Supplementary Appendix

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Table S1. Targeted gene panel

ASXL1
ATM
ATRX
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FLT3
GNAS
GNB1
IDH1
IDH2
JAK2
KDM6A
KRAS
LUC7L2
MYD88
NPM1
NRAS
PHF6
PPM1D
PTPN11
RAD21
RUNX1
SETBP1
SETD2
SF1
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STAG2
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TET2
TP53
U2AF1
ZBTB33
ZNF318
ZRSR2

Table S2. Clinical characteristics of patients with SCN or cyclic neutropenia

Study ID	SCN or Cyclic	HPC Exome Seq	Age (years)	Gender	*ELANE* Mutation	Median G-CSF (µg/kg/d)	ANC (x 10^{-3}/µl)	MDS/AML (age in years)	BMT/death (age in years)	Tissue
CN01	Cyclic	No	3	M	W241G	NA	NA	N		Bone Marrow
CN03	Cyclic	No	30	M	V190_F199del	1	0.2	N		Blood
CN08	Cyclic	No	31	M	S126L	3.6	NA	N		Bone Marrow
CN09	Cyclic	No	26	M	I60T	2.5	NA	N		Bone Marrow
CN10	Cyclic	No	17	M	Unknown	0.5	0.2	N		Blood
CN11	Cyclic	No	30	M	Unknown	N/A	N/A	N		Blood
CN12	Cyclic	No	47	M	Unknown	1.7	N/A	N		Blood
CN13	Cyclic	No	33	F	Unknown	N/A	N/A	N		Blood
CN14	Cyclic	No	18	M	P139L	2.8	0.4	N		Blood
CN15	Cyclic	No	47	M	R220Q	0.15	1.521	N		Bone Marrow
CN16	Cyclic	No	23	F	S126L	2.2	NA	N	BMT (24)	Bone Marrow
CN17	Cyclic	No	3	F	S46F	5	0.745	N		Bone Marrow
CN18	Cyclic	No	11	F	IVS4+5 G>A	17	NA	N		Bone Marrow
SCN01	SCN	Yes	14	F	S126L	1.2	0.1	N		Bone Marrow
SCN02	SCN	Yes	14	F	S126L	1.3	0	N		Bone Marrow
SCN03	SCN	Yes	7	M	V65F	12	0.1	N	BMT (13)	Bone Marrow
SCN04	SCN	Yes	6	F	IVS4+1 G>A	NA	NA	N		Bone Marrow
SCN05	SCN	Yes	18	M	S126L	1.6	0	N		Bone Marrow
SCN06	SCN	Yes	18	M	S126L	0.8	0	N		Bone Marrow
SCN07	SCN	Yes	18	F	S126L	2.2	0	N		Bone Marrow
SCN11	SCN	Yes	21	M	Unknown	3.6	0	N		Bone Marrow
SCN12	SCN	Yes	16	M	R103L	2.4	NA	N	BMT (16)	Bone Marrow
SCN13	SCN	Yes	15	M	S126L	2.1	0.3	N		Bone Marrow
SCN14	SCN	No	11	F	C223X	15	0.175	N	Died (18)	Bone Marrow
SCN15	SCN	No	10	M	Thr95_Ala100del	7.4	NA	N		Bone Marrow
SCN18	SCN	No	1	M	G125R	0	0.2	N		Bone Marrow
SCN22	SCN	Yes	36	F	C194ter	12	0.06	AML (37)		Bone Marrow
SCN24	SCN	No	10	F	R81P	5.8	0.141	N		Bone Marrow
SCN26	SCN	No	1	M	C55R	8.3	0.219	N		Bone Marrow
SCN27	SCN	No	3	F	S126L	1.7	0.094	N		Bone Marrow
SCN28	SCN	No	3	F	S126L	3.3	0.062	N		Bone Marrow
SCN30	SCN	No	1	M	G203R	20.6	0.081	N	BMT (3)	Bone Marrow
SCN33	SCN	No	22	F	S126L	11.5	0.038	N		Bone Marrow
SCN34	SCN	No	1	F	V101M	4.6	0	N	BMT (6)	Bone Marrow
SCN35	SCN	No	1	M	G203R	183	0.445	N	BMT (2)	Bone Marrow
SCN36	SCN	No	1	F	A57T	53	0	N	BMT (1)	Bone Marrow
SCN37	SCN	No	3 months	F	C71S	18	0.347	N	BMT (6)	Bone Marrow
SCN38	SCN	No	14	F	S126W	2.5	0.059	N		Bone Marrow
SCN39	SCN	No	13	M	I60T	19	0	N		Bone Marrow
SCN40	SCN	No	3	F	P139L	0.6	0.252	N		Bone Marrow
SCN41	SCN	No	7	F	V45M	4.2	0.175	N		Bone Marrow
SCN42	SCN	No	4	F	G221X	15	0.27	N		Bone Marrow
SCN43	SCN	No	6	M	S126L	12	0.1	N		Bone Marrow
SCN44	SCN	No	20	M	L84P	8.5	0.061	N		Bone Marrow
SCN45	SCN	No	25	F	I129DEL	2.7	0	N		Bone Marrow
SCN46	SCN	No	6	M	I129DEL	1.8	0.305	N		Bone Marrow
SCN47	SCN	No	2	F	Ins PQ123	22.5	0.124	N	BMT (2)	Bone Marrow
SCN48	SCN	No	32	F	M66R	3.4	0.648	N	Died (36)	Bone Marrow
SCN49	SCN	No	2	F	G214R	15.4	0.069	N	BMT (8)	Bone Marrow
SCN50	SCN	No	10	F	S126L	5	0	N		Bone Marrow
SCN51	SCN	No	45	F	S126L	5	0.173	AML (56)		Bone Marrow
SCN52	SCN	No	15	M	S126L	6	0.077	N		Bone Marrow
SCN53	SCN	No	13	M	S126L	1.9	0.403	N		Bone Marrow

HPC: hematopoietic progenitor cells; ANC:absolute neutrophil count: MDS: myelodyplastic syndrome

AML: acute myeloid leukemia; BMT: bone marrow transplantation.

The age at the time the sample was collected is shown.

Samples in which exome sequencing was performed on expanded hematopoeitic progenitors is indicated

Table S3. Clinical characteristics of patients with SDS

Study ID	Exome Seq	Age (years)	Gender	*SBDS* Mutation	G-CSF	ANC (x 10^{-}	Hb (gm/dL)	Platelet (x 10^{-}	MDS/ AML	Cytog Abnl	Tissue
SDS16	Yes	2	F	Homozygous IVS2+2 T>C	No	0.33	11.4	180	N	N	Bone Marrow
SDS35	Yes	12	M	Homozygous 258+2T>C	No	2.02	11.9	117	N	del20q	Bone Marrow
SDS05	No	12	M	183-184CT>TA,258+2T>C	Yes	2.4	12.8	158	N	del20q	Bone Marrow
SDS06	No	18	M	183-184CT>TA,258+2T>C	No	0.29	15.9	150	N	N	Bone Marrow
SDS07	No	19	F	183-184CT>TA,258+2T>C	Yes	0.78	12.1	111	N	N	Bone Marrow
SDS08	No	8	M	183-184CT>TA,258+2T>C	No	1.11	13.4	233	N	N	Bone Marrow
SDS09	No	5	M	183-184CT>TA,258+2T>C	No	0.77	12.7	203	N	N	Bone Marrow
SDS10	No	4	M	183-184CT>TA,258+2T>C	No	1.18	11.3	141	N	N	Bone Marrow
SDS11	No	11	M	183-184CT>TA,258+2T>C	Yes	3.24	11.0	42	N	N	Bone Marrow
SDS12	No	3	F	183-184CT>TA,258+2T>C	No	0.26	12.0	209	N	N	Bone Marrow
SDS13	No	11	F	183-184CT>TA,258+2T>C	Yes	2.54	14.0	141	N	N	Bone Marrow
SDS15	No	6	M	183-184CT>TA,258+2T>C	No	0.83	13.0	171	N	N	Bone Marrow
SDS17	No	10	M	183-184CT>TA,258+2T>C	Yes	1.78	11.5	197	N	N	Blood
SDS18	No	3	F	183-184CT>TA,258+2T>C	No	1.66	11.6	130	N	N	Blood
SDS19	No	13	M	183-184CT>TA,258+2T>C	No	0.25	12.2	90	N	N	Blood
SDS20	No	6	F	183-184CT>TA,258+2T>C	Yes	0.38	12.6	145	N	N	Blood
SDS21	No	5	F	183-184CT>TA,258+2T>C	No	4.49	12.9	224	N	N	Blood
SDS22	No	8	M	183-184CT>TA,258+2T>C	No	1.03	12.5	134	N	N	Blood
SDS23	No	9	M	183-184CT>TA,258+2T>C	NA	0.62	12.4	72	N	N	Blood
SDS24	No	5	M	183-184CT>TA,258+2T>C	NA	0.40	11.7	234	N	N	Blood
SDS25	No	10	M	183-184CT>TA,258+2T>C	No	1.98	13.3	96	N	N	Blood
SDS28	No	2	M	183_184TA>CT; 258+2T>C	Yes	0.00	7.3	112	N	N	Bone Marrow
SDS29	No	1	M	183_184TA>CT+258_2T>C; 258+2T>C	Yes	0.00	11.1	418	N	N	Bone Marrow
SDS30	No	2	F	258+2T>C; 613C>T (p.Q205*)	Yes	0.28	11.2	225	N	N	Bone Marrow
SDS31	No	16	F	183_184TA>CT; 258+2T>C	Yes	0.37	12.2	176	N	N	Bone Marrow
SDS32	No	5	F	183_184TA>CT; 258+2T>C	No	1.03	11.8	158	N	N	Bone Marrow
SDS33	No	14	M	183_184TA>CT; 258+2T>C	No	0.47	11.0	319	N	N	Bone Marrow
SDS34	No	6	F	98A>C (p.K33T); structural variant	No	0.70	10.0	121	N	N	Bone Marrow

