

**Supplementary Table S1.** Main clinical characteristics of patients enrolled as validation cohort.

Patient	Age (years)	BM fibrosis	Splenomegaly	Hb (g/dL)	WBC (x10 <sup>9</sup> /L)	PLTs (x10 <sup>9</sup> /L)
FI	71	II	grade II	9.6	10.57	126
PS	65	II	grade II	8.8	42	303
GG	75	II	grade II	9.2	4.7	152
CE	58	I	grade IV	12	15	150
CL	70	II	grade II	9.8	5.4	83
BV	81	I	grade II	12	13.5	196
VM	73	II	grade IV	8.7	26	170
DMFG	56	II	grade II	9.4	45	123
CGD	77	II	grade IV	9.5	10.5	398

**Supplementary Table S2.** General statistics for WES analysis

Sample	# Mate Pairs	% Mapped Reads (on total)	# base pair	Theoretical Coverage	Mean coverage	% of Exome covered at least 1X	% of Exome covered at least 10X	% base >Q30	Mean quality Score
GT_G	20.122.276	98,36%	4024455200	60X	31X	87,07%	76,93%	90,97	35,58
GT_L	15.862.059	98,35%	3172411800	48X	25X	86,94%	73,46%	90,87	35,55
GT_S	14.407.376	97,05%	2881475200	43X	24X	85,70%	71,17%	91,25	35,67
GB_G	10.306.336	98,11%	2061267200	31X	16X	86,39%	61,20%	94,37	36,64
GB_L	8.445.568	98,11%	1689113600	25X	14X	85,93%	52,12%	94,31	36,63
GB_S	7.025.907	96,24%	1405181400	21X	12X	85,60%	44,17%	94,56	36,69
CC_G	8.718.111	98,26%	1743622200	26X	14X	86,32%	52,37%	94,08	36,53
CC_L	7.754.762	98,30%	1550952400	23X	12X	85,94%	46,81%	94,15	36,59
CC_S	8.619.081	97,48%	1723816200	26X	14X	86,22%	54,18%	94,39	36,62

G: granulocytes  
L: lymphocytes  
S: saliva

**Supplementary Table S3.** SNVs recorded by WES

<b>SAMPLES</b>	<b># TOTAL SNV's called</b>	<b># PASSED FILTER SNV's</b>	<b>Ti/T v ratio</b>	<b># SOMATIC SNV's</b>	<b># EXONIC/SPLICING SNV's*</b>	<b># NON SYNONIMOUS SNV's</b>	<b># NOVEL SNV's *</b>
GT_G_L	415.579	126.369	1,82	45.969	3.173	1.540	80
GB_G_L	313.077	104.687	1,86	21.980	1.787	857	40
CC_G_L	282.391	102.479	1,94	17.077	1.283	603	26

\*NOT IN DBSNP AND  
1000GENOME

Supplementary Table S4. Novel SNVs

Chr	Start	End	Ref	Alt	Func.refGene	Gene.refGene	Recurrency/case	ExonicFunc.refGene	AAChange.refGene	cosmic64
5	1405545 71	1405545 71	C	T	exonic	<i>PCDHB7</i>	1	nonsynonymous SNV	PCDHB7:NM_018940:exon1:c.C2155T:p.P719S	ID=COSM370895;OCCURENCE=1(lung)
4	1201073 11	1201073 11	G	A	exonic	<i>MYOZ2</i>	1	nonsynonymous SNV	MYOZ2:NM_016599:exon6:c.G751A:p.E251K	ID=COSM202461;OCCURENCE=2(large_intestine)
2	2022524 70	2022524 70	C	T	exonic	<i>TRAK2</i>	1	nonsynonymous SNV	TRAK2:NM_015049:exon13:c.G1652A:p.R551Q	ID=COSM170962;OCCURENCE=1(large_intestine)
5	9485266 6	9485266 6	C	T	exonic	<i>TTC37</i>	1	nonsynonymous SNV	TTC37:NM_014639:exon22:c.G2390A:p.G797D	ID=COSM1230789;OCCURENCE=1(large_intestine)
3	1955120 42	1955120 42	T	C	exonic	<i>MUC4</i>	2	nonsynonymous SNV	MUC4:NM_018406:exon2:c.A6409G:p.T2137A	
3	1955120 42	1955120 42	T	C	exonic	<i>MUC4</i>	2	nonsynonymous SNV	MUC4:NM_018406:exon2:c.A6409G:p.T2137A	
2	2196028 30	2196028 30	G	A	exonic	<i>TTLL4</i>	2	nonsynonymous SNV	TTLL4:NM_014640:exon3:c.G431A:p.S144N	
2	2196028 30	2196028 30	G	A	exonic	<i>TTLL4</i>	2	nonsynonymous SNV	TTLL4:NM_014640:exon3:c.G431A:p.S144N	
20	3739622 0	3739622 0	C	G	exonic	<i>ACTR5</i>	1	nonsynonymous SNV	ACTR5:NM_024855:exon8:c.C1547G:p.P516R	
5	1289835 36	1289835 36	T	C	exonic	<i>ADAMTS19</i>	1	nonsynonymous SNV	ADAMTS19:NM_133638:exon12:c.T1933C:p.C645R	
15	8916989 1	8916989 1	A	T	exonic	<i>AEN</i>	1	nonsynonymous SNV	AEN:NM_022767:exon2:c.A451T:p.M151L	
16	5643692 2	5643692 2	G	A	exonic	<i>AMFR</i>	1	nonsynonymous SNV	AMFR:NM_001144:exon7:c.C949T:p.R317C	
11	5700434 8	5700434 8	G	A	exonic	<i>APLNR</i>	1	nonsynonymous SNV	APLNR:NM_005161:exon1:c.C131T:p.T44M	
3	1538402 24	1538402 24	G	C	exonic	<i>ARHGEF26</i>	1	nonsynonymous SNV	ARHGEF26:NM_001251962:exon2:c.G443C:p.R148P,ARHGEF26:	

