

SUPPLEMENTARY MATERIALS

**Homologous recombination DNA repair defects in *PALB2*-associated breast
cancers**

Li et al.

Supplementary Methods

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SUPPLEMENTARY METHODS

Massively parallel sequencing and bioinformatics analysis

DNA samples derived from microdissected tumor and normal tissue were subjected to WES (n=14) or MSK-IMPACT¹ (n=8), which targets all exons and selected introns of 410 cancer genes. Reads were aligned to the reference human genome GRCh37 using the Burrows-Wheeler Aligner (BWA, v0.7.10).² Local realignment, duplicate removal and base quality recalibration were performed using the Genome Analysis Toolkit (GATK, v3.1.1).³ *PALB2* and *BRCA1/2* germline variants were detected by HaplotypeCaller from GATK in the gvcf mode with default settings, and were retained if the germline variants were present in both normal and tumor samples.⁴ Somatic single nucleotide variants (SNVs) were detected by MuTect (v1.0),⁵ small insertions and deletions (indels) by Strelka (v2.0.15)⁶, VarScan 2 (v2.3.7)⁷, Lancet (v1.0.0)⁸ and Scalpel (v0.5.3)⁹, and further curated by manual inspection. SNVs and indels located outside of the target regions were disregarded.¹⁰ We excluded SNVs and indels for which the tumor mutant allele fraction (MAF) was <5 times that of the paired normal MAF, as well as SNVs and indels found at >5% global minor allele frequency of dbSNP (build 137) as previously described.^{10, 11}

The cancer cell fraction (CCF) of each mutation was inferred using ABSOLUTE (v1.0.6)¹² and manually reviewed¹¹⁻¹³ A mutation was classified as clonal if its probability of being clonal was >50%¹³ or if the lower bound of the 95% confidence interval of its CCF was >90%;^{11, 14}. Somatic LOH of the *PALB2* germline mutations was classified as subclonal if the CCF was lower than the tumor purity ($\delta \geq 0.2$) as determined by FACETS,¹⁵ as previously described.¹⁴ We employed a combination of mutation function predictors¹⁶ to infer whether a mutation was potentially pathogenic, as previously described.¹¹ Mutation hotspots were defined according to Chang et al.¹⁵

Comparisons with breast cancers from TCGA

Comparisons of mutation burden, mutation frequencies, CNAs and genomic features indicative of HRD were conducted between the *PALB2*-associated breast cancers, non-*BRCA1/2/PALB2*-

associated breast cancers with matched ER and HER2 status (n=683), and *BRCA1*- (n=17) and *BRCA2*-associated (n=16) breast cancers with bi-allelic inactivation from TCGA¹⁷. The bi-allelic inactivation status of *BRCA1* and *BRCA2* was retrieved from Riaz et al.⁴ TCGA breast cancers with available data to assess LST scores and numbers of somatic mutations adequate for mutational signature assessment were employed for comparative analyses of genomic features indicative of HRD. The recently updated publicly available MC3 dataset from the TCGA was obtained at <https://gdc.cancer.gov/about-data/publications/mc3-2017> and <https://gdac.broadinstitute.org> (01/28/2016).

Statistical analysis

Comparisons of the tumor mutation burden, LST scores, gene-level copy number states and mutational signatures between *PALB2*-associated breast cancers and non-*BRCA1/2/PALB2*-, *BRCA1*- and *BRCA2*-associated breast cancers were performed using the Mann-Whitney *U* test and Fisher's exact test, respectively. To account for differences in sample sizes, a bootstrap resampling analysis was performed for the comparisons of mutation burden, mutation frequencies, gene-level copy number states, LSTs and mutational signatures between the *PALB2*-associated and TCGA non-*BRCA1/2/PALB2*-associated breast cancers at a 1:3 ratio. In brief, 3 non-*BRCA1/2/PALB2*-associated breast cancers were randomly selected for each *PALB2*-associated breast cancer, allowing for a given non-*BRCA1/2/PALB2*-associated sample to be included in the distinct random subsets. We iterated this process 100 times and two-tailed *P*-values were calculated using either Mann-Whitney *U* test or Fisher's exact test for each iteration, as appropriate. A bootstrapping-corrected *P*-value was determined as the mean *P*-value of all iterations for a given comparison. Two-tailed *P*-values<0.05 were considered statistically significant. Statistical analyses were performed with R v3.1.2.

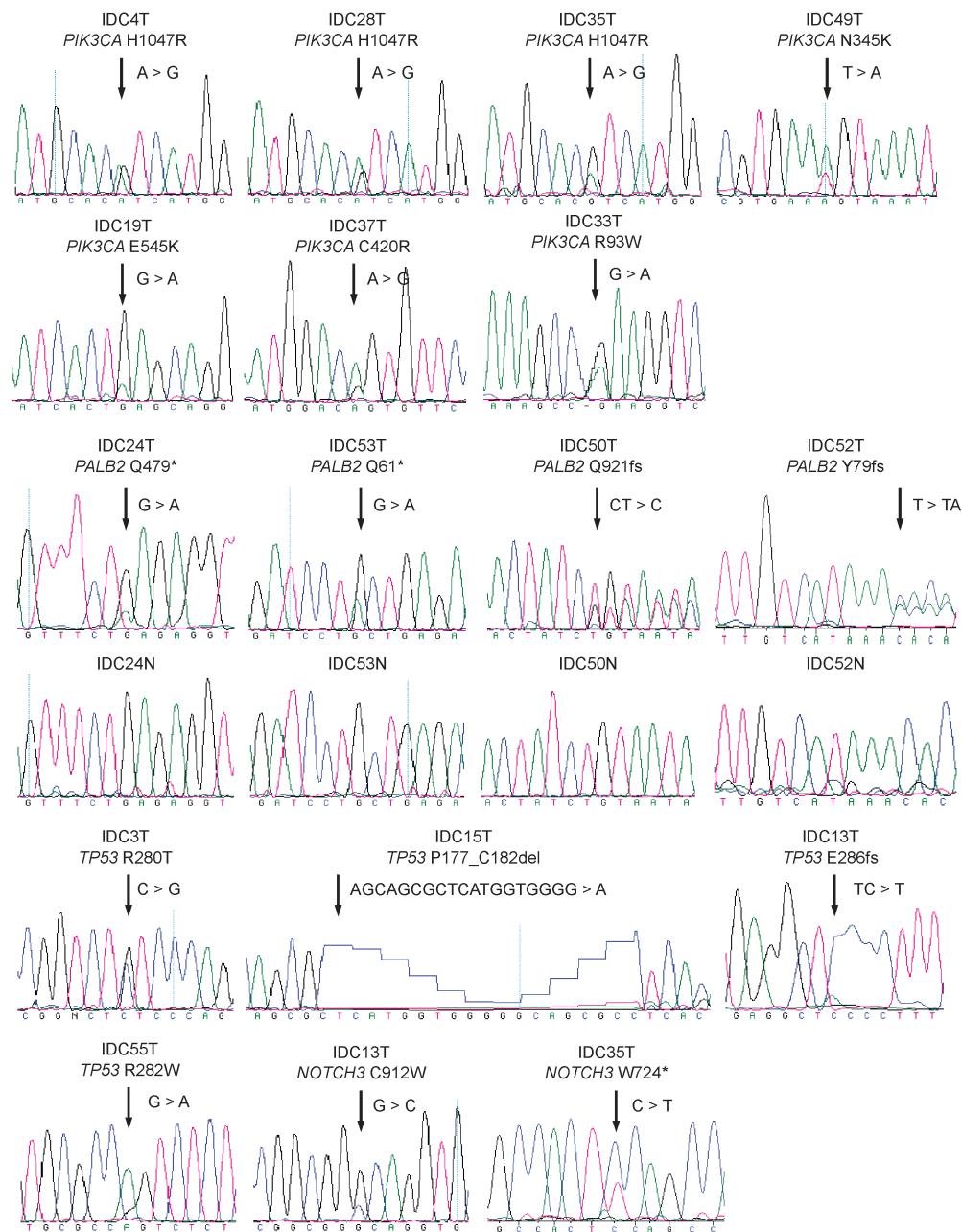
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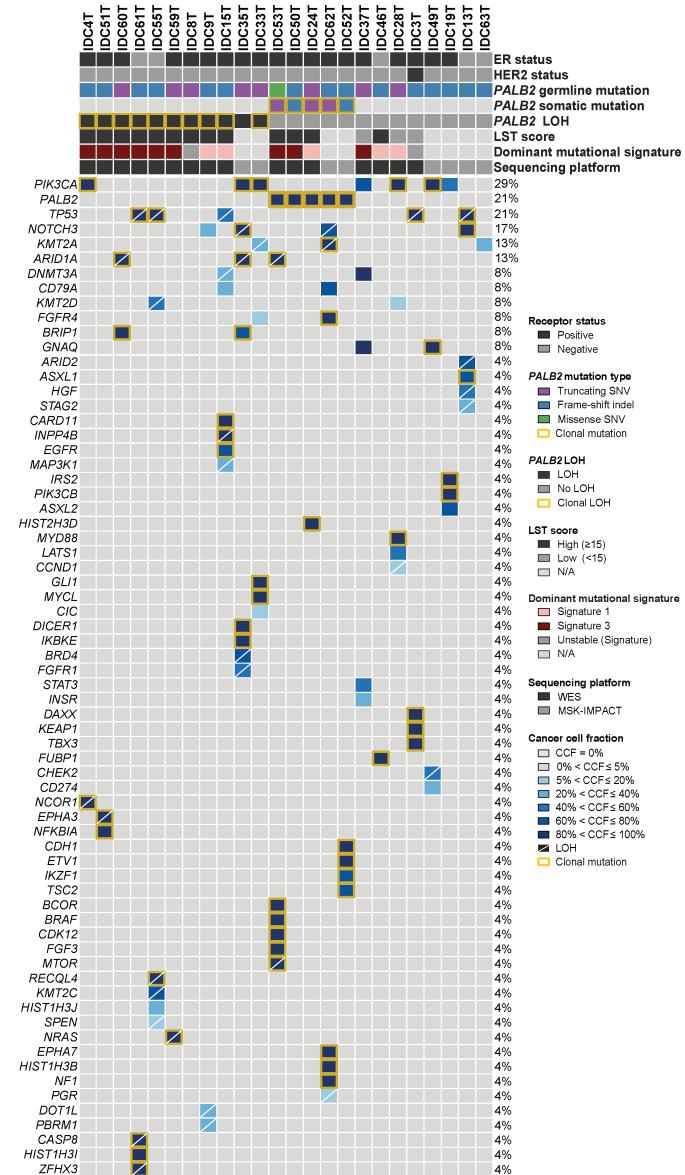
Supplementary Figure 1



Supplementary Figure 1. Validation of *PIK3CA*, *TP53*, *PALB2* and *NOTCH3* somatic mutations using Sanger sequencing.

Representative Sanger sequence electropherograms of *PIK3CA* (n=7), *TP53* (n=4) and *NOTCH3* (n=2) somatic mutations in index tumor samples, and somatic *PALB2* (n=4) mutations in both index tumor and matched normal samples. The presence of the mutation is indicated by a black arrow.

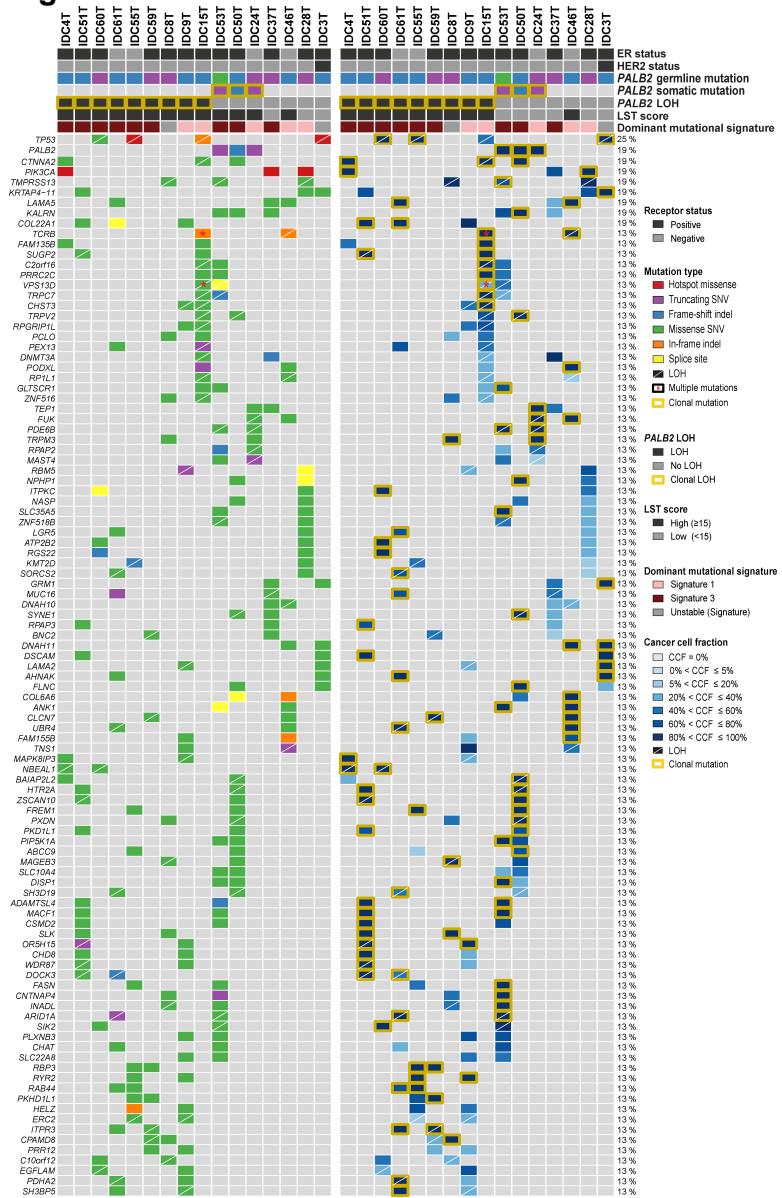
Supplementary Figure 2



Supplementary Figure 2. Cancer cell fractions of somatic mutations affecting 410 cancer genes in *PALB2*-associated breast cancers.