HPC: hematopoetic progenitor cell; ANC: absolute neutrophil count: MDS: myelodysplastic syndrome: AML: acute myeloid leukemia

Cytog abnl: cytogenetic abnormality

Table S4. Hematopoeitic progenitor clone somatic variants

Sample	HPC Clone #	Chr	start	stop	Ref	Var	type	Gene	Mutation type	cDNA position	amino acid change	WBC ref count	WBC var ount	WBC VAF	HPC ref count	HPC var count	HPC VAF
Cord01	1	3	10102079	10102079	G	A	SNP	FANCD2	missense	c.257	p.R86Q	107	0	0	36	46	54.76
Cord01	1	16	56509438	56509438	C	T	SNP	OGFOD1	silent	c.1428	p.G476	169	0	0	83	74	45.68
Cord01	2	5	73179614	73179614	C	T	SNP	ARHGEF28	missense	c.2960	p.A987V	152	0	0	68	58	44.62
Cord01	3	16	89661953	89661953	G	A	SNP	CPNE7	missense	c.1706	p.R569Q	90	0	0	64	61	46.56
Cord01	4	10	97402772	97402772	G	A	SNP	ALDH18A1	missense	c.280	p.R94C	37	0	0	13	18	58.06
Cord02	3	5	121188197	121188197	T	C	SNP	FTMT	missense	c.539	p.L180P	64	0	0	16	31	64.58
Cord02	3	11	5530122	5530122	A	T	SNP	UBQLN3	missense	c.667	p.F223I	50	0	0	17	19	47.5
Cord02	4	16	84907569	84907569	G	A	SNP	CRISPLD2	missense	c.1156	p.V386M	79	1	1.19	49	44	47.31
Cord02	4	7	107710283	107710283	C	T	SNP	LAMB4	silent	c.2172	p.K724	61	0	0	40	22	33.85
Cord02	4	15	59466007	59466007	C	T	SNP	MYO1E	missense	c.2272	p.V758M	62	0	0	32	29	46.77
Cord03	1	10	37441037	37441037	T	G	SNP	ANKRD30A	silent	c.1527	p.S509	179	3	1.6	65	86	54.78
Cord03	1	22	28194881	28194883	GCT	-	DEL	MN1	in_frame_del	c.1651_1649	p.Q550in_frame_del	34	1	2.86	23	7	23.33
Cord03	1	1	158607931	158607931	C	T	SNP	SPTA1	missense	c.5081	p.R1694H	89	0	0	39	40	49.38
Cord03	3	14	60585194	60585194	G	A	SNP	PCNXL4	missense	c.1726	p.V576M	173	2	1.13	44	45	49.45
Cord03	4	16	23535662	23535662	G	A	SNP	EARS2	rna	NULL	NULL	77	0	0	44	67	60.36
Cord03	4	1	70303967	70303967	G	T	SNP	LRRC7	splice_site	c.533+1	e5+1	125	0	0	81	76	47.8
Cord03	4	11	102667861	102667861	G	C	SNP	MMP1	missense	c.383	p.A128G	87	0	0	57	44	38.94
NM01	1	3	62478113	62478113	C	T	SNP	CADPS	missense	c.643	p.V215M	80	0	0	24	24	50
NM01	1	19	19338712	19338712	C	T	SNP	NCAN	silent	c.2283	p.F761	51	0	0	21	20	48.78
NM01	1	11	4936626	4936626	A	C	SNP	OR51G2	missense	c.268	p.F90V	46	0	0	13	21	61.76
NM01	1	19	3643312	3643312	C	T	SNP	PIP5K1C	silent	c.1578	p.T526	110	0	0	22	35	61.4
NM01	1	18	9887456	9887456	G	T	SNP	TXNDC2	missense	c.980	p.G327V	68	1	1.43	40	31	41.33
NM01	2	16	88873773	88873776	CTGC	-	DEL	CDT1	frame_shift_del	c.1360_1363	p.P455fs	52	0	0	20	18	47.37
NM01	2	7	74004235	74004235	G	A	SNP	GTF2IRD1	missense	c.508	p.G170R	53	1	1.85	37	38	50.67
NM01	2	9	91965660	91965661	-	TAAA	INS	SECISBP2	frame_shift_ins	c.2006_2007	p.R671fs	110	0	0	62	44	41.51
NM01	3	17	79517302	79517302	C	T	SNP	C17orf70	silent	c.1218	p.S406	49	0	0	5	16	76.19
NM01	3	19	41633855	41633856	-	T	INS	CYP2F1	frame_shift_ins	c.1344_1345	p.Y448fs	74	0	0	16	15	48.39
NM01	3	20	26061845	26061845	T	A	SNP	FAM182A	rna	NULL	NULL	74	1	1.33	12	7	36.84
NM01	4	4	83978461	83978461	A	T	SNP	COPS4	silent	c.615	p.A205	52	0	0	62	84	54.9
NM01	4	20	37653945	37653945	A	G	SNP	DHX35	missense	c.1744	p.R582G	90	0	0	80	89	50
NM01	4	9	136570105	136570105	G	T	SNP	SARDH	silent	c.1519	p.R507	203	1	0.49	132	135	48.74
NM02	1	2	208434919	208434919	T	A	SNP	CREB1	missense	c.421	p.L141I	84	0	0	70	75	50.68
NM02	2	11	83770418	83770418	G	A	SNP	DLG2	missense	c.859	p.R287C	59	0	0	47	40	45.98
NM02	2	11	17631829	17631829	C	T	SNP	OTOG	missense	c.5018	p.S1673L	42	0	0	27	31	51.67
NM02	2	2	220163827	220163827	A	T	SNP	PTPRN	missense	c.1610	p.V537E	49	1	2	27	24	46.15
NM02	2	22	25297913	25297913	C	T	SNP	SGSM1	silent	c.2889	p.N963	46	0	0	16	11	39.29
NM02	3	19	3905613	3905613	G	C	SNP	ATCAY	silent	c.318	p.S106	116	0	0	56	45	44.12