									NM_001251963:exon2:c.G443C:p.R148P,ARHGEF26:NM_015595:exon2:c.G443C:p.R148P	
2	3918517 7	3918517 7	C	T	exonic	ARHGEF33	1	nonsynonymous SNV	ARHGEF33:NM_001145451:exon13:c.C1373T:p.S458L	
X	1553939	1553939	C	T	exonic	ASMTL	1	nonsynonymous SNV	ASMTL:NM_001173474:exon4:c.G328A:p.A110T,ASMTL:NM_001173473:exon5:c.G202A:p.A68T,ASMTL:NM_004192:exon5:c.G376A:p.A126T	
3	1941677 12	1941677 12	T	C	exonic	ATP13A3	1	nonsynonymous SNV	ATP13A3:NM_024524:exon13:c.A1441G:p.I481V	
16	2884748 5	2884748 5	G	A	exonic	ATXN2L	1	nonsynonymous SNV	ATXN2L:NM_007245:exon22:c.G3127A:p.D1043N,ATXN2L:NM_017492:exon22:c.G3127A:p.D1043N,ATXN2L:NM_145714:exon22:c.G3127A:p.D1043N,ATXN2L:NM_148414:exon22:c.G3127A:p.D1043N	
10	1240506 80	1240506 80	C	A	exonic	BTBD16	1	nonsynonymous SNV	BTBD16:NM_144587:exon7:c.C561A:p.H187Q	
10	1240506 06	1240506 06	G	T	exonic	BTBD16	1	nonsynonymous SNV	BTBD16:NM_144587:exon7:c.G487T:p.A163S	
14	5995065 0	5995065 0	G	C	exonic	C14orf149	1	nonsynonymous SNV	C14orf149:NM_144581:exon1:c.C385G:p.R129G	
15	4067515 8	4067515 8	C	T	exonic	C15orf23	1	nonsynonymous SNV	C15orf23:NM_001142761:exon1:c.C122T:p.A41V,C15orf23:NM_001142762:exon1:c.C122T:p.A41V,C15orf23:NM_033286:exon1:c.C122T:p.A41V	
19	757886	757886	C	T	exonic	C19orf21	1	nonsynonymous SNV	C19orf21:NM_173481:exon2:c.C940T:p.R314W	
19	1010785	1010785	G	A	exonic	C19orf6	1	nonsynonymous SNV	C19orf6:NM_001033026:exon11:c.C1427T:p.T476M	
21	4758194 6	4758194 6	G	C	exonic	C21orf56	1	nonsynonymous SNV	C21orf56:NM_032261:exon3:c.C118G:p.R40G,C21orf56:NM_001142854:exon4:c.C580G:p.R194G	
2	2780440 3	2780440 3	G	A	exonic	C2orf16	1	nonsynonymous SNV	C2orf16:NM_032266:exon1:c.G4964A:p.R1655H	

5	1734163 34	1734163 34	C	T	exonic	<i>C5orf47</i>	1	nonsynonymous SNV	C5orf47:NM_001144954:exon1:c. C68T:p.S23L
17	4883619	4883619	T	C	exonic	<i>CAMTA2</i>	1	nonsynonymous SNV	CAMTA2:NM_001171166:exon8: c.A1004G:p.Q335R,CAMTA2:NM _001171168:exon8:c.A995G:p.Q3 32R,CAMTA2:NM_001171167:ex on9:c.A1067G:p.Q356R,CAMTA2 :NM_015099:exon9:c.A998G:p.Q 333R
12	7526150	7526150	T	C	exonic	<i>CD163L1</i>	1	nonsynonymous SNV	CD163L1:NM_174941:exon14:c. A3496G:p.S1166G
22	1760134 4	1760134 4	A	G	exonic	<i>CECR6</i>	1	nonsynonymous SNV	CECR6:NM_031890:exon1:c.T67 4C:p.L225P
22	4693113 7	4693113 7	T	G	exonic	<i>CELSR1</i>	1	nonsynonymous SNV	CELSR1:NM_014246:exon1:c.A1 931C:p.E644A
16	6699776 8	6699776 8	G	T	exonic	<i>CES3</i>	1	nonsynonymous SNV	CES3:NM_001185177:exon4:c.G 490T:p.A164S,CES3:NM_024922 :exon4:c.G490T:p.A164S
2	1898750 78	1898750 78	A	G	exonic	<i>COL3A1</i>	1	nonsynonymous SNV	COL3A1:NM_000090:exon49:c.A 3998G:p.D1333G
10	9968314 0	9968314 0	C	T	exonic	<i>CRTAC1</i>	1	nonsynonymous SNV	CRTAC1:NM_001206528:exon4:c .G439A:p.D147N,CRTAC1:NM_0 18058:exon4:c.G439A:p.D147N
10	1694270 8	1694270 8	A	G	exonic	<i>CUBN</i>	1	nonsynonymous SNV	CUBN:NM_001081:exon53:c.T83 26C:p.S2776P
14	2458908 5	2458908 5	A	C	exonic	<i>DCAF11</i>	1	nonsynonymous SNV	DCAF11:NM_001163484:exon11: c.A1072C:p.I358L,DCAF11:NM_0 25230:exon11:c.A1072C:p.I358L, DCAF11:NM_181357:exon11:c.A 994C:p.I332L
12	1326258 56	1326258 56	A	G	exonic	<i>DDX51</i>	1	nonsynonymous SNV	DDX51:NM_175066:exon8:c.T12 14C:p.L405P
9	1264336 16	1264336 16	T	C	exonic	<i>DENND1A</i>	1	nonsynonymous SNV	DENND1A:NM_020946:exon7:c. A407G:p.H136R,DENND1A:NM_ 024820:exon7:c.A407G:p.H136R
1	1539153 73	1539153 73	C	T	exonic	<i>DENND4B</i>	1	nonsynonymous SNV	DENND4B:NM_014856:exon3:c. G551A:p.R184H
8	1455417 70	1455417 70	G	T	exonic	<i>DGAT1</i>	1	nonsynonymous SNV	DGAT1:NM_012079:exon8:c.C73 9A:p.L247M

