

**Supplemental Table 1. Biallelic SBDS Sequence Variants in this Cohort of Subjects with SDS**

# of Subjects	DNA change		mRNA/Protein change		mRNA/Protein change Class		# of subjects per category
	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	
95	c.258+2T>C	c.183_184delTAinsCT	IVS2 splice donor+2	Lys62*	splice	stop	116
2		c.184A>T		Lys62*			
2		c.652C>T		Arg218*			
1		c.25C>T		Gln9*			
7		c.258+2T>C; c.183_184delTAinsCT		IVS2 splice donor+2; Lys62*		splice and stop	
2		c.120delIG		Ser41AlafsX18		frameshift	
2		c.523delIC		Arg175GlyfsX12			
1		c.18delIC		Thr7ProfsX6			
1		c.107delIT		Val36AlafsX23			
1		c.119delIG		Ser41AlafsX18			
1		c.123delIC		Ser41ArgfsX18			
1		c.297_300delAAGA		Glu99AspfsX21			
15		c.258+2T>C (homozygous)		IVS2 splice donor+2 (homozygous)		splice (homozygous)	
6	c.258+2T>C	c.258+1G>C	IVS2 splice donor+2	IVS2 splice donor +1	splice	compound heterozygous splice	9
1		c.259-2A>G		IVS2 splice acceptor-2			
1		c.460-1G>A		IVS3 splice acceptor-1			
1		c.624+1G>A		IVS4 splice donor +1			
2	c.258+2T>C	c.250T>C	IVS2 splice donor+2	Cys84Arg	splice	missense	15
2		c.355T>C		Cys119Arg			
2		c.505C>T		Arg169Cys			
2		c.653G>A		Arg218Gln			
1		c.3G>A		Met1Ile <sup>#</sup>			
1		c.41A>G		Asn14Ser			
1		c.95A>G		Tyr32Cys			
1		c.169T>C		Phe57Leu			
1		c.170T>C		Phe57Ser			
1		c.458A>G		Gln153Arg			
1		c.641C>T		Pro214Leu			
1		c.258+2T>C		deletion chr7:66,448,591-66,463,734			
1	deletion chr7:66,450,964-66,454,945		deletion chr7:66,450,964-66,454,945				
1	deletion 7q11.21q11.22		deletion 7q11.21q11.22				
1	c.183_184delTAinsCT	c.523C>T	Lys6*	Arg175Trp	stop	missense	1