NM02	3	7	71252771	71252771	C	T	SNP	CALN1	missense	c.775	p.G259S	53	0	0	26	16	38.1
NM02	3	12	56489515	56489515	C	A	SNP	ERBB3	silent	c.1980	p.G660	81	0	0	41	40	49.38
NM02	3	18	34324149	34324149	C	T	SNP	FHOD3	nonsense	c.559	p.Q187*	63	0	0	26	24	48
NM02	3	16	47941192	47941192	G	A	SNP	LOC101927132	rna	NULL	NULL	41	0	0	29	41	58.57
NM02	3	17	1386202	1386202	C	T	SNP	MYO1C	missense	c.499	p.G167R	51	0	0	21	22	51.16
NM02	3	6	160864668	160864668	C	T	SNP	SLC22A3	silent	c.1407	p.F469	48	0	0	40	26	38.81
NM02	3	10	106974293	106974293	C	T	SNP	SORCS3	silent	c.2469	p.G823	75	0	0	30	27	44.26
NM02	3	17	49075878	49075878	C	T	SNP	SPAG9	missense	c.1765	p.V589I	104	0	0	65	47	41.96
NM02	4	15	78401634	78401634	C	G	SNP	CIB2	missense	c.289	p.V97L	149	0	0	132	151	52.8
NM02	4	7	121636544	121636544	T	C	SNP	PTPRZ1	missense	c.1037	p.F346S	103	1	0.96	75	80	51.28
NM03	1	10	105236269	105236269	C	T	SNP	CALHM3	missense	c.325	p.A109T	69	0	0	46	48	49.48
NM03	1	3	58134511	58134511	A	G	SNP	FLNB	missense	c.6023	p.Y2008C	60	0	0	45	44	48.35
NM03	1	11	68115353	68115353	C	T	SNP	LRP5	missense	c.130	p.R44W	56	0	0	41	58	56.31
NM03	1	6	138417567	138417567	G	A	SNP	PERP	silent	c.279	p.I93	52	0	0	43	52	51.49
NM03	2	16	27556779	27556779	T	A	SNP	GTF3C1	missense	c.287	p.D96V	54	0	0	50	49	47.12
NM03	2	5	140348339	140348339	G	A	SNP	PCDHAC2	missense	c.1988	p.R663Q	77	0	0	109	77	40.31
NM03	2	6	4060872	4060872	G	C	SNP	PRPF4B	rna	NULL	NULL	102	0	0	79	77	47.53
NM03	3	1	159166757	159166757	C	T	SNP	CADM3	missense	c.961	p.P321S	50	0	0	76	67	46.53
NM03	3	5	176018323	176018323	C	T	SNP	CDHR2	nonsense	c.3652	p.R1218*	68	0	0	50	50	47.62
NM03	3	2	201853137	201853137	T	C	SNP	FAM126B	missense	c.839	p.N280S	77	0	0	100	97	47.32
NM03	3	9	133805003	133805003	A	T	SNP	FIBCD1	missense	c.503	p.L168Q	36	0	0	58	49	44.95
NM03	3	5	153056597	153056597	A	G	SNP	GRIA1	missense	c.935	p.E312G	81	0	0	98	80	43.24
NM03	3	13	101763031	101763031	C	T	SNP	NALCN	missense	c.2303	p.R768K	71	0	0	62	44	40
NM03	3	1	111931880	111931880	T	C	SNP	PGCP1	rna	NULL	NULL	120	0	0	143	117	44.15
NM03	3	12	42859012	42859012	G	A	SNP	PRICKLE1	missense	c.824	p.T275M	61	1	1.61	75	71	47.02
NM03	3	4	184605112	184605112	G	A	SNP	TRAPPC11	missense	c.10	p.V4I	68	0	0	74	78	49.06
NM03	4	11	75139508	75139508	G	C	SNP	KLHL35	missense	c.1045	p.R349G	87	0	0	48	59	53.64
NM03	4	6	167755081	167755081	G	A	SNP	TTLL2	missense	c.1693	p.A565T	56	0	0	28	27	45.76
NM04	1	9	114423923	114423923	C	T	SNP	GNG10	silent	c.9	p.S3	183	0	0	79	70	46.36
NM04	1	15	88420309	88420309	G	A	SNP	NTRK3	nonsense	c.2377	p.R793*	32	0	0	34	24	41.38
NM04	1	5	95735831	95735831	G	A	SNP	PCSK1	missense	c.1256	p.P419L	34	0	0	15	17	53.12
NM04	1	12	107155008	107155008	C	A	SNP	RFX4	missense	c.1996	p.P666T	63	0	0	34	23	40.35
NM04	1	1	45116726	45116728	GGG	-	DEL	RNF220	rna	NULL	NULL	74	0	0	43	24	35.82
NM04	2	6	151672215	151672215	G	A	SNP	AKAP12	missense	c.2689	p.A897T	46	1	2.13	34	35	49.3
NM04	2	2	217735635	217735635	T	C	SNP	ENSG00000223874	silent	c.66	p.C22	95	0	0	42	47	50.54
NM04	2	8	61471448	61471448	C	A	SNP	RAB2A	missense	c.84	p.D28E	35	0	0	41	26	37.68
NM04	2	17	3352408	3352408	G	A	SNP	SPATA22	missense	c.365	p.T122I	99	1	0.98	85	93	51.1
NM04	2	15	85113954	85113954	G	A	SNP	UBE2Q2P1	rna	NULL	NULL	157	0	0	73	66	44.9
NM04	3	10	50530617	50530617	A	T	SNP	C10orf71	silent	c.27	p.T9	64	2	2.99	39	41	50.62
NM04	3	14	37117121	37117121	C	A	SNP	ENSG00000258661	rna	NULL	NULL	111	0	0	93	95	48.72
NM04	3	21	19698860	19698860	C	G	SNP	TMPRSS15	missense	c.1810	p.D604H	97	0	0	74	60	42.25
NM04	4	5	7690959	7690959	C	T	SNP	ADCY2	rna	NULL	NULL	50	0	0	34	39	53.42

NM04	4	17	39633313	39633313	A	T	SNP	KRT35	missense	c.1363	p.F455I	128	1	0.78	98	109	52.15
NM04	4	16	2820440	2820440	C	G	SNP	SRRM2	missense	c.85	p.P29A	158	0	0	103	104	49.29
NM05	1	10	72434740	72434740	T	C	SNP	ADAMTS14	missense	c.511	p.C171R	36	0	0	32	32	50
NM05	1	9	139701454	139701454	C	T	SNP	KIAA1984	silent	c.1422	p.S474	79	0	0	33	22	40
NM05	1	1	200562799	200562799	C	T	SNP	KIF14	missense	c.2648	p.R883H	62	0	0	47	33	41.25
NM05	1	17	39156101	39156101	T	C	SNP	KRTAP3-2	missense	c.5	p.D2G	90	0	0	54	48	47.06
NM05	1	4	39290347	39290347	A	-	DEL	RFC1	rna	NULL	NULL	65	0	0	64	36	36
NM05	1	12	118469011	118469011	C	T	SNP	RFC5	silent	c.951	p.N317	49	0	0	35	41	53.25
NM05	1	19	52570086	52570086	C	T	SNP	ZNF841	missense	c.1049	p.C350Y	85	0	0	55	40	42.11
NM05	2	12	92814833	92814833	G	T	SNP	CLLU1OS	missense	c.259	p.H87N	43	0	0	31	51	62.2
NM05	2	21	33371202	33371202	C	A	SNP	HUNK	missense	c.1850	p.P617Q	40	0	0	35	24	40.68
NM05	2	11	55579055	55579055	C	T	SNP	OR5L1	missense	c.113	p.T38M	75	0	0	54	45	45
NM05	2	6	158924155	158924155	A	T	SNP	TULP4	missense	c.3460	p.M1154L	70	0	0	16	10	38.46
NM05	3	5	1216788	1216788	G	A	SNP	SLC6A19	missense	c.1003	p.D335N	50	0	0	21	35	61.4
NM05	3	3	33255452	33255452	G	T	SNP	SUSD5	nonsense	c.258	p.C86*	41	0	0	18	30	62.5
NM05	3	6	132892416	132892416	G	-	DEL	TAAR6	frame_shift_del	c.956	p.A321fs	53	0	0	32	24	42.86
NM05	4	15	51233728	51233728	G	A	SNP	AP4E1	missense	c.1033	p.V345I	91	0	0	86	80	47.9
NM06	1	7	75615063	75615063	G	T	SNP	POR	missense	c.1565	p.R522L	101	0	0	72	49	40.16
NM06	1	X	102192455	102192455	C	T	SNP	RAB40AL	missense	c.209	p.T70M	92	1	1.08	58	46	44.23
NM06	2	16	77469935	77469946	GTGTGTGTGTGG	-	DEL	ENSG00000216131	rna	NULL	NULL	46	0	0	18	6	25
NM06	2	17	45377915	45377915	G	A	SNP	ITGB3	missense	c.1985	p.R662H	107	0	0	52	49	48.04
NM06	3	14	78353575	78353575	G	A	SNP	ADCK1	missense	c.565	p.D189N	44	0	0	28	26	47.27
NM06	3	1	225492743	225492743	C	T	SNP	DNAH14	missense	c.8678	p.T2893I	87	0	0	50	55	52.38
NM06	3	5	158125661	158125661	T	A	SNP	EBF1	rna	NULL	NULL	35	1	2.78	12	6	33.33
NM06	3	19	5831554	5831554	T	C	SNP	FUT6	missense	c.1025	p.K342R	120	0	0	64	48	42.86
NM06	3	17	58040517	58040517	C	G	SNP	RNFT1	missense	c.185	p.C62S	70	0	0	25	28	52.83
NM06	3	19	41094601	41094601	G	A	SNP	SHKBP1	missense	c.1408	p.G470S	69	1	1.41	33	24	42.11
NM06	3	1	214503563	214503563	G	A	SNP	SMYD2	missense	c.758	p.R253Q	94	0	0	46	57	54.81
NM06	3	7	63537961	63537961	T	G	SNP	ZNF727	missense	c.534	p.C178W	37	0	0	10	27	72.97
NM06	4	16	1412110	1412110	G	A	SNP	GNPTG	missense	c.389	p.R130H	43	0	0	29	23	44.23
NM06	4	14	55518404	55518404	G	A	SNP	MAPK1IP1L	rna	NULL	NULL	59	1	1.67	42	33	43.42
SCN01	1	14	94725661	94725661	G	T	SNP	PPP4R4	missense	c.2082	p.L694F	66	0	0	36	23	36.51
SCN01	2	20	60912720	60912720	C	T	SNP	LAMA5	missense	c.2090	p.S697N	59	0	0	39	39	48.15
SCN01	2	10	21157652	21157652	C	T	SNP	NEBL	missense	c.625	p.A209T	39	0	0	42	49	51.58
SCN01	2	12	3389626	3389626	G	T	SNP	TSPAN9	missense	c.409	p.A137S	32	0	0	20	6	22.22
SCN01	3	17	61584467	61584467	C	T	SNP	ACE	nonsense	c.2020	p.Q674*	44	0	0	49	51	50.5
SCN01	3	5	149011589	149011589	G	A	SNP	ARHGEF37	silent	c.1863	p.V621	47	0	0	78	69	45.39
SCN01	3	22	40037034	40037034	C	T	SNP	CACNA1I	silent	c.903	p.Y301	35	0	0	47	43	46.74
SCN01	3	2	9568934	9568934	G	A	SNP	CPSF3	missense	c.91	p.E31K	111	1	0.88	144	121	43.84
SCN01	3	6	97247041	97247041	C	G	SNP	GPR63	missense	c.567	p.K189N	34	0	0	59	38	38.78
SCN01	3	1	37337837	37337837	G	A	SNP	GRIK3	silent	c.684	p.R228	34	0	0	38	31	43.66