8	1295720 6	1295720 6	G	T	exonic	<i>DLC1</i>	1	nonsynonymous SNV	DLC1:NM_001164271:exon5:c.C1107A:p.D369E,DLC1:NM_006094:exon5:c.C1329A:p.D443E,DLC1:NM_182643:exon9:c.C2640A:p.D880E	
3	5745729 7	5745729 7	G	C	exonic	<i>DNAH12</i>	1	nonsynonymous SNV	DNAH12:NM_178504:exon15:c.C1813G:p.L605V	
6	5636593 2	5636593 2	C	A	exonic	<i>DST</i>	1	nonsynonymous SNV	DST:NM_015548:exon61:c.G11973T:p.Q3991H	
1	1670954 44	1670954 44	G	A	exonic	<i>DUSP27</i>	1	nonsynonymous SNV	DUSP27:NM_001080426:exon5:c.G1076A:p.G359D	
20	2730050	2730050	G	A	exonic	<i>EBF4</i>	1	nonsynonymous SNV	EBF4:NM_001110514:exon9:c.G631A:p.V211M	
2	2122516 46	2122516 46	T	C	exonic	<i>ERBB4</i>	1	nonsynonymous SNV	ERBB4:NM_001042599:exon26:c.A3365G:p.E1122G,ERBB4:NM_005235:exon27:c.A3413G:p.E1138G	
11	1246266 02	1246266 02	G	A	exonic	<i>ESAM</i>	1	nonsynonymous SNV	ESAM:NM_138961:exon3:c.C286T:p.P96S	
16	6826518 0	6826518 0	C	T	exonic	<i>ESRP2</i>	1	nonsynonymous SNV	ESRP2:NM_024939:exon12:c.G1612A:p.V538M	
1	3271399 7	3271399 7	A	G	exonic	<i>FAM167B</i>	1	nonsynonymous SNV	FAM167B:NM_032648:exon2:c.A287G:p.Q96R	
7	208930	208930	A	G	exonic	<i>FAM20C</i>	1	nonsynonymous SNV	FAM20C:NM_020223:exon3:c.A817G:p.I273V	
22	2498223 1	2498223 1	C	G	exonic	<i>FAM211B</i>	1	nonsynonymous SNV	FAM211B:NM_207644:exon4:c.G571C:p.G191R	
1	1173383 1	1173383 1	G	A	exonic	<i>FBXO6</i>	1	nonsynonymous SNV	FBXO6:NM_018438:exon6:c.G805A:p.E269K	
12	4554391	4554391	T	C	exonic	<i>FGF6</i>	1	nonsynonymous SNV	FGF6:NM_020996:exon1:c.A346G:p.S116G	
10	1232633 68	1232633 68	T	C	exonic	<i>FGFR2</i>	1	nonsynonymous SNV	FGFR2:NM_001144914:exon7:c.A1039G:p.M347V,FGFR2:NM_001144916:exon7:c.A1030G:p.M344V,FGFR2:NM_001144917:exon8:c.A1027G:p.M343V,FGFR2:NM_001144918:exon8:c.A1024G:p.M342V,FGFR2:NM_001144913:exon9:c.A1378G:p.M460V,FGFR2:NM_001144915:exon9:c.A1108G:p	

									.M370V,FGFR2:NM_001144919:exon9:c.A1111G:p.M371V,FGFR2:NM_000141:exon10:c.A1375G:p.M459V,FGFR2:NM_022970:exon10:c.A1378G:p.M460V	
17	7413345 1	7413345 1	C	T	exonic	<i>FOXJ1</i>	1	nonsynonymous SNV	FOXJ1:NM_001454:exon3:c.G1249A:p.V417M	
4	4863637 3	4863637 3	T	G	exonic	<i>FRYL</i>	1	nonsynonymous SNV	FRYL:NM_015030:exon4:c.A55C:p.S19R	
6	5299334 4	5299334 4	G	A	exonic	<i>GCM1</i>	1	nonsynonymous SNV	GCM1:NM_003643:exon6:c.C971T:p.P324L	
1	2283466 99	2283466 99	C	G	exonic	<i>GJC2</i>	1	nonsynonymous SNV	GJC2:NM_020435:exon2:c.C1240G:p.R414G	
16	4385106	4385106	A	G	exonic	<i>GLIS2</i>	1	nonsynonymous SNV	GLIS2:NM_032575:exon4:c.A568G:p.N190D	
12	1109064 19	1109064 19	G	T	exonic	<i>GPN3</i>	1	nonsynonymous SNV	GPN3:NM_001164372:exon1:c.C23A:p.A8D	
12	6934685	6934685	C	T	exonic	<i>GPR162</i>	1	nonsynonymous SNV	GPR162:NM_014449:exon3:c.C52T:p.P18S,GPR162:NM_019858:exon3:c.C904T:p.P302S	
7	7392983 0	7392983 0	G	A	exonic	<i>GTF2IRD1</i>	1	nonsynonymous SNV	GTF2IRD1:NM_001199207:exon4:c.G421A:p.G141S,GTF2IRD1:NM_005685:exon4:c.G325A:p.G109S,GTF2IRD1:NM_016328:exon4:c.G325A:p.G109S	
8	1452233 39	1452233 39	A	G	exonic	<i>HEATR7A</i>	1	nonsynonymous SNV	HEATR7A:NM_001099281:exon3:c.A164G:p.D55G,HEATR7A:NM_001099280:exon4:c.A164G:p.D55G,HEATR7A:NM_032450:exon4:c.A164G:p.D55G	
19	5577726 3	5577726 3	G	A	exonic	<i>HSPBP1</i>	1	nonsynonymous SNV	HSPBP1:NM_012267:exon6:c.C884T:p.A295V,HSPBP1:NM_001130106:exon7:c.C884T:p.A295V	
16	7088381 8	7088381 8	G	T	exonic	<i>HYDIN</i>	1	nonsynonymous SNV	HYDIN:NM_032821:exon75:c.C12681A:p.F4227L	
8	3977564 2	3977564 2	C	G	exonic	<i>IDO1</i>	1	nonsynonymous SNV	IDO1:NM_002164:exon3:c.C219G:p.H73Q	
1	1589864 08	1589864 08	G	C	exonic	<i>IFI16</i>	1	nonsynonymous SNV	IFI16:NM_005531:exon4:c.G467C:p.G156A	

