0.03%	7	0	6		1	100.0%

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gene	count	freq	unreported	reported	unreported sub	unreported	indel	% unreported
<b>PHIP</b>	7	0.03%	7	0	2		5	100.0%
<b>PTPRB</b>	7	0.03%	7	0	0		7	100.0%
<b>PRR14L</b>	7	0.03%	7	0	0		7	100.0%
<b>TP53BP1</b>	7	0.03%	7	0	6		1	100.0%
<b>ASH1L</b>	7	0.03%	7	0	7		0	100.0%
<b>AHNAK</b>	6	0.02%	6	0	0		6	100.0%
<b>PIGA</b>	6	0.02%	6	0	3		3	100.0%
<b>SF3A1</b>	6	0.02%	6	0	6		0	100.0%
<b>PRPF40B</b>	6	0.02%	6	0	6		0	100.0%
<b>STAT5B</b>	6	0.02%	1	5	1		0	16.7%
<b>SRRM2</b>	6	0.02%	6	0	0		6	100.0%
<b>STAG3</b>	5	0.02%	5	0	4		1	100.0%
<b>DOT1L</b>	5	0.02%	5	0	5		0	100.0%
<b>ZBTB33</b>	5	0.02%	5	0	0		5	100.0%
<b>ELF1</b>	5	0.02%	5	0	0		5	100.0%
<b>MRE11A</b>	5	0.02%	5	0	2		3	100.0%
<b>HUWE1</b>	5	0.02%	5	0	0		5	100.0%
<b>SRRM1</b>	4	0.02%	4	0	1		3	100.0%
<b>SYNE2</b>	4	0.02%	4	0	0		4	100.0%
<b>H3F3A</b>	4	0.02%	4	0	4		0	100.0%
<b>SMG1</b>	4	0.02%	4	0	4		0	100.0%
<b>STAT3</b>	4	0.02%	0	4	0		0	0.0%
<b>TG</b>	4	0.02%	4	0	0		4	100.0%
<b>WHSC1</b>	4	0.02%	4	0	4		0	100.0%
<b>PHF8</b>	4	0.02%	4	0	4		0	100.0%
<b>NIPBL</b>	4	0.02%	4	0	4		0	100.0%
<b>NF2</b>	4	0.02%	4	0	4		0	100.0%
<b>FANCD2</b>	4	0.02%	4	0	4		0	100.0%
<b>CMYA5</b>	4	0.02%	4	0	0		4	100.0%
<b>NOTCH2</b>	4	0.02%	4	0	3		1	100.0%
<b>HNRNPK</b>	4	0.02%	4	0	0		4	100.0%
<b>ZFHX4</b>	3	0.01%	3	0	0		3	100.0%
<b>YLPM1</b>	3	0.01%	3	0	3		0	100.0%
<b>CDKN1B</b>	3	0.01%	3	0	0		3	100.0%
<b>ALAS2</b>	3	0.01%	3	0	3		0	100.0%
<b>DICER1</b>	3	0.01%	3	0	0		3	100.0%
<b>ABCG8</b>	3	0.01%	3	0	3		0	100.0%
<b>ASXL3</b>	3	0.01%	3	0	2		1	100.0%
<b>NRD1</b>	3	0.01%	3	0	0		3	100.0%
<b>HIPK2</b>	3	0.01%	3	0	0		3	100.0%
<b>ITPR1</b>	3	0.01%	3	0	0		3	100.0%
<b>FARS2</b>	3	0.01%	3	0	0		3	100.0%
<b>CSF2RB</b>	3	0.01%	3	0	2		1	100.0%
<b>RAD50</b>	3	0.01%	3	0	1		2	100.0%
<b>KIAA1549</b>	3	0.01%	3	0	0		3	100.0%
<b>GRID1</b>	3	0.01%	3	0	3		0	100.0%
<b>SARDH</b>	2	0.01%	2	0	0		2	100.0%
<b>MLH3</b>	2	0.01%	2	0	1		1	100.0%
<b>FANCB</b>	2	0.01%	2	0	0		2	100.0%
<b>FAT4</b>	2	0.01%	2	0	0		2	100.0%
<b>C17orf74</b>	2	0.01%	2	0	2		0	100.0%
<b>CSMD1</b>	2	0.01%	2	0	0		2	100.0%
