Clinical Heme Panel Optimisation - Annex

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1 Protein variant summary table (with count ≥ 10)

protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
JAK2_617	1622	129	1385	108	1622	0	0	0	0	1622	0	0
NPM1_288	1396	1340	0	56	0	1396	0	0	0	1396	0	0
SRSF2_95	1203	574	36	593	1070	0	0	133	0	1070	0	133
DNMT3A_882	739	617	31	91	739	0	0	0	0	739	0	0
IDH2_140	573	445	16	112	573	0	0	0	0	573	0	0
NRAS_12	550	423	8	119	550	0	0	0	0	550	0	0
SF3B1_700	457	61	13	383	457	0	0	0	0	457	0	0
IDH1_132	404	337	7	60	404	0	0	0	0	404	0	0
FLT3_835	290	280	0	10	289	0	0	1	0	290	0	0
U2AF1_157	262	82	24	156	262	0	0	0	0	262	0	0
CALR_367	256	27	225	4	0	256	0	0	0	256	0	0
NRAS_61	225	208	0	17	225	0	0	0	0	225	0	0
NRAS_13	220	185	0	35	220	0	0	0	0	220	0	0
U2AF1_34	212	120	6	86	212	0	0	0	0	212	0	0
ASXL1_635	211	90	20	101	0	208	3	0	0	208	3	0
CALR_385	198	1	197	0	0	196	2	0	0	196	2	0
KRAS_12	183	130	4	49	183	0	0	0	0	183	0	0
SF3B1_666	156	39	20	97	156	0	0	0	0	156	0	0
IDH2_172	144	133	0	11	144	0	0	0	0	144	0	0
KIT_816	126	107	3	16	126	0	0	0	0	126	0	0
MPL_515	102	11	78	13	89	0	13	0	0	102	0	0
SF3B1_662	90	8	0	82	90	0	0	0	0	90	0	0
RUNX1_166	72	51	0	21	45	3	24	0	0	72	0	0
SF3B1_625	69	12	0	57	69	0	0	0	0	69	0	0
KRAS_13	69	59	0	10	69	0	0	0	0	69	0	0
ASXL1_693	67	27	6	34	0	2	65	0	0	67	0	0
PTPN11_72	67	57	0	10	67	0	0	0	0	67	0	0
PTPN11_76	66	60	1	5	66	0	0	0	0	66	0	0
RUNX1_201	59	42	0	17	35	1	23	0	0	59	0	0
SETBP1_870	57	30	0	27	57	0	0	0	0	57	0	0
PTPN11_61	54	48	0	6	54	0	0	0	0	54	0	0
SETBP1_868	53	25	0	28	53	0	0	0	0	53	0	0
TP53_273	53	32	1	20	52	1	0	0	0	53	0	0
FLT3_676	52	49	0	3	52	0	0	0	0	52	0	0
RUNX1_162	49	36	2	11	49	0	0	0	0	49	0	0
RUNX1_198	45	42	0	3	45	0	0	0	0	45	0	0
MYC_74	45	42	0	3	45	0	0	0	0	45	0	0
SF3B1_622	44	0	2	42	44	0	0	0	0	44	0	0
CEBPA_313	44	43	0	1	3	0	0	35	6	41	3	0

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protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
TP53_248	44	28	0	16	44	0	0	0	0	44	0	0
ASXL1_591	44	14	9	21	0	25	19	0	0	25	19	0
KRAS_146	43	24	3	16	43	0	0	0	0	43	0	0
PRPF8_1598	42	29	0	13	42	0	0	0	0	0	42	0
NPM1_287	42	35	0	7	0	42	0	0	0	42	0	0
CEBPA_24	42	34	0	8	0	42	0	0	0	42	0	0
TET2_1261	41	16	4	21	41	0	0	0	0	41	0	0
FLT3_598	41	41	0	0	21	0	2	17	1	20	21	0
RUNX1_346	39	20	0	19	0	39	0	0	0	39	0	0
ASXL1_646	39	5	0	34	0	39	0	0	0	39	0	0
CBL_404	38	12	1	25	38	0	0	0	0	38	0	0
PTPN11_503	37	33	0	4	37	0	0	0	0	37	0	0
RUNX1_204	37	21	1	15	17	0	20	0	0	37	0	0
PTPN11_502	36	32	0	4	36	0	0	0	0	36	0	0
TET2_275	36	13	6	17	0	36	0	0	0	36	0	0
TET2_1216	36	16	5	15	1	0	35	0	0	36	0	0
TET2_1271	36	10	10	16	12	21	3	0	0	36	0	0
JAK2_1108	35	5	14	16	35	0	0	0	0	0	35	0
TP53_175	35	19	2	14	35	0	0	0	0	35	0	0
PTPN11_60	35	33	1	1	35	0	0	0	0	35	0	0
EZH2_690	34	16	3	15	34	0	0	0	0	34	0	0
RUNX1_107	33	17	0	16	30	1	0	2	0	33	0	0
TET2_764	33	13	0	20	0	32	1	0	0	32	1	0
WT1_381	33	32	0	1	1	14	18	0	0	0	19	14
FLT3_592	32	30	0	2	28	0	3	1	0	4	28	0
TET2_1359	32	10	5	17	31	1	0	0	0	32	0	0
TET2_1380	31	10	2	19	31	0	0	0	0	31	0	0
FLT3_836	31	28	0	3	0	0	0	31	0	31	0	0
WT1_462	31	30	0	1	31	0	0	0	0	31	0	0
PHF6_274	31	21	0	10	18	0	13	0	0	31	0	0
TET2_1873	30	11	6	13	30	0	0	0	0	30	0	0
TP53_220	30	19	1	10	28	0	2	0	0	30	0	0
ETNK1_244	29	8	0	21	29	0	0	0	0	29	0	0
WT1_380	29	27	0	2	12	16	1	0	0	0	12	17
FBXW7_15	29	24	0	5	0	0	0	29	0	0	0	29
DNMT3A_543	29	20	4	5	29	0	0	0	0	29	0	0
TET2_550	29	11	3	15	0	0	29	0	0	29	0	0
FLT3_491	29	28	0	1	29	0	0	0	0	29	0	0
FLT3_590	28	28	0	0	19	0	0	9	0	9	19	0
KRAS_117	28	16	0	12	28	0	0	0	0	28	0	0
TET2_1516	28	16	2	10	0	0	28	0	0	28	0	0
GNAS_844	28	10	5	13	28	0	0	0	0	28	0	0
CSF3R_618	27	16	0	11	27	0	0	0	0	27	0	0
TET2_916	27	11	0	16	0	0	27	0	0	27	0	0
MPL_591	27	8	3	16	26	0	1	0	0	27	0	0
TP53_152	26	10	0	16	5	21	0	0	0	26	0	0
RUNX1_141	26	13	1	12	12	1	11	2	0	26	0	0
FLT3_597	25	24	0	1	3	0	0	20	2	22	3	0
TP53_238	25	14	0	11	24	0	1	0	0	25	0	0
CBL_371	25	13	2	10	25	0	0	0	0	25	0	0
FLT3_839	25	25	0	0	25	0	0	0	0	25	0	0
DNMT3A_736	24	16	2	6	24	0	0	0	0	24	0	0
U2AF1_156	24	14	0	10	24	0	0	0	0	24	0	0

