

Supplementary Table 2: First validation study

Student's t-test of Ψ (PSI) values for each AS event in each group (p-value) with a correction for multiple testing using the false discovery rate (q-value)

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From the results of this first validation step, we selected 21 genes with significant p-value<0.05 (highlighted in yellow) in at least one comparison

and acceptable q-value (less than 55% false positive)

PBD ^{wt}	PDB no mutation	(n=5)
PBD ^{P392L}	PDB with SQSTM1/p62 P392L mutation	(n=5)
HD ^{wt}	HD no mutation	(n=5)
HD ^{P392L}	HD with SQSTM1/p62 P392L mutation	(n=5)

Gene	Primer Pair	Paget vs No Paget p-value	Paget vs No Paget q-value	Mutation vs No Mutation p-value	Mutation vs No Mutation q-value	PDB ^{wt} vs PDB ^{P392L} p-value	PDB ^{wt} vs PDB ^{P392L} q-value	PDB ^{wt} vs HD ^{wt} p-value	PDB ^{wt} vs HD ^{wt} q-value	PDB ^{wt} vs HD ^{P392L} p-value	PDB ^{wt} vs HD ^{P392L} q-value	PDB ^{P392L} vs HD ^{wt} p-value	PDB ^{P392L} vs HD ^{wt} q-value	PDB ^{P392L} vs HD ^{P392L} p-value	PDB ^{P392L} vs HD ^{P392L} q-value	HD ^{wt} vs HD ^{P392L} p-value	HD ^{wt} vs HD ^{P392L} q-value
ABI1	rs.ABI1.F1 rs.ABI1.R1	2,364E-1	4,786E-1	7,504E-1	9,923E-1	2,275E-1	8,969E-1	1,808E-2	1,774E-1	2,981E-1	6,435E-1	5,548E-1	8,401E-1	8,708E-1	8,853E-1	4,349E-1	7,584E-1
ABTB1	refseq_ABTB1.F1 refseq_ABTB1.R1	5,035E-1	4,861E-1	4,636E-1	9,923E-1	2,797E-1	8,969E-1	8,212E-2	4,028E-1	9,650E-1	7,156E-1	7,176E-2	7,446E-1	3,386E-2	5,220E-1	2,614E-3	2,214E-1
AKT2	AKT2.e02.F1 AKT2.e02.R1	7,419E-1	9,953E-1	7,588E-1	9,923E-1	1,745E-1	5,964E-1	3,570E-1	7,899E-1	5,988E-1	6,020E-1	9,881E-1	9,916E-1	1,435E-1	7,950E-1	2,471E-1	9,005E-1
ANXA6	refseq_ANXA6.F1 refseq_ANXA6.R1	3,750E-1	4,786E-1	4,480E-1	9,923E-1	7,784E-1	9,757E-1	6,699E-1	6,479E-1	2,718E-1	6,435E-1	9,355E-1	9,130E-1	4,693E-1	8,743E-1	4,783E-1	7,981E-1
APAF1	APAF1.F8 APAF1.R8	5,599E-1	9,953E-1	6,795E-1	7,788E-1	2,634E-1	5,964E-1	2,201E-1	7,899E-1	6,109E-1	6,020E-1	9,916E-1	6,069E-1	8,089E-1	5,238E-1	9,312E-1	9,312E-1
APAF1	rs.APAF1.F1 rs.APAF1.R1	7,554E-1	9,953E-1	7,174E-1	7,788E-1	2,787E-1	5,964E-1	2,922E-1	7,899E-1	7,180E-1	9,920E-1	9,916E-1	4,050E-1	7,950E-1	4,275E-1	9,205E-1	9,205E-1
APC	APC.F16 APC.R14	5,180E-1	9,953E-1	8,702E-1	7,788E-1	7,760E-1	7,073E-1	3,963E-1	7,899E-1	7,870E-1	6,020E-1	9,916E-1	9,923E-1	8,929E-1	5,082E-1	9,205E-1	9,205E-1
ARNT	refseq_ARNT.F1 refseq_ARNT.R1	8,722E-1	9,953E-1	7,128E-1	7,788E-1	5,133E-1	6,145E-1	4,117E-1	7,899E-1	7,224E-1	6,020E-1	9,916E-1	2,844E-1	7,950E-1	2,011E-1	9,005E-1	9,005E-1
ARNTL	refseq_ARNTL.F1 refseq_ARNTL.R1	6,700E-1	9,953E-1	9,413E-1	7,788E-1	5,456E-1	6,216E-1	7,470E-1	6,691E-1	8,474E-1	6,020E-1	9,916E-1	2,432E-1	7,950E-1	4,476E-1	9,205E-1	9,205E-1
ARRB1	refseq_ARRB1.F1 refseq_ARRB1.R1	9,044E-1	9,953E-1	5,567E-1	7,788E-1	7,565E-1	7,073E-1	9,918E-1	9,942E-1	6,524E-1	6,020E-1	9,916E-1	8,945E-1	7,898E-1	6,443E-1	9,757E-1	9,757E-1