2	2769512 8	2769512 8	G	A	exonic	<i>IFT172</i>	1	nonsynonymous SNV	IFT172:NM_015662:exon15:c.C1513T;p.R505W	
15	6562709 2	6562709 2	C	T	exonic	<i>IGDCC3</i>	1	nonsynonymous SNV	IGDCC3:NM_004884:exon5:c.G797A;p.R266H	
16	8772383 0	8772383 0	G	A	exonic	<i>JPH3</i>	1	nonsynonymous SNV	JPH3:NM_020655:exon4:c.G1864A;p.E622K	
14	8872973 7	8872973 7	C	T	exonic	<i>KCNK10</i>	1	nonsynonymous SNV	KCNK10:NM_021161:exon2:c.G196A;p.V66I,KCNK10:NM_138317:exon2:c.G211A;p.V71I,KCNK10:NM_138318:exon2:c.G211A;p.V71I	
14	1053599 81	1053599 81	C	T	exonic	<i>KIAA0284</i>	1	nonsynonymous SNV	KIAA0284:NM_015005:exon14:c.C4055T;p.T1352M,KIAA0284:NM_001112726:exon15:c.C4160T;p.T1387M	
6	1386575 95	1386575 95	T	G	exonic	<i>KIAA1244</i>	1	nonsynonymous SNV	KIAA1244:NM_020340:exon34:c.T6506G;p.V2169G	
20	6088971 5	6088971 5	C	T	exonic	<i>LAMA5</i>	1	nonsynonymous SNV	LAMA5:NM_005560:exon61:c.G8263A;p.D2755N	
4	1827316	1827316	T	C	exonic	<i>LETM1</i>	1	nonsynonymous SNV	LETM1:NM_012318:exon7:c.A1175G;p.D392G	
15	1015237 54	1015237 54	G	A	exonic	<i>LRRK1</i>	1	nonsynonymous SNV	LRRK1:NM_024652:exon4:c.G283A;p.A95T	
11	6393339 0	6393339 0	G	A	exonic	<i>MACROD1</i>	1	nonsynonymous SNV	MACROD1:NM_014067:exon1:c.C77T;p.P26L	
6	1192528 77	1192528 77	A	C	exonic	<i>MCM9</i>	1	nonsynonymous SNV	MCM9:NM_017696:exon1:c.T12G;p.D4E,MCM9:NM_153255:exon2:c.T12G;p.D4E	
19	5032162 1	5032162 1	C	A	exonic	<i>MED25</i>	1	nonsynonymous SNV	MED25:NM_030973:exon1:c.C23A;p.P8Q	
19	3622431 6	3622431 6	C	T	exonic	<i>MLL4</i>	1	nonsynonymous SNV	MLL4:NM_014727:exon28:c.C6866T;p.P2289L	
20	4957589 9	4957589 9	G	T	exonic	<i>MOCS3</i>	1	nonsynonymous SNV	MOCS3:NM_014484:exon1:c.G520T;p.D174Y	
2	7136066 6	7136066 6	A	T	exonic	<i>MPHOSPH10</i>	1	nonsynonymous SNV	MPHOSPH10:NM_005791:exon2:c.A728T;p.D243V	
10	2840914 7	2840914 7	A	C	exonic	<i>MPP7</i>	1	nonsynonymous SNV	MPP7:NM_173496:exon12:c.T863G;p.I288S	
22	3040951 7	3040951 7	A	G	exonic	<i>MTMR3</i>	1	nonsynonymous SNV	MTMR3:NM_021090:exon14:c.A1490G;p.N497S,MTMR3:NM_1530	