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protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
FLT3_594	24	24	0	0	19	0	0	5	0	5	19	0
ASXL1_775	23	6	2	15	0	11	12	0	0	11	12	0
KIT_822	23	23	0	0	23	0	0	0	0	23	0	0
KRAS_61	23	18	0	5	22	0	1	0	0	23	0	0
CBL_420	23	11	1	11	23	0	0	0	0	23	0	0
TET2_1404	23	9	1	13	0	0	23	0	0	23	0	0
SRSF2_94	22	11	2	9	0	0	0	22	0	0	0	22
TET2_1440	22	8	1	13	0	22	0	0	0	22	0	0
CBL_380	22	10	3	9	22	0	0	0	0	22	0	0
RUNX1_98	22	12	0	10	2	20	0	0	0	20	2	0
STAG2_216	22	12	0	10	0	0	22	0	0	0	22	0
TET2_1221	21	5	4	12	16	4	1	0	0	4	17	0
DNMT3A_770	20	12	3	5	15	1	4	0	0	1	19	0
DNMT3A_732	20	7	4	9	8	3	0	9	0	12	8	0
TET2_615	20	8	0	12	0	20	0	0	0	20	0	0
FLT3_680	20	19	0	1	20	0	0	0	0	20	0	0
TET2_1452	20	6	2	12	0	0	20	0	0	20	0	0
TET2_1465	20	8	2	10	0	0	20	0	0	20	0	0
NFE2_261	19	2	13	4	4	15	0	0	0	0	4	15
DNMT3A_904	19	13	2	4	19	0	0	0	0	19	0	0
STAG2_259	19	10	0	9	0	0	19	0	0	19	0	0
ASXL1_417	19	9	1	9	0	0	19	0	0	19	0	0
DNMT3A_714	19	15	1	3	18	1	0	0	0	19	0	0
TET2_1298	18	7	1	10	14	4	0	0	0	4	14	0
U2AF2_191	18	1	1	16	0	0	0	18	0	0	0	18
TET2_1881	18	8	1	9	18	0	0	0	0	0	18	0
GATA2_321	18	16	0	2	18	0	0	0	0	18	0	0
PTPN11_73	18	16	0	2	18	0	0	0	0	18	0	0
RUNX1_165	18	14	0	4	9	7	0	2	0	9	9	0
CEBPA_312	18	17	0	1	3	0	1	14	0	14	4	0
TET2_544	18	7	2	9	0	0	18	0	0	18	0	0
SRSF2_97	18	15	0	3	0	18	0	0	0	0	0	18
PTPN11_308	17	12	0	5	17	0	0	0	0	0	17	0
GATA2_362	17	16	0	1	17	0	0	0	0	17	0	0
GATA2_390	17	5	3	9	5	0	0	12	0	0	5	12
FLT3_596	17	17	0	0	6	0	3	6	2	11	6	0
SRSF2_96	17	9	0	8	6	0	0	11	0	0	6	11
TET2_1255	17	6	1	10	0	12	5	0	0	12	5	0
ASXL1_630	17	1	1	15	0	17	0	0	0	17	0	0
ZRSR2_r.851+1g>a	17	3	1	13	0	0	12	0	5	0	17	0
KMT2C_1689	17	1	11	5	0	17	0	0	0	0	0	17
CBL_384	17	7	1	9	17	0	0	0	0	17	0	0
TET2_1167	16	7	0	9	16	0	0	0	0	0	16	0
MPL_592	16	6	3	7	9	0	7	0	0	0	16	0
DNMT3A_635	16	9	1	6	15	1	0	0	0	16	0	0
RUNX1_110	16	16	0	0	16	0	0	0	0	0	16	0
TET2_1884	16	6	6	4	16	0	0	0	0	16	0	0
TP53_245	16	8	2	6	16	0	0	0	0	16	0	0
CEBPA_309	16	14	0	2	0	0	1	14	1	15	1	0
TET2_1214	16	4	4	8	16	0	0	0	0	16	0	0
PHF6_314	16	6	1	9	16	0	0	0	0	0	16	0
STAG2_1012	15	6	1	8	0	0	15	0	0	15	0	0
NRAS_64	15	3	0	12	15	0	0	0	0	0	15	0