SCN01	3	16	11570754	11570754	C	T	SNP	LOC101929274	silent	c.2493	p.G831	56	0	0	45	40	46.51
SCN01	3	22	38470364	38470364	C	A	SNP	PICK1	silent	c.885	p.R295	68	0	0	61	78	54.55
SCN02	1	2	27800163	27800163	C	T	SNP	C2orf16	missense	c.724	p.R242W	61	1	1.56	28	18	39.13
SCN02	1	17	78175584	78175584	G	A	SNP	CARD14	silent	c.1893	p.L631	101	0	0	49	44	46.81
SCN02	1	14	100375781	100375781	G	A	SNP	EML1	missense	c.1261	p.G421R	80	0	0	23	44	61.97
SCN02	1	1	155209460	155209460	G	A	SNP	GBA	missense	c.401	p.A134V	79	1	1.16	27	26	47.27
SCN02	1	9	115480348	115480348	T	C	SNP	INIP	rna	NULL	NULL	145	2	1.27	55	56	48.7
SCN02	1	14	23884612	23884612	T	A	SNP	MYH7	missense	c.5261	p.K1754M	62	0	0	23	27	50.94
SCN02	1	2	179528826	179528826	C	A	SNP	TTN	splice_site	c.36281-1	e168-1	63	1	1.52	37	21	32.31
SCN02	3	4	114274853	114274853	G	A	SNP	ANK2	silent	c.5079	p.Q1693	36	1	2.7	26	21	42.86
SCN02	4	21	28296460	28296460	C	T	SNP	ADAMTS5	missense	c.2705	p.C902Y	44	0	0	14	9	39.13
SCN02	4	10	111879261	111879261	G	A	SNP	ADD3	missense	c.899	p.G300E	144	0	0	43	65	60.19
SCN02	4	9	730244	730244	C	T	SNP	KANK1	silent	c.2892	p.L964	84	1	1.1	43	40	47.62
SCN02	4	8	23177505	23177505	T	G	SNP	LOXL2	missense	c.1363	p.N455H	135	0	0	37	25	39.06
SCN03	1	15	83395628	83395628	G	A	SNP	LOC283693	rna	NULL	NULL	57	0	0	33	17	34
SCN03	1	4	90857914	90857914	G	A	SNP	MMRN1	missense	c.3083	p.R1028Q	73	0	0	17	24	58.54
SCN03	1	14	72431527	72431527	G	T	SNP	RGS6	missense	c.19	p.D7Y	70	0	0	25	19	42.22
SCN03	2	19	15807256	15807256	G	A	SNP	CYP4F12	missense	c.1331	p.R444H	54	0	0	16	25	60.98
SCN03	2	2	232389899	232389899	C	T	SNP	NMUR1	missense	c.1136	p.R379H	74	0	0	47	29	38.16
SCN03	3	11	130286861	130286861	G	A	SNP	ADAMTS8	missense	c.1070	p.A357V	79	1	1.19	51	55	50.93
SCN03	3	2	27320515	27320515	G	A	SNP	KHK	missense	c.562	p.V188M	48	0	0	32	45	58.44
SCN03	3	11	121448064	121448064	G	-	DEL	SORL1	frame_shift_del	c.3535	p.D1180fs	109	0	0	54	76	58.46
SCN03	4	19	37618828	37618828	G	A	SNP	ZNF420	missense	c.935	p.C312Y	46	1	2.13	16	15	46.88
SCN04	2	17	66915591	66915591	G	A	SNP	ABCA8	missense	c.1639	p.L547F	79	0	0	38	18	32.14
SCN04	2	12	31249675	31249675	T	C	SNP	DDX11	silent	c.1626	p.T542	124	0	0	47	40	45.98
SCN04	2	1	74507363	74507363	G	A	SNP	LRRIQ3	nonsense	c.1252	p.R418*	93	0	0	43	50	53.19
SCN04	2	11	58207266	58207266	C	T	SNP	OR5B12	missense	c.359	p.R120H	97	0	0	57	46	44.66
SCN04	2	1	153043246	153043246	G	A	SNP	SPRR2B	missense	c.70	p.P24S	74	0	0	54	30	35.29
SCN04	2	1	47767942	47767942	A	G	SNP	STIL	missense	c.229	p.S77P	218	1	0.44	85	102	53.97
SCN04	3	6	70866083	70866083	G	A	SNP	COL19A1	missense	c.2144	p.G715D	96	0	0	33	36	51.43
SCN04	3	1	24484116	24484116	G	A	SNP	IFNLR1	missense	c.1067	p.A356V	72	0	0	33	26	42.62
SCN04	3	20	44641940	44641940	G	A	SNP	MMP9	silent	c.1377	p.P459	111	0	0	38	27	40.3
SCN05	1	19	18053472	18053474	AGG	-	DEL	CCDC124	in_frame_del	c.167_169	p.E57in_frame_del	63	1	1.56	94	83	46.89
SCN05	1	11	66358159	66358159	G	C	SNP	CCDC87	silent	c.2328	p.L776	59	0	0	52	48	45.71
SCN05	1	3	195609379	195609379	G	A	SNP	TNK2	missense	c.395	p.A132V	36	1	2.7	42	28	37.84
SCN05	2	2	148672767	148672767	G	T	SNP	ACVR2A	missense	c.536	p.G179V	64	1	1.33	37	31	45.59
SCN05	2	19	14006213	14006213	C	T	SNP	C19orf57	missense	c.178	p.G60R	67	2	2.6	46	43	48.31
SCN05	3	19	9071998	9071998	C	T	SNP	MUC16	missense	c.15448	p.A5150T	31	0	0	23	26	52
SCN05	3	1	233122144	233122144	C	G	SNP	PCNXL2	missense	c.5934	p.Q1978H	34	0	0	36	47	55.29
SCN05	3	15	25448455	25448455	C	T	SNP	SNORD115-18	rna	NULL	NULL	52	0	0	73	61	44.85
SCN05	4	1	201868546	201868546	G	A	SNP	LMOD1	missense	c.1595	p.A532V	92	0	0	61	60	49.59