Cancer cell fractions (i.e. bioinformatically inferred percentage of cancer cells harboring a given somatic mutation) of non-synonymous somatic mutations affecting 410 cancer genes as defined by ABSOLUTE in the 24 *PALB2*-associated breast cancers sequenced by whole-exome (n=16) or MSK-IMPACT (n=8) sequencing. Immunohistochemical features, *PALB2* germline mutation type, presence of a second somatic *PALB2* mutation or loss of heterozygosity (LOH) of the *PALB2* wild-type allele, LST score, dominant mutational signature and sequencing platform are shown in the phenobar (top), color-coded according to the legend. Clonal somatic mutations or clonal LOH of the *PALB2* wild-type allele are indicated by a yellow box. Cancer cell fractions are color-coded according to the legend; the presence of LOH of the wild-type allele of mutated genes other than *PALB2* is represented by a diagonal bar. CCF, cancer cell fraction; indel, small insertion/deletion; LOH, loss of heterozygosity; LST, large-scale transition; N/A, not assessed; SNV, single nucleotide variant; WES, whole-exome sequencing.

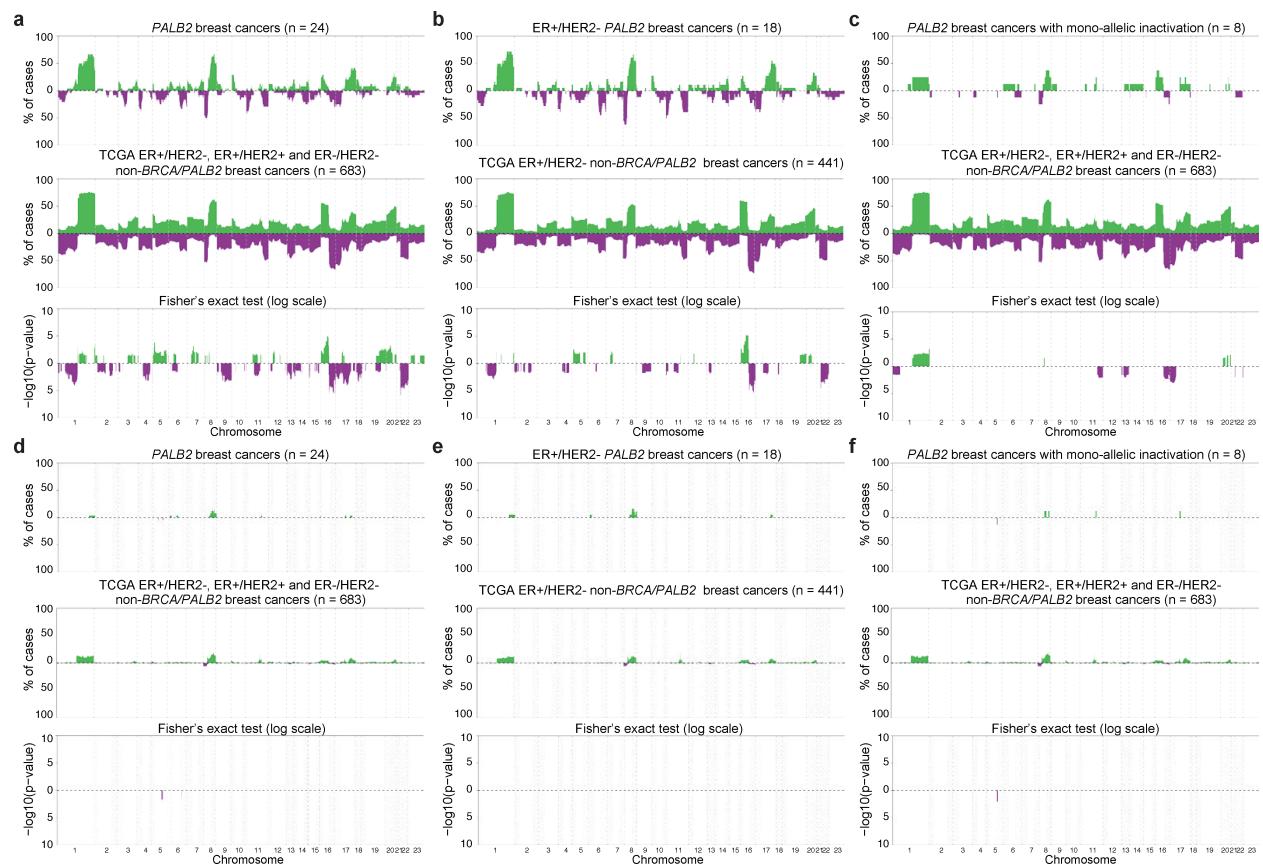
Supplementary Figure 3



Supplementary Figure 3. Repertoire of recurrent somatic mutations and corresponding cancer cell fractions in *PALB2*-associated breast cancer subjected to whole-exome sequencing.

Repertoire of recurrent ($n \geq 2$) non-synonymous somatic mutations (left), and their corresponding cancer cell fraction (i.e. bioinformatically inferred percentage of cancer cells harboring a given somatic mutation; right) identified in the 16 *PALB2*-associated breast cancer profiled by whole-exome sequencing. Immunohistochemical features, *PALB2* germline mutation type, presence of a second somatic *PALB2* mutation or loss of heterozygosity (LOH) of the *PALB2* wild-type allele, LST score and dominant mutational signature are shown in the phenobar (top), color-coded according to the legend. Clonal somatic mutations or clonal LOH of the *PALB2* wild-type allele are indicated by a yellow box. The presence of multiple somatic mutations affecting the same gene is indicated by a red asterisk. CCF, cancer cell fraction; indel, small insertion/deletion; LOH, loss of heterozygosity; LST, large-scale state transition; SNV, single nucleotide variant.

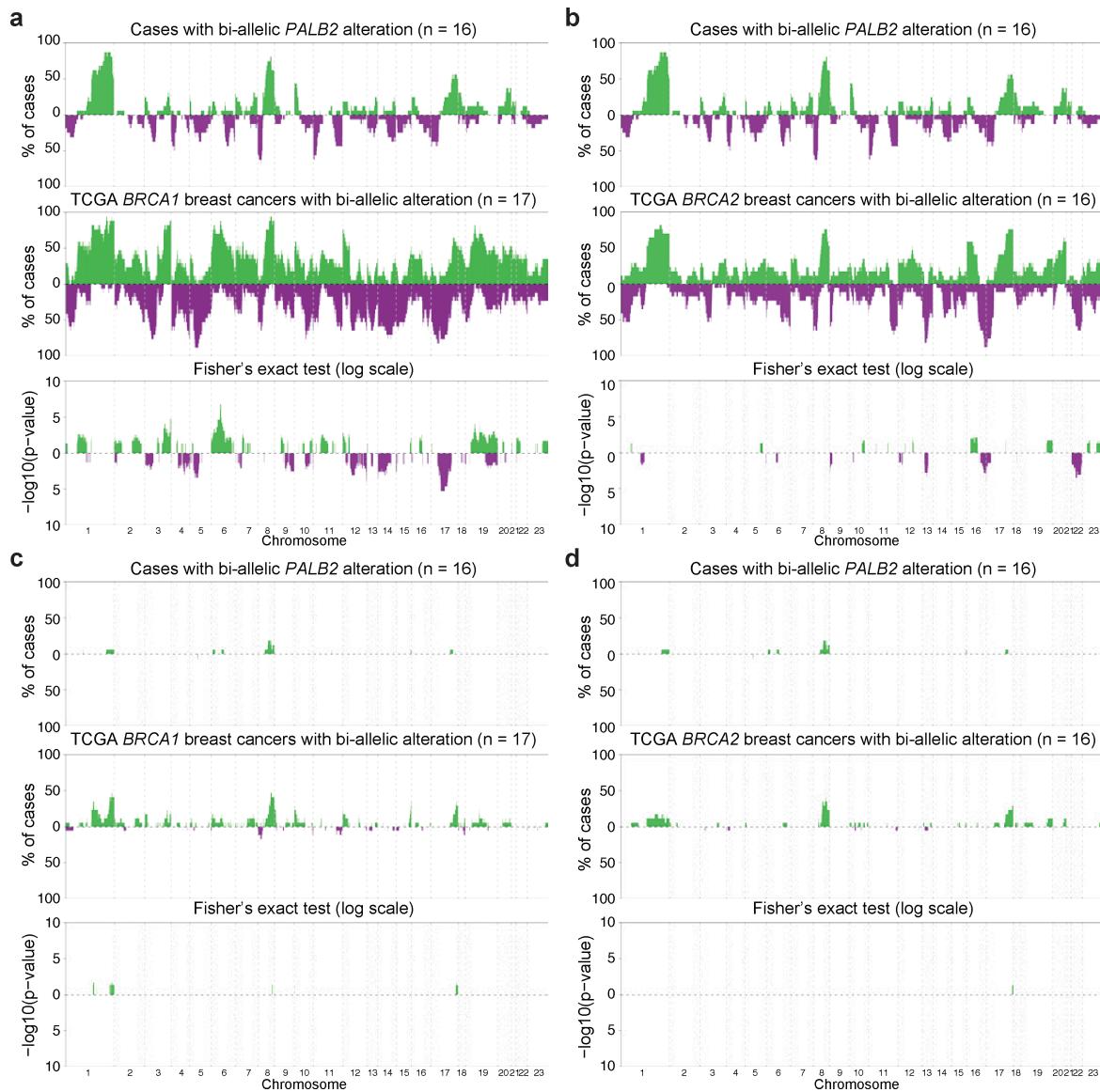
Supplementary Figure 4



Supplementary Figure 4. Comparisons of the frequencies of copy number alterations in *PALB2*-associated breast cancers and non-*BRCA1/2/PALB2* breast cancers from TCGA.

Frequency plots and multi-Fisher's exact test comparisons of gains and losses (**a-c**), amplifications and homozygous deletions (**d-f**) were performed between (**a, d**) the 24 *PALB2*-associated breast cancers and 683 non-*BRCA1/2/PALB2*-associated breast cancers (ER+/HER2-, ER+/HER2+ and ER-/HER2-), (**b, e**) between the 18 ER+/HER2- *PALB2*-associated breast cancers and 441 ER+/HER2- non-*BRCA1/2/PALB2*-associated breast cancers, (**c, f**) between the eight *PALB2*-associated breast cancers with mono-allelic *PALB2* alterations and 683 non-*BRCA1/2/PALB2*-associated breast cancers (ER+/HER2-, ER+/HER2+ and ER-/HER2-) from TCGA. The frequency of gains/amplifications (green bars) or losses/homozygous deletions (purple bars) for each gene is plotted on the y-axis, according to their genomic position on the x-axis. Inverse Log₁₀ values of the Fisher's exact test P-values are plotted according to genomic location (x-axis).