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protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
PTPN11_71	15	14	0	1	15	0	0	0	0	15	0	0
DNMT3A_860	15	8	3	4	11	1	3	0	0	15	0	0
STAG2_1045	15	9	0	6	0	0	15	0	0	15	0	0
NRAS_60	15	5	1	9	15	0	0	0	0	15	0	0
GNB1_57	15	4	5	6	15	0	0	0	0	0	15	0
CEBPA_23	15	10	0	5	0	15	0	0	0	15	0	0
TP53_272	15	7	1	7	15	0	0	0	0	15	0	0
TET2_1245	15	6	0	9	0	15	0	0	0	15	0	0
CEBPA_83	15	14	0	1	0	15	0	0	0	15	0	0
BCOR_839	15	12	0	3	0	14	1	0	0	0	1	14
RUNX1_320	15	8	0	7	0	0	15	0	0	15	0	0
SMC1A_586	15	14	0	1	15	0	0	0	0	15	0	0
FLT3_841	15	14	0	1	15	0	0	0	0	15	0	0
SMC1A_711	15	8	2	5	15	0	0	0	0	0	15	0
WT1_371	14	12	0	2	0	14	0	0	0	0	0	14
DNMT3A_771	14	10	0	4	8	1	5	0	0	14	0	0
TET2_810	14	6	1	7	0	1	13	0	0	1	13	0
PTPN11_69	14	11	0	3	14	0	0	0	0	14	0	0
GATA2_318	14	14	0	0	14	0	0	0	0	14	0	0
STAG2_1033	14	8	1	5	0	0	14	0	0	14	0	0
GATA2_317	14	13	0	1	14	0	0	0	0	0	14	0
MYC_73	14	14	0	0	13	0	0	1	0	0	13	1
WT1_458	14	13	0	1	2	1	10	1	0	0	12	2
RB1_137	14	4	6	4	14	0	0	0	0	14	0	0
FLT3_600	14	13	0	1	0	0	0	12	2	14	0	0
TP53_173	14	8	1	5	13	1	0	0	0	14	0	0
GATA2_359	14	11	1	2	14	0	0	0	0	14	0	0
CBL_416	14	3	2	9	14	0	0	0	0	0	14	0
DNMT3A_749	14	7	4	3	14	0	0	0	0	14	0	0
TP53_234	13	8	0	5	13	0	0	0	0	13	0	0
RUNX1_170	13	8	0	5	3	10	0	0	0	9	3	1
CEBPA_300	13	12	0	1	11	1	0	1	0	2	11	0
STAG2_604	13	10	0	3	0	0	13	0	0	0	13	0
DNMT3A_326	13	5	4	4	13	0	0	0	0	0	13	0
STAG2_636	13	6	0	7	0	11	2	0	0	0	2	11
NF1_1276	13	5	0	8	11	0	2	0	0	13	0	0
FLT3_593	13	13	0	0	7	0	0	0	6	6	7	0
DNMT3A_735	13	9	0	4	13	0	0	0	0	13	0	0
WT1_464	13	13	0	0	12	1	0	0	0	0	12	1
DNMT3A_581	13	12	0	1	11	1	1	0	0	1	12	0
MYC_75	13	13	0	0	11	0	0	2	0	0	11	2
TP53_242	13	5	2	6	12	1	0	0	0	13	0	0
TET2_642	13	3	2	8	0	10	3	0	0	10	3	0
TP53_237	13	9	0	4	13	0	0	0	0	13	0	0
TET2_1962	13	8	3	2	13	0	0	0	0	13	0	0
CBL_381	13	6	0	7	13	0	0	0	0	0	13	0
TET2_1274	13	1	1	11	1	7	5	0	0	7	6	0
GATA2_372	13	11	0	2	13	0	0	0	0	0	13	0
PTPN11_510	12	10	0	2	12	0	0	0	0	0	12	0
BRAF_600	12	10	0	2	12	0	0	0	0	12	0	0
RAD21_461	12	7	0	5	0	0	0	12	0	0	0	12
FLT3_599	12	11	1	0	4	0	1	4	3	8	4	0
FLT3_167	12	4	1	7	12	0	0	0	0	0	12	0

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protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
STAG2_110	12	8	0	4	0	0	12	0	0	0	12	0
DNMT3A_501	12	6	5	1	12	0	0	0	0	0	12	0
TET2_218	12	3	2	7	0	12	0	0	0	12	0	0
ASXL1_808	12	1	4	7	0	12	0	0	0	12	0	0
STAG2_953	12	7	0	5	0	0	12	0	0	0	12	0
RUNX1_338	12	9	0	3	1	11	0	0	0	11	1	0
TET2_1282	12	6	2	4	10	1	0	0	1	2	10	0
CBL_396	12	8	0	4	12	0	0	0	0	0	12	0
TET2_1904	12	7	0	5	12	0	0	0	0	12	0	0
TP53_193	12	8	0	4	12	0	0	0	0	12	0	0
TP53_179	12	8	0	4	12	0	0	0	0	12	0	0
FLT3_451	11	11	0	0	11	0	0	0	0	11	0	0
FLT3_842	11	10	0	1	11	0	0	0	0	0	11	0
RUNX1_105	11	7	0	4	7	4	0	0	0	4	7	0
ZRSR2_295	11	1	1	9	0	0	11	0	0	0	11	0
TET2_1894	11	3	1	7	11	0	0	0	0	0	11	0
RUNX1_425	11	6	0	5	9	2	0	0	0	2	9	0
GATA2_320	11	10	0	1	10	0	0	1	0	10	0	1
DNMT3A_729	11	11	0	0	11	0	0	0	0	11	0	0
TP53_241	11	9	0	2	11	0	0	0	0	11	0	0
TP53_195	11	6	0	5	11	0	0	0	0	11	0	0
WT1_434	11	11	0	0	9	0	0	1	1	0	9	2
TP53_216	11	7	0	4	11	0	0	0	0	11	0	0
SMC1A_807	11	10	0	1	11	0	0	0	0	0	11	0
TET2_1288	11	3	0	8	11	0	0	0	0	0	11	0
KRAS_60	11	3	0	8	11	0	0	0	0	11	0	0
SRSF2_103	11	0	0	11	0	11	0	0	0	0	0	11
PTPN11_285	11	9	0	2	11	0	0	0	0	0	11	0
SRSF2_57	11	6	0	5	11	0	0	0	0	0	11	0
CEBPA_195	11	4	0	7	0	0	0	11	0	11	0	0
MPL_204	11	1	7	3	11	0	0	0	0	0	11	0
BRAF_594	11	7	0	4	11	0	0	0	0	11	0	0
STAG2_614	11	5	0	6	0	0	11	0	0	11	0	0
RUNX1_123	11	8	0	3	0	11	0	0	0	11	0	0
GATA2_330	10	10	0	0	10	0	0	0	0	0	10	0
EP300_2268	10	0	0	10	0	0	0	10	0	0	0	10
PTPN11_491	10	9	0	1	10	0	0	0	0	0	10	0
DNMT3A_547	10	8	0	2	10	0	0	0	0	0	10	0
TET2_r.4814+5g>a	10	4	3	3	0	0	3	0	7	0	10	0
EZH2_288	10	3	2	5	7	0	3	0	0	10	0	0
STAG2_305	10	5	0	5	1	0	9	0	0	0	10	0
TET2_1262	10	4	0	6	10	0	0	0	0	0	10	0
ASXL1_796	10	2	3	5	0	4	6	0	0	4	6	0
TET2_1333	10	3	2	5	1	9	0	0	0	9	1	0
TET2_1322	10	2	0	8	9	1	0	0	0	10	0	0
RUNX1_207	10	7	0	3	7	2	0	1	0	3	7	0
EZH2_730	10	5	0	5	1	9	0	0	0	0	1	9
NPM1_290	10	10	0	0	0	10	0	0	0	10	0	0
TP53_205	10	6	1	3	7	0	3	0	0	10	0	0
TP53_275	10	7	0	3	10	0	0	0	0	10	0	0
PPM1D_484	10	0	10	0	0	7	3	0	0	0	3	7
CBL_383	10	2	0	8	10	0	0	0	0	0	10	0
TET2_413	10	4	1	5	0	10	0	0	0	10	0	0