<b>CTNNA1</b>	2	0.01%	2	0	1		1	100.0%

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gene	count	freq	unreported	reported	unreported sub	unreported	indel	% unreported
<b>GPR116</b>	2	0.01%	2	0	0	2	100.0%	
<b>PTPN14</b>	2	0.01%	2	0	0	2	100.0%	
<b>NUP98</b>	2	0.01%	2	0	0	2	100.0%	
<b>FAM175A</b>	2	0.01%	2	0	1	1	100.0%	
<b>DNAH9</b>	2	0.01%	2	0	0	2	100.0%	
<b>CSF1R</b>	2	0.01%	2	0	0	2	100.0%	
<b>SRCAP</b>	2	0.01%	2	0	1	1	100.0%	
<b>ZEB2</b>	2	0.01%	2	0	0	2	100.0%	
<b>ERG</b>	2	0.01%	2	0	0	2	100.0%	
<b>GPR112</b>	2	0.01%	2	0	0	2	100.0%	
<b>PPFIA2</b>	2	0.01%	2	0	2	0	100.0%	
<b>ABL1</b>	2	0.01%	1	1	1	0	50.0%	
<b>BRCA1</b>	2	0.01%	2	0	2	0	100.0%	
<b>SVEP1</b>	2	0.01%	2	0	0	2	100.0%	
<b>BEND6</b>	2	0.01%	2	0	0	2	100.0%	
<b>FAT2</b>	2	0.01%	2	0	1	1	100.0%	
<b>RRAS</b>	2	0.01%	2	0	2	0	100.0%	
<b>SMC1B</b>	2	0.01%	2	0	2	0	100.0%	
<b>STAG1</b>	2	0.01%	2	0	2	0	100.0%	
<b>CBLB</b>	2	0.01%	2	0	2	0	100.0%	
<b>ME1</b>	2	0.01%	2	0	0	2	100.0%	
<b>TCF4</b>	2	0.01%	2	0	0	2	100.0%	
<b>MAP2K5</b>	2	0.01%	2	0	0	2	100.0%	
<b>CD101</b>	2	0.01%	2	0	0	2	100.0%	
<b>SPTA1</b>	2	0.01%	2	0	0	2	100.0%	
<b>STK17B</b>	2	0.01%	2	0	0	2	100.0%	
<b>RINT1</b>	2	0.01%	2	0	0	2	100.0%	
<b>GRIN2B</b>	2	0.01%	2	0	0	2	100.0%	
<b>DIAPH2</b>	2	0.01%	2	0	2	0	100.0%	
<b>NXF1</b>	2	0.01%	2	0	2	0	100.0%	
<b>ABCC9</b>	2	0.01%	2	0	0	2	100.0%	
<b>CACNA1G</b>	1	0.00%	1	0	0	1	100.0%	
<b>ARHGAP32</b>	1	0.00%	1	0	0	1	100.0%	
<b>SPRED2</b>	1	0.00%	1	0	1	0	100.0%	
<b>RASGRF1</b>	1	0.00%	1	0	1	0	100.0%	
<b>FANCE</b>	1	0.00%	1	0	0	1	100.0%	
<b>SF3A3</b>	1	0.00%	1	0	1	0	100.0%	
<b>MYB</b>	1	0.00%	1	0	0	1	100.0%	
<b>DRC1</b>	1	0.00%	1	0	0	1	100.0%	
<b>SPP1</b>	1	0.00%	1	0	0	1	100.0%	
<b>RP1L1</b>	1	0.00%	1	0	0	1	100.0%	
<b>PRDX2</b>	1	0.00%	1	0	0	1	100.0%	
<b>AHNAK2</b>	1	0.00%	1	0	0	1	100.0%	
<b>CRLF2</b>	1	0.00%	1	0	1	0	100.0%	
<b>MGA</b>	1	0.00%	1	0	1	0	100.0%	
<b>CDCA5</b>	1	0.00%	1	0	0	1	100.0%	
<b>BTNL8</b>	1	0.00%	1	0	0	1	100.0%	
<b>SRPK2</b>	1	0.00%	1	0	0	1	100.0%	
<b>IL31RA</b>	1	0.00%	1	0	0	1	100.0%	
<b>HIF1A</b>	1	0.00%	1	0	0	1	100.0%	
<b>CUL2</b>	1	0.00%	1	0	0	1	100.0%	