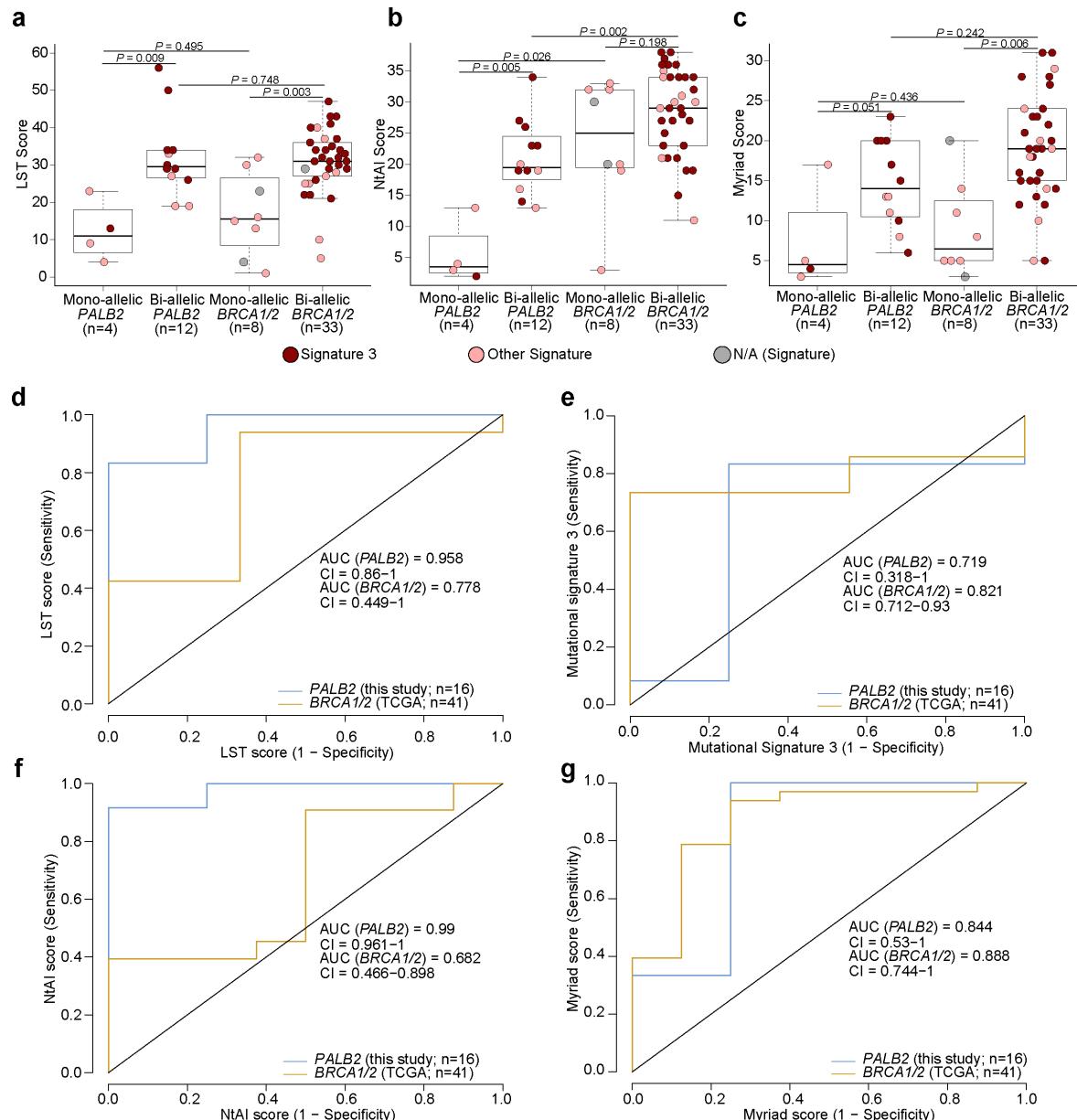
Supplementary Figure 5



Supplementary Figure 5. Comparisons of the frequencies of copy number gains and losses in *PALB2*-associated breast cancers with bi-allelic *PALB2* alterations and *BRCA1*- and *BRCA2*-associated breast cancers with bi-allelic alterations from TCGA.

(a-d) Frequency plots and multi-Fisher's exact test comparisons of copy number gains and losses between (a) the 16 *PALB2*-associated breast cancers with bi-allelic *PALB2* alterations and the 17 *BRCA1*-associated breast cancers with bi-allelic *BRCA1* alterations, and (b) the 16 *BRCA2*-associated breast cancers with bi-allelic *BRCA2* alterations from TCGA. (c-d) Frequency plots and multi-Fisher's exact test comparisons of high-level gains/amplifications and homozygous deletions between (c) the 16 *PALB2*-associated breast cancers with bi-allelic *PALB2* alterations and the 17 *BRCA1*-associated breast cancers with bi-allelic *BRCA1* alterations, and (d) the 16 *BRCA2*-associated breast cancers with bi-allelic *BRCA2* alterations. The frequency of gains/amplifications (green bars) or losses/homozygous deletions (purple bars) for each gene is plotted on the y-axis, according to their genomic position on the x-axis. Inverse \log_{10} values of the Fisher's exact test P-values are plotted according to genomic location (x-axis).

Supplementary Figure 6



Supplementary Figure 6. Comparisons of measurements of homologous recombination deficiency in *PALB2*-associated breast cancers and *BRCA1/2*-associated breast cancers with mono-allelic and bi-allelic *PALB2* or *BRCA1/2* inactivation.

(a-c) Comparisons of **(a)** Large-scale transition (LST) score, **(b)** NtAI scores and **(c)** Myriad scores of the four *PALB2*-associated breast cancers with mono-allelic *PALB2* alterations, the 12 *PALB2*-associated breast cancers with bi-allelic *PALB2* alterations, the eight *BRCA1/2*-associated breast cancers with mono-allelic *BRCA1* or *BRCA2* alterations and the 33 *BRCA1/2*-associated breast cancers with bi-allelic *BRCA1* or *BRCA2* alterations. **(d-g)** Receiver operating characteristic (ROC) curves comparing performance of **(d)** LST scores, **(e)** mutational signature 3 (%), **(f)** NtAI scores and **(g)** Myriad scores in *PALB2*-associated breast cancers and *BRCA1/2*-associated breast cancers for the identification of *PALB2* and *BRCA1/2* bi-allelic inactivation. AUC, area under the ROC curve; CI, confidence interval.

Supplementary Table 1. Sequencing statistics of tumor and paired normal samples of *PALB2*-associated breast cancers sequenced by whole-exome or MSK-IMPACT targeted massively parallel sequencing.

Case ID	Sequencing type	Target Territory	Total Reads	Percent Selected Bases	Mean Target Coverage	Percent Target Bases 2X	Percent Target Bases 10X	Percent Target Bases 20X	Percent Target Bases 30X	Percent Target Bases 40X	Percent Target Bases 50X	Percent Target Bases 100X
IDC13N	MSK-IMPACT	1362579	17,587,599	67.86%	316	99.50%	99.25%	99.04%	98.79%	98.49%	98.04%	92.95%
IDC13T	MSK-IMPACT	1362579	21,890,639	54.60%	128	98.95%	98.25%	97.65%	96.58%	94.91%	92.47%	65.62%
IDC19N	MSK-IMPACT	1362579	104,539,241	75.41%	789	99.53%	99.43%	99.35%	99.28%	99.19%	99.12%	98.77%
IDC19T	MSK-IMPACT	1362579	38,378,568	51.96%	227	99.25%	98.77%	98.34%	97.75%	96.73%	95.05%	78.22%
IDC33N	MSK-IMPACT	1522367	22,171,814	84.58%	585	99.46%	99.34%	99.23%	99.12%	98.98%	98.83%	97.67%
IDC33T	MSK-IMPACT	1522367	15,369,766	82.02%	324	99.16%	98.76%	98.50%	98.29%	98.04%	97.66%	93.05%
IDC35N	MSK-IMPACT	1362579	14,260,504	67.93%	172	99.19%	98.58%	96.96%	93.67%	89.14%	83.72%	53.91%
IDC35T	MSK-IMPACT	1362579	6,712,966	55.84%	124	99.02%	97.84%	94.32%	88.10%	80.93%	73.46%	42.67%
IDC49N	MSK-IMPACT	1362579	54,791,577	77.98%	1452	99.51%	99.45%	99.40%	99.35%	99.31%	99.25%	99.02%
IDC49T	MSK-IMPACT	1362579	36,941,697	62.09%	236	98.87%	98.26%	97.89%	97.39%	96.65%	95.65%	87.28%
IDC52N	MSK-IMPACT	1362579	30,113,913	68.94%	572	99.53%	99.44%	99.33%	99.21%	99.11%	99.00%	97.77%
IDC52T	MSK-IMPACT	1362579	9,519,069	62.84%	73	98.35%	93.74%	83.48%	71.71%	60.98%	51.78%	24.03%
IDC62N	MSK-IMPACT	1522367	15,173,050	84.93%	502	99.42%	99.15%	98.95%	98.75%	98.55%	98.35%	97.58%
IDC62T	MSK-IMPACT	1522367	32,271,761	84.50%	904	99.46%	99.30%	99.16%	99.06%	98.96%	98.86%	98.41%
IDC63N	MSK-IMPACT	1522367	15,767,985	89.13%	518	99.46%	99.26%	99.09%	98.96%	98.82%	98.65%	97.89%
IDC63T	MSK-IMPACT	1522367	21,077,959	89.66%	678	99.28%	99.05%	98.85%	98.67%	98.53%	98.40%	97.72%
IDC15N	Whole exome	51542852	133,574,121	85.80%	146	93.49%	91.44%	89.75%	87.45%	84.53%	81.20%	60.98%
IDC15T	Whole exome	51542852	67,920,645	81.58%	40	87.47%	74.55%	61.01%	47.91%	36.45%	27.26%	6.62%
IDC24N	Whole exome	51542852	75,039,053	85.83%	88	91.64%	85.64%	79.57%	73.71%	67.56%	60.98%	30.49%
IDC24T	Whole exome	51542852	109,782,207	87.43%	130	92.15%	87.81%	83.63%	79.78%	76.02%	72.20%	50.30%
IDC28N	Whole exome	51542852	88,885,131	89.73%	106	92.14%	88.21%	83.49%	78.94%	74.43%	69.75%	43.19%
IDC28T	Whole exome	51542852	100,553,535	87.98%	105	91.69%	86.60%	81.46%	76.63%	71.71%	66.44%	38.35%
IDC37N	Whole exome	51189318	233,784,412	82.85%	247	99.67%	99.28%	98.92%	98.52%	97.98%	97.22%	88.66%
IDC37T	Whole exome	51189318	250,533,924	78.34%	268	99.79%	99.57%	99.24%	98.70%	97.80%	96.52%	85.10%
IDC3N	Whole exome	51542852	86,590,460	82.29%	85	92.30%	86.29%	77.82%	68.33%	58.12%	48.31%	21.15%
IDC3T	Whole exome	51542852	110,163,811	90.09%	136	92.18%	88.46%	84.66%	81.29%	78.07%	74.87%	57.27%
IDC46N	Whole exome	51542852	139,890,230	83.78%	155	93.59%	91.31%	89.46%	86.99%	84.05%	80.79%	61.75%
IDC46T	Whole exome	51542852	44,683,165	88.43%	33	85.88%	70.18%	52.42%	37.15%	26.07%	18.72%	5.14%
IDC4N	Whole exome	51542852	107,232,015	85.31%	122	93.31%	91.13%	88.67%	85.40%	81.61%	77.58%	54.44%
IDC4T	Whole exome	51542852	114,347,035	89.63%	139	92.32%	88.89%	85.18%	81.81%	78.57%	75.34%	57.72%
IDC50N	Whole exome	51542852	123,171,969	85.09%	136	93.25%	90.97%	88.94%	86.21%	82.96%	79.38%	58.75%
IDC50T	Whole exome	51542852	72,671,924	84.91%	77	91.18%	83.69%	75.58%	67.74%	60.03%	52.73%	26.06%
IDC51N	Whole exome	51542852	112,888,954	81.16%	118	93.54%	90.84%	88.22%	84.66%	80.34%	75.45%	48.79%
IDC51T	Whole exome	51542852	66,148,714	87.59%	70	90.09%	81.77%	73.48%	64.98%	56.15%	47.52%	19.19%
IDC53N	Whole exome	51542852	186,199,465	84.58%	193	93.87%	91.33%	89.56%	87.52%	85.14%	82.40%	66.03%
IDC53T	Whole exome	51542852	68,290,444	80.92%	69	91.90%	85.02%	75.96%	66.47%	57.10%	48.43%	20.94%
IDC55N	Whole exome	51189318	165,293,498	62.27%	139	99.69%	99.17%	98.38%	96.97%	94.63%	91.24%	64.21%
IDC55T	Whole exome	51189318	267,310,420	81.28%	289	99.77%	99.53%	99.18%	98.69%	97.94%	96.88%	87.42%
IDC59N	Whole exome	51189318	185,081,923	84.63%	173	99.80%	99.49%	98.86%	97.81%	96.22%	94.06%	75.86%
IDC59T	Whole exome	51189318	175,236,053	91.76%	213	99.80%	99.51%	99.00%	98.20%	97.04%	95.45%	81.82%
IDC60N	Whole exome	51189318	73,994,558	65.70%	46	96.57%	91.44%	81.89%	68.30%	52.88%	38.47%	4.96%
IDC60T	Whole exome	51189318	89,242,346	57.28%	46	96.68%	92.05%	83.10%	69.18%	52.84%	37.56%	4.33%
IDC61N	Whole exome	51189318	152,388,926	57.82%	86	94.27%	86.65%	81.91%	76.44%	70.40%	64.04%	33.92%
IDC61T	Whole exome	51189318	143,124,548	50.78%	77	94.81%	85.35%	78.76%	71.65%	64.33%	57.05%	27.50%
IDC8N	Whole exome	51542852	101,656,174	86.68%	121	93.09%	90.79%	87.94%	84.45%	80.75%	76.96%	55.65%
IDC8T	Whole exome	51542852	124,287,664	89.61%	151	92.46%	89.37%	86.03%	83.02%	80.17%	77.37%	62.44%
IDC9N	Whole exome	51542852	122,188,395	85.89%	142	93.35%	91.31%	89.22%	86.56%	83.52%	80.31%	62.76%
IDC9T	Whole exome	51542852	107,445,459	86.92%	119	91.72%	86.91%	82.76%	79.16%	75.63%	72.00%	51.24%