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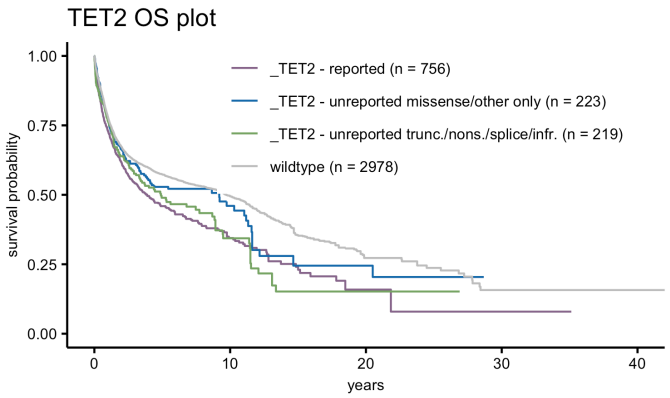
protein variant	count	AML	MPN	MDS	miss.	trunc.	nons./splice	infr.	other	✓	× sub	× indel
CEBPA_321	10	7	0	3	9	0	0	0	1	1	9	0
WT1_379	10	9	0	1	0	10	0	0	0	0	0	10
CEBPA_68	10	8	0	2	0	10	0	0	0	10	0	0
ASXL1_687	10	4	0	6	0	8	2	0	0	8	2	0
TET2_1193	10	5	5	0	10	0	0	0	0	0	10	0
ASXL1_1213	10	0	5	5	0	10	0	0	0	10	0	0
ASXL2_698	10	4	0	6	0	10	0	0	0	0	0	10
DNMT3A_733	10	7	0	3	1	9	0	0	0	9	1	0
CEBPA_311	10	10	0	0	2	1	1	3	3	7	3	0
TET2_1414	10	1	5	4	5	3	2	0	0	3	7	0
DNMT3A_893	10	10	0	0	10	0	0	0	0	0	10	0

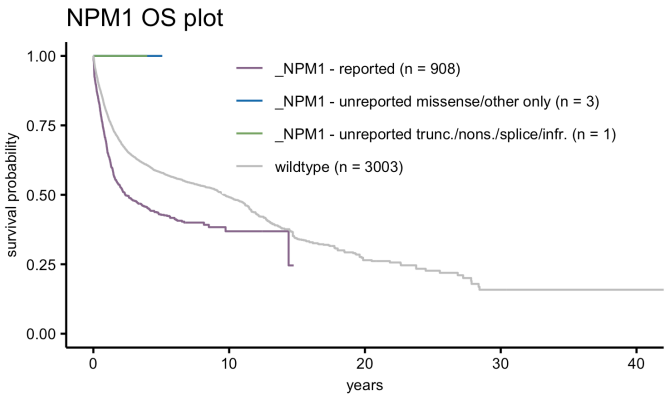
2 Detailed OS plots by gene

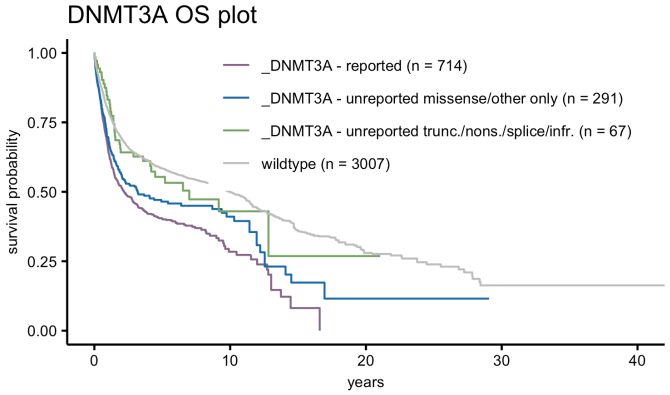
OS (overall survival) plot for the 33 most recurrent mutated genes between:

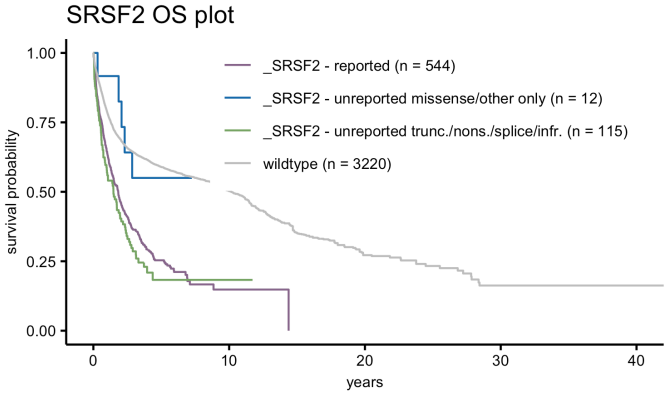
- **wildtype**: patients without any mutation in the studied gene
- **gene name - reported**: patients with at least one reported mutation in the studied gene
- **gene name - unreported missense/other only**: patients with mutations in the studied gene but not a single reported mutation **and** all the mutation consequences in this gene are missense/other
- **gene name - unreported trunc./nons./splice/infr.**: patients with mutations in the studied gene but not a single reported mutation **and** at least one of the mutation consequences in this gene is trunc./nons./splice/infr.