SCN05	4	6	76582975	76582975	C	T	SNP	MYO6	missense	c.2035	p.H679Y	74	0	0	60	47	43.52
SCN05	4	8	71068901	71068901	C	A	SNP	NCOA2	splice_site	c.NULL	NULL	36	0	0	36	32	47.06
SCN06	1	15	56665656	56665656	T	C	SNP	TEX9	missense	c.137	p.L46S	185	0	0	151	114	42.54
SCN06	2	4	110788917	110788917	G	A	SNP	LRIT3	missense	c.710	p.R237H	92	0	0	38	42	50.6
SCN06	2	8	57079060	57079060	T	C	SNP	PLAG1	silent	c.1245	p.A415	103	0	0	37	34	46.58
SCN06	2	3	58351615	58351615	A	C	SNP	PXK	silent	c.117	p.R39	138	0	0	76	56	40.58
SCN06	2	4	186544635	186544635	G	A	SNP	SORBS2	missense	c.1936	p.H646Y	38	0	0	12	10	45.45
SCN06	2	4	183714508	183714508	G	A	SNP	TENM3	missense	c.6683	p.R2228H	55	0	0	21	31	55.36
SCN06	2	2	179641019	179641019	C	T	SNP	TTN	missense	c.5572	p.A1858T	72	1	1.3	42	34	41.98
SCN06	3	1	150621135	150621135	T	C	SNP	GOLPH3L	missense	c.520	p.T174A	49	0	0	25	33	53.23
SCN06	3	20	61711352	61711352	G	C	SNP	LOC63930	rna	NULL	NULL	55	0	0	49	71	58.2
SCN06	3	10	105331334	105331334	C	T	SNP	NEURL	missense	c.404	p.P135L	82	0	0	64	91	56.88
SCN06	3	6	169629697	169629697	G	A	SNP	THBS2	silent	c.2229	p.D743	61	0	0	49	67	55.83
SCN06	4	6	109854608	109854608	C	T	SNP	AK9	missense	c.3416	p.R1139H	77	0	0	45	57	55.34
SCN06	4	4	123664650	123664650	G	A	SNP	BBS12	missense	c.1603	p.V535I	61	0	0	24	32	57.14
SCN06	4	4	2446991	2446991	A	T	SNP	ENSG00000206113	rna	NULL	NULL	167	0	0	127	95	42.04
SCN06	4	1	62574141	62574141	A	C	SNP	INADL	missense	c.4410	p.E1470D	122	0	0	88	71	44.38
SCN06	4	14	65438790	65438790	G	A	SNP	RAB15	missense	c.5	p.A2V	68	1	1.39	55	50	47.17
SCN06	4	9	125752384	125752384	A	G	SNP	RABGAP1	missense	c.815	p.K272R	87	1	1.06	59	42	40
SCN06	4	3	77147316	77147316	G	A	SNP	ROBO2	silent	c.213	p.V71	56	0	0	37	29	43.94
SCN06	4	22	24896172	24896172	C	T	SNP	UPB1	nonsense	c.202	p.R68*	42	0	0	27	27	50
SCN06	4	3	44489660	44489660	C	T	SNP	ZNF445	silent	c.1503	p.A501	44	0	0	25	35	57.38
SCN06	4	10	44112575	44112575	G	C	SNP	ZNF485	missense	c.1084	p.A362P	42	0	0	23	41	62.12
SCN07	1	17	65014375	65014375	G	A	SNP	CACNG4	silent	c.291	p.S97	43	0	0	16	28	60.87
SCN07	1	12	3131018	3131018	G	A	SNP	TEAD4	missense	c.499	p.A167T	57	0	0	31	31	47.69
SCN07	2	1	110173061	110173061	C	T	SNP	AMPD2	nonsense	c.2263	p.R755*	55	0	0	19	19	50
SCN07	3	17	48268769	48268769	C	A	SNP	COL1A1	missense	c.2210	p.G737V	70	0	0	56	49	46.23
SCN07	3	X	129205311	129205311	C	A	SNP	ELF4	missense	c.616	p.G206C	46	0	0	37	27	42.19
SCN07	3	19	18572457	18572457	C	G	SNP	ELL	silent	c.675	p.L225	127	0	0	85	70	44.87
SCN07	3	X	135289344	135289344	T	A	SNP	FHL1	missense	c.326	p.V109E	31	0	0	19	18	48.65
SCN07	3	16	29899008	29899008	C	A	SNP	SEZ6L2	missense	c.185	p.C62F	64	0	0	39	33	44.59
SCN07	4	14	24898127	24898127	C	A	SNP	CBLN3	missense	c.134	p.C45F	76	0	0	27	28	48.28
SCN07	4	10	7627894	7627894	C	A	SNP	ITIH5	missense	c.1078	p.V360L	116	0	0	72	42	35.59
SCN07	4	10	74806764	74806764	C	T	SNP	P4HA1	nonsense	c.996	p.W332*	105	0	0	28	41	56.16
SCN07	4	17	58259083	58259083	C	T	SNP	USP32	missense	c.4150	p.G1384R	133	1	0.7	59	48	42.11
SCN11	1	17	3336673	3336673	C	T	SNP	OR1E2	missense	c.463	p.A155T	111	0	0	39	49	55.68
SCN11	1	19	43349207	43349207	C	A	SNP	PSG10P	rna	NULL	NULL	373	0	0	153	148	49.17
SCN11	1	8	145096220	145096220	G	A	SNP	SPATC1	missense	c.1394	p.R465H	131	1	0.75	48	39	44.32
SCN11	2	2	234160505	234160505	C	T	SNP	ATG16L1	missense	c.32	p.P11L	98	0	0	25	37	56.92
SCN11	2	5	41904477	41904477	C	T	SNP	C5orf51	missense	c.8	p.A3V	141	0	0	56	46	43.4
SCN11	2	9	108380266	108380266	C	T	SNP	FKTN	missense	c.937	p.P313S	111	1	0.88	93	83	46.63
SCN11	2	12	70273192	70273192	A	T	SNP	MYRFL	silent	c.423	p.I141	87	0	0	43	37	45.68
SCN11	2	8	144887433	144887433	C	T	SNP	SCRIB	missense	c.2519	p.R840Q	45	0	0	22	19	44.19
SCN11	2	2	187703831	187703831	G	A	SNP	ZSWIM2	nonsense	c.349	p.Q117*	172	0	0	55	54	47.37

