

SUPPLEMENTAL MATERIAL

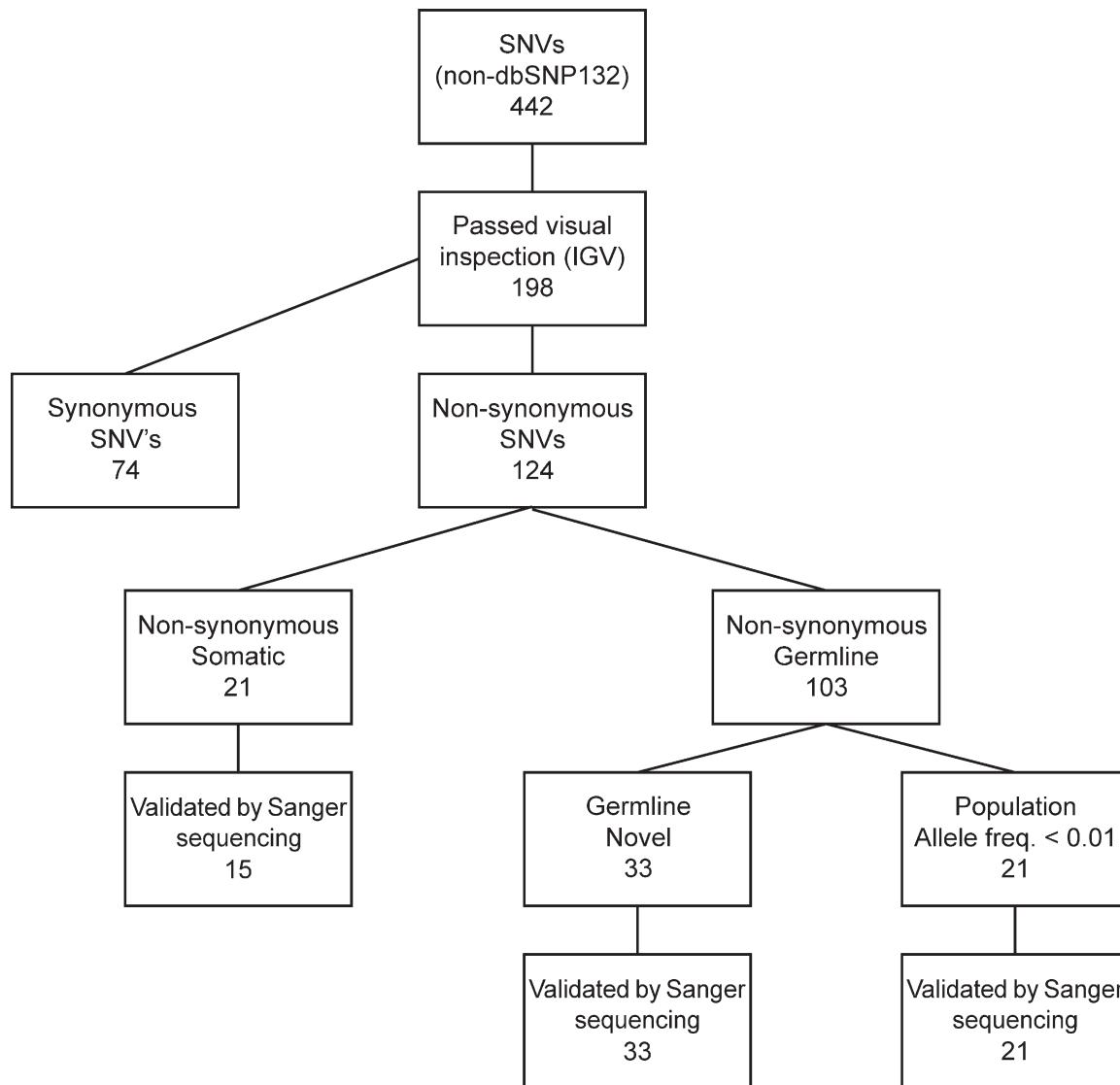
de Miranda et al., <http://www.jem.org/cgi/content/full/jem.20122842/DC1>

Figure S1. Single nucleotide variant (SNV) detection was performed by comparing heterozygous positions in tumor and normal samples against the human reference genome (hg18). Additionally, specific detection of somatic variants was performed by comparing sequencing data between paired tumor and normal samples. Analysis thresholds were set at a low level to maximize the detection of variants which partially explains the abundance of variants rejected by visual inspection.

