Supplementary Table 5. Individual mutations per SCNA group

Caucasian

GROUP 1 (n=173)				GROUP 2 (n=40)				GROUP 3 (n=67)			
Gene	**Type**	**HGVS**	**n (%)**	**Gene**	**Type**	**HGVS**	**n (%)**	**Gene**	**Type**	**HGVS**	**n (%)**
SETD1B	Frame-shift deletion	p.His8ThrfsTer27	28 (16.18)	*FGFR2*	Missense	p.Ser252Trp	7 (17.5)	*PIK3CA*	Missense	p.His1047Arg	7 (10.45)
RNF43	Frame-shift deletion	p.Gly659ValfsTer41	27 (15.61)	*PTEN*	Missense	p.Arg130Gly	6 (15.0)	*PTEN*	Missense	p.Arg130Gly	6 (8.96)
RPL22	Frame-shift deletion	p.Lys15ArgfsTer5	25 (14.45)	*PIK3CA*	Missense	p.His1047Arg	4 (10.0)	*CAMSAP2*	Frame-shift deletion	p.Met1437Ter	6 (8.96)
JAK1	Frame-shift deletion	p.Lys860AsnfsTer16	25 (14.45)	*CTNNB1*	Missense	p.Ser37Cys	4 (10.0)	*RPL22*	Frame-shift deletion	p.Lys15ArgfsTer5	6 (8.96)
PTEN	Missense	p.Arg130Gly	23 (13.29)	*PTEN*	Nonsense	p.Arg233Ter	3 (7.5)	*ACVR2A*	Frame-shift deletion	p.Lys437ArgfsTer5	5 (7.46)
PTEN	Missense	p.Arg130Gln	17 (9.83)	*KRAS*	Missense	p.Gly12Val	3 (7.5)	*NIPBL*	Frame-shift deletion	p.Ile2776LeufsTer3	5 (7.46)
PIK3CA	Missense	p.Arg88Gln	17 (9.83)	*KRAS*	Missense	p.Gly12Cys	3 (7.5)	*BCOR*	Missense	p.Asn1459Ser	5 (7.46)
DOCK3	Frame-shift deletion	p.Pro1852GlnfsTer45	15 (8.67)	*OR14K1*	Missense	p.Arg14Ser	3 (7.5)	*SETD1B*	Frame-shift deletion	p.His8ThrfsTer27	4 (5.97)
PTEN	Nonsense	p.Arg233Ter	13 (7.51)	*SOS1*	Missense	p.Asn233Tyr	3 (7.5)	*KRAS*	Missense	p.Gly12Asp	4 (5.97)
BCOR	Missense	p.Asn1459Ser	13 (7.51)	*PIK3R1*	Missense	p.Gly376Arg	3 (7.5)	*ARID1A*	Nonsense	p.Arg1989Ter	4 (5.97)
KRAS	Missense	p.Gly12Asp	12 (6.94)	*SMC3*	Frame-shift deletion	p.Tyr43MetfsTer69	2 (5.0)	*UPF3A*	Frame-shift deletion	p.Glu267ArgfsTer13	4 (5.97)
CTCF	Frame-shift insertion	p.Thr204AsnfsTer26	12 (6.94)	*FGFR2*	Missense	p.Cys382Arg	2 (5.0)	*ZNF609*	Frame-shift deletion	p.Lys734ArgfsTer12	4 (5.97)
FGFR2	Missense	p.Ser252Trp	11 (6.36)	*MYO3A*	Frame-shift deletion	p.Met1192Ter	2 (5.0)	*XYLT2*	Frame-shift deletion	p.Gly529AlafsTer78	4 (5.97)
ZBTB20	Frame-shift deletion	p.Pro692LeufsTer43	11 (6.36)	*CTGLF11P*	Missense	p.His132Arg	2 (5.0)	*SMAD7*	Frame-shift deletion	p.Pro209LeufsTer115	4 (5.97)
SLC3A2	Frame_Shift_Del	p.Lys300ArgfsTer31	10 (5.78)	*PTEN*	Nonsense	p.Arg130Ter	2 (5.0)	*BZW1*	Frame-shift deletion	p.Asn285ThrfsTer11	4 (5.97)