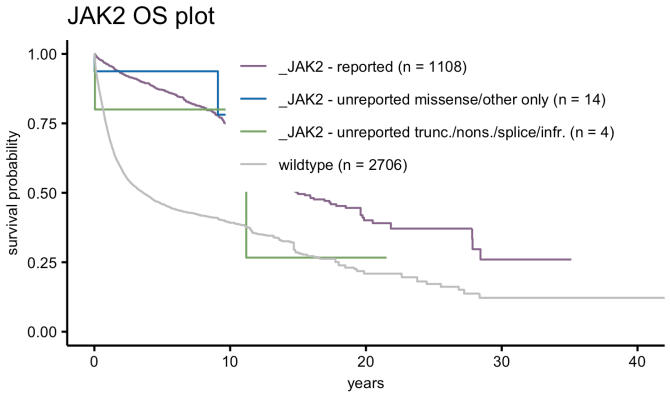
So the old orange curve is split in two between **gene name - unreported missense/other only** (blue curve) and **gene name - unreported trunc./nons./splice/infr.** (green curve).

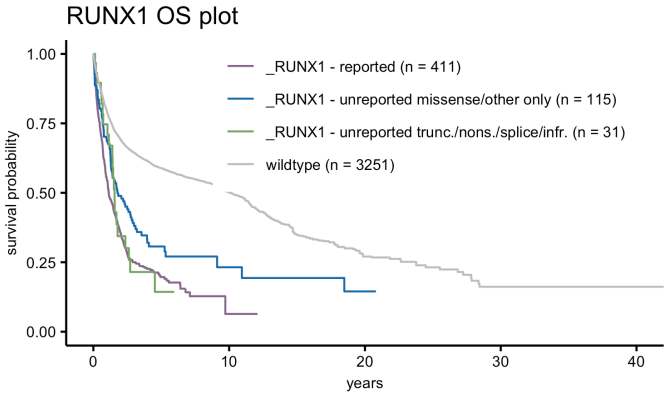




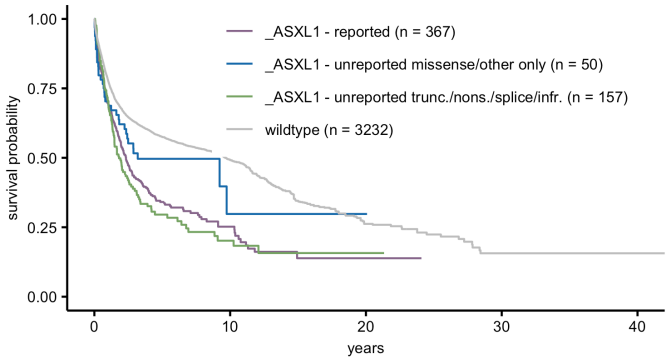




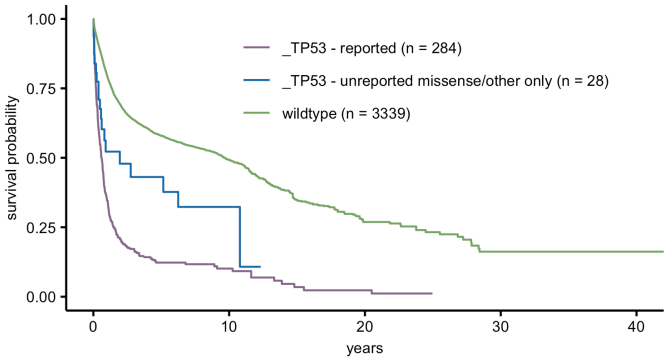




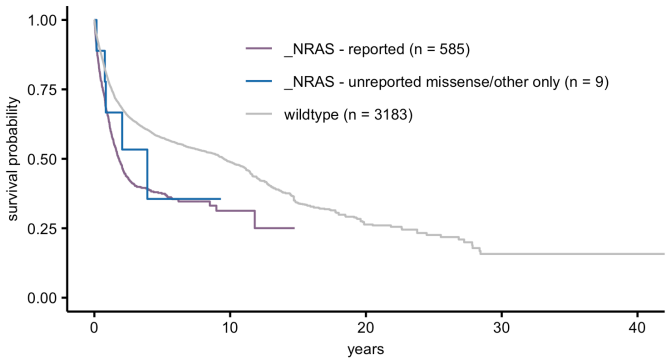
ASXL1 OS plot



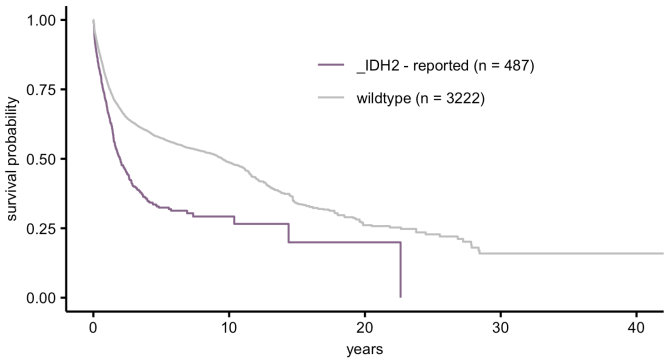
TP53 OS plot



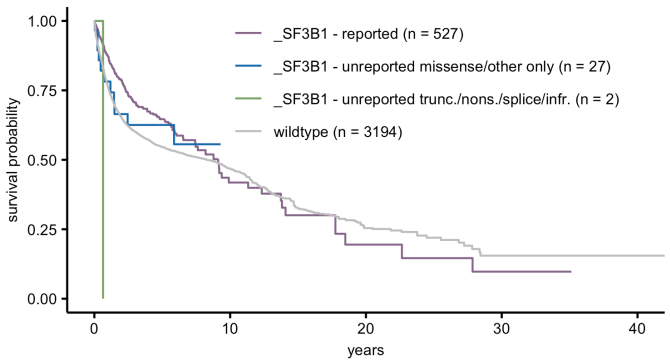
NRAS OS plot



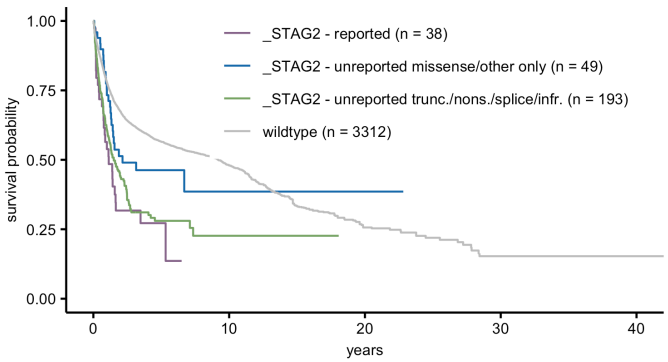
IDH2 OS plot