Supplementary Table 2. Clonal and subclonal variants, variant allele fraction range and tumor purity of the *PALB2*-associated breast cancers in this study

Sample ID	# Clonal Variants	# Subclonal Variants	# Total Variants	VAF Min	VAF Max	Purity	<i>PALB2</i> Status
IDC13T	3	3	6	6.38%	29.63%	0.47	Monoallelic
IDC15T	73	82	155	4.17%	75%	0.76	Biallelic
IDC19T	2	2	4	10.48%	26.09%	0.47	Monoallelic
IDC24T	32	22	54	4.94%	38.46%	0.43	Biallelic
IDC28T	9	58	67	4.11%	23.33%	0.52	Monoallelic
IDC33T	3	3	6	4.64%	40.93%	0.71	Biallelic
IDC35T	6	2	8	17.02%	64.52%	0.25	Biallelic
IDC37T	9	70	79	2.92%	23.08%	0.41	Monoallelic
IDC3T	27	10	37	3.45%	43.71%	0.45	Monoallelic
IDC46T	55	18	73	5.75%	77.65%	0.69	Monoallelic
IDC49T	2	2	4	5.71%	35%	0.51	Monoallelic
IDC4T	41	35	76	3.55%	76.09%	0.68	Biallelic
IDC50T	110	37	147	3.07%	81.82%	0.7	Biallelic
IDC51T	109	22	131	5.68%	64.71%	0.55	Biallelic
IDC52T	5	0	5	14.29%	41.94%	0.56	Biallelic
IDC53T	124	71	195	3.28%	67.57%	0.55	Biallelic
IDC55T	65	40	105	2.84%	73.51%	0.79	Biallelic
IDC59T	33	39	72	3.72%	77.78%	0.68	Biallelic
IDC60T	64	39	103	4.96%	47.69%	0.44	Biallelic
IDC61T	47	39	86	2.84%	86.67%	0.68	Biallelic
IDC62T	7	3	10	3.41%	53.45%	0.67	Biallelic
IDC63T	0	1	1	6.99%	6.99%	0.3	Monoallelic
IDC8T	15	26	41	5.14%	40%	0.4	Biallelic
IDC9T	25	136	161	3.55%	83.33%	0.6	Biallelic

VAF, variant allele fraction

Supplementary Table 3
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Supplementary Table 3. List of non-synonymous somatic mutations identified in the *PALB2*-associated breast cancers sequenced by whole-exome sequencing or MSK-IMPACT targeted massively parallel sequencing.

IC041	YES9B	p.262D	Misense Mutation	11	6493386	G	A	904	5.77%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.27	0.00014	12%	Subnormal		WES
IC041	ATXN7	p.R179C	Nonsense Mutation	7	2584846	G	A	247	18.0%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.77	0.0358	58%	Subnormal		WES
IC041	ATXN7	p.R129C	Nonsense Mutation	9	3465150	G	A	73	8.8%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.27	0.00015	14%	Subnormal		WES
IC041	MARFA	p.S38R	Nonsense Mutation	5	6598926	G	A	143	5.59%	Stop	PASSANGER/ROTHER	Passenger	stop	0.2	0.0001	16%	Subnormal		WES
IC041	F11T5	p.D177T	Nonsense Mutation	9	1659695	G	A	5	21.50%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.71	0.0001	10%	Subnormal	TRUE	WES
IC041	ANKG010	p.X5_S6del	Splice Site	5	7409268	G	C	49	22.40%	Stop	PASSANGER/ROTHER	Passenger	stop	0.62	0.00013	47%	Normal		WES
IC041	ASXL1	p.R164K	Misense Mutation	5	165447701	G	C	1	100.0%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.64	0.00011	24%	Subnormal		WES
IC041	ATXN7	p.R179C	Nonsense Mutation	5	2584846	G	A	247	18.0%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.77	0.0358	58%	Subnormal		WES
IC041	CHMP7Z	p.S27L	Misense Mutation	1	27278605	G	A	186	4.84%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.19	0.00099	10%	Subnormal		WES
IC041	CHMP7Z	p.D217N	Misense Mutation	5	31188503	G	A	44	27.27%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.18	0.00016	95%	Normal		WES
IC041	CHMP7Z	p.D217N	Misense Mutation	5	31188503	G	A	44	27.27%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.18	0.00016	95%	Normal	TRUE	WES
IC041	CNSP8	p.K305Q	Misense Mutation	8	87656003	G	T	141	4.26%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.34	0.0001	100%	Subnormal		WES
IC041	DHCR4	p.R118Q	Misense Mutation	2	3906343	G	A	55	9.30%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.43	0.00029	21%	Subnormal		WES
IC041	DHCR4	p.R118Q	Misense Mutation	2	3906343	G	A	55	9.30%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.43	0.00029	21%	Subnormal		WES
IC041	FAM3C	p.V78I	Misense Mutation	10	8119219	G	A	55	9.05%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.35	0.0008	95%	Subnormal		WES
IC041	GT18C2	p.S37L	Misense Mutation	2	2759294	G	A	50	10.00%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.4	0.00011	17%	Subnormal		WES
IC041	GT18C2	p.R25Q	Misense Mutation	22	4675102	G	A	97	7.25%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.22	0.0003	14%	Subnormal		WES
IC041	GUO702	p.Y439H	Misense Mutation	17	7989697	G	A	161	5.79%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.23	0	11%	Subnormal		WES
IC041	HMOX2	p.E146K	Misense Mutation	15	4597490	G	A	760	14.83%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.65	0.00042	57%	Subnormal		WES
IC041	HOMD12	p.V223I	Misense Mutation	2	17695342	G	A	233	4.51%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.18	0.0008	87%	Subnormal		WES
IC041	ITPKC	p.R55Q	Misense Mutation	19	4129215	G	A	29	13.75%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.65	0.0179	21%	Subnormal		WES
IC041	KIAA0400	p.R460H	Misense Mutation	17	2654848	G	A	72	8.94%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.23	0.00027	12%	Subnormal		WES
IC041	KIAA1102	p.V486M	Misense Mutation	12	12577180	G	A	34	14.71%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.59	0.0177	25%	Subnormal		WES
IC041	KMT2A	p.Q448L	Misense Mutation	10	4869145	G	A	480	10.00%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.75	0.0001	100%	Subnormal	TRUE	WES
IC041	ART4A/F11	p.S83R	Misense Mutation	17	9924719	G	C	16	19.70%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.75	0.00048	25%	Subnormal		WES
IC041	ATXN7	p.L105P	Misense Mutation	12	7105602	G	A	93	5.38%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.21	0	95%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES
IC041	ATXN7	p.R54Q	Misense Mutation	18	5979500	G	A	134	4.48%	Delsite	PASSANGER/ROTHER	Passenger	passenger	0.25	0.0001	20%	Subnormal		WES

ISOCAT	C50407	p.R602	Misense Mutation	5	37246265	G	A	76	9.85%	Neutral	PASSANGER	passenger	inh	0.26	0.00074	15%	Subnormal		WES		
ISOCAT	C50407	p.L117T	Misense Mutation	5	34112106	G	A	76	9.85%	Neutral	PASSANGER	ROTHHER	Dwarf, body hypotonic	inh	1	0.00702	55%	Normal		WES	
ISOCAT	C50407	p.G49E	Misense Mutation	11	34112106	G	C	50	49.40%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00532	62%	Normal		WES	
ISOCAT	C50407	p.T737S	Misense Mutation	3	42593501	G	T	30	30.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.C100R	Misense Mutation	17	20590955	G	T	35	11.43%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.L120W	Misense Mutation	6	49956252	G	C	17	17.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.D54A	Misense Mutation	14	73418526	G	A	18	21.22%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00707	35%	Normal		WES	
ISOCAT	C50407	p.Q100P	Misense Mutation	12	72438897	G	A	60	8.05%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00707	35%	Normal		WES	
ISOCAT	C50407	p.DNAH10	Misense Mutation	1	35337274	G	A	33	27.27%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00707	35%	Normal		WES	
ISOCAT	C50407	p.E117K	Misense Mutation	16	36099108	G	A	40	10.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00796	21%	Subnormal		WES	
ISOCAT	C50407	p.F904Q	p.P53L	Misense Mutation	9	70571261	G	T	30	30.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00531	64%	Normal	TRUE	WES
ISOCAT	C50407	p.F411L	Misense Mutation	17	20590955	G	T	35	11.43%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal	TRUE	WES	
ISOCAT	C50407	p.C180T	Misense Mutation	17	20590955	G	C	15	17.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal	TRUE	WES	
ISOCAT	C50407	p.S93Y	Misense Mutation	7	15590172	G	C	25	49.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.A120T	Misense Mutation	2	15594520	G	C	20	42.86%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00719	55%	Normal		WES	
ISOCAT	C50407	p.L481L	Misense Mutation	20	80988868	G	A	20	9.85%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00719	55%	Normal		WES	
ISOCAT	C50407	p.L482C	Misense Mutation	15	16020269	G	A	48	7.80%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00719	55%	Normal		WES	
ISOCAT	C50407	p.M71I	Misense Mutation	12	72438897	G	A	50	8.05%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.47	0.00480	15%	Subnormal		WES	
ISOCAT	C50407	p.E350L	Misense Mutation	16	8718451	G	A	33	27.27%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00707	35%	Normal		WES	
ISOCAT	C50407	p.R51Q	Misense Mutation	15	48470542	G	C	21	37.14%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00531	64%	Normal		WES	
ISOCAT	C50407	p.K171K	Misense Mutation	16	36099108	G	A	40	10.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00796	21%	Subnormal		WES	
ISOCAT	C50407	p.D119S	Misense Mutation	11	34112106	G	C	26	39.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00531	64%	Normal	YES	WES	
ISOCAT	C50407	p.N69L	Misense Mutation	5	154654733	G	C	17	17.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00548	86%	Normal		WES	
ISOCAT	C50407	p.P68L	Misense Mutation	5	154654733	G	C	15	15.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.P219R	Misense Mutation	16	70571261	G	C	29	24.14%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00736	45%	Normal		WES	
ISOCAT	C50407	p.D101R	Misense Mutation	16	70571261	G	C	20	21.43%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00736	45%	Normal		WES	
ISOCAT	C50407	p.I59G	Misense Mutation	17	20590955	G	C	25	49.00%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.K191T	Misense Mutation	2	15594520	G	A	19	21.05%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00722	27%	Normal		WES	
ISOCAT	C50407	p.R200T	Misense Mutation	19	1882725	G	A	25	49.11%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00493	63%	Normal		WES	
ISOCAT	C50407	p.R201T	Misense Mutation	22	30775002	G	C	18	20.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00501	30%	Normal		WES	
ISOCAT	C50407	p.R202T	Misense Mutation	22	30775002	G	C	17	21.05%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00501	30%	Normal		WES	
ISOCAT	C50407	p.S240L	Misense Mutation	5	48505151	G	C	27	14.81%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.S240L	Misense Mutation	5	48505151	G	C	25	14.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.S240H	Misense Mutation	1	24093174	G	C	31	77.90%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.S240H	Misense Mutation	1	24093174	G	C	30	73.40%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.E33Q	Misense Mutation	18	4469628	G	A	48	9.85%	Neutral	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00524	71%	Normal		WES	
ISOCAT	C50407	p.T340M	Misense Mutation	18	4469628	G	A	45	8.80%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.48	0.00420	15%	Subnormal		WES	
ISOCAT	C50407	p.T340M	Misense Mutation	3	3688468	G	A	45	8.80%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.48	0.00420	15%	Subnormal		WES	
ISOCAT	C50407	p.U69R	Misense Mutation	1	19499522	G	A	22	18.85%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00489	86%	Normal		WES	
ISOCAT	C50407	p.R170Q	Misense Mutation	16	70571261	G	A	18	18.70%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00736	45%	Normal		WES	
ISOCAT	C50407	p.R170Q	Misense Mutation	16	70571261	G	A	17	18.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00736	45%	Normal		WES	
ISOCAT	C50407	p.Y189V	Misense Mutation	1	19499522	G	A	22	18.85%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00489	86%	Normal		WES	
ISOCAT	C50407	p.A191T	Misense Mutation	5	48505151	G	C	27	14.81%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00524	71%	Normal		WES	
ISOCAT	C50407	p.C192L	Misense Mutation	12	10515172	G	C	32	24.41%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00736	45%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	11	22201059	G	C	24	19.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00531	64%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	11	22201059	G	C	23	19.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	1	0.00531	64%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	22	30811030	G	C	18	20.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	22	30811030	G	C	17	19.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	18	20.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	17	19.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	16	18.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	15	18.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	14	17.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	13	17.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	12	16.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	11	16.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	10	15.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	9	15.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	8	14.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	7	14.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	6	13.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	5	13.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	4	12.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	3	12.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	2	11.50%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal		WES	
ISOCAT	C50407	p.A205L	Misense Mutation	1	20590955	G	C	1	11.00%	Deleterious	PASSANGER	ROTHHER	Dwarf	passenger	0.58	0.00581	86%	Normal	</td		