SCN11	3	11	6578093	6578093	C	T	SNP	DNHD1	missense	c.7568	p.T2523I	40	0	0	11	18	62.07
SCN11	3	7	123509187	123509187	G	T	SNP	HYAL4	missense	c.860	p.R287M	112	0	0	54	57	51.35
SCN11	3	8	42742870	42742870	C	G	SNP	RNF170	splice_site	c.137+1	e1+1	116	0	0	72	64	47.06
SCN11	4	3	119305233	119305233	C	A	SNP	ADPRH	silent	c.400	p.R134	57	0	0	21	27	56.25
SCN11	4	4	113362249	113362249	A	G	SNP	ALPK1	missense	c.3715	p.M1239V	106	0	0	34	52	60.47
SCN11	4	11	2411735	2411735	G	A	SNP	CD81	missense	c.160	p.A54T	44	0	0	24	13	35.14
SCN11	4	10	22022938	22022938	T	C	SNP	MLLT10	missense	c.2738	p.V913A	107	0	0	27	32	54.24
SCN11	4	14	64989741	64989741	A	C	SNP	ZBTB1	missense	c.1519	p.M507L	80	0	0	36	30	45.45
SCN12	1	17	35578722	35578722	G	A	SNP	ACACA	silent	c.3606	p.S1202	125	0	0	60	61	50
SCN12	1	1	183084662	183084662	A	G	SNP	LAMC1	silent	c.1218	p.L406	76	0	0	54	47	45.63
SCN12	1	15	48427121	48427121	C	G	SNP	SLC24A5	missense	c.530	p.A177G	85	0	0	42	35	44.87
SCN12	1	10	123976384	123976384	G	A	SNP	TACC2	silent	c.327	p.S109	53	0	0	32	33	50.77
SCN12	2	17	58678206	58678206	A	G	SNP	PPM1D	missense	c.431	p.K144R	40	0	0	51	31	36.05
SCN12	2	14	94733300	94733300	A	T	SNP	PPP4R4	missense	c.2383	p.S795C	103	0	0	99	84	43.75
SCN12	2	9	112207555	112207555	T	A	SNP	PTPN3	missense	c.431	p.N144I	68	0	0	75	75	48.39
SCN12	2	11	67401832	67401832	C	T	SNP	TBX10	splice_site	c.378-1	e4-1	113	0	0	68	79	51.63
SCN12	3	9	72459533	72459533	G	C	SNP	C9orf135	missense	c.253	p.E85Q	102	0	0	48	32	35.56
SCN12	3	8	62212688	62212688	G	A	SNP	CLVS1	missense	c.302	p.R101H	68	0	0	31	28	43.75
SCN12	3	2	206605325	206605325	G	A	SNP	NRP2	missense	c.1229	p.R410H	77	0	0	36	35	45.45
SCN12	3	3	146171929	146171929	C	G	SNP	PLSCR2	missense	c.343	p.G115R	70	0	0	24	36	60
SCN12	3	7	38812163	38812163	C	T	SNP	VPS41	missense	c.1087	p.D363N	65	0	0	31	30	43.48
SCN12	4	19	51022148	51022148	G	A	SNP	LRRC4B	silent	c.822	p.D274	84	0	0	78	52	39.69
SCN12	4	17	40939796	40939796	C	T	SNP	WNK4	missense	c.1742	p.S581L	59	0	0	47	33	41.25
SCN13	1	7	92098567	92098567	C	A	SNP	ERVW-1	missense	c.1129	p.A377S	45	1	2.08	24	8	22.86
SCN13	1	12	31135513	31135513	C	T	SNP	TSPAN11	missense	c.503	p.T168M	84	0	0	36	39	50
SCN13	1	9	96000542	96000542	G	A	SNP	WNK2	missense	c.1261	p.V421M	94	0	0	67	38	34.23
SCN13	2	15	49716584	49716584	A	G	SNP	FGF7	silent	c.90	p.L30	52	0	0	24	27	48.21
SCN13	2	2	176996134	176996134	A	C	SNP	HOXD8	missense	c.667	p.T223P	66	0	0	21	18	42.86
SCN13	2	1	237732457	237732457	C	T	SNP	RYR2	missense	c.3430	p.H1144Y	104	0	0	40	35	43.21
SCN13	3	11	26677951	26677951	A	G	SNP	ANO3	missense	c.2686	p.S896G	48	0	0	40	46	52.27
SCN13	3	20	20232324	20232324	C	T	SNP	C20orf26	nonsense	c.2245	p.R749*	41	0	0	38	23	37.1
SCN13	3	15	79229752	79229752	T	C	SNP	CTSH	missense	c.137	p.Y46C	49	0	0	56	58	49.57
SCN13	3	16	70690880	70690880	C	G	SNP	IL34	missense	c.258	p.S86R	63	0	0	32	33	50.77
SCN13	3	9	6007253	6007253	A	C	SNP	KIAA2026	missense	c.535	p.S179A	42	0	0	12	18	60
SCN22	1	2	220038190	220038190	C	T	SNP	CNPPD1	splice_site	c.573-1	e7-1	55	0	0	3	9	75
SCN22	1	2	165404193	165404193	T	C	SNP	GRB14	missense	c.458	p.E153G	83	0	0	18	12	38.71
SCN22	1	2	24297015	24297015	G	A	SNP	SF3B14	missense	c.80	p.P27L	71	0	0	6	13	65
SCN22	1	1	205890872	205890872	G	A	SNP	SLC26A9	missense	c.1877	p.T626I	106	0	0	25	10	28.57
SCN22	2	2	238243319	238243319	C	T	SNP	COL6A3	missense	c.9179	p.C3060Y	112	0	0	40	57	58.76
SCN22	2	6	110110879	110110879	G	A	SNP	FIG4	missense	c.2179	p.G727R	209	1	0.48	96	93	48.44
SCN22	2	14	106641726	106641726	C	G	SNP	IGHV1-18	missense	c.188	p.G63A	141	1	0.7	65	65	49.24
SCN22	2	6	39282954	39282954	T	C	SNP	KCNK16	silent	c.915	p.P305	163	1	0.61	57	55	48.25
SCN22	2	1	169762650	169762659	GTTCCCACAA	-	DEL	METTL18	frame_shift_del	c.187_178	p.L60fs	89	0	0	54	33	37.93

SCN22	2	2	43973102	43973102	G	A	SNP	PLEKHH2	missense	c.3653	p.R1218K	105	0	0	56	67	54.03
SCN22	2	11	48166638	48166638	G	A	SNP	PTPRJ	missense	c.2873	p.S958N	90	0	0	60	50	45.05
SCN22	2	4	183710350	183710350	G	A	SNP	TENM3	silent	c.5409	p.T1803	85	2	2.22	46	46	50
SCN22	2	19	38377660	38377660	A	G	SNP	WDR87	silent	c.6651	p.D2217	73	1	1.33	39	32	45.07
SCN22	3	1	26517240	26517240	G	A	SNP	CATSPER4	missense	c.122	p.R41H	52	0	0	24	22	47.83
SCN22	3	20	61950462	61950462	C	T	SNP	COL20A1	missense	c.2716	p.R906C	133	0	0	52	56	51.85
SCN22	3	20	62198835	62198835	C	G	SNP	HELZ2	missense	c.1876	p.D626H	111	0	0	37	30	44.78
SCN22	3	16	84695191	84695191	T	G	SNP	KLHL36	missense	c.1303	p.Y435D	68	0	0	30	42	58.33
SCN22	3	6	51798909	51798909	G	A	SNP	PKHD1	silent	c.6120	p.H2040	63	0	0	18	15	45.45
SCN22	3	12	130015716	130015716	C	T	SNP	TMEM132D	missense	c.1003	p.V335M	79	0	0	24	14	36.84
SCN22	4	11	65319581	65319581	T	G	SNP	LTBP3	missense	c.1393	p.T465P	71	0	0	25	24	48
SCN22	4	X	26235696	26235696	T	C	SNP	MAGEB5	missense	c.278	p.F93S	59	0	0	27	27	50
SCN22	4	15	52539658	52539658	G	A	SNP	MYO5C	silent	c.1878	p.S626	51	0	0	22	16	40
SCN22	4	14	77697962	77697962	G	A	SNP	TMEM63C	missense	c.382	p.D128N	75	0	0	35	41	52.56
SCN22	4	6	142490776	142490776	G	A	SNP	VTA1	silent	c.297	p.L99	213	0	0	96	100	50.51
SDS16	1	19	44832052	44832052	C	G	SNP	ZFP112	missense	c.2327	p.S776T	142	0	0	91	90	49.7
SDS16	1	2	179410354	179410354	C	T	SNP	TTN	nonsense	c.68864	p.W22955*	134	0	0	87	76	46.6
SDS16	2	10	101985714	101985714	C	T	SNP	CHUK	missense	c.166	p.E56K	51	0	0	22	29	56.9
SDS16	3	12	7864241	7864241	C	A	SNP	DPPA3	missense	c.75	p.D25E	104	0	0	52	50	49
SDS35	1	9	990686	990686	G	A	SNP	DMRT3	missense	c.1100	p.R367Q	182	0	0	79	60	43.2
SDS35	2	1	117122352	117122352	C	T	SNP	IGSF3	missense	c.3056	p.G1019E	124	0	0	53	47	47
SDS35	2	16	56875711	56875711	C	A	SNP	NUP93	missense	c.2315	p.S772Y	57	0	0	22	19	46.3
SDS35	2	20	34089737	34089737	A	G	SNP	CEP250	missense	c.3964	p.M1322V	32	0	0	15	17	53.1
SDS35	2	3	119445086	119445086	G	A	SNP	C3orf15	missense	c.751	p.E251K	41	0	0	19	18	48.6
SDS35	3	8	135669924	135669924	C	A	SNP	ZFAT	nonsense	c.76	p.E26*	41	0	0	19	30	61.2
SDS35	4	1	158641152	158641152	G	T	SNP	SPTA1	missense	c.1580	p.A527D	12	0	0	9	7	43.8
SDS35	4	19	10247890	10247890	C	T	SNP	DNMT1	missense	c.4360	p.D1454N	19	0	0	13	10	43.5
SDS35	4	6	86351101	86351101	A	T	SNP	SYNCRIP	silent	c.57	p.S19	15	0	0	5	5	50

HPC: hematopoietic progenitor cell; Chr: chromosome; Ref: reference allele; Var: variant allele; VAF: variant allele frequency