Table S1. Genes targeted by Selector technology

Gene	Chr	Pathways	Gene	Chr	Pathways
<i>AICDA</i>	12p13	CSR/SHM initiator	<i>MSH2</i>	2p21	MMR
<i>APEX1</i>	14q11.2	BER	<i>MSH3</i>	5q11-q12	MMR
<i>APEX2</i>	Xp11.21	BER	<i>MSH4</i>	1p31	MMR
<i>ATM</i>	11q22-q23	DDR	<i>MSH5</i>	6p21.3	MMR
<i>ATR</i>	3q23	DDR	<i>MSH6</i>	2p16	MMR
<i>BLM</i>	15q26.1	HR/FA	<i>MUS81</i>	11q13	HR/FA
<i>BRCA1</i>	17q21	HR	<i>NBN</i>	8q21	DDR
<i>BRCA2</i>	13q12.3	HR/FA	<i>NEIL2</i>	8p23.1	BER
<i>CHEK1</i>	11q24.2	DDR	<i>NHEJ1</i>	2q35	NHEJ
<i>CHEK2</i>	22q12.1	DDR	<i>PARP1</i>	1q41-q42	DDR
<i>DCLRE1C</i>	10p13	NHEJ	<i>PMS1</i>	2q31.1	MMR
<i>DDB1</i>	11q12-q13	NER	<i>PMS2</i>	7p22.2	MMR
<i>DNTT</i>	10q23-q24	BCR/TCR maturation	<i>PNKP</i>	19q13.3-q13.4	BER; NHEJ
<i>ERCC1</i>	19q13.32	NER	<i>POLB</i>	8p11.2	BER
<i>ERCC2</i>	19q13.3	NER	<i>PRKDC</i>	8q11	NHEJ
<i>ERCC3</i>	2q21	NER	<i>RAD23B</i>	9q31.2	NER
<i>ERCC4</i>	16p13.12	NER	<i>RAD50</i>	5q31	DDR; NHEJ
<i>ERCC5</i>	13q33	NER	<i>RAD51</i>	15q15.1	HR
<i>ERCC6</i>	10q11.23	NER	<i>RAD51B</i>	14q23-q24.2	HR
<i>ERCC8</i>	5q12.1	NER	<i>RAD51C</i>	17q25.1	HR
<i>EXO1</i>	1q42-q43	MMR	<i>RAD51D</i>	17q11	HR
<i>FANCA</i>	16q24.3	FA	<i>RAD52</i>	12p13-p12.2	HR
<i>FANCC</i>	9q22.3	FA	<i>RAD54B</i>	8q22.1	HR
<i>FANCD2</i>	3p26	HR/FA	<i>RAD54L</i>	1p32	HR
<i>FANCE</i>	6p22-p21	FA	<i>RPA1</i>	17p13.3	DDR
<i>FANCF</i>	11p15	FA	<i>TNFAIP3</i>	6q23	NF-κB
<i>FANCG</i>	9p13	FA	<i>TP53</i>	17p13.1	DDR
<i>GTF2H4</i>	6p21.3	NER	<i>TP53BP1</i>	15q15-q21	DDR
<i>GTF2H5</i>	6q25.3	NER	<i>UNG</i>	12q23-q24.1	BER
<i>H2AFX</i>	11q23.3	DDR	<i>XPA</i>	9q22.3	NER
<i>LIG1</i>	19q13.2-q13.3	BER/NER	<i>XPC</i>	3p25	NER
<i>LIG3</i>	17q11.2-q12	BER/NER	<i>XRCC1</i>	19q13.2	BER
<i>LIG4</i>	13q33-q34	NHEJ	<i>XRCC2</i>	7q36.1	HR
<i>MDC1</i>	6p21.3	DDR	<i>XRCC3</i>	14q32.3	HR
<i>MLH1</i>	3p21.3	MMR	<i>XRCC4</i>	5q14.2	NHEJ
<i>MLH3</i>	14q24.3	MMR	<i>XRCC5</i>	2q35	NHEJ
<i>MRE11A</i>	11q21	DDR; HR	<i>XRCC6</i>	22q13.2	NHEJ

Generalized pathway mapping was based on KEGG (<http://www.genome.jp/kegg/>), NCBI (<https://www.ncbi.nlm.nih.gov/>), and UniProt (<http://www.uniprot.org/>) databases.

Table S2. Selector design parameters and capture and sequencing performance

Design parameters	Design 1	Design 2
Genes	74	74
Exons	1,223	1,223
ROI (bp)	196,933	196,933
Mean amplicon length (bp)	614	260
Number of selector amplicons	2,958	6,045
Base coverage by selector amplicons	2.1×	3.3×
Covered ROI by design (%)	98.8	96.7
Covered ROI (excluding repeat rich regions; %)	99.0	97.5
Covered ROI (only repeat rich regions; %)	84.3	54.2
Bases covered outside ROI (%)	77.3	60.3
Performance (mean per sample)		
Aligned bp in ROI	83,304,825	51,819,177
Specificity (%)	90.8	94.2
Average read depth in ROI	317	228
ROI covered by at least 1x (%)	96.1	97.8
ROI covered by at least 5x (%)	90.2	93.7
ROI covered by at least 10x (%)	84.9	89.9
ROI covered by at least 20x (%)	77.3	83.4

Table S3. Somatic and germline (novel and rare) mutations discover by SOLiD targeted sequencing