Caucasian (continued)

GROUP 4 (n=39)				GROUP 5 (n=49)			
Gene	**Type**	**HGVS**	**n (%)**	**Gene**	**Type**	**HGVS**	**n (%)**
TP53	Missense	p.Cys141Tyr	3 (7.69)	*PPP2R1A*	Missense	p.Pro179Arg	12 (24.49)
PPP2R1A	Missense	p.Pro179Arg	3 (7.69)	*GOLGA6L6*	Missense	p.Glu519Val	4 (8.16)
PIK3CA	Missense	p.Glu542Lys	3 (7.69)	*OR14K1*	Missense	p.Arg14Ser	3 (6.12)
FGFR2	Missense	p.Ser252Trp	2 (5.13)	*TP53*	Missense	p.Arg273His	3 (6.12)
PTEN	Missense	p.Arg130Pro	2 (5.13)	*TP53*	Missense	p.Arg273Cys	3 (6.12)
CHD4	Missense	p.Arg1162Trp	2 (5.13)	*TP53*	Missense	p.Tyr220Cys	3 (6.12)
TP53	Missense	p.Arg282Trp	2 (5.13)	*PIK3CA*	Missense	p.Glu542Lys	3 (6.12)
TP53	Missense	p.Arg248Trp	2 (5.13)	*PCNXL3*	Missense	p.Pro403Ser	2 (4.08)
PPP2R1A	Missense	p.Ser256Phe	2 (5.13)	*KRAS*	Missense	p.Gly12Val	2 (4.08)
PPP2R1A	Missense	p.Trp257Leu	2 (5.13)	*CHD4*	Missense	p.Arg975His	2 (4.08)
PIK3CA	Missense	p.Gln546Pro	2 (5.13)	*GOLGA6L6*	Missense	p.Met472Ile	2 (4.08)
PIK3CA	Missense	p.His1047Arg	2 (5.13)	*GOLGA6L6*	Missense	p.Lys425Glu	2 (4.08)
TLX1NB	Missense	p.His81Arg	1 (2.56)	*GOLGA6L6*	Missense	p.Glu376Lys	2 (4.08)
POLL	Missense	p.Pro231Leu	1 (2.56)	*GOLGA6L9*	Missense	p.Ala373Pro	2 (4.08)
KCNIP2	Missense	p.Pro44Ser	1 (2.56)	*TP53*	Missense	p.Arg248Gln	2 (4.08)