									50:exon14:c.A1490G:p.N497S,M TMR3:NM_153051:exon14:c.A14 90G:p.N497S	
7	1006921 68	1006921 68	C	T	exonic	<i>MUC17</i>	1	nonsynonymous SNV	MUC17:NM_001040105:exon5:c. C12578T:p.T4193I	
10	2644367 8	2644367 8	G	A	exonic	<i>MYO3A</i>	1	nonsynonymous SNV	MYO3A:NM_017433:exon25:c.G2 719A:p.D907N	
10	5158661 9	5158661 9	A	G	exonic	<i>NCOA4</i>	1	nonsynonymous SNV	NCOA4:NM_001145260:exon11:c .A1897G:p.K633E	
8	2481374 4	2481374 4	C	T	exonic	<i>NEFL</i>	1	nonsynonymous SNV	NEFL:NM_006158:exon1:c.G286 A:p.D96N	
11	284304	284304	C	T	exonic	<i>NLRP6</i>	1	nonsynonymous SNV	NLRP6:NM_138329:exon6:c.C22 76T:p.T759M	
1	1190724 4	1190724 4	G	A	exonic	<i>NPPA</i>	1	nonsynonymous SNV	NPPA:NM_006172:exon2:c.C376 T:p.R126W	
2	2765654 6	2765654 6	C	G	exonic	<i>NRBP1</i>	1	nonsynonymous SNV	NRBP1:NM_013392:exon3:c.C21 7G:p.Q73E	
12	2944085	2944085	G	A	exonic	<i>NRIP2</i>	1	nonsynonymous SNV	NRIP2:NM_031474:exon1:c.C65T :p.T22M	
1	2285663 24	2285663 24	G	T	exonic	<i>OBSCN</i>	1	nonsynonymous SNV	OBSCN:NM_001098623:exon105 :c.G23735T:p.R7912L	
1	2284025 83	2284025 83	C	A	exonic	<i>OBSCN</i>	1	nonsynonymous SNV	OBSCN:NM_001098623:exon5:c. C1612A:p.H538N,OBSCN:NM_05 2843:exon5:c.C1612A:p.H538N	
13	5361731 0	5361731 0	G	A	exonic	<i>OLFM4</i>	1	nonsynonymous SNV	OLFM4:NM_006418:exon4:c.G64 1A:p.R214Q	
11	6221100	6221100	T	C	exonic	<i>OR52W1</i>	1	nonsynonymous SNV	OR52W1:NM_001005178:exon1: c.T647C:p.I216T	
8	1077260 58	1077260 58	C	G	exonic	<i>OXR1</i>	1	nonsynonymous SNV	OXR1:NM_181354:exon9:c.C178 0G:p.H594D,OXR1:NM_0011985 32:exon10:c.C1804G:p.H602D,O XR1:NM_001198533:exon11:c.C 1801G:p.H601D,OXR1:NM_0180 02:exon11:c.C1801G:p.H601D	
11	7405362 0	7405362 0	C	G	exonic	<i>PGM2L1</i>	1	nonsynonymous SNV	PGM2L1:NM_173582:exon12:c.G 1518C:p.R506S	
3	1705625 1	1705625 1	C	G	exonic	<i>PLCL2</i>	1	nonsynonymous SNV	PLCL2:NM_015184:exon3:c.C24 88G:p.L830V	
22	5071416 5	5071416 5	C	T	exonic	<i>PLXNB2</i>	1	nonsynonymous SNV	PLXNB2:NM_012401:exon37:c.G 5485A:p.A1829T	

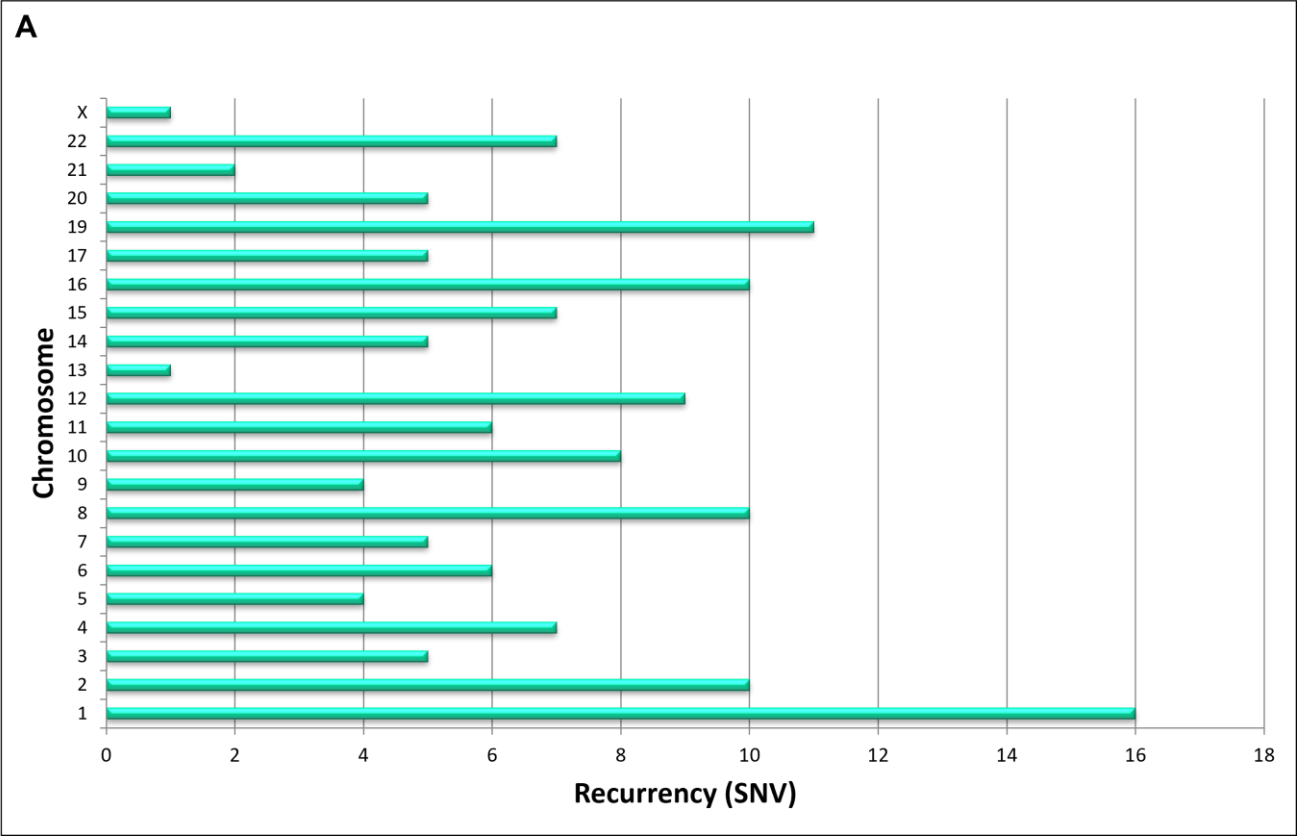
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20	4812972 6	4812972 6	G	A	exonic	<i>PTGIS</i>	1	nonsynonymous SNV	PTGIS:NM_000961:exon8:c.C1097T:p.A366V	
1	8421196	8421196	G	A	exonic	<i>RERE</i>	1	nonsynonymous SNV	RERE:NM_001042682:exon8:c.C709T:p.P237S,RERE:NM_001042681:exon18:c.C2371T:p.P791S,RERE:NM_012102:exon19:c.C2371T:p.P791S	
1	1143545 75	1143545 75	C	A	exonic	<i>RSBN1</i>	1	nonsynonymous SNV	RSBN1:NM_018364:exon1:c.G460T:p.A154S	
21	4389602 5	4389602 5	C	T	exonic	<i>RSPH1</i>	1	nonsynonymous SNV	RSPH1:NM_080860:exon8:c.G860A:p.R287H	
9	1392985 97	1392985 97	T	G	exonic	<i>SDCCAG3</i>	1	nonsynonymous SNV	SDCCAG3:NM_001039708:exon7:c.A899C:p.Q300P,SDCCAG3:NM_006643:exon8:c.A1049C:p.Q350P,SDCCAG3:NM_001039707:exon9:c.A1118C:p.Q373P	
22	3089137 2	3089137 2	C	T	exonic	<i>SEC14L4</i>	1	nonsynonymous SNV	SEC14L4:NM_001161368:exon5:c.G292A:p.V98M,SEC14L4:NM_174977:exon5:c.G292A:p.V98M	
9	1393589 54	1393589 54	C	A	exonic	<i>SEC16A</i>	1	nonsynonymous SNV	SEC16A:NM_014866:exon9:c.G4364T:p.G1455V	
1	1561466 34	1561466 34	G	A	exonic	<i>SEMA4A</i>	1	nonsynonymous SNV	SEMA4A:NM_001193302:exon13:c.G1736A:p.R579Q,SEMA4A:NM_001193300:exon15:c.G2132A:p.R711Q,SEMA4A:NM_001193301:exon15:c.G2132A:p.R711Q,SEMA4A:NM_022367:exon15:c.G2132A:p.R711Q	
15	4806389 7	4806389 7	C	T	exonic	<i>SEMA6D</i>	1	nonsynonymous SNV	SEMA6D:NM_020858:exon17:c.C2951T:p.P984L,SEMA6D:NM_153616:exon17:c.C2912T:p.P971L,SEMA6D:NM_153617:exon18:c.C2969T:p.P990L,SEMA6D:NM_153618:exon19:c.C3137T:p.P1046L	

