SUPPLEMENTARY INFORMATION

A comprehensive custom panel design for routine hereditary cancer testing: preserving control, improving diagnostics and revealing a complex variation landscape

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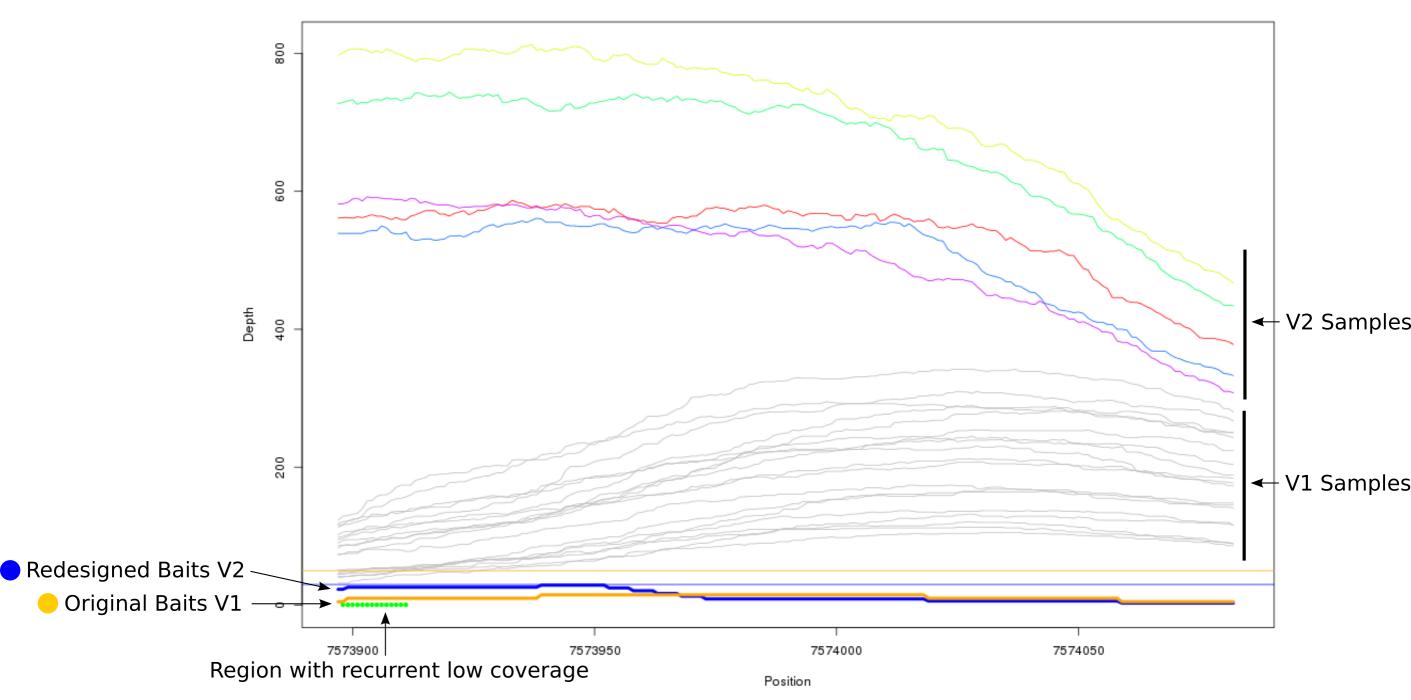
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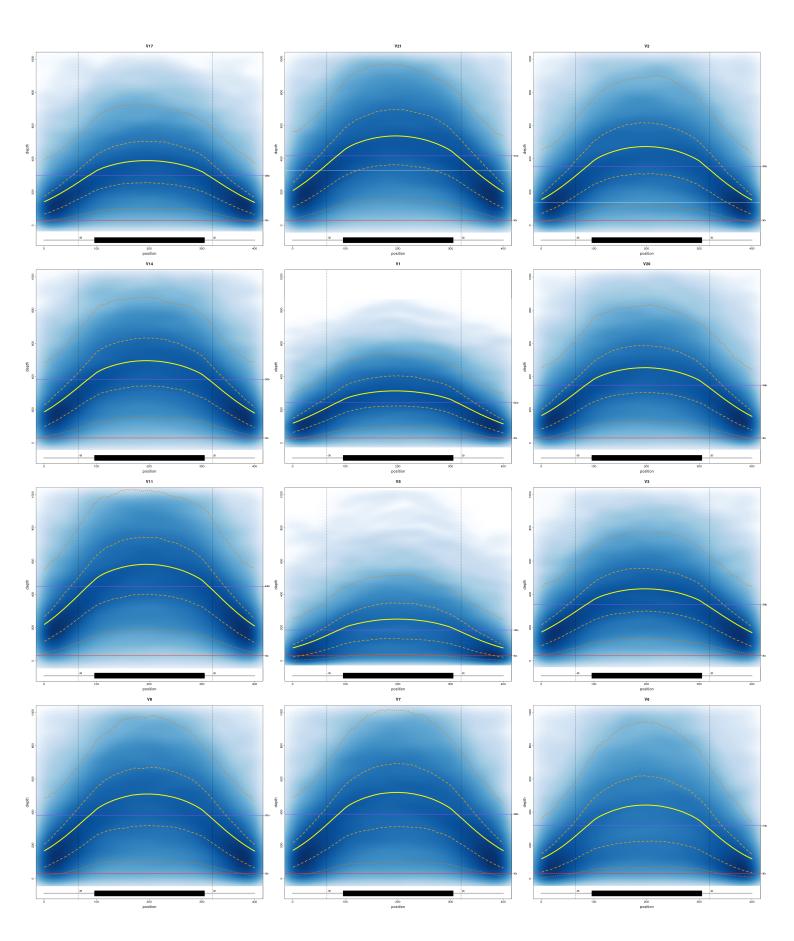
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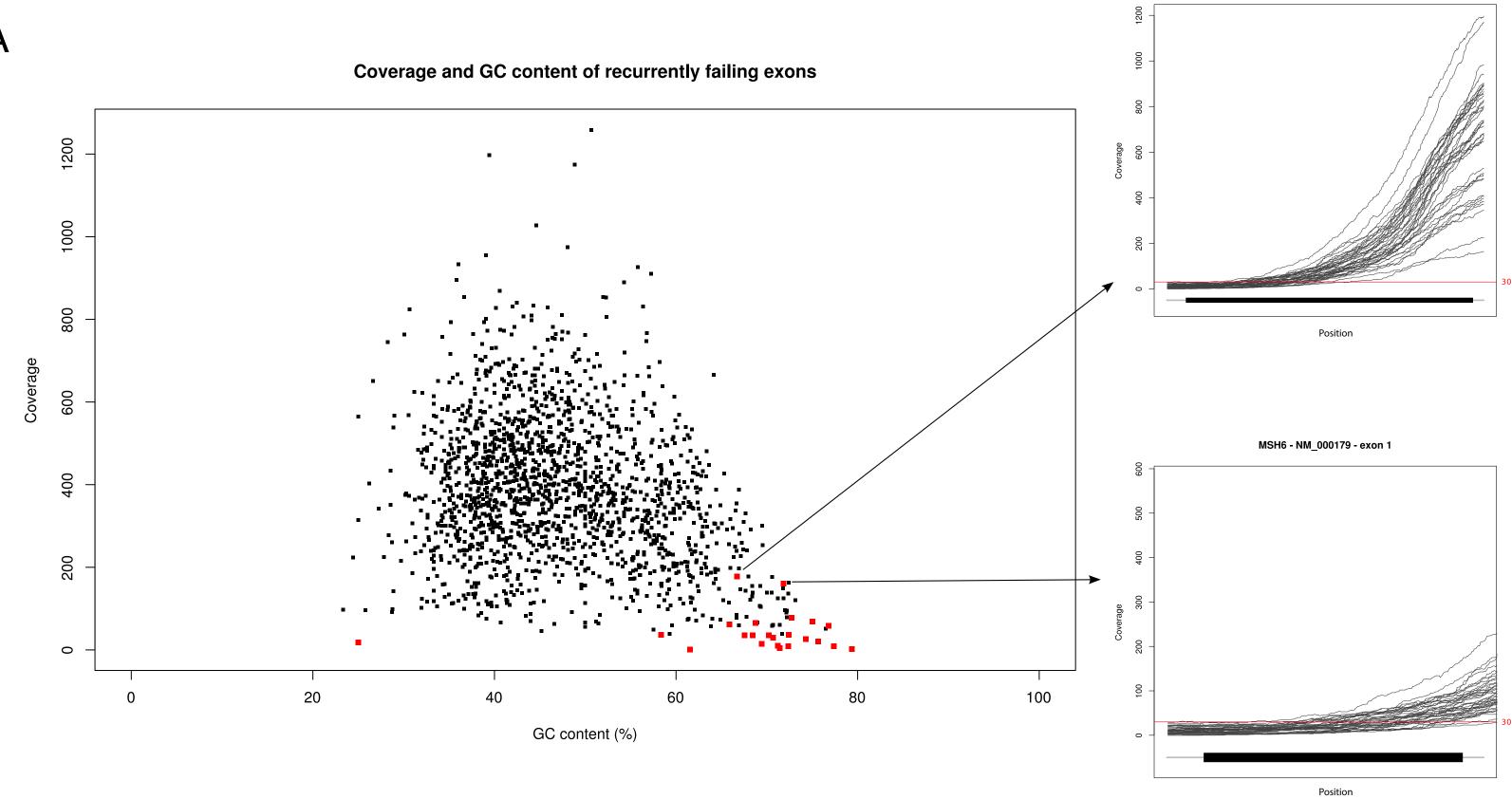
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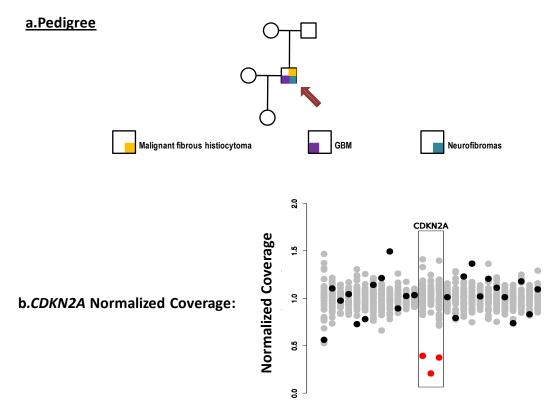