<sup>#</sup> Start codon variant

**Supplemental Table 2. Disease Characteristics and Outcomes of subjects with SDS and MDS or AML**

Patient number	Sex	Age at MDS/AML diagnosis	Cytogenetics	Blast % IHC (^Flow)	Initial therapy	Survival
<b>AML</b>						
42	F	14.76	43-46,XX,-5,-6,-8,add(20)9q13.3),add(21)(11.1),-22,del(22)(q12),+1-4mar[cp20] del5d, del20q and del8q by FISH	not available	Chemo	Deceased
16	F	39.07	46,XX,add(3)(q27),del(8)(q13q22),der(10)t(10;15)(p11.2q15)[1], 43-44,der(2)t(2;12)(q33;q13),add(3)(q27),add(4)(q21),-5,-6,add(6)(q25),-7,del(8)(q13q22),-11,-12,der(18)t(11;19)(q13;p13.3)+2-3mar[6], 46,XX[13]	25(19)	Chemo	Deceased
43	M	19.53	47,XY,+21,dic(21;21)(p11.2;p11.2),dic(22;22)(p11.2;p11.2),+2mar [1]/ 47-48,sl,-16,+1-2mar[cp13][13] / 47,sl,del(6)(q21q27),dic(13;16)(p13.p11.2),+mar[3] / 48,sl,-18,+2mar [1]/42,XY,-8,-10,-14,-18,dic(21;21)(p11.2;p11.2),dic(21;22)(p11.2,p11.2),+mar[1]/46,XY[1]	34(20)	Chemo	Deceased
44	F	22.97	47-48,XX,add(5)(q14)[5],add(7)(q22)[5],+16[8],add(19)(q13.3)[7],-20[4], del(20)(q11.2q13.1)[5],+21[13], add(21)(p11.2)x2[12], +add(21)(p11.2)[4][cp4]/46,XX[6]. Del20q and del7q by FISH	60-70 (10)	HSCT	Deceased
45	F	37.88	44,XX,add(2)(q35),del(5)(q22q35),add(6)(p21.3),del(7)(q11.2),-9,del(12)(p11.2p13),-13,add(15)(p11.2)[2] / 44,XX,add(2)(q35),der(3;7)(q10;q10),-5,add(6)(p21.3), der(9;12)(q10;q10),add(15)(p11.2),+mar1[9] / 44,XX,+1,idel(1)(p13), add(2)(q35), der(3;7)(q10;q10),-5,add(6)(p21.3),der(9;12), (q10;q10),add(15)(p11.2),+mar1[2] / 46,XX [6] . Mosaic 5 and 7 by FISH	20 (31)	Chemo	Deceased
46	F	36.96	not available	not available	Chemo	Deceased
47	F	47.34	46,XX,del(5)(q11.2q33),add(22)(q13)[9]/46-49, idem, add(3)(p14),add(13)(p11.2),+19,der(20)t(3;20)(p14.q11.2),-21,+mar13[cp6]/46,XX[3]. Del5q, del20q, del21 by FISH	30-40(30)	Chemo	Deceased
48	M	14.44	46,XY[20]	56 (73)	Chemo	Alive in remission
49	M	29.17	46,XY,add(2)(q12-14),-5,r(11)(p11.2q23),+der(15)t(5;15)(p13;q22)[4]/46-48,sl,-add(2)(q12-14),+2,-3,del(7)(q21),-r(11)(p11.2q23),+der(11)add(11)(p11.2)hsr(11)(q23),+del(19)(p13.1p13.3),+r,+mar1[cp6]/47,sl,-Y,add(X)(p11.2),-add(2)(q12-14),+2,add(10)(q26),-r(11)(p11.1q23),+dic r(11;11)(p11.2q23;p.11.2q23),+13,-der(15)t(5;15)(p13;q22),+del(19)(p13.1p13.3),+mar2[8]/46,XY,ins(5;?)(p13;?) [5] ; del5q and del7q by FISH	52 (57)	Chemo	Deceased
7	M	27.7	not available	20-40	Chemo	Deceased
<b>MDS</b>						
50	M	10.75	42/R/44,XY,der(5)del(5)(q15q33)psu dic(5;12)(q35;p13), -7[cp19]/46, Xy[1]; monosomy 5 and 7 by FISH	5	HSCT	Deceased
51	F	0.48	not available	0	HSCT	Alive
22	M	4.82	47,XYc,del(20)(q11.2q13.2)[15]/47,XYc[5]. Del20q by FISH	0	HSCT	Alive
52	F	45.05	46,XX,der(7)t(1;7)(q12;q11.2)[4]/46,XX[16]	0	<b>None</b>	Deceased
53	F	21.27	46, XX [20]; trisomy 8 by FISH	1 (0.5)	HSCT	Deceased
25	M	5.43	46, XY, del(20)(q11.2q13.1)[10]/46,XY,add(7)(q22)[7]/45, XY-7[3]	not available	HSCT	Alive
54	M	10.34	46,XY,i(7)9q10[10]/46,XY[1]. Trisomy 7q by FISH, low level del20q by FISH	2	HSCT	Alive
14	M	2.19	46, XY, +der(1;17)(q10;q10)[18]/46, XY[2]	1(3)	HSCT	N/A

4	F	13.84	46,XX,der(3;6)(q25;q13)[20]/45, dic(6;7)(q13;p11.2)[4]/46,XX[1]. Add 3q27 by FISH	5(3)	Chemo	Deceased
55	M	9.39	92, XXYY, add(7)9q32)x2, add(9)9q21)[5]/46,XY[15]	16(3)	HSCT	Alive
56	F	22.57	not available	not available	Chemo	Deceased
33	F	30.15	46XX,del(20)(q11.2q13.3)[9]/46,XX[11]	1	HSCT	Alive
57	F	18.62	44,XX,-5,-7,del(9)(p22),der(13)r(13:13)(?p13q34;q12q34),-18,+mar1-3,2dmin[3]/44idem,add(3)(q27),-15,+mar1,+mar2,+mar3[7]/43,idem,der(6)dic(6,-17)(p?23;p13),-17[6]	not available	Chemo	Deceased
58	M	18.03	not available	not available	HSCT	Deceased
37	M	21.47	46XY[5]/46, XY, del(12)(p11.1)[5]/35,idem,del(5)(q?22q?31),-7,del(9)(q21)[3]/44, idem, add(1)(p32),add(3)(p24),-5,-7,del(9)(q21)[7]/45, idem, add(1)(p32),add(3)(p24),-5,-7,del(9)(q21),=mar[4]; 77%monosomy 7 by FISH	not available	Chemo	N/A
59	F	1.34	46,XX	not available	HSCT	Alive

^ blast percentage by immunophenotyping when available; IHC, immunohistochemistry