DCS01	NASP	p.A289V	Misense Mutation	1	46932099	G	T	28	97.7%	Neutral	PASSANGER OTHER	Passenger	passenger	0.44	0.29926	15%	Subnormal		WES
DCS01	NPBP1	p.S349K	Misense Mutation	2	110901069	G	T	27	92.2%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.92	0.85292	39%	Clinical		WES
DCS01	ARCC2	p.T717T	Misense Mutation	3	150625254	TACAGGAAACTCTTGATCTGATGATGTTG	C	18	14.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.27	0.57548	24%	Clinical		WES
DCS01	DTF3	p.S620R	Misense Mutation	10	74992058	G	A	223	14.5%	Neutral	PASSANGER OTHER	Passenger	passenger	0.89	0.71598	87%	Clinical		WES
DCS01	PANK2	p.D241I	Misense Mutation	20	388975	G	T	67	93.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.78397	55%	Clinical		WES
DCS01	PCMV42	p.S207R	Misense Mutation	5	146937442	G	A	12	85.7%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.95553	86%	Clinical		WES
DCS01	PCMV42	p.R207Q	Misense Mutation	11	18152058	G	A	58	97.1%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.83	0.80779	57%	Clinical		WES
DCS01	RASGRIP	p.E104Q	Misense Mutation	2	37474956	A	C	28	92.2%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.79	0.5983	32%	Clinical		WES
DCS01	PTEN	p.L111P	Misense Mutation	19	49119158	G	T	58	82.7%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85242	25%	Clinical		WES
DCS01	SECBP1	p.T120R	Misense Mutation	4	110498683	T	G	32	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.94453	85%	Clinical		WES
DCS01	SEMA3B	p.G58T	Misense Mutation	3	52472056	G	A	58	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.9582	75%	Clinical		WES
DCS01	SH2D1A	p.R887W	Misense Mutation	4	162640451	G	A	24	17.2%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.92	0.85207	33%	Clinical		WES
DCS01	SLC27A1	p.Q429Y	Misense Mutation	15	50519158	G	A	16	95.0%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.39	0.31544	14%	Subnormal		WES
DCS01	SLC27A1	p.A392S	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SLC27A1	p.Q429Y	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SLC27A1	p.A392S	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SLC27A1	p.Q429Y	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SLC27A1	p.Q429Y	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SLC27A1	p.Q429Y	Misense Mutation	1	43593390	G	A	74	22.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.95	0.79593	57%	Clinical		WES
DCS01	SUMF1	p.R469H	Misense Mutation	17	2703658	G	A	71	29.9%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.92959	77%	Clinical		WES
DCS01	SYNE1	p.K2205	Misense Mutation	6	152793254	A	T	11	81.8%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.94110	88%	Clinical		WES
DCS01	FA2M2	p.P2121L	Misense Mutation	9	1741520	G	A	47	24.3%	Neutral	PASSANGER OTHER	Passenger	passenger	1	0.93646	61%	Clinical		WES
DCS01	TMSM25	p.G35A	Misense Mutation	14	21959591	G	A	49	24.4%	Neutral	PASSANGER OTHER	Passenger	passenger	1	0.92867	60%	Clinical		WES
DCS01	TSHZ9	p.I219Y	Misense Mutation	14	2254777	G	A	87	97.0%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.92445	95%	Clinical		WES
DCS01	TRIP12	p.A435D	Misense Mutation	2	23068379	G	C	42	45.4%	Deleterious	PASSANGER OTHER	Passenger	passenger	1	0.93693	77%	Clinical		WES
DCS01	TRIP12	p.E351D	Misense Mutation	12	1859541	G	A	64	93.6%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85381	52%	Clinical		WES
DCS01	TRIP12	p.E351D	Misense Mutation	16	1859541	G	A	64	93.6%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85381	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	9	48486509	G	C	100	100%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.97	0.77994	95%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	10	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93	0.85201	52%	Clinical		WES
DCS01	WDR33	p.Y363C	Misense Mutation	15	48486509	G	C	104	94.3%	Deleterious	PASSANGER OTHER	Passenger	passenger	0.93					

LOCID	FAMCF	p.V257_P2126a	In Frame Del	11	2296629	GCGGCGCTCACAGCGCGAC	G	588	5.16%	Indel pathogenic	0.25	0	12%	Subclinical	TRUE	WES			
LOCID1	IPNCH1	p.3814del	In Frame Del	7	44837912	GGAT	G	124	25.81%	passenger	1	0.00507	76%	Clinical		WES			
LOCID2	TEB444	p.L285_P278del	In Frame Del	11	78891652	TGAGAO	G	23	21.70%	passenger	1	0.01745	35%	Clinical		WES			
LOCID3	SEPT9	p.R80Q	Misense Mutation	22	42290339	G	A	33	30.30%	Deleterious	PASSANGER ROTHER	Passenger	inh	1	0.00486	54%	Clinical		WES
LOCID4	ASBT	p.Y95R	Misense Mutation	17	67116959	G	A	50	30.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.04	0.02097	57%	Clinical		WES
LOCID5	ADAMTSL2	p.R136G	Misense Mutation	5	33549431	G	C	213	43.63%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.0039	94%	Clinical		WES
LOCID6	AKM9301	p.D272C	Misense Mutation	5	74443057	G	C	100	25.00%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.02693	30%	Subclinical		WES
LOCID7	CDH1	p.198L	Misense Mutation	18	24447208	A	G	58	30.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00167	84%	Clinical		WES
LOCID8	APGP4	p.R97K	Misense Mutation	18	24447208	A	G	58	30.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00167	41%	Clinical	TRUE	WES
LOCID9	ARH14	p.477M	Misense Mutation	22	42290339	G	A	12	41.67%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00167	41%	Clinical	TRUE	WES
LOCID10	ARH14N	p.477M	Misense Mutation	22	42290339	G	A	12	41.67%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00167	41%	Clinical	TRUE	WES
LOCID11	BMP9B	p.P95A	Misense Mutation	4	96576242	G	C	40	35.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.00279	76%	Clinical		WES
LOCID12	CDH17	p.T12V	Misense Mutation	12	27295052	T	A	72	8.94%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.01190	14%	Subclinical		WES
LOCID13	CDH21	p.E54Y	Misense Mutation	19	13372373	A	C	47	31.51%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00120	70%	Subclinical		WES
LOCID14	CDH2	p.E39Q	Misense Mutation	18	2507198	C	Q	38	23.88%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00173	45%	Clinical		WES
LOCID15	CDH2	p.E39Q	Misense Mutation	18	2507198	C	Q	38	23.88%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00173	45%	Clinical		WES
LOCID16	CDH4	p.A46D	Misense Mutation	20	8053384	G	A	514	8.81%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00210	48%	Subclinical		WES
LOCID17	CDH20	p.R15H	Misense Mutation	9	9059518	G	C	7	27.40%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00227	70%	Clinical		WES
LOCID18	CDH2	p.S34R	Misense Mutation	18	80718462	G	C	114	12.20%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00293	42%	Subclinical		WES
LOCID19	CDH2	p.G54A	Misense Mutation	5	8134930	G	C	6	30.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.00342	31%	Clinical		WES
LOCID20	CHRNA4	p.A50D	Misense Mutation	20	6181675	G	C	313	20.50%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00271	54%	Clinical		WES
LOCID21	CHS27	p.P28R	Misense Mutation	1	46344205	G	C	179	15.64%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00287	52%	Subclinical		WES
LOCID22	CHS27	p.P28R	Misense Mutation	1	46344205	G	C	179	15.64%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00287	52%	Subclinical		WES
LOCID23	CHS27	p.Q78R	Misense Mutation	3	12691538	G	Q	44	21.40%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.00367	79%	Clinical		WES
LOCID24	CHS27	p.Q78R	Misense Mutation	3	12691538	G	Q	44	21.40%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00287	52%	Subclinical		WES
LOCID25	CHS27	p.L167I	Misense Mutation	1	34481454	G	T	509	11.60%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00350	47%	Subclinical		WES
LOCID26	CHS27	p.R187H	Misense Mutation	14	45077054	G	C	43	18.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00348	45%	Clinical		WES
LOCID27	CHS27	p.R187H	Misense Mutation	14	45077054	G	C	43	18.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00348	45%	Clinical		WES
LOCID28	CHS27	p.A77T	Misense Mutation	14	59113734	G	C	136	20.30%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00319	88%	Clinical		WES
LOCID29	CHS27	p.A77T	Misense Mutation	14	59113734	G	C	136	20.30%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00319	88%	Clinical		WES
LOCID30	CHS27	p.K227V	Misense Mutation	6	74154631	A	T	25	16.00%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00282	26%	Clinical		WES
LOCID31	CHS27	p.A428P	Misense Mutation	19	3818173	G	A	101	18.80%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00274	27%	Subclinical		WES
LOCID32	CHS27	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID33	CHS27	p.D42P	Misense Mutation	17	27068770	G	A	132	17.40%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00251	95%	Clinical		WES
LOCID34	CHS27	p.A42P	Misense Mutation	17	27068770	G	A	132	17.40%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00251	95%	Clinical		WES
LOCID35	CHS27	p.R187P	Misense Mutation	6	38817146	G	C	27	16.70%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.00354	85%	Clinical		WES
LOCID36	CHS27	p.R195C	Misense Mutation	2	2517954	G	A	34	18.82%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00348	45%	Subclinical		WES
LOCID37	CHS27	p.R195C	Misense Mutation	17	198921	G	C	66	18.70%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00274	46%	Subclinical		WES
LOCID38	CHS27	p.F155L	Misense Mutation	6	80381745	G	C	57	22.31%	Deleterious	PASSANGER ROTHER	Passenger	passenger	1	0.00372	57%	Clinical		WES
LOCID39	ENPP2	p.S62G	Misense Mutation	8	12694465	G	C	92	5.43%	Neutral	PASSANGER ROTHER	Passenger	passenger	0.05	0.00248	26%	Subclinical		WES
LOCID40	FAM120C	p.A271	Misense Mutation	9	54409553	G	C	14	24.55%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	52%	Clinical		WES
LOCID41	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID42	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID43	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID44	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID45	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID46	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID47	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID48	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID49	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID50	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID51	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID52	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID53	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID54	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID55	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID56	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID57	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID58	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID59	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID60	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID61	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID62	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID63	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID64	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID65	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.00281	51%	Clinical		WES
LOCID66	FAM120B	p.N40D	Misense Mutation	17	27068770	G	C	179	18.30%	Deleterious	PASSANGER ROTHER	Passenger	passenger	0.05	0.002				