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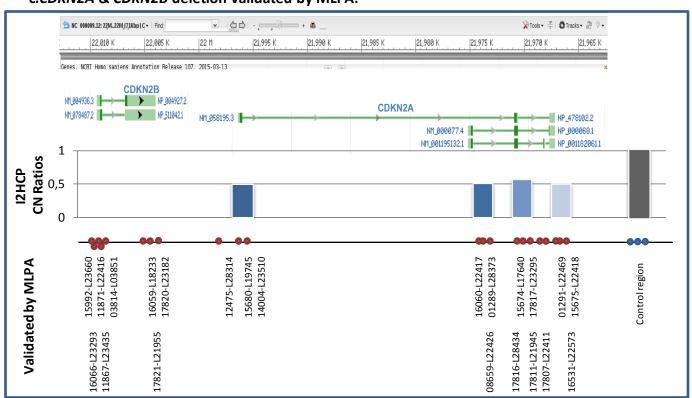
Gene	Transcript	Exon Rank	Ensemblid	CDS Start	CDS End
BAP1	NM 004656		ENSE00001879355	1	37
BARD1	NM 000465		ENSE00002311538	1	158
BRAF	NM_004333		ENSE00001154485	1	138
CBL	NM_005188	1	ENSE00001240819	1	195
CDH1	NM_004360	1	ENSE00001859184	1	48
CDKN1C	NM_000076	1	ENSE00001691292	1	820
CDKN1C	NM_000076	2	ENSE00003634161	821	951
ERCC2	NM_000400	1	ENSE00001510142	1	5
FANCA	NM_000135	1	ENSE00002618162	1	79
FANCE	NM_021922	1	ENSE00000849631	1	248
FH	NM_000143	1	ENSE00001442032	1	132
MAP2K2	NM_030662	1	ENSE00001147081	1	92
MN1	NM_002430	1	ENSE00001150385	1	3781
MSH6	NM_000179	1	ENSE00002724201	1	260
NF1	NM_000267	1	ENSE00003598164	1	60
PDGFB	NM_002608	4	ENSE00000654697	251	456
PTCH1	NM_000264	1	ENSE00001630740	1	201
PTPN11	NM_002834	1	ENSE00001202658	1	14
RASA1	NM_002890	1	ENSE00001505330	1	539
RET	NM_020975	1	ENSE00001948565	1	73
TMEM127	NM_017849	2	ENSE00000921763	1	244
WT1	NM_024426	1	ENSE00001906049	1	646
XPA	им_000380	1	ENSE00001465835	1	172