BoAA

GROUP 1 (n=23)				GROUP 2 (n=31)				GROUP 3 (n=30)			
Gene	**Type**	**HGVS**	**n (%)**	**Gene**	**Type**	**HGVS**	**n (%)**	**Gene**	**Type**	**HGVS**	**n (%)**
TP53	Missense	p.Arg273His	3 (13.04)	*TP53*	Missense	p.Arg273His	3 (9.68)	*PTEN*	Nonsense	p.Arg233Ter	4 (13.3)
SPOP	Missense	p.Met117Val	2 (8.70)	*KMT2D*	Frame-shift deletion	p.Gly1235ValfsTer95	2 (6.45)	*POLE*	Missense	p.Pro286Arg	4 (13.3)
TP53	Missense	p.Arg248Gln	2 (8.70)	*GOLGA6L6*	Missense	p.Met472Ile	2 (6.45)	*ACVR2A*	Frame-shift deletion	p.Lys437ArgfsTer5	4 (13.3)
PPP2R1A	Missense	p.Pro179Arg	2 (8.70)	*CHGB*	Missense	p.Val351Ile	2 (6.45)	*PIK3CA*	Missense	p.Arg88Gln	4 (13.3)
DSCAM	Missense	p.Asn1363Lys	2 (8.70)	*FBXW7*	Missense	p.Arg465His	2 (6.45)	*BCOR*	Missense	p.Asn1459Ser	4 (13.3)
FBXW7	Missense	p.Arg505Gly	2 (8.70)	*SRP72*	Frame-shift deletion	p.Val9CysfsTer10	2 (6.45)	*PTEN*	Missense	p.Arg130Gln	3 (10.0)
GTPBP4	Missense	p.Asn234Tyr	1 (4.35)	*PKD2L1*	Missense	p.Leu374Phe	1 (3.23)	*PIWIL1*	Missense	p.Arg275Gln	3 (10.0)
CHUK	Missense	p.Leu152Phe	1 (4.35)	*SCD*	Missense	p.Gly31Val	1 (3.23)	*KRAS*	Missense	p.Gly12Asp	3 (10.0)
WNT8B	Missense	p.Arg79His	1 (4.35)	*NDUFB8*	Missense	p.Asp110His	1 (3.23)	*DIO1*	Missense	p.Arg241Cys	3 (10.0)
HPS6	Missense	p.Asp714His	1 (4.35)	*FGF8*	Missense	p.Ser138Ile	1 (3.23)	*RPL22*	Frame-shift deletion	p.Lys15ArgfsTer5	3 (10.0)
GBF1	Missense	p.Gln1467Lys	1 (4.35)	*IDI2*	Missense	p.Arg142Lys	1 (3.23)	*CDH1*	Frame-shift deletion	p.Pro126ArgfsTer89	3 (10.0)
ACTR1A	Missense	p.Arg46Cys	1 (4.35)	*PPRC1*	Missense	p.Gln863His	1 (3.23)	*NF1*	Nonsense	p.Arg2450Ter	3 (10.0)
NEURL1	Missense	p.Ser493Cys	1 (4.35)	*GBF1*	Missense	p.Ser289Cys	1 (3.23)	*RNF43*	Frame-shift deletion	p.Gly659ValfsTer41	3 (10.0)
SORCS3	Missense	p.Glu378Asp	1 (4.35)	*GBF1*	Missense	p.Glu626Asp	1 (3.23)	*SMAD7*	Frame-shift deletion	p.Pro209LeufsTer115	3 (10.0)
SORCS3	Missense	p.Gln980His	1 (4.35)	*NFKB2*	Missense	p.Asp561Tyr	1 (3.23)	*ZNF490*	Missense	p.Arg382Ile	3 (10.0)

BoAA (cont.)

GROUP 4 (n=22)			
Gene	**Type**	**HGVS**	**n (%)**
SETD1B	Frame-shift deletion	p.His8ThrfsTer27	4 (18.18)
KRAS	Missense	p.Gly12Asp	4 (18.18)
RPL22	Frame-shift deletion	p.Lys15ArgfsTer5	4 (18.18)
JAK1	Frame-shift deletion	p.Lys860AsnfsTer16	4 (18.18)
PAX2	Frame-shift deletion	p.Arg403GlyfsTer37	3 (13.64)
MVK	Frame-shift deletion	p.Ala141ArgfsTer18	3 (13.64)
ZNF124	Frame-shift deletion	p.Thr339LeufsTer31	3 (13.64)
KMT2D	Frame-shift deletion	p.Pro2354LeufsTer30	3 (13.64)
SPECC1	Frame-shift deletion	p.Asn303ThrfsTer63	3 (13.64)
RNF43	Frame-shift deletion	p.Gly659ValfsTer41	3 (13.64)
KMT2B	Frame-shift deletion	p.Lys553AsnfsTer52	3 (13.64)
PHF2	Frame-shift deletion	p.Lys492ArgfsTer6	3 (13.64)
USP26	Frame-shift deletion	p.Phe348LeufsTer7	3 (13.64)
TUBGCP2	Frame-shift deletion	p.Gln883LysfsTer25	2 (9.09)
CTGLF11P	Missense	p.His132Arg	2 (9.09)