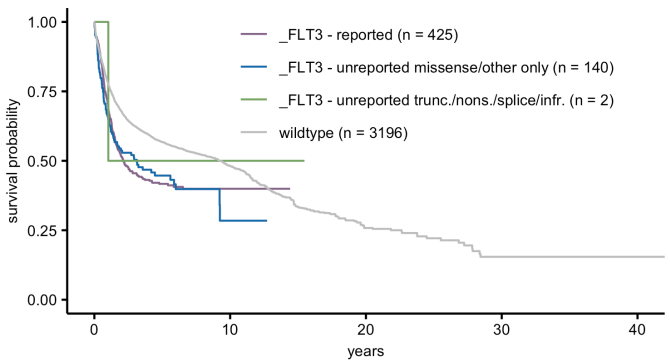
SF3B1 OS plot



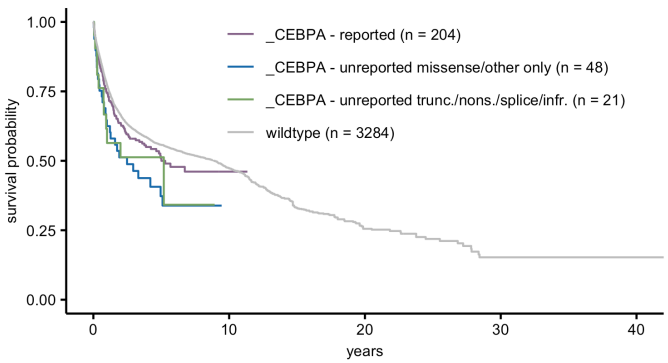
STAG2 OS plot



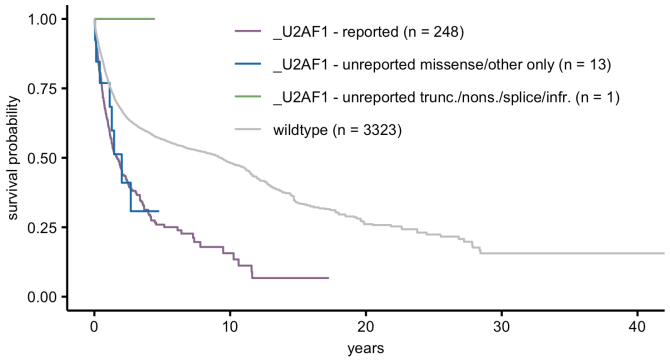
FLT3 OS plot



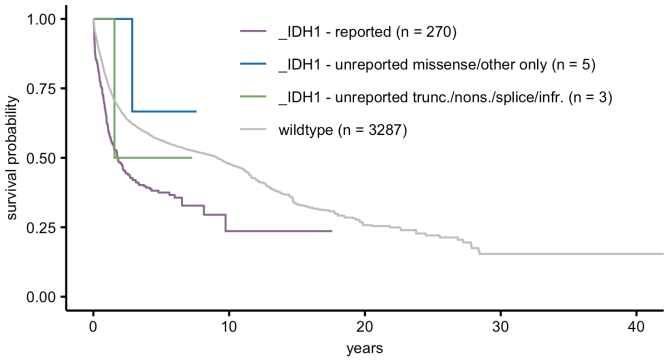
CEBPA OS plot



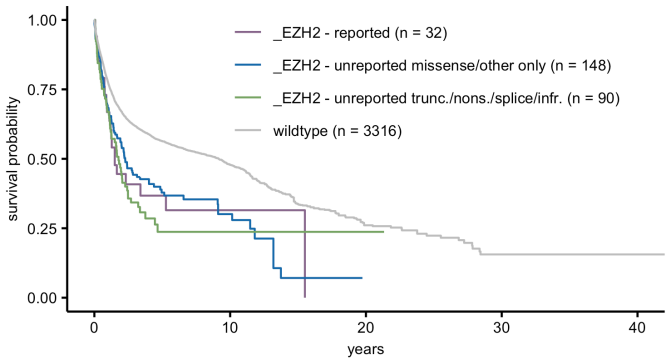
U2AF1 OS plot



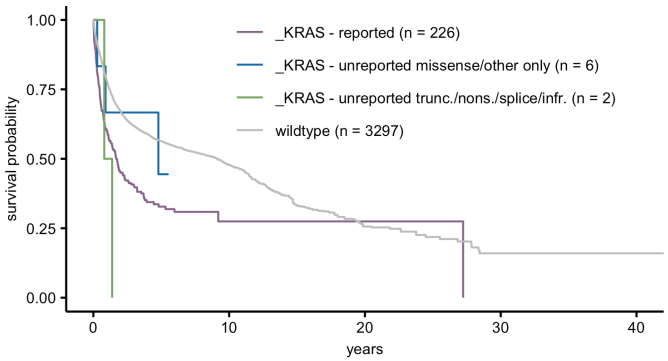
IDH1 OS plot



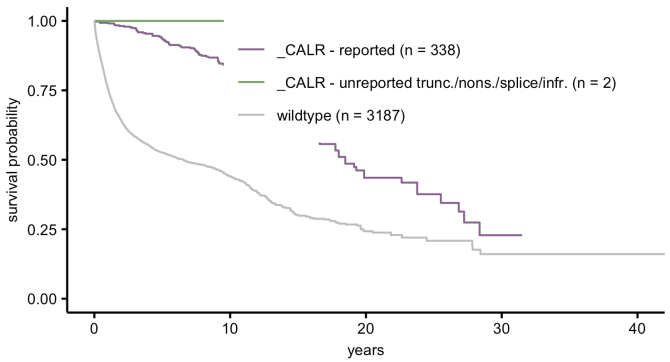
EZH2 OS plot



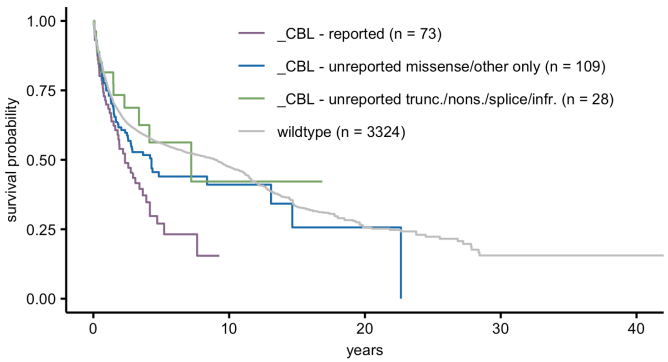
KRAS OS plot



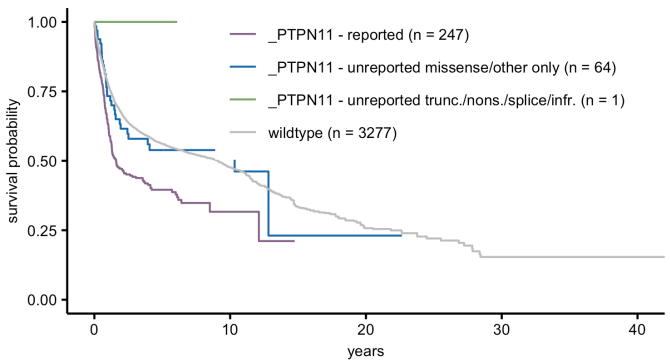
CALR OS plot



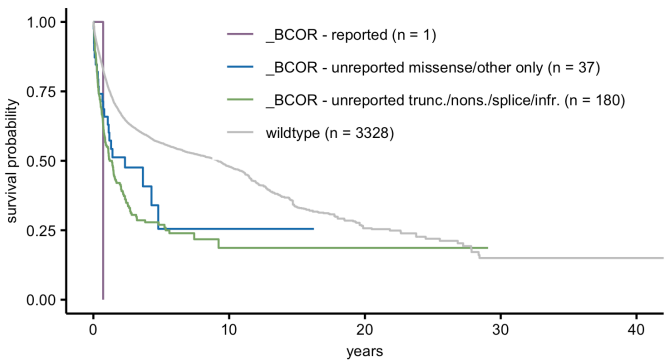
CBL OS plot



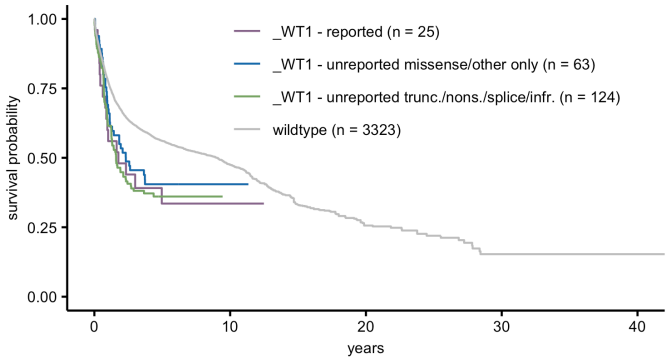