SUPPLEMENTARY FIGURE 5

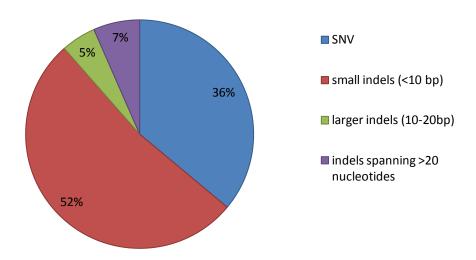
Sample R2



c.CDKN2A & CDKN2B deletion validated by MLPA:



PATHOGENIC VARIANT TYPES



	НВОС	FAP	HNPCC	NF1	NF2
BRCA1	•		•		
BRCA2	•		•		
APC		•	•		
MUTYH		•	•		
MLH1	•		•		
MSH2	•		•		
MSH6	•		•		
PMS2	•		•		
NF1				•	
NF2					•
PTEN	•	•	•		
STK11	•	•	•		
EPCAM	•		•		
TP53	•		•		
ATM	•				
CDH1	•				
CHEK2	•				
PALB2	•				
RAD51C	•				
RAD51D	•				
POLD1		•	•		
POLE		•	•		
BMPR1A		•			
SMAD4		•			
SPRED1				•	
MN1					•
PDGFB					•
SMARCB1					•
SMARCE1					•
SUFU					•

Other Genes Included in the I2HCP:

AIP	ELAC2	FANCM	PDGFRA	SDHC
ALK	ERCC2	FH	PHOX2B	SDHD
			PMS1	SHOC2
ARAF	ERCC3	FLCN		
BAP1	ERCC4	GPC3	POLH	SLX4
BARD1	ERCC5	HNF1A	PPM1D	SOS1
BLM	ERCC6	HRAS	PRKAR1A	TGFBR2
BRAF	ERCC8	KIT	PRSS1	TMEM127
BRIP1	EXO1	KLLN	PTCH1	TSC1
BUB1B	EXT1	KRAS	PTPN11	TSC2
CBL	EXT2	MAP2K1	RAD50	TSHR
CDC73	FANCA	MAP2K2	RAD51	VHL
CDK4	FANCB	MAX	RAF1	WRN
CDKN1C	FANCC	MEN1	RASA1	WT1
CDKN2A	FANCD2	MET	RB1	XPA
CDKN2C	FANCE	MLH3	RET	XPC
CYLD	FANCF	MRE11A	RNASEL	XRCC2
DDB1	FANCG	MSH3	SBDS	
DDB2	FANCI	NBN	SDHAF2	
DICER1	FANCL	NRAS	SDHB	

<u>Table S2</u>: Results from Training set (n=23)

Sample	Genes	Pathogenic Variant	Detected in 1st Approach	Detected in 2nd Approach	Variant Context
T1	BRCA1	NM_007294.2: c.2397T>A	YES	YES	
T2	APC	NM_001127510.2: c.3927_3931delAAAGA	YES	YES	
Т3	NF1	NM_000267.3:c.7739C>G	YES	YES	
T4	NF1	NM_000267.3: c.3860_3861insTT	YES	YES	
T5	BRCA1	NM_007294.2:c.1961delA	NO	YES	HomoP
Т6	MLH1	NM_000249.3: c.306+5A>G	YES	YES	
Т7	BRCA2	NM_000059.3:c.956dupA	NO	YES	HomoP
Т8	MSH6	NM_000179.2:c.3261delC	NO	YES	HomoP
Т9	NF1	NM_000267.3:c.4306A>G	YES	YES	
T10	MSH6	NM_000179.2:c.255dupC	NO	NO	Low coverage
T11	NF1	NM_000267.3: Del exons 33-36	YES	YES	
T12	NF2	NM_000268.3: Del exons 1-5	YES	YES	
T13	NF1	NM_000267.3:c.6365-2A>G	YES	YES	
T14	APC	NM_001127510.2:c.423-3T>A	YES	YES	
T15	MSH2	NM_000251.2:c.602_603insT	NO	YES	HomoP
T16	BRCA2	NM_000059.3:c.8946delA	NO	YES	HomoP
T17	BRCA2	NM_000059.3:c.2402_2420del19	YES	YES	
T18	BRCA2	NM_000059.3:c.6275_6276delTT	YES	YES	
T19	MSH6	NM_000179.2:c.3150_3161dup12	YES	YES	
T20	MSH6	NM_000179.2:c.3261dupC	NO	YES	HomoP
T21	MLH1	NM_000249.3:c.1590_1598dup9	YES	YES	
T22	NF1	NM_000267.3:c.6226delG	YES	YES	
T23	BRCA1	NM_007294.2:c.1953_1956delGAAA	YES	YES	