025671	GUCY1A3	p.858R	I	Misseense_Mutation	4	158861217	G	D	155	54.1%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.81	0.7887	77%	asbestose		WES
025672	HNRNCA	p.73Q	I	Missense_Mutation	12	69703860	T	G	25	60.0%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88487	73%	Cancer		WES
025673	PTEN	p.1792L	I	Missense_Mutation	3	128981972	G	C	141	56.74%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.95	0.80231	57%	cancer		WES
025674	KIF19	p.2529I	I	Missense_Mutation	17	223412053	G	A	328	34.7%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.93	0.81533	79%	Cancer		WES
025675	MEF2D	p.1349T	I	Missense_Mutation	15	60319989	A	T	211	36.87%	Dileukosid	CANCER	skler. pathogene		0.98	0.88484	80%	Cancer		WES	
025676	MYOZ2	p.205M	I	Missense_Mutation	10	17384338	G	C	265	4.67%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.11	0.90035	86%	Cancer		WES
025677	NAPRA	p.5434N	I	Missense_Mutation	18	10546262	G	A	265	3.95%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.14	0	7%	asbestose		WES
025678	MAP3K1	p.530I	I	Missense_Mutation	2	18768234	C	A	259	31.46%	Neutral	CANCER	skler. pathogene		0.91148	0.8475	93%	Cancer		WES	
025679	MTORC1B	p.1350I	I	Missense_Mutation	11	128114880	T	C	261	35.25%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.11	0.88487	73%	Cancer		WES
025680	CTGZ	p.2330R	I	Missense_Mutation	11	17853851	T	C	267	37.34%	Neutral	CANCER	skler. pathogene		0.99	0.80138	83%	Cancer		WES	
025681	PTEN	p.1254S	I	Missense_Mutation	6	110469863	G	T	101	18.5%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.82	0.11952	39%	Cancer		WES
025682	PTCHD3	p.276L	I	Missense_Mutation	10	27881104	C	G	177	38.8%	Neutral	CANCER	skler. pathogene		0.02	0.85233	4%	Cancer		WES	
025683	RAB44	p.471T	I	Missense_Mutation	6	28895078	G	A	463	40.9%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.95912	91%	Cancer		WES
025684	PTEN	p.1588R	I	Missense_Mutation	18	9385813	G	C	204	21.57%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.79	0.3098	80%	Cancer		WES
025685	RALBP1	p.576P	I	Missense_Mutation	19	13525460	G	A	264	57.94%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.12	0.88484	80%	Cancer		WES
025686	RLB1	p.2257L	I	Missense_Mutation	20	35695405	G	T	144	43.06%	Neutral	CANCER	skler. pathogene		0.92081	0.85%	93%	Cancer		WES	
025687	RB144	p.2854H	I	Missense_Mutation	11	48822010	C	A	551	34.35%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.92	0.70502	81%	Cancer		WES
025688	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.08	0.1	4%	asbestose		WES
025689	RECO4	p.9893S	I	Missense_Mutation	8	14578308	G	A	274	52.8%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.90035	95%	Cancer	TRUE	WES
025690	PTEN	p.1719C	I	Missense_Mutation	10	17750551	A	G	185	34.85%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.08	0.1	4%	asbestose		WES
025691	RYRD2	p.1910W	I	Missense_Mutation	1	287711840	C	T	86	34.88%	Dileukosid	CANCER	skler. pathogene		0.89813	0.86133	77%	Cancer		WES	
025692	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.29	0.88484	80%	Cancer		WES
025693	SP100	p.103Y	I	Missense_Mutation	19	16335564	T	A	358	40.30%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.00113	95%	Cancer		WES
025694	SAMAD3	p.148M	I	Missense_Mutation	6	13925460	C	A	133	36.95%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.74	0.89507	94%	Cancer		WES
025695	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.92	0.70502	81%	Cancer		WES
025696	SSLC1CT	p.1518A	I	Missense_Mutation	19	62023208	T	C	58	51.7%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.87	0.71883	84%	Cancer		WES
025697	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	CANCER	skler. pathogene		0.86	0.1	5%	asbestose		WES	
025698	TEK15	p.480S	I	Missense_Mutation	6	30704728	C	A	193	8.22%	Neutral	PASSENGER/ROTHER	Pasenager	pasenager	het	0.51	0	5%	Cancer		WES
025699	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.00113	95%	Cancer	TRUE	WES
025700	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.87569	86%	Cancer	TRUE	WES
025701	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.12	0	15%	asbestose	TRUE	WES
025702	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	0.22	0	15%	asbestose	TRUE	WES
025703	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	CANCER	skler. pathogene		0.86	0.1	5%	asbestose	TRUE	WES	
025704	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025705	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025706	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025707	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025708	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025709	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025710	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025711	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025712	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025713	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025714	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025715	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025716	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025717	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025718	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025719	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025720	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025721	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025722	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025723	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025724	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025725	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025726	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025727	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025728	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025729	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025730	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025731	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025732	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025733	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025734	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025735	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025736	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34%	Dileukosid	PASSENGER/ROTHER	Pasenager	pasenager	het	1	0.88484	80%	Cancer	TRUE	WES
025737	PTEN	p.1719L	I	Missense_Mutation	10	17853851	C	G	261	37.34											

DO001	TMAT	p.S205S	Maisenne Mutation	21	3299231	T	C	36	32.22%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.25492	46%	Clonal		WES
DO001	TP53	p.D280H	Maisenne Mutation	7	6551514	G	C	38	18.4%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.85	0.86315	39%	Clonal		WES
DO001	UBR4	p.G434F	Maisenne Mutation	1	1948267	G	A	28	28.92%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.97	0.72787	47%	Clonal		WES
DO001	VIPRP1	p.R131V	Maisenne Mutation	5	5146607	G	A	19	17.58%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.87099	44%	Clonal		WES
DO001	ZFATND4	p.S311C	Maisenne Mutation	10	4817228	G	C	21	14.29%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.65	0.62551	21%	Clonal		WES
DO001	ZNF19	p.E26Q	Maisenne Mutation	6	4352509	G	O	88	19.32%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.89	0.72617	54%	Clonal		WES
DO001	ZNF19	p.T200P	Maisenne Mutation	1	1948268	G	A	29	17.76%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.85	0.72617	54%	Clonal		WES
DO001	CALCA	p.Q97L	Maisenne Mutation	2	188345422	G	O	29	17.76%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.64	0.67873	24%	Subclonal		WES
DO001	RSB2	p.G59K	Maisenne Mutation	8	1919653	CT	C	65	16.50%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.85939	70%	Clonal		WES
DO001	AKT1	p.A219P	Maisenne Mutation	7	6552317	G	T	27	13.33%	Neutral	CANCER	Passenger	passenger	1	0.87072	65%	Clonal		WES
DO001	ATP52B	p.S380R	Maisenne Mutation	3	16378101	G	D	116	31.53%	Neutral	CANCER	Passenger	passenger	1	0.87032	75%	Clonal		WES
DO001	BSX10	p.R246S	Maisenne Mutation	10	7874102	G	O	62	33.87%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86115	68%	Clonal		WES
DO001	C10orf172	p.D415Y	Maisenne Mutation	10	9874259	G	T	24	20.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.65	0.67098	29%	Subclonal		WES
DO001	C10orf172	p.K465I	Maisenne Mutation	1	7507220	G	A	29	17.76%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.15	0.69444	7%	Subclonal		WES
DO001	CADPR	p.Y259D	Maisenne Mutation	2	20214168	A	A	91	17.14%	Neutral	CANCER	Passenger	passenger	1	0.86739	91%	Clonal		WES
DO001	CDC2BPB	p.M167N	Maisenne Mutation	14	10340160	G	O	66	13.94%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.35	0.67078	35%	Clonal		WES
DO001	CDC2BPB	p.M167N	Maisenne Mutation	23	13947402	G	O	44	22.92%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86737	70%	Clonal		WES
DO001	CED9	p.H120N	Maisenne Mutation	2	1808981	G	A	45	22.22%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86262	76%	Clonal		WES
DO001	CDKN2A	p.P470A	Maisenne Mutation	14	1469501	G	C	43	20.00%	Neutral	CANCER	Passenger	passenger	0.85	0.62449	76%	Clonal		WES
DO001	CDKN2A	p.P470A	Maisenne Mutation	23	1311353	G	C	116	31.33%	Neutral	CANCER	Passenger	passenger	1	0.86155	68%	Clonal		WES
DO001	CDKN2A	p.P470A	Maisenne Mutation	23	1311353	G	C	116	31.33%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.85	0.62449	76%	Clonal		WES
DO001	ESPA1	p.R49W	Maisenne Mutation	5	3847224	G	T	120	7.50%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.25	0.67129	15%	Subclonal		WES
DO001	FAM187	p.S135P	Maisenne Mutation	22	4047146	G	T	88	19.47%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86485	74%	Clonal		WES
DO001	FAM187	p.S135P	Maisenne Mutation	22	4047146	G	T	88	19.47%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86485	74%	Clonal		WES
DO001	FAM187	p.S135P	Maisenne Mutation	17	43114962	G	A	58	16.07%	Neutral	CANCER	Passenger	passenger	1	0.86074	89%	Clonal		WES
DO001	FSTL2	p.K75R	Maisenne Mutation	4	9521055	G	C	125	24.40%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.23	0.62451	52%	Subclonal		WES
DO001	GPR192	p.T725A	Maisenne Mutation	6	14273051	A	G	80	13.75%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86952	91%	Clonal		WES
DO001	GSDT	p.F173P	Maisenne Mutation	10	8813718	A	T	50	11.17%	Neutral	PASSANGER/ROTHER	Passenger	passenger	0.11	0.67074	64%	Clonal		WES
DO001	HISTH2H	p.A28P	Maisenne Mutation	6	27460108	G	O	24	20.83%	Neutral	CANCER	Passenger	passenger	0.89	0.62023	36%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001	ISG13C	p.Y394C	Maisenne Mutation	23	130416483	G	C	61	16.85%	Neutral	PASSANGER/ROTHER	Passenger	passenger	1	0.86469	90%	Clonal		WES
DO001																			