Unfractionated bone marrow mononuclear cells (WBC) were sequenced and represent the "normal" sample to call somatic variants

Table S5. Somatic variants associated with clonal hematopoiesis

Study ID	Chr	start	stop	Ref	Var	Gene	Mutation type	cDNA position	amino acid change	Ref count	Variant count	VAF	ExAC adj AF	Cosmic
CN09	1	36932296	36932296	G	A	*CSF3R*	nonsense	c.2173	p.Q702*	3179	16	0.005	0	COSM4219286
CN09	1	36932209	36932209	G	A	*CSF3R*	nonsense	c.2260	p.Q731*	3162	22	0.0069	0	COSM4173043
CN09	1	36932281	36932281	G	A	*CSF3R*	nonsense	c.2188	p.Q707*	3171	38	0.0118	0	COSM4172543
CN09	1	36932254	36932254	G	A	*CSF3R*	nonsense	c.2215	p.Q716*	3004	59	0.0192	0	COSM1168011
CN09	12	25398281	25398281	C	T	*KRAS*	missense	c.38	p.G13D	1084	21	0.019	0	COSM532
CN10	6	43305245	43305245	C	A	*ZNF318*	missense	c.6491	p.G2164V	693	3	0.0043	0	
CN14	2	198264986	198264986	G	T	*SF3B1*	missense	c.2891	p.T964N	1543	4	0.0026	0	
CN16	X	129149759	129149759	C	A	*BCORL1*	missense	c.3011	p.P1004H	1878	4	0.0021	0	
CN18	4	106164792	106164792	C	A	*TET2*	missense	c.3569	p.P1190H	2727	4	0.0015	0	
NM01	2	25470548	25470548	C	T	*DNMT3A*	missense	c.926	p.R309H	637	32	0.0478	0	
NM14	11	64535613	64535613	G	A	*SF1*	missense	c.1033	p.R345C	2836	8	0.0028	0	COSM1509537
NM15	7	101892152	101892152	C	A	*CUX1*	missense	c.4381	p.P1461T	1955	4	0.002	0	
NM19	4	106180840	106180840	T	C	*TET2*	missense	c.3868	p.S1290P	4032	5	0.0012	0	COSM4383924
NM20	11	108165749	108165749	T	A	*ATM*	missense	c.4872	p.H1624Q	3819	4	0.001	0	
SCN01	1	36932213	36932213	A	C	*CSF3R*	nonsense	c.2256	p.Y729*	7199	111	0.0152	0	COSM4172579
SCN02	1	36932224	36932224	G	A	*CSF3R*	nonsense	c.2245	p.Q726*	6949	9	0.0013	0	COSM1724982
SCN02	1	36932213	36932213	A	T	*CSF3R*	nonsense	c.2256	p.Y729*	6917	50	0.0072	0	COSM4172579
SCN04	X	76939445	76939445	T	C	*ATRX*	missense	c.1303	p.I435V	2878	5	0.0017	0.00018	
SCN04	1	36932224	36932224	G	A	*CSF3R*	nonsense	c.2245	p.Q726*	4163	10	0.0024	0	COSM1724982
SCN05	11	108181037	108181037	G	T	*ATM*	missense	c.5913	p.E1971D	1740	44	0.0247	0	
SCN05	1	36932224	36932224	G	A	*CSF3R*	nonsense	c.2245	p.Q726*	3622	13	0.0036	0	COSM1724982
SCN05	3	47163303	47163303	C	A	*SETD2*	missense	c.2823	p.K941N	2557	5	0.002	0	
SCN06	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	1198	20	0.0164	0	COSM4172565
SCN07	1	36932242	36932242	G	A	*CSF3R*	nonsense	c.2227	p.Q720*	4284	290	0.0634	0	COSM4172525
SCN13	11	108205783	108205783	A	G	*ATM*	missense	c.8098	p.K2700E	2241	4	0.0018	0	
SCN14	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	7231	19	0.0026	0	COSM4172565
SCN15	1	36932325	36932325	G	C	*CSF3R*	nonsense	c.2144	p.S692*	7879	11	0.0014	0	COSM4340576
SCN15	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	8271	1410	0.1456	0	COSM4172565
SCN22	20	31023667	31023667	G	A	*ASXL1*	missense	c.3152	p.R1051H	1429	6	0.0042	8.1E-06	COSM5486129
SCN22	8	117870603	117870603	C	A	*RAD21*	nonsense	c.469	p.E157*	870	252	0.2246	0	
SCN24	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	4723	204	0.0414	0	COSM4172565
SCN24	1	36932209	36932209	G	A	*CSF3R*	nonsense	c.2260	p.Q731*	4884	43	0.0087	0	COSM4173043
SCN26	7	101848448	101848448	C	T	CUX1	missense	c.3161	p.S1054L	3033	188	0.0584	0.00086	
SCN27	11	108137904	108137904	T	G	*ATM*	missense	c.2473	p.F825V	815	6	0.0073	0	
SCN28	11	108137904	108137904	T	G	*ATM*	missense	c.2473	p.F825V	1794	6	0.0033	0	
SCN36	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	5253	23	0.0044	0	COSM4172565
SCN38	2	198288636	198288636	C	A	*SF3B1*	missense	c.91	p.V31L	1352	4	0.0029	0	

SCN39	2	25505399	25505399	C	A	*DNMT3A*	missense	c.359	p.G120V	2860	5	0.0017	0	
SCN39	12	112926951	112926951	C	A	*PTPN11*	missense	c.1571	p.T524K	830	4	0.0048	0	
SCN42	1	36932209	36932209	G	A	*CSF3R*	nonsense	c.2260	p.Q731*	3698	102	0.0268	0	COSM4173043
SCN45	12	25398281	25398281	C	T	*KRAS*	missense	c.38	p.G13D	939	4	0.0042	0	COSM532
SCN47	12	112926884	112926884	T	G	*PTPN11*	missense	c.1504	p.S502A	3418	4	0.0012	0	COSM20901
SCN48	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	4620	6	0.0013	0	COSM4172565
SCN48	1	36932224	36932224	G	A	*CSF3R*	nonsense	c.2245	p.Q726*	4688	12	0.0026	0	COSM1724982
SCN48	1	36932209	36932209	G	A	*CSF3R*	nonsense	c.2260	p.Q731*	4559	10	0.0022	0	COSM4173043
SCN48	20	57430276	57430276	C	A	*GNAS*	missense	c.1766	p.P589H	2568	4	0.0016	0	
SCN48	12	112915455	112915455	T	C	*PTPN11*	missense	c.854	p.F285S	1511	19	0.0124	0	COSM5424306
SCN50	1	36932224	36932224	G	A	*CSF3R*	nonsense	c.2245	p.Q726*	6601	419	0.0597	0	COSM1724982
SCN51	20	31022589	31022589	C	T	*ASXL1*	nonsense	c.2074	p.Q692*	2554	44	0.0169	0	COSM1155830
SCN51	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	3428	7	0.002	0	COSM4172565
SCN51	1	36932242	36932242	G	A	*CSF3R*	nonsense	c.2227	p.Q720*	2090	1338	0.3903	0	COSM4172525
SCN51	2	25462018	25462018	T	C	*DNMT3A*	missense	c.2389	p.N797D	7497	202	0.0262	0	COSM5878752
SCN52	1	36932248	36932248	G	A	*CSF3R*	nonsense	c.2221	p.Q718*	2753	717	0.2066	0	COSM4172565
SCN53	1	36932209	36932209	G	A	*CSF3R*	nonsense	c.2260	p.Q731*	3025	72	0.0232	0	COSM4173043
SDS05	2	25457290	25457290	C	G	*DNMT3A*	splice_site	c.2598-1	e22-1	1986	8	0.004	8.3E-06	
SDS05	1	115256445	115256445	G	A	*NRAS*	missense	c.266	p.S89L	3420	15	0.0044	0	
SDS05	16	30745826	30745826	C	T	*SRCAP*	nonsense	c.6619	p.R2207*	6900	28	0.004	0	
SDS05	17	7578479	7578479	G	A	*TP53*	missense	c.451	p.P151S	3127	12	0.0038	0	COSM10905
SDS05	17	7578406	7578406	C	T	*TP53*	missense	c.524	p.R175H	4898	9	0.0018	8.1E-06	COSM10648
SDS05	17	7578403	7578403	C	T	*TP53*	missense	c.527	p.C176Y	4862	45	0.0092	8.1E-06	COSM10687
SDS05	17	7578394	7578394	T	C	*TP53*	missense	c.536	p.H179R	5020	30	0.0059	0	COSM10889
SDS05	17	7577544	7577544	A	T	*TP53*	missense	c.737	p.M246K	7325	4	0.0005	0	COSM44103
SDS06	17	7578404	7578404	A	T	*TP53*	missense	c.526	p.C176S	2846	7	0.0025	0	COSM44146
SDS06	17	7578271	7578271	T	C	*TP53*	missense	c.578	p.H193R	2879	10	0.0035	0	COSM10742
SDS06	17	7578190	7578190	T	C	*TP53*	missense	c.659	p.Y220C	2781	5	0.0018	2.5E-05	COSM10758
SDS06	17	7577568	7577568	C	A	*TP53*	missense	c.713	p.C238F	3623	11	0.003	0	COSM43778
SDS07	16	3786748	3786748	G	A	*CREBBP*	missense	c.4463	p.P1488L	3463	13	0.0037	0	COSM3356828
SDS07	17	7579350	7579350	A	C	*TP53*	missense	c.337	p.F113V	5242	76	0.0143	0	COSM11498
SDS07	17	7578190	7578190	T	C	*TP53*	missense	c.659	p.Y220C	4659	9	0.0019	2.5E-05	COSM10758
SDS08	7	148515059	148515059	C	A	*EZH2*	missense	c.1150	p.D384Y	2196	4	0.0018	0	
SDS08	17	7577580	7577580	T	C	*TP53*	missense	c.701	p.Y234C	5398	3	0.0006	8.1E-06	COSM10725
SDS09	16	30732222	30732222	C	T	SRCAP	missense	c.3176	p.P1059L	700	5	0.0071	0.00013	
SDS11	16	30732222	30732222	C	T	SRCAP	missense	c.3176	p.P1059L	873	7	0.008	0.00013	
SDS13	17	7573009	7573009	C	T	*TP53*	splice_site	c.1101-1	e10-1	2049	15	0.0073	0	likely pathogenic
SDS13	17	7578532	7578532	A	T	*TP53*	missense	c.398	p.M133K	1400	19	0.0134	0	COSM11781
SDS13	17	7577539	7577539	G	A	*TP53*	missense	c.742	p.R248W	1683	6	0.0036	8.1E-06	COSM6545
SDS15	X	76939445	76939445	T	C	ATRX	missense	c.1303	p.I435V	2867	5	0.0017	0.00018	
SDS15	17	7577538	7577538	C	T	*TP53*	missense	c.743	p.R248Q	7691	8	0.001	5.7E-05	COSM10662