	totalDaada	netMennedDeede	anne de la Decido de Decido				C1	C10	C30	C60	C100	U20	U50
sample T1	totalReads 2700557	pctMappedReads 99.89%	properlyPairedReads 98.48%	meanCoverage 458.3894277	medianCoverage 442	maxCoverage 1443	99.93%	99.78%	99.35%	98.42%	96.72%	0.66%	0.28%
T2	3620541	99.90%	98.90%	601.7126241	580	2052	99.96%	99.82%	99.41%	98.89%	97.53%	0.67%	0.28%
T3	2971530	99.90%	98.87%	496.6900251	476	1741	99.97%	99.75%	99.15%	98.11%	96.55%	0.68%	0.30%
T4	3080176	99.90%	98.97%	557.5283568	535	1886	99.90%	99.73%	99.28%	98.46%	97.08%	0.69%	0.31%
T5	2932666	99.22%	96.18%	336.7794168	325	1236	99.92%	99.68%	98.75%	96.89%	93.72%	0.68%	0.31%
T6	2948395	99.89%	97.81%	270.3864092	256	1087	99.59%	98.82%	97.39%	94.58%	88.94%	0.70%	0.34%
T7	2974347	99.80%	97.34%	483.3309917	465	1679	99.94%	99.69%	99.18%	98.21%	96.27%	0.68%	0.30%
T8	4030929	98.80%	95.68%	579.8089141	558	2152	99.97%	99.69%	99.27%	98.24%	96.68%	0.69%	0.31%
Т9	6529562	99.04%	97.86%	1086.624322	1051	3512	99.99%	99.83%	99.61%	99.21%	98.53%	0.69%	0.30%
T10	2937637	99.75%	97.56%	398.1105709	382	1500	99.87%	99.68%	99.03%	97.58%	95.35%	0.68%	0.31%
T11	3455356	99.95%	98.73%	155.0875564	149	579	99.65%	98.52%	96.55%	90.25%	75.71%	0.68%	0.31%
T12	3454764	99.91%	98.69%	526.7052471	508	1792	99.98%	99.78%	99.26%	98.42%	96.87%	0.68%	0.29%
T13	3157978	99.95%	98.83%	438.9549286	422	1609	99.55%	98.83%	97.91%	96.69%	94.56%	0.70%	0.34%
T14	3040733	99.38%	95.45%	481.469552	463	1725	99.86%	99.69%	99.12%	98.28%	96.28%	0.68%	0.30%
T15	2491293	99.55%	97.02%	409.6304417	395	1465	99.89%	99.66%	99.12%	97.69%	95.47%	0.68%	0.30%
T16	4103602	99.37%	97.17%	447.420466	426	1672	99.93%	99.60%	98.93%	97.58%	95.60%	0.69%	0.31%
T17	4101591	99.32%	97.40%	646.1095886	621	2398	99.92%	99.79%	99.37%	98.63%	97.24%	0.69%	0.31%
T18	2823557	99.55%	97.95%	440.1877041	424	1643	99.95%	99.69%	98.93%	97.72%	95.78%	0.69%	0.31%
T19	4366424	99.70%	98.02%	635.1975054	605	2428	99.99%	99.77%	99.33%	98.52%	97.20%	0.69%	0.32%
T20	3517122	99.94%	98.95%	512.7125471	496	1768	99.92%	99.76%	99.25%	98.36%	96.79%	0.67%	0.30%
T21	3414029	99.84%	98.44%	489.5334775	472	1741	99.90%	99.76%	99.26%	98.20%	96.49%	0.67%	0.30%
T22	2852050	99.41%	98.01%	506.8352003	484	2014	99.90%	99.64%	99.17%	98.19%	96.49%	0.69%	0.32%
T23	2702987	99.59%	97.87%	432.9446815	413	1649	99.91%	99.70%	99.05%	97.76%	95.75%	0.69%	0.31%
V1	3120052	99.94%	98.80%	295.6665951	290	842	99.95%	99.52%	98.55%	96.75%	93.33%	0.66%	0.27%
V2 V3	3102542 3184166	99.91%	98.89%	437.7827133 412.2868723	414 400	1910 1354	99.82% 99.95%	99.33% 99.75%	98.53% 99.16%	97.27% 97.93%	94.89% 95.95%	0.70% 0.68%	0.32% 0.29%
V3 V4	3564282	99.93% 99.73%	98.94% 98.73%	566.3987475	534	2428	99.83%	99.75%	98.60%	97.57%	96.07%	0.06%	0.29%
V4 V5	2324799	99.66%	98.38%	234.0922369	217	1019	99.84%	98.74%	95.74%	89.85%	80.52%	0.71%	0.44%
V6	2762570	99.71%	98.75%	409.2610649	376	1933	99.81%	99.08%	97.43%	95.03%	90.66%	0.77%	0.44 %
V7	3389493	99.36%	97.71%	490.9416272	460	2102	99.81%	99.19%	98.19%	96.77%	94.31%	0.72%	0.38%
V8	2898965	99.68%	98.58%	480.3903083	454	1917	99.81%	99.24%	98.23%	96.98%	94.55%	0.72%	0.36%
V9	3022540	99.66%	97.68%	180.8694414	174	645	99.79%	99.28%	97.32%	92.81%	82.18%	0.68%	0.30%
V10	3142118	99.82%	98.55%	412.5336949	402	1243	99.89%	99.59%	99.07%	97.95%	96.13%	0.67%	0.28%
V11	3348711	99.61%	97.56%	539.0104176	523	1576	99.98%	99.84%	99.38%	98.67%	97.39%	0.67%	0.28%
V12	3815649	99.73%	97.82%	114.4278238	111	390	99.82%	98.79%	95.11%	83.63%	56.84%	0.68%	0.30%
V13	3202154	99.68%	97.92%	483.2600551	473	1444	99.96%	99.77%	99.24%	98.34%	96.67%	0.67%	0.28%
V14	3118424	99.15%	96.59%	463.5094917	451	1366	99.97%	99.70%	99.25%	98.31%	96.60%	0.66%	0.28%
V15	3310405	99.77%	97.91%	515.3704581	493	1820	99.94%	99.66%	99.12%	98.18%	96.73%	0.69%	0.31%
V16	2675671	99.86%	98.01%	393.4459817	377	1546	99.95%	99.63%	98.73%	97.32%	94.81%	0.69%	0.31%
V17	3121897	99.88%	97.46%	366.6039986	349	1422	99.87%	99.48%	98.62%	97.06%	94.23%	0.69%	0.32%
V18	3209613	99.88%	98.22%	497.52831	478	1793	99.96%	99.61%	99.01%	97.96%	96.28%	0.68%	0.31%
V19	1655353	99.83%	97.31%	237.1547783	226	836	99.89%	99.23%	97.88%	94.76%	87.70%	0.70%	0.33%
V20	2735448	99.76%	97.67%	425.3410158	409	1547	99.88%	99.57%	98.91%	97.62%	95.57%	0.68%	0.31%
V21	3343120	99.85%	97.90%	509.2725969	494	1663	99.95%	99.68%	99.22%	98.21%	96.82%	0.68%	0.30%
V22	5416213	98.65%	96.47%	272.6839881	263	1017	99.92%	99.51%	98.33%	96.14%	91.34%	0.68%	0.30%
V23	4263438	99.94%	98.48%	208.4556151	200	725	99.84%	99.32%	97.58%	93.85%	85.93%	0.68%	0.31%
V24	3250856	99.92%	98.46%	401.8487658	386	1443	99.85%	99.65%	99.26%	97.93%	95.44%	0.68%	0.29%
V25 V26	3511443	99.93%	98.70%	445.2353951	426 602	1590 2206	99.97% 99.99%	99.77% 99.83%	99.19% 99.39%	98.06%	96.08%	0.67%	0.30% 0.29%
V26 V27	3815216 3403384	99.94% 99.94%	98.73% 98.40%	624.4534936 397.4319897	382	1351	99.99%	99.83%	99.39%	98.84% 97.99%	97.75% 95.54%	0.67% 0.67%	0.29%
V27 V28	4403274	99.92%	98.60%	727.3731052	704	2487	99.93%	99.71%	99.12%	99.10%	98.20%	0.67%	0.29%
V29	2993755	99.94%	98.13%	209.4945422	202	704	99.85%	99.36%	97.89%	94.44%	86.80%	0.68%	0.29%
V29 V30	3135958	99.94%	98.64%	465.2157651	448	1693	99.93%	99.74%	99.12%	98.16%	96.31%	0.67%	0.29%
V30	2925995	99.93%	98.61%	273.4182949	256	1057	99.94%	99.41%	98.08%	95.46%	89.72%	0.70%	0.29%
V32	3160863	99.93%	97.03%	235.1366617	221	934	99.84%	99.20%	97.47%	94.15%	86.72%	0.71%	0.35%
V33	3427158	99.85%	92.68%	493.8036751	475	1685	99.92%	99.75%	99.18%	98.22%	96.56%	0.68%	0.30%
V34	3248449	99.92%	98.92%	493.7740374	475	1732	99.94%	99.74%	99.31%	98.32%	96.51%	0.68%	0.30%
V35	2826057	99.94%	99.05%	387.3789909	373	1447	99.85%	99.45%	98.83%	97.71%	95.29%	0.67%	0.29%
V36	2551495	99.89%	98.71%	253.8151548	244	1015	99.78%	99.21%	98.14%	95.76%	90.67%	0.68%	0.29%
V37	3427675	99.91%	98.60%	135.1547646	131	469	99.44%	98.35%	95.37%	86.84%	68.07%	0.69%	0.32%
V38	2648692	99.67%	98.59%	397.9490951	378	1483	99.71%	99.10%	98.08%	96.71%	94.14%	0.71%	0.34%
V39	2771123	99.94%	99.22%	459.7732505	435	1881	99.83%	99.24%	98.35%	97.28%	95.34%	0.70%	0.33%
V40	3422016	99.93%	99.23%	575.9554321	548	2453	99.90%	99.48%	98.78%	98.00%	96.60%	0.70%	0.32%