PTPN11 OS plot



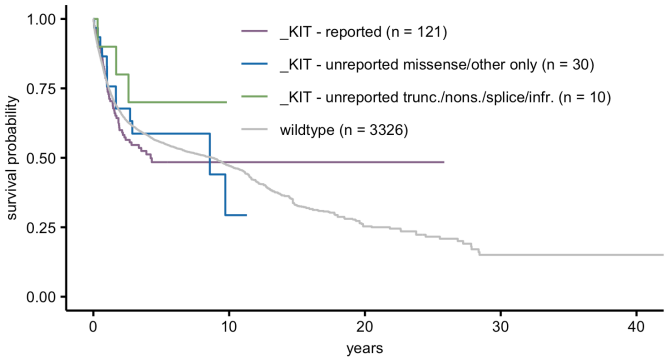
BCOR OS plot



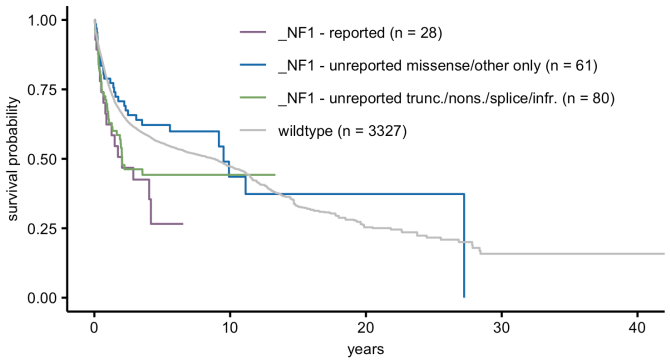
WT1 OS plot



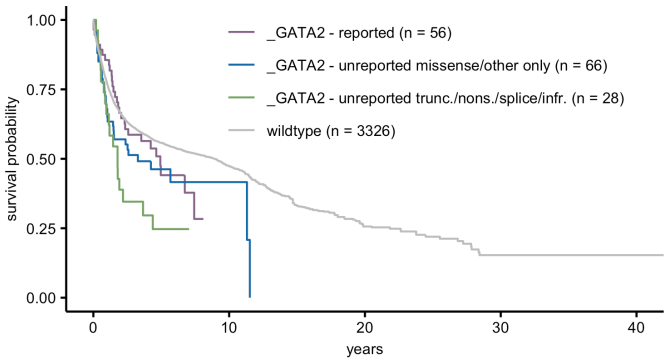
KIT OS plot



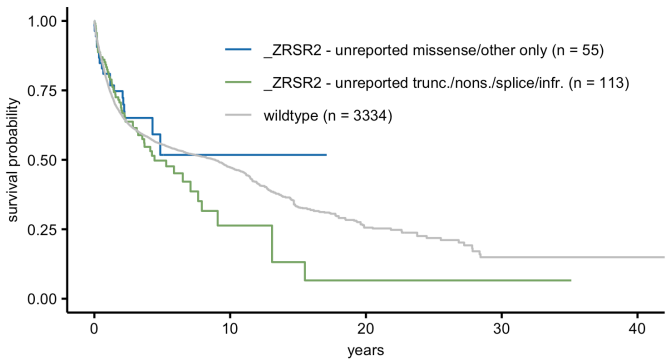
NF1 OS plot



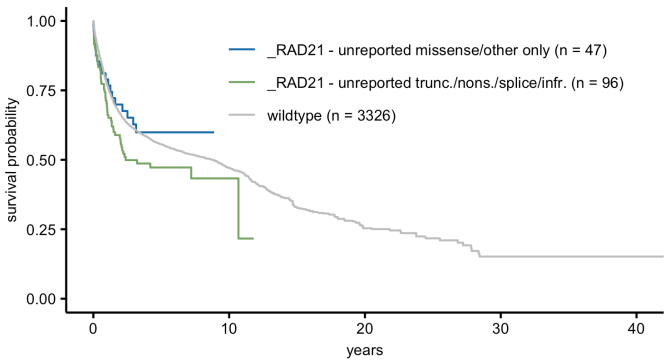
GATA2 OS plot



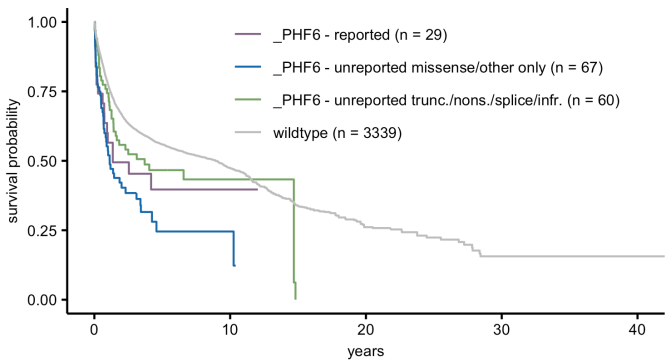
ZRSR2 OS plot



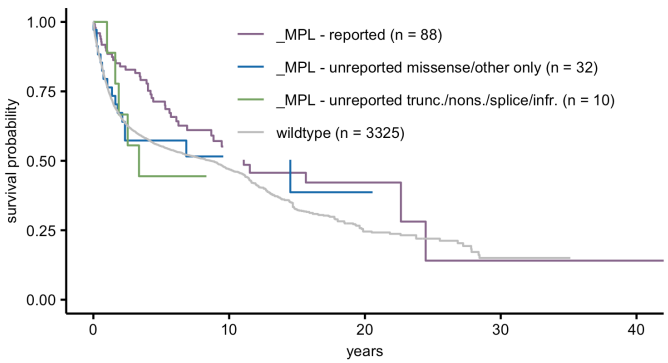
RAD21 OS plot

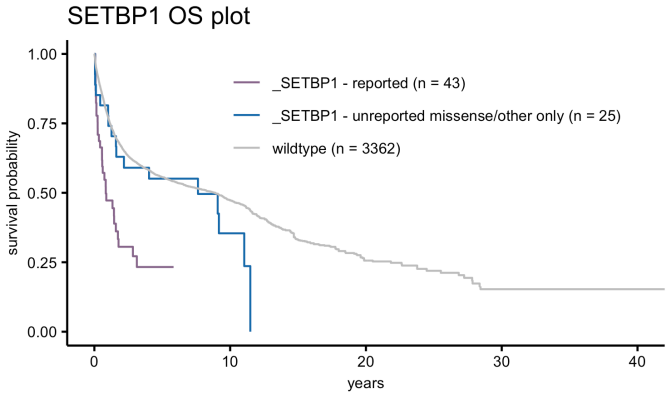


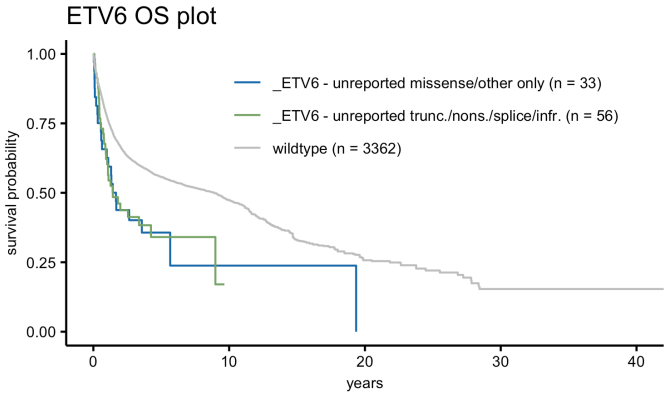
PHF6 OS plot

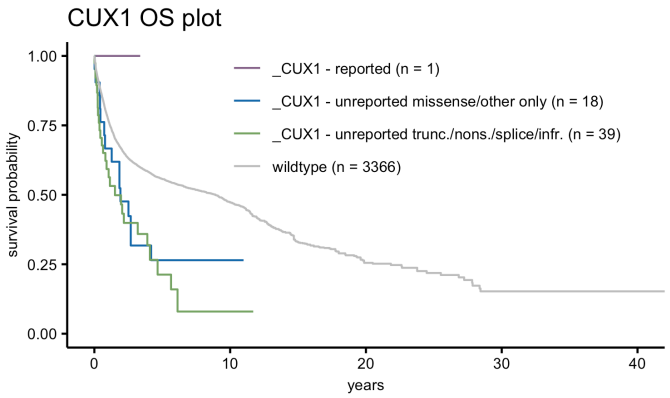


MPL OS plot




SETBP1 OS plot
survival probability
1.00
0.75
0.50
0.25
0.00
0
10
20
30
40
years
_SETBP1 - reported (n = 43)
_SETBP1 - unreported missense/other only (n = 25)
wildtype (n = 3362)


ETV6 OS plot
survival probability
1.00
0.75
0.50
0.25
0.00
0
10
20
30
40
years
_ETV6 - unreported missense/other only (n = 33)
_ETV6 - unreported trunc./nons./splice/infr. (n = 56)
wildtype (n = 3362)


CUX1 OS plot
survival probability
1.00
0.75
0.50
0.25
0.00
0
10
20
30
40
years
_CUX1 - reported (n = 1)
_CUX1 - unreported missense/other only (n = 18)
_CUX1 - unreported trunc./nons./splice/infr. (n = 39)
wildtype (n = 3366)