SDS19	17	7578478	7578478	G	T	*TP53*	missense	c.452	p.P151H	1464	33	0.022	0	COSM11476
SDS19	17	7578203	7578203	C	T	*TP53*	missense	c.646	p.V216M	3818	6	0.0016	0	COSM10667
SDS19	17	7577550	7577550	C	A	*TP53*	missense	c.731	p.G244V	2591	104	0.0386	0	COSM43652
SDS21	15	90645507	90645507	C	A	*IDH2*	splice_site	c.115+1	e1+1	2536	5	0.002	0	
SDS22	17	7578393	7578393	A	T	*TP53*	missense	c.537	p.H179Q	3748	31	0.0082	0	COSM44214
SDS25	13	28602366	28602366	G	T	*FLT3*	missense	c.2002	p.L668M	693	56	0.0748	8.2E-06	
SDS25	17	7578394	7578394	T	C	*TP53*	missense	c.536	p.H179R	1321	5	0.0038	0	COSM10889
SDS31	X	53426564	53426564	T	C	*SMC1A*	missense	c.2509	p.M837V	1690	7	0.0041	0	
SDS31	17	7573009	7573009	C	T	*TP53*	splice_site	c.1101-1	e10-1	3066	88	0.0279	0	likely pathogenic
SDS31	17	7579310	7579310	A	C	*TP53*	splice_site	c.375+2	e3+2	1522	7	0.0046	0	COSM46043
SDS31	17	7578532	7578532	A	T	*TP53*	missense	c.398	p.M133K	1630	50	0.0298	0	COSM11781
SDS31	17	7578395	7578395	G	A	*TP53*	missense	c.535	p.H179Y	2398	15	0.0062	0	COSM10768
SDS31	17	7577610	7577610	T	C	*TP53*	splice_site	c.673-2	e6-2	2129	3	0.0014	0	COSM25225
SDS31	17	7577574	7577574	T	C	*TP53*	missense	c.707	p.Y236C	3234	4	0.0012	0	COSM10731
SDS31	17	7577539	7577539	G	A	*TP53*	missense	c.742	p.R248W	2606	16	0.0061	8.1E-06	COSM6545
SDS31	17	7577511	7577511	A	T	*TP53*	missense	c.770	p.L257Q	3637	6	0.0016	0	COSM43530
SDS32	17	7577094	7577094	G	A	*TP53*	missense	c.844	p.R282W	2762	88	0.0309	1.6E-05	COSM10704
SDS33	17	7578406	7578406	C	T	*TP53*	missense	c.524	p.R175H	9016	12	0.0013	8.1E-06	COSM10648
SDS33	17	7577610	7577610	T	C	*TP53*	splice_site	c.673-2	e6-2	5707	7	0.0012	0	COSM25225
SDS33	17	7577580	7577580	T	C	*TP53*	missense	c.701	p.Y234C	6529	14	0.0021	8.1E-06	COSM10725
SDS34	17	7577538	7577538	C	T	*TP53*	missense	c.743	p.R248Q	3909	130	0.0322	5.7E-05	COSM10662

Chr: chromosome; Ref: reference; Var: variant; VAF: variant allele frequency

ExAC adj AF: Exome Aggregation Consortium adjusted allele frequency (http://exac.broadinstitute.org)

Cosmic: Shown is the identification number (if present) in the Catalogue of somatic mutations in cancer (COSMIC) database (http://cancer.sanger.ac.uk/cosmic)

For *CSF3R*, transcript *CSF3R*-205 (ENT00000373106.5) was used to align mutations. Per convention, the 23 amino acid signal peptide was removed to generate the final amino acid position of the mutation

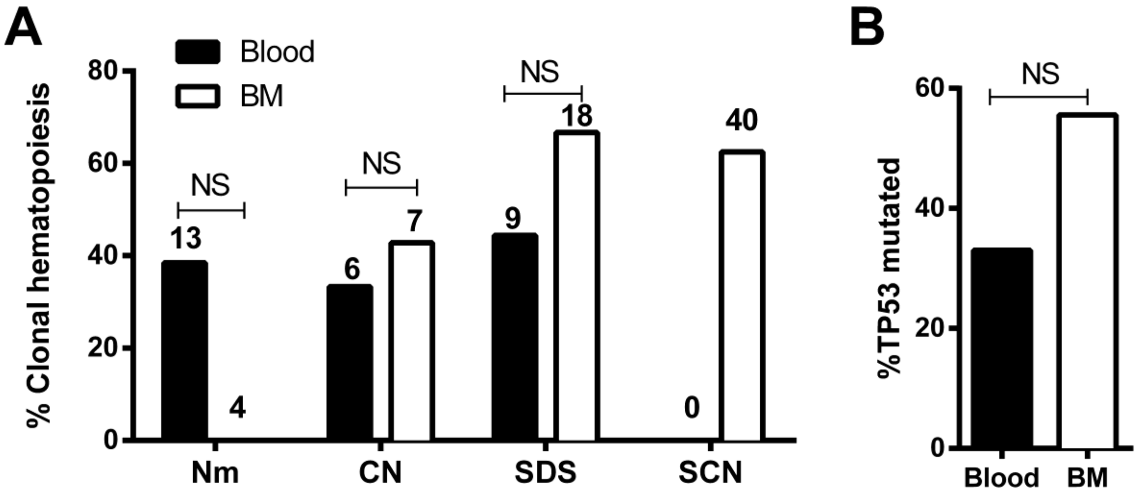

Figure S1 Tissue origin has no consistent effect on the prevalence of clonal hematopoiesis. (A) The percentage of cases with clonal hematopoiesis due to any mutation is shown based on whether a blood or bone marrow sample was analyzed. The numbers above the bars indicate the number of patients analyzed. All of the SCN samples were from the bone marrow. Nm: healthy donors; CN: cyclic neutropenia; SDS: Shwachman Diamond syndrome; and SCN: severe congenital neutropenia. (B) Percentage of patients with SDS who had one or more *TP53* mutation is shown.