Sample Name	Variants in analyzed genes detected with I2HCP V2	Rs (Snp138)	Variant Freq	Status	Reason to be undetected by pre-NGS methods	Validated by Sanger	Variants also detected in I2HCP V1 (PGM)
TRAINING							
T1							
	BRCA2:NM_000059:c.425+67A>C	rs11571610	56,27	Known			YES
	BRCA2:NM_000059:c.425+147G>T	rs4942423	44,83		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.426-89T>C	rs3783265	55,11	Known			YES
	BRCA2:NM_000059:c.631+183T>A	rs3752451	48,57		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.681+56C>T	rs2126042	49,3		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,24		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.865A>C	rs766173	46,41	Known			YES
	BRCA2:NM_000059:c.1365A>G	rs1801439	36,47	Known			YES
	BRCA2:NM_000059:c.1910-74T>C	rs2320236	48,62		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1910-51G>T	rs11571651	49,87		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.2229T>C	rs1801499	48,6	Known			YES
	BRCA2:NM_000059:c.2971A>G	rs1799944	52,22	Known			YES
	BRCA2:NM_000059:c.4563A>G	rs206075	100		Could not be detected by CSCE		
	BRCA2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE		
	BRCA2:NM_000059:c.6937+151A>G	rs4942439	41,38		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.6937+177G>A	rs4942440	41,67		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7397T>C	rs169547	100		Could not be detected by CSCE		
	BRCA2:NM 000059:c.7435+53C>T	rs11147489	48,15	Known			YES
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		. = 0
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	100	Known	out of profitor mounds covered region		YES
	BRCA2:NM 000059:c.8755-66T>C	rs4942486	99,57	Talowii	Out of pre-NGS method covered region		120
	BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	62,86		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5278-191A>T	rs8176297	33,78		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5152+66G>A	rs3092994	49,71		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5144G>A	rs45444999	51,29	Known	Out of pre-1403 method covered region		YES
	BRCA1:NM_007294:c.5075-237C>A	rs8176257	66,67	KIIOWII	Out of pre-NGS method covered region		120
	BRCA1:NM_007294:c.5074+65G>A	rs8176235	51,77	Known	Out of pre-1403 method covered region		YES
	BRCA1:NM_007294:C:3074+03G>A BRCA1:NM_007294:C:4987-68A>G	rs8176234	48,32	KIIOWII	Out of pre-NGS method covered region		TES
	BRCA1:NM 007294:c.4987-96A2G	rs8176233	46,79				
		rs3092987	71,43		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4986+222A>G			I/o access	Out of pre-NGS method covered region	+	VEC
	BRCA1:NM_007294:c.4837A>G	rs1799966	53,83	Known		+	YES YES
	BRCA1:NM_007294:c.4485-63C>G	rs8176212	52,48	Known	Out of any NOO mother discussed as sign	-	YES
	BRCA1:NM_007294:c.4485-137T>A	rs2236762	47,32		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4358-2590T>G	rs8176194	45,41		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4358-2885G>A	rs8176193	47,98	14	Out of pre-NGS method covered region		\/F0
	BRCA1:NM_007294:c.4308T>C	rs1060915	50,43	Known			YES
	BRCA1:NM_007294:c.4097-141A>C	rs799916	50,83	1,	Out of pre-NGS method covered region		1/50
	BRCA1:NM_007294:c.3548A>G	rs16942	41,54	Known			YES
	BRCA1:NM_007294:c.3113A>G	rs16941	50,27	Known			YES
	BRCA1:NM_007294:c.2612C>T	rs799917	50,78	Known			YES
	BRCA1:NM_007294:c.2397T>A	rs80357203	52,11	PAT			YES
	BRCA1:NM_007294:c.2311T>C	rs16940	51,76	Known			YES
	BRCA1:NM_007294:c.2082C>T	rs1799949	49,38	Known			YES
	BRCA1:NM_007294:c.594-225G>A	rs8176147	45,83		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.593+167T>C	rs8176145	45,45	Known			YES
	BRCA1:NM_007294:c.547+146A>T	rs8176140	36,67		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.213-161A>G	rs799912	55,56		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.1-134T>C	rs3765640	46,97	Known			YES
	BRCA1:NM 007294:c.4485-203 4485-199delAACCC	rs34250703	30.77		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.548-58 548-58delT	rs8176144	45.14		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.441+64 441+64delT	rs368252296	43.55		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.441+52 441+63delCTTTTTTTTTT		81.33		Out of pre-NGS method covered region		