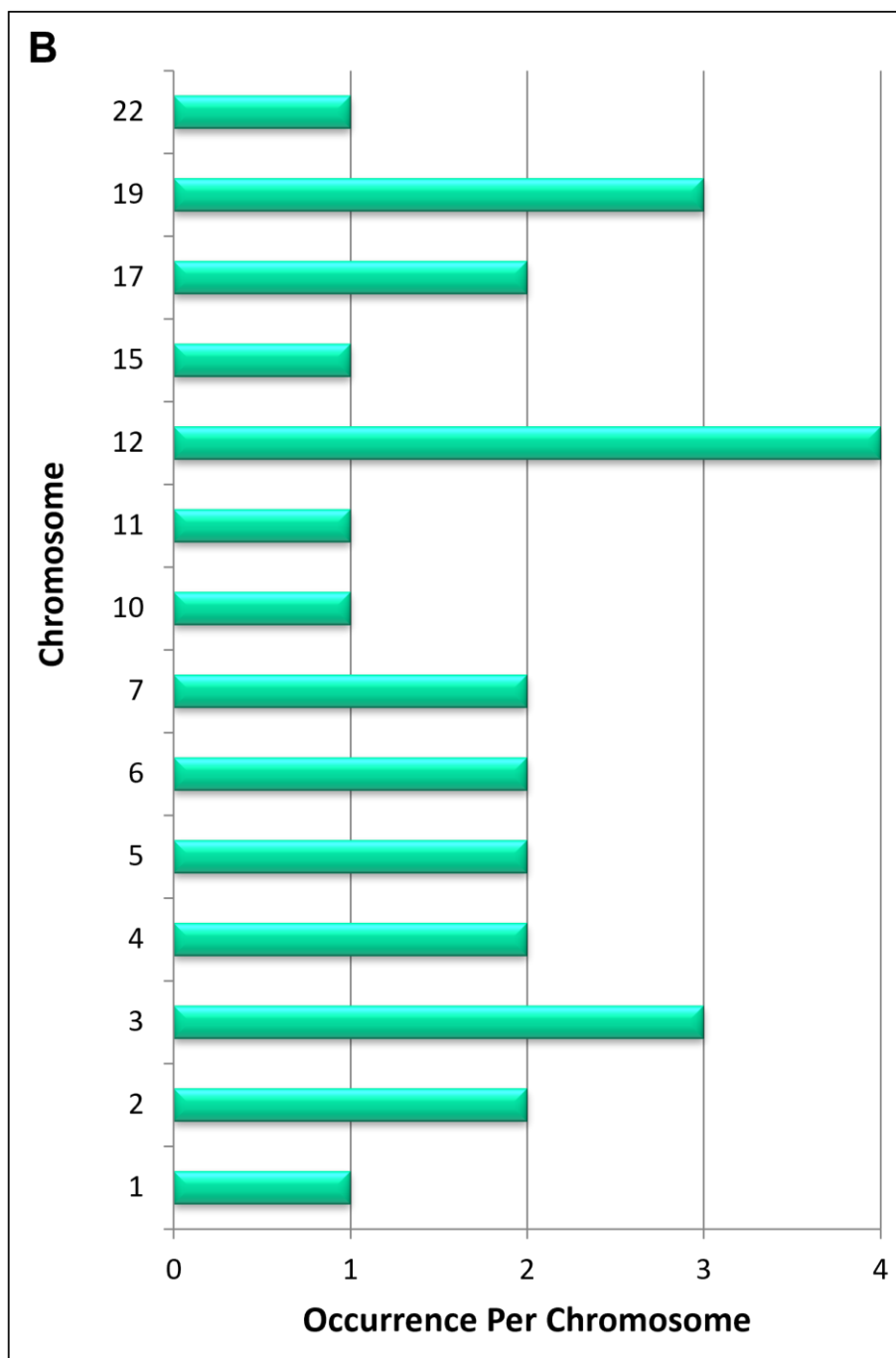
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8	8260605 4	8260605 4	T	C	exonic	SLC10A5	1	nonsynonymous SNV	SLC10A5:NM_001010893:exon1: c.A1154G:p.Q385R	
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12	346452	346452	G	A	exonic	SLC6A13	1	nonsynonymous SNV	SLC6A13:NM_001190997:exon4: c.C292T:p.R98W,SLC6A13:NM_0 16615:exon6:c.C568T:p.R190W	
17	1687713	1687713	A	T	exonic	SMYD4	1	nonsynonymous SNV	SMYD4:NM_052928:exon8:c.T19 27A:p.S643T	
4	5292703 5	5292703 5	C	G	exonic	SPATA18	1	nonsynonymous SNV	SPATA18:NM_145263:exon3:c.C 281G:p.S94C	
15	9176981 0	9176981 0	G	A	exonic	SV2B	1	nonsynonymous SNV	SV2B:NM_014848:exon3:c.G317 A:p.R106H	
16	2999883 1	2999883 1	G	C	exonic	TAOK2	1	nonsynonymous SNV	TAOK2:NM_016151:exon16:c.G3 238C:p.G1080R,TAOK2:NM_001 252043:exon17:c.G2899C:p.G96 7R	
4	1070167 90	1070167 90	C	G	exonic	TBCK	1	nonsynonymous SNV	TBCK:NM_033115:exon23:c.G22 31C:p.R744P,TBCK:NM_001163 435:exon25:c.G2420C:p.R807P,T BCK:NM_001163436:exon25:c.G 2420C:p.R807P,TBCK:NM_0011 63437:exon25:c.G2303C:p.R768 P	
14	9043756 8	9043756 8	C	T	exonic	TDP1	1	nonsynonymous SNV	TDP1:NM_001008744:exon5:c.C 709T:p.H237Y,TDP1:NM_018319 :exon6:c.C709T:p.H237Y	