<b>GATA3</b>	1	0.00%	1	0	1	0	100.0%	
<b>EGFR</b>	1	0.00%	1	0	0	1	100.0%	

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gene	count	freq	unreported	reported	unreported sub	unreported	indel	% unreported
<b>DDX18</b>	1	0.00%	1	0	0		1	100.0%
<b>OCA2</b>	1	0.00%	1	0	0		1	100.0%
<b>ATXN7L1</b>	1	0.00%	1	0	0		1	100.0%
<b>PHF12</b>	1	0.00%	1	0	0		1	100.0%
<b>NR3C1</b>	1	0.00%	1	0	0		1	100.0%
<b>SLC25A38</b>	1	0.00%	1	0	0		1	100.0%
<b>FANCI</b>	1	0.00%	1	0	1		0	100.0%
<b>HNRNPCL1</b>	1	0.00%	1	0	0		1	100.0%
<b>GRIN2A</b>	1	0.00%	1	0	0		1	100.0%
<b>SPAG9</b>	1	0.00%	1	0	0		1	100.0%
<b>PTPN21</b>	1	0.00%	1	0	0		1	100.0%
<b>FAT1</b>	1	0.00%	1	0	1		0	100.0%
<b>S100B</b>	1	0.00%	1	0	1		0	100.0%
<b>KSR2</b>	1	0.00%	1	0	0		1	100.0%
<b>PTPN2</b>	1	0.00%	1	0	0		1	100.0%
<b>OTOP1</b>	1	0.00%	1	0	0		1	100.0%
<b>KIAA1377</b>	1	0.00%	1	0	0		1	100.0%
<b>RPS6KA6</b>	1	0.00%	1	0	1		0	100.0%
<b>PTPRC</b>	1	0.00%	1	0	0		1	100.0%
<b>ZNF717</b>	1	0.00%	1	0	0		1	100.0%
<b>IL12B</b>	1	0.00%	1	0	0		1	100.0%
<b>EPHA2</b>	1	0.00%	1	0	0		1	100.0%
<b>NAAA</b>	1	0.00%	1	0	0		1	100.0%
<b>PIK3CA</b>	1	0.00%	0	1	0		0	0.0%
<b>WAPAL</b>	1	0.00%	1	0	1		0	100.0%
<b>IKZF1</b>	1	0.00%	1	0	1		0	100.0%
<b>NFKB2</b>	1	0.00%	1	0	0		1	100.0%
<b>SLC12A1</b>	1	0.00%	1	0	0		1	100.0%
<b>PTCH1</b>	1	0.00%	1	0	1		0	100.0%
<b>ESCO2</b>	1	0.00%	1	0	0		1	100.0%
<b>MSH6</b>	1	0.00%	1	0	0		1	100.0%
<b>CNTN5</b>	1	0.00%	1	0	0		1	100.0%
<b>DDX41</b>	1	0.00%	1	0	0		1	100.0%
<b>KCNMA1</b>	1	0.00%	1	0	0		1	100.0%
<b>IFNGR1</b>	1	0.00%	1	0	0		1	100.0%
<b>HLA-C</b>	1	0.00%	1	0	1		0	100.0%
<b>DTNA</b>	1	0.00%	1	0	0		1	100.0%
<b>TRPM4</b>	1	0.00%	1	0	0		1	100.0%
<b>FANCG</b>	1	0.00%	1	0	1		0	100.0%
<b>CDH1</b>	1	0.00%	1	0	0		1	100.0%
<b>FAM47C</b>	1	0.00%	1	0	0		1	100.0%
<b>CSNK1A1</b>	1	0.00%	1	0	1		0	100.0%
<b>RAD51</b>	1	0.00%	1	0	0		1	100.0%
<b>COLCA1</b>	1	0.00%	1	0	0		1	100.0%
<b>CTNNA2</b>	1	0.00%	1	0	0		1	100.0%
<b>SH3RF3</b>	1	0.00%	1	0	0		1	100.0%
<b>CSMD3</b>	1	0.00%	1	0	0		1	100.0%
<b>IL6ST</b>	1	0.00%	1	0	0		1	100.0%
<b>ALK</b>	1	0.00%	1	0	1		0	100.0%
<b>PTPRF</b>	1	0.00%	1	0	0		1	100.0%