ASPH	refseq_ASPH.F1 refseq_ASPH.R1	3,763E-1	4,786E-1	8,294E-1	9,923E-1	9,689E-1	9,764E-1	1,398E-1	4,672E-1	6,459E-1	6,435E-1	8,768E-1	8,287E-1	8,852E-1	6,188E-1	8,798E-1	8,798E-1
ATG16L1	refseq_ATG16L1.F1 refseq_ATG16L1.R1	6,201E-1	9,953E-1	2,262E-1	7,788E-1	4,786E-1	6,145E-1	7,086E-1	9,617E-1	3,958E-1	6,020E-1	9,916E-1	7,568E-1	8,448E-1	2,300E-1	9,005E-1	9,005E-1
ATG4C	refseq_ATG4C.F1 refseq_ATG4C.R1	6,970E-1	9,953E-1	5,897E-2	7,788E-1	1,195E-1	5,964E-1	3,659E-1	7,899E-1	1,406E-1	6,020E-1	9,916E-1	9,937E-1	8,849E-1	3,658E-1	9,005E-1	9,005E-1
ATG5	APG5L.u.f.9 APG5L.u.r.5	5,565E-1	9,953E-1	9,686E-1	7,788E-1	8,054E-1	7,162E-1	7,634E-1	9,771E-1	9,937E-1	9,986E-1	9,916E-1	7,069E-1	8,351E-1	6,349E-1	9,757E-1	9,757E-1
ATG5	ATG5.e06.F1 ATG5.e06.R1	5,319E-2	9,953E-1	4,328E-1	7,788E-1	3,958E-1	6,145E-1	1,640E-2	5,608E-1	2,721E-1	6,020E-1	9,916E-1	8,310E-2	8,929E-1	4,430E-2	8,506E-1	8,506E-1
ATXN3	refseq_ATXN3.F1 refseq_ATXN3.R1	2,468E-1	9,953E-1	3,440E-1	7,788E-1	9,550E-2	5,964E-1	1,451E-1	7,128E-1	2,104E-1	6,020E-1	9,916E-1	9,902E-1	8,929E-1	9,091E-1	9,757E-1	9,757E-1
ATXN3	rs.ATXN3.F1 rs.ATXN3.R1	4,586E-1	9,953E-1	5,618E-1	7,788E-1	3,871E-1	6,145E-1	4,531E-1	7,899E-1	3,741E-1	6,020E-1	9,916E-1	4,371E-2	7,505E-1	6,244E-2	8,888E-1	8,888E-1
AXIN1	AXIN1.u.f.11 AXIN1.R5	3,079E-1	9,953E-1	8,277E-1	9,923E-1	2,267E-1	5,964E-1	6,239E-2	5,608E-1	6,642E-1	6,020E-1	9,916E-1	5,673E-1	8,089E-1	1,948E-1	9,005E-1	9,005E-1
AXIN1	rs.AXIN1.F1 rs.AXIN1.R1	9,940E-1	9,953E-1	7,039E-1	7,788E-1	1,730E-1	5,964E-1	2,320E-1	7,899E-1	8,318E-1	6,020E-1	9,916E-1	4,904E-1	7,950E-1	2,019E-1	9,005E-1	9,005E-1
BAG6	refseq_BAT3.F1 refseq_BAT3.R1	5,714E-1	9,953E-1	2,058E-1	7,788E-1	2,074E-1	5,964E-1	8,685E-1	9,809E-1	6,391E-1	6,020E-1	9,916E-1	2,236E-1	7,950E-1	7,524E-1	9,757E-1	9,757E-1
BCAS1	BCAS1.F9 BCAS1.R4	3,996E-1	4,786E-1	7,753E-2	9,923E-1				2,088E-1		6,435E-1						
BCL11B	refseq_BCL11B.F2 refseq_BCL11B.R2	6,283E-1	4,972E-1	2,442E-1	9,923E-1	7,463E-1	9,757E-1	1,084E-1	4,654E-1	6,136E-1	6,435E-1	2,162E-1	7,542E-1	5,186E-1	8,743E-1	1,754E-1	7,559E-1
BCL2L1	bc12l1.f.2 bc12l1.r.2	9,268E-1	9,953E-1	7,274E-2	7,788E-1	2,517E-1	5,964E-1	8,646E-1	9,809E-1	3,476E-1	6,020E-1	9,916E-1	1,114E-1	8,075E-1	1,725E-1	9,005E-1	9,005E-1
BCL2L12	BCL2L12-3 BCL2L12-4	3,636E-1	4,786E-1	7,226E-1	9,923E-1	1,352E-1	7,253E-1	3,627E-1	5,877E-1	2,875E-1	6,435E-1	8,768E-1	9,990E-3	4,874E-1	7,884E-2	6,709E-1	6,709E-1
BCL2L15*	refseq_C1orf178.F1 refseq_C1orf178.R1	9,128E-3	3,493E-1	4,193E-1	9,923E-1	7,921E-1	9,757E-1	5,224E-2	2,990E-1	3,561E-1	6,435E-1	4,448E-3	3,838E-1	2,875E-1	8,366E-1	6,562E-1	8,798E-1
BCL2L2andPABPN1	BCL2L2andPABPN1.e02.F1 BCL2L2andPABPN1.R1	3,613E-1	4,786E-1	7,686E-1	9,923E-1	9,709E-1	5,964E-1	3,120E-1	5,790E-1	5,054E-1	6,435E-1	8,401E-1	7,469E-1	8,743E-1	6,436E-1	8,798E-1	8,798E-1
BCLAF1	rs.BCLAF1.F1 rs.BCLAF1.R1	9,504E-1	9,953E-1	6,965E-1	7,788E-1	8,490E-1	6,145E-1	3,187E-1	7,899E-1	8,680E-1	6,089E-1	9,380E-1	2,759E-1	7			