9	1030708 06	1030708 06	C	T	exonic	<i>TEX10</i>	1	nonsynonymous SNV	TEX10:NM_001161584:exon13:c. G2450A:p.G817E,TEX10:NM_01 7746:exon13:c.G2441A:p.G814E	
17	3296487 1	3296487 1	G	A	exonic	<i>TMEM132E</i>	1	nonsynonymous SNV	TMEM132E:NM_207313:exon10: c.G2575A:p.E859K	
19	1679327 4	1679327 4	G	A	exonic	<i>TMEM38A</i>	1	nonsynonymous SNV	TMEM38A:NM_024074:exon4:c. G509A:p.R170Q	
1	1149477	1149477	C	T	exonic	<i>TNFRSF4</i>	1	nonsynonymous SNV	TNFRSF4:NM_003327:exon1:c.G 31A:p.G11R	
6	1677537 08	1677537 08	G	T	exonic	<i>TTLL2</i>	1	nonsynonymous SNV	TTLL2:NM_031949:exon3:c.G320 T:p.G107V	
2	1794285 20	1794285 20	C	T	exonic	<i>TTN</i>	1	nonsynonymous SNV	TTN:NM_003319:exon154:c.G55 144A:p.V18382M,TTN:NM_13343 2:exon155:c.G55519A:p.V18507 M,TTN:NM_133437:exon155:c.G 55720A:p.V18574M,TTN:NM_133 378:exon275:c.G74635A:p.V2487 9M,TTN:NM_001256850:exon276 :c.G77416A:p.V25806M	
19	3494297 3	3494297 3	G	A	exonic	<i>UBA2</i>	1	nonsynonymous SNV	UBA2:NM_005499:exon10:c.G95 9A:p.R320H	
1	2297727 58	2297727 58	C	G	exonic	<i>URB2</i>	1	nonsynonymous SNV	URB2:NM_014777:exon4:c.C239 8G:p.L800V	
4	8560010 4	8560010 4	A	G	exonic	<i>WDFY3</i>	1	nonsynonymous SNV	WDFY3:NM_014991:exon65:c.T1 0115C:p.I3372T	
7	1586944 46	1586944 46	A	G	exonic	<i>WDR60</i>	1	nonsynonymous SNV	WDR60:NM_018051:exon9:c.A10 76G:p.E359G	
8	1075562 3	1075562 3	T	C	exonic	<i>XKR6</i>	1	nonsynonymous SNV	XKR6:NM_173683:exon3:c.A176 5G:p.M589V	
6	4349662 2	4349662 2	G	A	exonic	<i>XPO5</i>	1	nonsynonymous SNV	XPO5:NM_020750:exon24:c.C27 19T:p.H907Y	
16	1186818 7	1186818 7	T	C	exonic	<i>ZC3H7A</i>	1	nonsynonymous SNV	ZC3H7A:NM_014153:exon9:c.A8 08G:p.M270V	
19	4468169 3	4468169 3	T	A	exonic	<i>ZNF226</i>	1	nonsynonymous SNV	ZNF226:NM_001032372:exon6:c. T2278A:p.C760S,ZNF226:NM_00 1032373:exon6:c.T2278A:p.C760 S	
7	6438923 5	6438923 5	G	A	exonic	<i>ZNF273</i>	1	nonsynonymous SNV	ZNF273:NM_021148:exon4:c.G1 529A:p.G510E	