	BRCA1:NM_007294:c.441+36_441+38delCTT	rs147856441	45.96		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.134+224_134+224delT	rs35149296	52.94		Out of pre-NGS method covered region	
T2						
	APC:NM_001127510:c.136-230C>A	rs2464805	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.423-256G>A	rs392179	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.645+173A>G	rs2289484	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.933+261G>T	rs12656359	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-132G>T	rs12521276	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1458T>C	rs2229992	99,76		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1635G>A	rs351771	99,91		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1743+193G>A	rs351772	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.4479G>A	rs41115	99,81		Out of pre-NGS method covered region	
	APC:NM_001127510:c.5034G>A	rs42427	99,82		Out of pre-NGS method covered region	
	APC:NM_001127510:c.5268T>G	rs866006	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.5465T>A	rs459552	99,55		Out of pre-NGS method covered region	
	APC:NM 001127510:c.5880G>A	rs465899	100		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-132_934-132delG	rs141013063	58.82		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-120_934-119insG	rs138003874	78.99		Out of pre-NGS method covered region	
	APC:NM 001127510:c.1959-145 1959-144insAGAA	rs3839284	98.57		Out of pre-NGS method covered region	
	APC:NM_001127510:c.3921_3925delAAAAG		44.81	PAT		YES
T3						
	NF1:NM 000267:c.61-300C>T	rs2269856	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.61-123G>A	rs2269855	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.288+41G>A	rs2952976	99,74		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.480-90C>T	rs2905807	99,17		Out of pre-NGS method covered region	
	NF1:NM 000267:c.702G>A	rs1801052	98,16	Known		YES
	NF1:NM 000267:c.888+108C>T	rs2953000	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.888+118G>T	rs2952999	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.1393-130A>T	rs2905875	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.1393-32T>C	rs2905876	99,69		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1641+39T>C	rs2905880	99,76		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.5205+23T>C	rs9894648	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.5546+19T>A	rs2285894	99,42		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.5546+117G>A	rs3815154	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.6085-29T>A	rs7406038	90,72		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-28T>A	rs7406039	99,32		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.7126+37C>G	rs7405740	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7127-167T>A	rs2525569	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.7395-29G>A	rs964288	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.7739C>G		48,72	PAT	,	YES
	NF1:NM 000267:c.7907+166C>T	rs7350943	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.730+15 730+16insT		25.39		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.889-191 889-190insT	rs35966725	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.1528-37 1528-36insT	rs67472948	94.06		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.2002-118 2002-117insAC	rs141114572	39.71		Out of pre-NGS method covered region	
T4						
	NF1:NM_000267:c.61-300C>T	rs2269856	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.61-123G>A	rs2269855	99,86		Out of pre-NGS method covered region	
	NF1:NM_000267:c.288+41G>A	rs2952976	99,53		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.480-90C>T	rs2905807	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.702G>A	rs1801052	98,91	Known		 YES
	NF1:NM_000267:c.888+108C>T	rs2953000	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.888+118G>T	rs2952999	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.1393-130A>T	rs2905875	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.1393-32T>C	rs2905876	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1641+39T>C	rs2905880	99,82		Could not be detected by RNA-based analysis	
	1		40.55	Known		YES
	NF1:NM_000267:c.1726C>G NF1:NM_000267:c.5205+23T>C	rs9894648	48,55 100		Could not be detected by RNA-based analysis	TLO