Sample	Diagnosis	Primary/ relapse	GCB/ non-GCB	Somatic	Germline (novel)	Germline (rare)	Sequenced exome
DL-1	DLBCL	Primary	Non-GCB	-	<i>FANCA</i> (p.R1321H) <i>LIG1</i> (p.T311M) <i>PMS1</i> (p.L369P)	-	Yes
DL-30	DLBCL	Primary	Non-GCB	<i>MSH6</i> (p.K543R) <i>TNFAIP3</i> ((p.C627Ffs*44))	-	<i>ATM</i> (p.R924W)	Yes
DL-31	DLBCL	Primary	Non-GCB	-	<i>FANCA</i> (p.T620I) <i>RAD54B</i> (p.P55L)	<i>RAD50</i> (p.L1125F)	Yes
DL-36	DLBCL	Primary	Non-GCB	-	<i>FANCG</i> (p.M431R) <i>LIG3</i> (p.R343Q)	<i>DDB1</i> (p.Y517C)	Yes
DL-37	DLBCL	Primary	Non-GCB	-	-	<i>MLH3</i> (p.I988M) <i>TP53</i> (p.V31I) <i>PMS2</i> (p.S128L)	Yes
DL-40	DLBCL	Primary	GCB	<i>ATM</i> (p.L1206V, p.Y1216H)	-	<i>RAD54B</i> (p.I778V)	Yes
DL-41	DLBCL	Primary	Non-GCB	-	<i>MDC1</i> p.(H35L) <i>MLH3</i> (p.L1111F) <i>MSH6</i> (p.T563N)	-	Yes
DL-42	DLBCL	Primary	Non-GCB	<i>TP53</i> (p.R273C)	<i>MSH3</i> (p.P657S)	<i>TP53BP1</i> (p.T519A)	Yes
DL-43	DLBCL	Primary	GCB	<i>XRCC5</i> (p.T283A)	<i>PRKDC</i> (p.D566N)	-	Yes
DL-44	DLBCL	Primary	GCB	-	-	-	Yes
DL-45	DLBCL	Primary	GCB	<i>DDB1</i> (p.A9G) <i>TP53</i> (p.F134V)	<i>POLB</i> (p.R333Q)	<i>MLH3</i> (p.S946F)	Yes
DL-46	DLBCL	Primary	Non-GCB	-	<i>DNTT</i> (p.R335W) <i>MSH3</i> (p.R1061G) <i>PMS2</i> (p.M362K)	<i>CHEK2</i> (p.E528K)	No
DL-47	DLBCL	Primary	non-GCB	-	-	-	Yes
DL-48	DLBCL	Primary	non-GCB	<i>CHEK2</i> (p.Q336L) <i>DCLRE1C</i> (p.[I543Rfs*12]) <i>EXO1</i> (p.K471R) <i>MSH2</i> (p.S540G, p.L833V) <i>PARP1</i> (p.[K637Sfs*13])	<i>RPA1</i> (p.G160R)	<i>BLM</i> (p.V765I)	Yes
DL-49	DLBCL	Primary	non-GCB	-	<i>BLM</i> (p.S580P) <i>UNG</i> (p.E121Q)	<i>DNTT</i> (p.V37I) <i>TP53BP1</i> (p.A1714S)	Yes
DL-50	DLBCL	Primary	GCB	<i>BRCA2</i> (p.V849A, p.M3322I)	<i>BRCA2</i> (p.S1744I)	-	No
DL-51	DLBCL	Primary	GCB	-	<i>PARP1</i> (p.S776G) <i>PRKDC</i> (p.K1984N)	<i>BRCA2</i> (p.C315S)	No
DL-52	DLBCL	Primary	non-GCB	-	<i>MLH3</i> (p.C40Y) <i>MSH3</i> (p.R1061G)	-	No
DL-53	DLBCL	Relapse	non-GCB	<i>XRCC6</i> (p.I267Kfs*2)	<i>PMS1</i> (p.D397E)	-	Yes
DL-54	DLBCL	Relapse	non-GCB	-	-	<i>BRCA2</i> (p.I1929V)	Yes
DL-55	DLBCL	Primary	non-GCB	<i>PRKDC</i> (p.F3418L)	-	<i>TP53BP1</i> (p.T519A) <i>XRCC1</i> (p.A121T)	No
DL-56	DLBCL	Relapse	GCB	<i>TP53</i> (p.R273H)	<i>RAD51B</i> (p.E346D) <i>TNFAIP3</i> (p.P714S)	<i>BRCA1</i> (p.Y856H)	Yes
FL-15	FL	Primary	-	-	<i>MDC1</i> (p.N280D) <i>RAD50</i> (p.V400E)	-	No
FL-16	FL	Primary	-	-	<i>ERCC5</i> (p.D387N) <i>PNKP</i> (p.P112R)	<i>DNTT</i> (p.V37I)	No
FL-19	FL	Primary	-	-	<i>MSH2</i> (p.G820D)	<i>PMS1</i> (p.R919C)	No
FL-20	FL	Primary	-	-	-	-	No
FL-21	FL	Primary	-	-	<i>MDC1</i> (p.P965L)	-	No
BL-6	BL	Primary	-	-	-	<i>TP53BP1</i> (p.A1714S)	No
BL-7	BL	Primary	-	-	-	-	No

Germline (rare): MAF < 0.01.