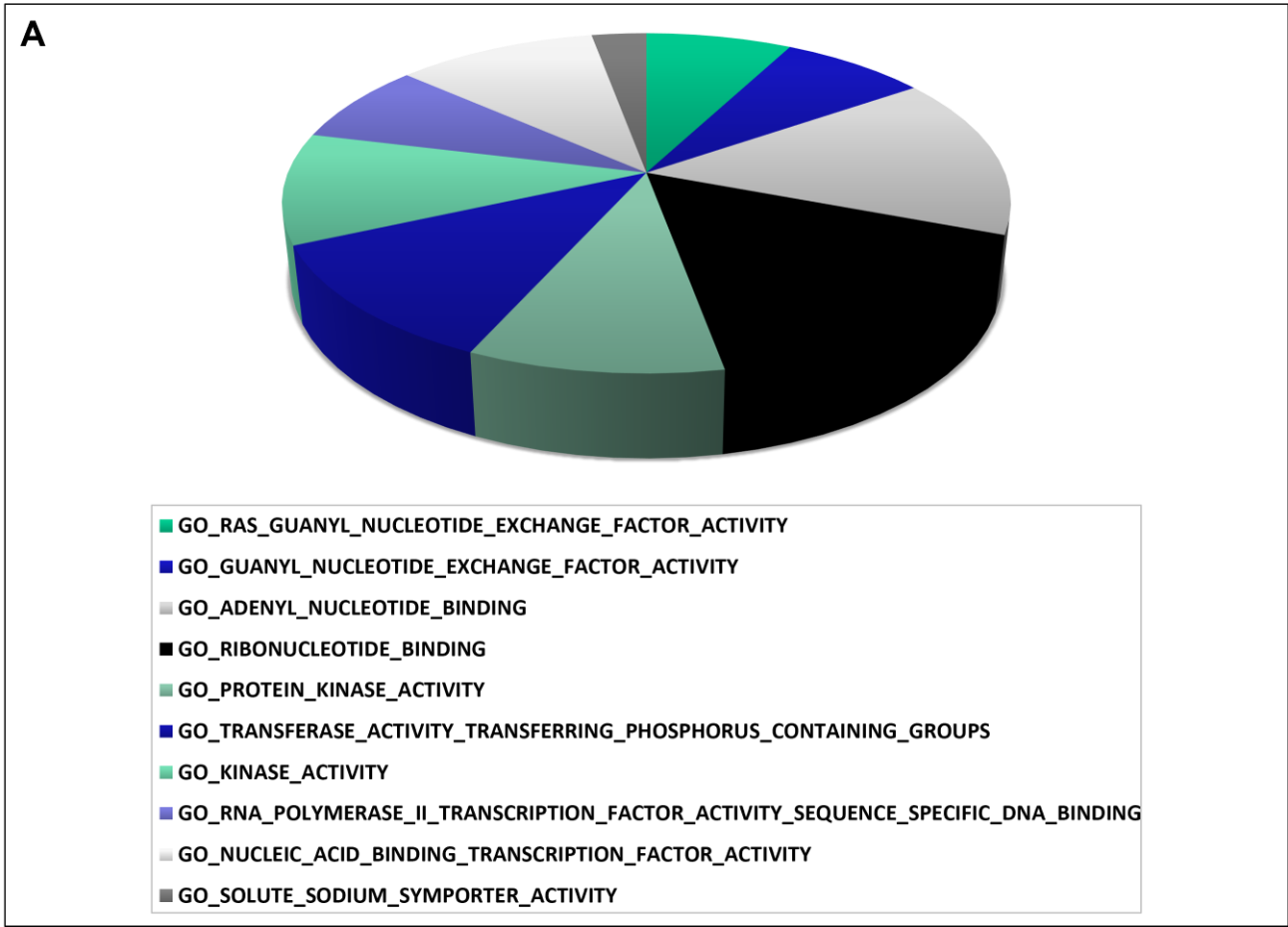
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16	3434804	3434804	G	C	exonic	<i>ZNF434</i>	1	nonsynonymous SNV	ZNF434:NM_017810:exon5:c.C253G:p.Q85E	
19	5899169 9	5899169 9	C	T	exonic	<i>ZNF446</i>	1	nonsynonymous SNV	ZNF446:NM_017908:exon7:c.C959T:p.T320M	
19	5790997 5	5790997 5	C	T	exonic	<i>ZNF548</i>	1	nonsynonymous SNV	ZNF548:NM_152909:exon3:c.C320T:p.P107L,ZNF548:NM_001172773:exon4:c.C356T:p.P119L	
12	1336335 70	1336335 70	G	T	exonic	<i>ZNF84</i>	1	nonsynonymous SNV	ZNF84:NM_001127372:exon5:c.G269T:p.W90L,ZNF84:NM_003428:exon5:c.G269T:p.W90L	
8	1054634 84	1054634 84	C	T	exonic	<i>DPYS</i>	1	stopgain SNV	DPYS:NM_001385:exon2:c.G413A:p.W138X	
1	1112971 2	1112971 2	G	A	exonic	<i>EXOSC10</i>	1	stopgain SNV	EXOSC10:NM_002685:exon21:c.C2317T:p.R773X,EXOSC10:NM_001001998:exon22:c.C2392T:p.R798X	
17	3988402 8	3988402 8	G	A	exonic	<i>HAP1</i>	1	stopgain SNV	HAP1:NM_177977:exon8:c.C1261T:p.Q421X	
22	2513182 0	2513182 0	T	A	exonic	<i>PIWIL3</i>	1	stopgain SNV	PIWIL3:NM_001008496:exon13:c.A1489T:p.R497X,PIWIL3:NM_001255975:exon13:c.A1489T:p.R497X	

**Supplementary Figure S1. Occurrence of novel SNVs (A) and novel InDels (B).** The X-axis graphs the number of cases. The Y-axis displays the chromosomes.





**Supplementary Figure S2. Functional analysis of genes affected by SNVs.** The affected genes were analyzed in terms of enrichment of Gene Ontology Biological Processes (A). Pathways analysis aimed to recognize actionable genes was performed by Cognoscente (B).



B