	NF1:NM_000267:c.5546+19T>A	rs2285894	99,54		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5546+117G>A	rs3815154	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.6085-29T>A	rs7406038	90,16		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-28T>A	rs7406039	95,56		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7126+37C>G	rs7405740	99,51		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.7127-167T>A	rs2525569	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.7395-29G>A	rs964288	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.7907+166C>T	rs7350943	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.730+15 730+16insT		27.34		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.889-191 889-190insT	rs35966725	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.1528-37 1528-36insT	rs67472948	96.02		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.2002-118 2002-117insAC	rs141114572	44.44		Out of pre-NGS method covered region	
	NF1:NM_000267:c.3860_3861insTT		44.39		Could not be detected by RNA-based analysis	YES
	NF1:NM 000267:c.6085-31 6085-30insA		34.49		Could not be detected by RNA-based analysis	
T5					, , , , , , , , , , , , , , , , , , , ,	
-	BRCA2:NM 000059:c.793+98G>A	rs206073	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.3807T>C	rs543304	99,27	Known		YES
	BRCA2:NM 000059:c.4563A>G	rs206075	99,42		Could not be detected by CSCE	·
	BRCA2:NM 000059:c.6513G>C	rs206076	99,62		Could not be detected by CSCE	
	BRCA2:NM 000059:c.6938-120T>C	rs206080	98,97		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7397T>C	rs169547	99,81		Could not be detected by CSCE	
	BRCA2:NM_000059:c.7435+207T>A	10100041	57,14		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.9501+178A>G	rs206343	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.10257+105A>C	rs15869	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5333-153A>G	rs8176305	40,54		Out of pre-NGS method covered region	
	BRCA1:NM 007294:C:5278-191A>T	rs8176297	100		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.5152+66G>A	rs3092994	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5075-53C>T	rs8176258	57,73		Out of pre-NGS method covered region	YES
	BRCA1:NM_007294:c.5075-237C>A	rs8176257	60	Known	Out of pre-NGS method covered region	1123
	BRCA1:NM 007294:c.5075-2376-A	rs8176256	35		Out of pre-NGS method covered region	
		rs8176235	44,9		Out of pre-NGS method covered region	VEC
	BRCA1:NM_007294:c.5074+65G>A	rs8176234		Known	Out of are NCC method sovered region	YES
	BRCA1:NM_007294:c.4987-68A>G		100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4987-92A>G BRCA1:NM_007294:c.4986+222A>G	rs8176233	97,96		Out of pre-NGS method covered region	
		rs3092987	100		Out of pre-NGS method covered region	VEC
	BRCA1:NM_007294:c.4837A>G	rs1799966	99,3	Known	0.1.5	YES
	BRCA1:NM_007294:c.4675+245G>A	rs141660808	53,85		Out of pre-NGS method covered region	VEC
	BRCA1:NM_007294:c.4485-63C>G	rs8176212	98,75	Known	O to form NOO coefficient construction	YES
	BRCA1:NM_007294:c.4485-137T>A	rs2236762	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4358-2590T>G	rs8176194	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4358-2885G>A	rs8176193	99,51		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4357+117G>A	rs3737559	49,25		Out of pre-NGS method covered region	V/50
	BRCA1:NM_007294:c.4308T>C	rs1060915	99,54	Known	0 11 11 11 1200	YES
	BRCA1:NM_007294:c.4097-141A>C	rs799916	99,14		Could not be detected by CSCE	VEO
	BRCA1:NM_007294:c.3548A>G	rs16942	99,28	Known		YES
	BRCA1:NM_007294:c.3113A>G	rs16941	99,14	Known		YES
	BRCA1:NM_007294:c.2612C>T	rs799917	99,61	Known		YES
	BRCA1:NM_007294:c.2311T>C	rs16940	99,28	Known		YES
	BRCA1:NM_007294:c.2082C>T	rs1799949	99,73	Known		YES
	BRCA1:NM_007294:c.2077G>A	rs4986850	49,54	Known		YES
	BRCA1:NM_007294:c.594-225G>A	rs8176147	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.593+167T>C	rs8176145	100	Known		YES
	BRCA1:NM_007294:c.547+146A>T	rs8176140	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.441+36C>T	rs45569832	62,5	ļ	Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.213-161A>G	rs799912	100		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.1-134T>C	rs3765640	100	Known		YES
	BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	54.39		Out of pre-NGS method covered region	