Supplementary Table 3

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DOID	Gene	Mutation Type	Position	Allele	Frequency	Phenotype	Pathogenicity	Passenger	Host Pathogenic	Host	Frequency	Subcellular	TRUE	WES				
DOID_1	p.R172BC	Misense Mutation	12	40758488	G	T	99	9.05%	Diseaseless	CANCER	Passenger	Weak pathogenic	0.32	0.00296	1%	Subcellular		
DOID_2	p.R172C	Misense Mutation	12	40758489	G	A	82	13.41%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.00601	32%	Subcellular	WES	
DOID_3	MCR3	Misense Mutation	18	13808440	G	A	84	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.01482	15%	Subcellular	WES	
DOID_4	MCR3	Misense Mutation	3	12732558	G	A	100	100%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.01482	15%	Subcellular	WES	
DOID_5	MCR3	Misense Mutation	9	13808441	G	A	87	9.57%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.34	0.00382	41%	Subcellular	WES	
DOID_6	MMAA	Misense Mutation	4	14657635	G	T	105	7.55%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00342	15%	Subcellular	WES	
DOID_7	MNH	p.L119V	Misense Mutation	22	28790237	G	C	115	5.22%	Diseaseless	CANCER	Passenger	Weak pathogenic	0.22	0.00208	10%	Subcellular	TRUE
DOID_8	MNTD1	Misense Mutation	10	10449509	G	A	94	9.75%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.24	0.00279	24%	Subcellular	WES	
DOID_9	MUC17	p.T180N	Misense Mutation	7	100580120	G	A	63	13.21%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.69	0.25686	3%	Subcellular	TRUE
DOID_10	MUC17	p.T180W	Misense Mutation	7	100580121	G	A	99	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.46	0.00208	21%	Subcellular	WES
DOID_11	NPATL5	p.R97TC	Misense Mutation	16	69981208	G	T	65	7.69%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00198	21%	Subcellular	WES
DOID_12	NPFLR	p.Y187M	Misense Mutation	17	4793933	G	A	229	9.95%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.44	0.00171	27%	Subcellular	WES
DOID_13	NPFLR	p.Y187M	Misense Mutation	17	4793934	G	A	100	9.95%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00171	27%	Subcellular	WES
DOID_14	NLRN	p.V31H	Misense Mutation	5	65591722	G	A	67	8.77%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.00167	16%	Subcellular	WES
DOID_15	NLRN	p.V31H	Misense Mutation	5	65591723	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.00167	16%	Subcellular	YES
DOID_16	NLRN	p.V23M	Misense Mutation	2	51254106	G	A	98	5.21%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.32	0.00415	14%	Subcellular	WES
DOID_17	NTRK2	p.D120W	Misense Mutation	10	10449509	G	A	75	8.47%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.28	0.00208	11%	Subcellular	TRUE
DOID_18	NTRK2	p.D120W	Misense Mutation	10	10449510	G	A	99	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.32	0.00202	15%	Subcellular	WES
DOID_19	NYX	p.D198N	Misense Mutation	X	4153308	G	A	83	8.62%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.34	0.00198	21%	Subcellular	WES
DOID_20	OMMBRD	p.G230M	Misense Mutation	19	3658605	G	A	105	8.87%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00137	15%	Subcellular	WES
DOID_21	PABPN1	p.P27L	Misense Mutation	16	8889171	G	A	48	10.4%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.55	0.10592	23%	Subcellular	WES
DOID_22	PCPQ2	p.R18H	Misense Mutation	14	2487759	G	A	129	14.87%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.24	0.00501	11%	Subcellular	WES
DOID_23	PDZ3	p.D24G	Misense Mutation	11	7236865	T	C	23	94.43%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.1	0.00236	56%	Clonal	WES
DOID_24	PDZK2	p.R26T	Misense Mutation	4	9873055	G	T	144	3.47%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.23	0.00501	15%	Subcellular	WES
DOID_25	PDZC1	p.E54K	Misense Mutation	18	15126764	G	A	99	8.95%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00882	17%	Subcellular	WES
DOID_26	PDZC1	p.E54K	Misense Mutation	18	15126765	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.44	0.00203	31%	Subcellular	WES
DOID_27	PLA2G4B	p.C92P	Misense Mutation	15	42139653	G	T	192	84.95%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.36	0.00389	92%	Clonal	WES
DOID_28	PLA2G4B	p.C92P	Misense Mutation	15	42139654	G	A	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.36	0.00389	92%	Clonal	WES
DOID_29	PLXNC1	p.A39S	Misense Mutation	X	16533259	G	A	71	15.46%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.65	0.19778	37%	Subcellular	WES
DOID_30	PMV22	p.P122L	Misense Mutation	17	15134374	G	A	15	20.0%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.81	0.30951	25%	Subcellular	WES
DOID_31	PMV22	p.P122L	Misense Mutation	17	15134375	G	A	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.42	0.00213	15%	Subcellular	WES
DOID_32	PRH12	p.G90S	Misense Mutation	19	90150479	G	A	81	8.17%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00997	17%	Subcellular	WES
DOID_33	PRH12	p.G90S	Misense Mutation	19	90150480	G	A	99	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00997	17%	Subcellular	WES
DOID_34	PTSK	p.G57V	Misense Mutation	10	12474041	G	T	91	10.56%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00220	37%	Subcellular	WES
DOID_35	PTSK	p.G57V	Misense Mutation	10	12474042	G	T	80	12.26%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00203	31%	Subcellular	WES
DOID_36	R1PF1	p.E171K	Misense Mutation	17	3374820	G	A	85	7.79%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.44	0.00381	20%	Subcellular	WES
DOID_37	R1PF1	p.E171K	Misense Mutation	17	3374821	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00381	20%	Subcellular	WES
DOID_38	R1PF1	p.E171K	Misense Mutation	17	3374822	G	A	72	8.94%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00382	20%	Subcellular	WES
DOID_39	R1PF1	p.E171K	Misense Mutation	17	3374823	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00382	20%	Subcellular	WES
DOID_40	R1PF1	p.E171Q	Misense Mutation	17	3374824	G	A	38	16.87%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	1	0.08126	45%	Clonal	WES
DOID_41	R1PF1	p.E171Q	Misense Mutation	17	3374825	G	A	78	14.87%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.41	0.00213	15%	Subcellular	WES
DOID_42	R1PF1	p.E171Q	Misense Mutation	17	3374826	G	A	84	14.87%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.41	0.00213	15%	Subcellular	WES
DOID_43	R1PF1	p.E171Q	Misense Mutation	17	3374827	G	A	48	8.70%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.43	0.00462	15%	Subcellular	WES
DOID_44	SLC24A1	p.R26Q	Misense Mutation	1	15392344	G	A	138	9.35%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.31	0.00114	14%	Subcellular	WES
DOID_45	SLC24A1	p.R26Q	Misense Mutation	1	15392345	G	A	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00201	17%	Subcellular	WES
DOID_46	SLC24A1	p.R26Q	Misense Mutation	2	4469259	G	C	80	12.26%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00201	17%	Subcellular	WES
DOID_47	SLC24A1	p.R26Q	Misense Mutation	2	4469260	G	C	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.39	0.00201	17%	Subcellular	WES
DOID_48	SLC24A1	p.R26Q	Misense Mutation	2	4469261	G	C	209	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.2	0.00188	100%	Subcellular	WES
DOID_49	SLC24A1	p.R26Q	Misense Mutation	2	4469262	G	C	93	9.35%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.34	0.00376	14%	Subcellular	WES
DOID_50	SLC24A1	p.R26Q	Misense Mutation	2	4469263	G	C	80	9.35%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.34	0.00376	14%	Subcellular	WES
DOID_51	SLC24A1	p.R26Q	Misense Mutation	2	4469264	G	C	117	7.89%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.4	0.00242	21%	Subcellular	WES
DOID_52	SLC24A1	p.R26Q	Misense Mutation	2	4469265	G	C	141	9.92%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.52	0.01146	31%	Subcellular	WES
DOID_53	SLC24A1	p.R26Q	Misense Mutation	2	4469266	G	C	95	9.26%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.28	0.00407	12%	Subcellular	WES
DOID_54	SLC24A1	p.R26Q	Misense Mutation	2	4469267	G	C	95	9.26%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.28	0.00407	12%	Subcellular	WES
DOID_55	SLC24A1	p.R26Q	Misense Mutation	2	4469268	G	C	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.01339	16%	Subcellular	WES
DOID_56	SLC24A1	p.R26Q	Misense Mutation	2	4469269	G	C	98	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.37	0.01339	16%	Subcellular	WES
DOID_57	SLC24A1	p.R26Q	Misense Mutation	2	4469270	G	C	98	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.3	0.01732	13%	Subcellular	WES
DOID_58	SLC24A1	p.R26Q	Misense Mutation	2	4469271	G	C	98	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.25	0.00004	12%	Subcellular	WES
DOID_59	SLC24A1	p.R26Q	Misense Mutation	2	4469272	G	C	83	8.02%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.26	0.00311	11%	Subcellular	WES
DOID_60	SLC24A1	p.R26Q	Misense Mutation	2	4469273	G	C	113	14.9%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.83	0.38888	50%	Clonal	WES
DOID_61	SLP450	p.G165D	Misense Mutation	5	7174306	G	A	164	4.88%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_62	SLP450	p.G165D	Misense Mutation	5	7174307	G	A	164	4.88%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_63	SLP450	p.G165D	Misense Mutation	5	7174308	G	A	113	14.87%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_64	SLP450	p.G165D	Misense Mutation	5	7174309	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_65	SLP450	p.G165D	Misense Mutation	5	7174310	G	A	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_66	SLP450	p.G165D	Misense Mutation	5	7174311	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_67	SLP450	p.G165D	Misense Mutation	5	7174312	G	A	100	9.85%	Diseaseless	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular	WES
DOID_68	SLP450	p.G165D	Misense Mutation	5	7174313	G	A	100	9.85%	Neutral	PASSENGER ROTHER	Passenger	Weak pathogenic	0.35	0.00362	16%	Subcellular</	

Supplementary Table 4. Clinicopathologic characteristics of the *PALB2*-associated breast cancers according to bi-allelic inactivation of the *PALB2* wild-type allele.

	All cases		Bi-allelic <i>PALB2</i> alterations		Mono-allelic <i>PALB2</i> alterations		<i>P</i> value
	No.	Percentage	No.	Percentage	No.	Percentage	
Study							
Li et al.	24	62%	16	62%	8	62%	
Lee et al.	15	38%	10	38%	5	38%	
Age (years)							
<50	21	54%	11	42%	10	77%	0.051
≥50	18	46%	15	58%	3	23%	
Receptor status							
ER+/HER2-	21	54%	16	62%	5	38%	0.383
ER+/HER2+	5	13%	3	12%	2	15%	
ER-/HER2-	13	33%	7	27%	6	46%	
Histologic grade							
I/II	13	33%	8	31%	5	38%	0.725
III	26	67%	18	69%	8	62%	
Germline mutation type							
Frameshift	22	56%	12	46%	10	77%	0.175
Truncating	16	41%	13	50%	3	23%	
Missense	1	3%	1	4%	0	0%	
TP53 gene							
Mutant	10	26%	5	19%	5	38%	0.254
Wild-type	29	74%	21	81%	8	62%	
LSTs*,¹							
High	27	87%	22	100	5	62.5	0.004
Low	4	13%	0	0	4	37.5	

Abbreviations: Age, age at diagnosis; ER, estrogen receptor; LST, large-scale state transition; *, LST high, ≥15; LST low, <15; No., number of cases.

¹LSTs were investigated in 16 *PALB2*-associated breast cancers from this study and 15 *PALB2* breast cancers from Lee et al, J Pathol 2018.

Supplementary Table 5

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Supplementary Table 5: Comparisons of frequencies of somatic mutations affecting 410 cancer genes between PALB2-associated breast cancers and non-BRCA1/2/PALB2-, BRCA1- and BRCA2-associated breast cancers from TCGA.

	ER+/HER2-, ER+/HER2+ and ER-/HER2-						ER+/HER2-						PALB2 with bi-allelic alterations (n=16)						TCGA BRCA1 with bi-allelic alterations (n=17)		PALB2 vs. TCGA BRCA1		PALB2 with bi-allelic alterations (n=16)		TCGA BRCA2 with bi-allelic alterations (n=16)		PALB2 vs. TCGA BRCA2	
	PALB2 (n=24)		TCGA non-BRCA1/2/PALB2 (n=683)		PALB2 vs. TCGA non-BRCA1/2/PALB2		PALB2 (n=18)		TCGA non-BRCA1/2/PALB2 (n=441)		PALB2 vs. TCGA non-BRCA1/2/PALB2 (n=16)		Fisher's exact test P value		Fisher's exact test P value		Fisher's exact test P value		Fisher's exact test P value		Fisher's exact test P value		Fisher's exact test P value		Fisher's exact test P value			
	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Fisher's exact test P value	Fisher's exact test P value of bootstrapping comparison	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Fisher's exact test P value	Fisher's exact test P value of bootstrapping comparison	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes	Number of cases with somatic mutations affecting 410 cancer genes	% of cases with somatic mutations affecting 410 cancer genes		
MSK-IMPACT 410 cancer genes	0	4%	1	0%	1	1	0	0%	2	0.5%	1	1	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ABL1	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ACVR1	0	0%	16	2.3%	1	0.94	0	0%	7	1.6%	1	0.99	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AKT1	0	0%	20	2.9%	1	0.87	0	0%	18	4.1%	1	0.85	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AKT2	0	0%	3	0%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AKT3	0	0%	4	0.6%	1	0.99	0	0%	3	0.7%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ALK	0	0%	7	1.0%	1	0.98	0	0%	5	1.1%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ALOX12B	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AMER1	0	0%	12	1.8%	1	0.93	0	0%	7	1.6%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ANKRD11	0	0%	16	2.3%	1	0.94	0	0%	7	1.6%	1	0.99	0	0%	1	0	0	0%	2	11.8%	0.48	0	0	0	0	0	0%	1
APC	0	0%	9	1.3%	1	0.97	0	0%	4	0.9%	1	1	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AR	0	0%	5	0.7%	1	0.99	0	0%	3	0.7%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ARAF	0	0%	6	0.9%	1	0.99	0	0%	4	0.9%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ARID1A	3	12.5%	26	3.8%	0.07	0.19	3	16.7%	15	3.4%	0.03	0.11	3	18.8%	0	0%	0.10	0.3	18.8%	1	6.2%	0.60						
ARID1B	0	0%	16	2.3%	1	0.92	0	0%	6	1.4%	1	0.98	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
ARID2	1	4.2%	11	1.6%	0.34	0.58	0	0%	7	1.6%	1	0.43	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
ARID5B	0	0%	3	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	2	12.5%	0.48			
ASXL1	1	4.2%	7	1.0%	0.24	0.43	0	0%	4	0.9%	1	0.34	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
ASXL2	1	4.2%	14	2.0%	0.41	0.66	1	5.6%	6	1.4%	0.25	0.40	0	0%	2	11.8%	0.48	0	0	0	0%	0	0	0	0%	1		
ATM	0	0%	20	2.9%	1	0.89	0	0%	7	1.6%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
ATR	0	0%	8	1.2%	1	0.98	0	0%	5	1.1%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
ATRX	0	0%	21	3.1%	1	0.84	0	0%	13	2.9%	1	0.89	0	0%	1	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AURKA	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
AURKB	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
AXIN1	0	0%	4	0.6%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0	0	0%	1	0	0	0%	1	0	0	0%	1	
AXIN2	0	0%	5	0.7%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
AXL	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	2	11.8%	0.48	0	0	0	0%	0	0	0	0%	1		
B2M	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BAP1	0	0%	7	1.0%	1	0.99	0	0%	3	0.7%	1	0.15	0	0%	1	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BARD1	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BBC3	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BCL10	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	1	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BCL11	0	0%	3	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BCL12	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BCL6	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BCOR	1	4.2%	10	1.5%	0.32	0.49	1	5.6%	4	0.9%	0.18	0.37	1	6.2%	0	0%	0.48	1	6.2%	1	6.2%	1	6.2%	1	6.2%	1	6.2%	1
BIRC3	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BLM	0	0%	10	1.5%	1	0.96	0	0%	5	1.1%	1	0.99	0	0%	1	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BMPR1A	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BRAF	1	4.2%	6	0.9%	0.22	0.41	1	5.6%	3	0.9%	0.18	0.37	0	6.2%	0	0%	0.48	1	6.2%	0	0%	0	0%	1	6.2%	0	0%	1
BRCA1	0	0%	9	1.3%	0.29	0.49	1	5.6%	5	1.1%	0.21	0.40	1	6.2%	0	0%	0.48	1	6.2%	1	6.2%	1	6.2%	1	6.2%	1	6.2%	1
BRCA2	0	0%	20	2.9%	1	0.85	0	0%	11	2.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BRCA1D1	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	1	0	0	0%	1	0	0	0%	0	0	0	0%	1	
BRCA2D1	0	0%	11	1.6%	1	0.97	0	0%	8	1.8%	1	0.97	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
CTCF	0	0%	11	1.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
CTTNB1	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
CUL3	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0	0	0%	1	0	0	0%	0	0	0	0%	1	
CXCR4	0	0%	3	0%	1	1	0	0%	3	0.7%	1	1	0	0%	1	0	0	0%	1	0	0	0%	0	0	0	0%	1	
DAXX	1	4.2%	2	0%	0.10	0.30	0	0%	2</td																			

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FBXW7	0	0%	14	2.0%	1	0.90	0	0%	6	1.4%	1	0.99	0	0%	2	11.8%	0.48	0	0%	0	0%	1
FGF19	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FGF3	1	4.2%	1	0%	0.07	0.27	1	5.6%	1	0.2%	0.08	0.27	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1
FGF4	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FGFR1	1	4.2%	5	0.7%	0.19	0.38	1	5.6%	5	1.1%	0.21	0.39	1	6.2%	1	5.9%	1	1	6.2%	0	0%	1
FGFR2	0	0%	12	1.8%	1	0.96	0	0%	8	1.8%	1	0.97	0	0%	1	5.9%	1	0	0%	0	0%	1
FGFR3	0	0%	1	0%	1	1	0	0%	1	0%	1	0.2%	0	0%	0	0%	1	0	0%	0	0%	1
FGFR4	2	8.3%	4	0.6%	0.02	0.11	2	11.1%	2	0.5%	0.008	0.08	2	12.5%	0	0%	0.23	2	12.5%	0	0%	0.48
FH	0	0%	3	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FLCN	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FLT1	0	0%	6	0.9%	1	1	0	0%	0	0%	1	1	0	0%	1	5.9%	1	0	0%	0	0%	1
FLT3	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FLT4	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FOXA1	0	0%	22	3.2%	1	0.86	0	0%	18	4.1%	1	0.83	0	0%	0	0%	1	0	0%	0	0%	1
FOXJ2	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FOXO1	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FOXO1P1	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
FUBP1	1	4.2%	4	0.6%	0.16	0.35	0	0%	2	0.5%	1	0.31	0	0%	0	0%	1	0	0%	0	0%	1
FU	0	0%	2	0%	1	1	0	0%	2	0.5%	1	1	0	0%	1	0	0%	0	0%	1		
GATA1	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	1	0	0%	0	0%	1		
GATA2A	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
GATA3	0	0%	89	13.0%	0.06	0.15	0	0%	70	15.9%	0.09	0.22	0	0%	0	0%	1	0	0%	2	12.5%	0.48
GLI1	1	4.2%	4	0.6%	0.16	0.36	1	5.6%	3	0.7%	0.15	0.34	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1
GNA11	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
GNAQ	2	8.3%	2	0%	0.006	0.08	2	11.1%	1	0.2%	0.004	0.07	0	0%	0	0%	1	0	0%	0	0%	1
GNAS	0	0%	11	1.6%	1	0.97	0	0%	10	2.3%	1	0.96	0	0%	0	0%	1	0	0%	0	0%	1
GPS2	0	0%	8	1.2%	1	0.99	0	0%	6	1.4%	1	0.99	0	0%	0	0%	1	0	0%	0	0%	1
GREM1	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
GRIN2A	0	0%	17	2.5%	1	0.91	0	0%	12	2.7%	1	0.95	0	0%	0	0%	1	0	0%	0	0%	1
GSK3B	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
H2AFY2	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
H2AFY2B	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
H2AFY3C	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HGF	1	4.2%	4	0.6%	0.16	0.35	0	0%	3	0.7%	1	0.33	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H1C	0	0%	5	0.7%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H2BD	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3A	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3B	1	4.2%	11	1.6%	0.34	0.56	1	5.6%	5	1.1%	0.21	0.40	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1
HIST1H3C	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3D	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3E	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3F	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3G	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3H	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST1H3I	1	4.2%	1	0%	0.07	0.27	0	0%	0	0%	1	0.25	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1
HIST1H3J	1	4.2%	1	0%	0.07	0.27	0	0%	1	0.2%	1	0.27	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1
HIST2H3C	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HIST2H3D	1	4.2%	3	0%	0.13	0.33	1	5.6%	2	0.5%	0.11	0.30	1	6.2%	1	5.9%	1	1	6.2%	0	0%	1
HIST3H3	0	0%	3	0%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN1	0	0%	2	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN2A	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN2B	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN3A	0	0%	0	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN3B	0	0%	0	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
HNRN4A	0	0%	6	1.2%	0.27	0.46	1	5.6%	7	1.5%	0.28	0.46	0	0%	1	5.9%	1	1	6.2%	0	0%	1
HNRN4B	1	4.2%	8	1.2%	0.27	0.46	1	5.6%	7	1.5%	0.28	0.46	0	0%	1	5.9%	1	1	6.2%	0	0%	1
HNRN5A	0	0%	15	2.2%	1	0.89	0	0%	10	2.3%	1	0.95	0	0%	1	5.9%	1	0	0%	0	0%	1
HNRN5B	0	0%	7	1.0%	1	0.98	0	0%	5	1.1%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KEAP1	1	4.2%	2	0%	0.10	0.30	0	0%	2	0.5%	1	0.30	0	0%	0	0%	1	0	0%	0	0%	1
KIF5A	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5B	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5C	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5D	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5E	0	0%	6	0.9%	1	1	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5F	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5G	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5H	0	0%	4	0.6%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5I	0	0%	6	0.9%	1	1	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5L	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5M	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5N	0	0%	7	1.0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5T1	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1
KIF5T2	0	0%	3	0%</td																		

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NTRK2	0	0%	5	0.7%	1	0.99	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	1	6.2%	1	
NTRK3	0	0%	8	1.2%	1	0.98	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
NUP93	0	0%	3	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PAK1	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PAK7	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PALB2	5	20.8%	4	0.6%	0.000003	0.002	5	27.5%	1	0.2%	0.000003	0.001	5	31.2%	0	0%	0.02	5	31.2%	0	0%	0.04	
PAPOL2	0	0%	5	0.7%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PAPOL3	0	0%	5	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PAX6	0	0%	4	0.6%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PBRM1	1	4.2%	8	1.2%	0.27	0.48	1	5.6%	4	0.9%	0.18	0.36	1	6.2%	1	5.9%	1	1	6.2%	0	0%	1	
POCD1	0	0%	3	0%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PDGFRα	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	1	6.2%	1	
PDGFRβ	0	0%	7	1.0%	1	0.99	0	0%	7	1.6%	1	0.98	0	0%	0	0%	1	0	0%	0	0%	1	
PDPK1	0	0%	2	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PGR	1	4.2%	4	0.6%	0.16	0.35	1	5.6%	3	0.7%	0.15	0.32	1	6.2%	0	0%	0.48	1	6.2%	0	0%	1	
PHOX2B	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PIK3CG	0	0%	8	1.2%	1	0.97	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PIK3C3	0	0%	5	0.7%	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1		
PIK3C6	7	29.2%	246	3.6%	0.67	0.77	7	38.5%	187	42.4%	0.81	0.81	3	18.8%	0	0%	0.10	3	18.8%	1	6.2%	0.80	
PIK3CB	1	4.2%	7	1.0%	0.24	0.43	5	1.5%	1	1.1%	0.21	0.44	1	5.9%	1	0	0%	0	0%	1	0%	1	
PIK3CD	0	0%	3	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PIK3CG	0	0%	5	0.7%	1	0.99	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PIK3R1	0	0%	21	3.1%	1	0.82	0	0%	14	3.2%	1	0.89	0	0%	2	11.8%	0.48	0	0%	0	0%	1	
PIK3R2	0	0%	3	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PIK3R3	0	0%	4	0.6%	1	1	0	0%	4	0.9%	1	1	0	0%	1	5.9%	1	0	0%	0	0%	1	
PIM1	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PLCG2	0	0%	5	0.7%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	1	6.2%	1	
PLK2	0	0%	8	1.2%	1	0.98	0	0%	5	1.1%	1	1	0	0%	0	0%	1	0	0%	1	6.2%	1	
PLMA1P1	0	0%	0	1	0	0%	1	0	0%	1	0	0%	1	0	0%	0	0%	1	0	0%	0	0%	1
PMID	0	0%	8	0.2%	1	0.98	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PP2R2A	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PPP6C	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PRDM1	0	0%	10	1.5%	1	0.95	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PRKAR1A	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PTCH1	0	0%	7	1.0%	1	0.98	0	0%	4	0.9%	1	1	0	0%	2	11.8%	0.48	0	0%	0	0%	1	
PTEN	0	0%	42	6.1%	0.39	0.49	0	0%	26	5.9%	0.61	0.88	0	0%	3	17.6%	0.23	0	0%	1	6.2%	1	
PTPN1	0	0%	0	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PTPRD	0	0%	19	2.8%	1	0.82	0	0%	12	2.7%	1	0.96	0	0%	1	5.9%	1	0	0%	0	0%	1	
PTPRS	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
PTPRT	0	0%	7	1.0%	1	0.99	0	0%	5	1.1%	1	0.99	0	0%	0	0%	1	0	0%	0	0%	1	
RAB35	0	0%	2	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAC1	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD21	0	0%	7	1.0%	1	0.98	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD50	0	0%	7	1.0%	1	0.99	0	0%	5	1.1%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD51	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD51B	0	0%	3	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	2	12.5%	0.48	
RAD51C	0	0%	3	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD51D	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD51L	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAD54L	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAF1	0	0%	5	0.7%	1	1	0	0%	3	0.7%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RARA	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RAAS1	0	0%	4	0.6%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RB1	0	0%	16	2.3%	1	0.90	0	0%	6	1.4%	1	0.99	0	0%	0	0%	1	0	0%	3	18.8%	0.23	
RBML0	0	0%	6	0.9%	1	0.99	0	0%	4	0.9%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RECQL	1	4.2%	3	0%	0.13	0.32	0	0%	2	0.5%	1	0.30	1	6.2%	1	5.9%	1	1	6.2%	0	0%	1	
REL	0	0%	1	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RET	0	0%	6	0.9%	1	1	0	0%	5	1.1%	1	1	0	0%	1	5.9%	1	0	0%	0	0%	1	
REV3L2	0	0%	3	0%	1	1	0	0%	1	0.2%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RHEB	0	0%	1	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RHOA	0	0%	6	0.9%	1	0.99	0	0%	5	1.1%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RICTOR	0	0%	6	0.9%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RTT1	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
RUNX1	0	0%	33	4.8%	0.62	0.63	0	0%	25	5.7%	0.61	0.69	0	0%	0	0%	1	0	0%	0	0%	1	
RXRA	0	0%	3	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SDHA4	0	0%	3	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SDHA4F2	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SDHB	0	0%	2	0%	1	1	0	0%	2	0.5%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SDHD	0	0%	0	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SETD2	0	0%	17	2.5%	1	0.89	0	0%	11	2.5%	1	0.96	0	0%	2	11.8%	0.48	0	0%	0	0%	1	
SF3B1	0	0%	14	2.0%	1	0.94	0	0%	11	2.5%	1	0.94	0	0%	0	0%	1	0	0%	0	0%	1	
SH2B3	0	0%	2	0%	1	1	0	0%	0	0%	1	1	0	0%	0	0%	1	0	0%	0	0%	1	
SH2D1A	0	0%	3	0%	1	1	0	0%	3														