BRC	A1:NM_007294:c.1961_1961delA	rs80357522	43.32	PAT		NO, variant in homopolymer reg
	:A1:NM 007294:c.548-58 548-58delT	rs8176144	92.95		Out of pre-NGS method covered region	. , ,
	A1:NM 007294:c.441+52 441+63delCTTTTTTTTT		95.71		Out of pre-NGS method covered region	
	A1:NM 007294:c.441+36 441+38delCTT	rs147856441	97.97		Out of pre-NGS method covered region	
T6	7 (1.14M)_00120 1.0.111 - 00_111 - 00001011	10111000111	07.07		Cat of profited mounds severed region	
	11:NM 000249:c.306+5G>A	rs267607735	46,33	PAT		YES
	1:NM 000249:c.453+79A>G	rs4234259	100	I AI	Out of pre-NGS method covered region	120
	11:NM 000249:c.655A>G	rs1799977	45,41	Known	Out of pre-NOS method covered region	YES
	11:NM 000249:c.1038+86T>C	rs2286939	100	Known		YES
	11:NM_000249:c.1668-19A>G		100			YES
	11:NM_000249:c.1968-19A>G 1:NM_000249:c.1990-121C>T	rs9876116		Known		
		rs2241031	100	Known	O tofour NOO well adversed as in	NO, variant in low covered region
	1:NM_000249:c.1897-167_1897-167delT	rs11290150	100		Out of pre-NGS method covered region	
T7	440 NH4 0000E0 = T00 000 A	000070	00.50		0.4.6. 1100 # 1	VEO
	:A2:NM_000059:c.793+98G>A	rs206073	99,59		Out of pre-NGS method covered region	YES
	:A2:NM_000059:c.3807T>C	rs543304	99,78	Known		
	:A2:NM_000059:c.4563A>G	rs206075	100		Could not be detected by CSCE	
	:A2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE	
	:A2:NM_000059:c.6938-120T>C	rs206080	100		Could not be detected by CSCE	
	:A2:NM_000059:c.7397T>C	rs169547	100		Could not be detected by CSCE	
	:A2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
BRC	:A2:NM_000059:c.9501+178A>G	rs206343	100		Out of pre-NGS method covered region	
BRC	:A2:NM_000059:c.10257+105A>C	rs15869	98,06		Out of pre-NGS method covered region	
BRC	A2:NM_000059:c.949_950insA		43.17	PAT		NO, variant in homopolymer re
BRC	A1:NM 007294:c.5407-193A>G	rs8176310	100		Out of pre-NGS method covered region	
BRC	A1:NM 007294:c.5333-153A>G	rs8176305	46,56		Out of pre-NGS method covered region	
	:A1:NM 007294:c.5278-191A>T	rs8176297	98,96		Out of pre-NGS method covered region	
	A1:NM 007294:c.5152+66G>A	rs3092994	100		Out of pre-NGS method covered region	
	A1:NM 007294:c.5075-237C>A	rs8176257	41,18		Out of pre-NGS method covered region	
	A1:NM_007294:c.5075-238A>G	rs8176256	58,82		Out of pre-NGS method covered region	
	A1:NM 007294:c.5074+65G>A	rs8176235	44,33	Known	Cut of profited mounds severed region	YES
	A1:NM 007294:c.4987-68A>G	rs8176234	99,49	TUIOWII	Out of pre-NGS method covered region	120
	A1:NM 007294:c.4987-92A>G	rs8176233	97,84		Out of pre-NGS method covered region	
	A1:NM 007294:c.4986+222A>G	rs3092987	100		Out of pre-NGS method covered region	
	A1:NM 007294:c.4837A>G	rs1799966	99,29	Known	Out of pre-NGS method covered region	YES
	A1:NM 007294:c.4485-63C>G	rs8176212	99,46	Known		YES
	A1:NM 007294:c.4465-137T>A	rs2236762	99,40	KIIOWII	Out of pre-NGS method covered region	163
	A1:NM 007294:c.4465-15712A A1:NM 007294:c.4358-2590T>G					
		rs8176194	98,65		Out of pre-NGS method covered region	
	A1:NM_007294:c.4358-2885G>A	rs8176193	100		Out of pre-NGS method covered region	
	A1:NM_007294:c.4357+117G>A	rs3737559	75,36	17	Out of pre-NGS method covered region	VE0
	A1:NM_007294:c.4308T>C	rs1060915	99,48	Known		YES
	A1:NM_007294:c.4097-141A>C	rs799916	100	14	Out of pre-NGS method covered region	VE2
	A1:NM_007294:c.3548A>G	rs16942	100	Known		YES
	:A1:NM_007294:c.3113A>G	rs16941	99,67		Could not be detected by CSCE	
	A1:NM_007294:c.2612C>T	rs799917	99,02	Known		YES
BRC	:A1:NM_007294:c.2311T>C	rs16940	99,46	Known		YES
	A1:NM_007294:c.2082C>T	rs1799949	98,81	Known		YES
	:A1:NM_007294:c.2077G>A	rs4986850	46,48	Known		YES
	:A1:NM_007294:c.594-225G>A	rs8176147	100	1	Out of pre-NGS method covered region	
	:A1:NM_007294:c.593+167T>C	rs8176145	100		Out of pre-NGS method covered region	
BRC	:A1:NM_007294:c.547+146A>T	rs8176140	100		Out of pre-NGS method covered region	
BRC	A1:NM_007294:c.441+36C>T	rs45569832	55,56		Out of pre-NGS method covered region	
	:A1:NM 007294:c.213-161A>G	rs799912	100		Out of pre-NGS method covered region	
	A1:NM_007294:c.1-134T>C	rs3765640	90,91		Out of pre-NGS method covered region	
	A1:NM 007294:c.5277+59 5277+60insGTATTCCACTCC		46.04	Known		YES
	A1:NM 007294:c.4485-203 4485-199delAACCC	rs34250703	70.21	1	Out of pre-NGS method covered region	1
	A1:NM 007294:c.548-58 548-58delT	rs8176144	93.54	+	Out of pre-NGS method covered region	
	A1:NM 007294:c.441+52 441+63delCTTTTTTTTTT	100110177	93.23		Out of pre-NGS method covered region	
	A1:NM 007294:c.441+36 441+38delCTT	rs147856441	96.59		Out of pre-NGS method covered region	

	BRCA1:NM 007294:c.134+224 134+224delT	rs35149296	62.5		Out of pre-NGS method covered region	
Т8	BRCAT.NW_007294.C.134+224_134+2240e11	1830149290	62.5		Out of pre-NGS method covered region	
10	MSH6:NM 000179:c.186C>A	rs1042820	100		Could not be detected by D-HPLC	
	MSH6:NM 000179:c.180C>A MSH6:NM 000179:c.260+22C>G	rs55927047	100		Could not be detected by D-HPLC	
	MSH6:NM 000179:c.276A>G	rs1800932	98,05	Known	Codid not be detected by D-HFLC	YES
	MSH6:NM_000179:c.457+52T>A	rs3136282	98,54	Known		YES
	MSH6:NM 000179:c.457+521>A MSH6:NM 000179:c.457+217G>T	rs3136283	100	KIIOWII	Out of pro NCS method sovered region	1E3
		rs3136315	100	+	Out of pre-NGS method covered region	
	MSH6:NM_000179:c.458-273A>G			I/	Out of pre-NGS method covered region	NOinstinction law.
	MSH6:NM_000179:c.458-133A>G	rs3136316	99,15	Known		NO, variant in low covered regior
	MSH6:NM_000179:c.540T>C	rs1800935	99,09	Known		YES YES
	MSH6:NM_000179:c.3173-101G>C	rs2072447	100	Known	Osvild mat has datasted by DUDLO	YES
	MSH6:NM_000179:c.3438+14A>T	rs2020911	99,57	17	Could not be detected by D-HPLC	NO selection of the control of the c
	MSH6:NM_000179:c.3556+146G>A	rs7562048	100	Known		NO, variant in low covered regior
	MSH6:NM_000179:c.3646+91T>C	rs3136359	100	Known		YES
	MSH6:NM_000179:c.3802-40C>G	rs3136367	99,6	Known		YES
	MSH6:NM_000179:c.3254_3254delC		46.39	PAT		NO, variant in homopolymer region
	MSH6:NM_000179:c.3557-18_3557-17insT		90.24		Could not be detected by D-HPLC	
	MSH6:NM_000179:c.3646+29_3646+32delCTAT	rs2234731	88.42	Known		YES
	MSH6:NM_000179:c.4002-27_4002-27delT		34.74		Out of pre-NGS method covered region	
T9						
	NF1:NM_000267:c.61-300C>T	rs2269856	51,35		Out of pre-NGS method covered region	
	NF1:NM_000267:c.2034G>A	rs2285892	100	Known		YES
	NF1:NM_000267:c.3197+139T>C	rs2071009	100		Out of pre-NGS method covered region	
	NF1:NM_000267:c.3315-130G>C	rs2072131	99,75		Out of pre-NGS method covered region	
	NF1:NM_000267:c.3496+33C>A	rs2066736	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3974+46T>G	rs72813695	83,3		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.4306A>G		48,37	PAT		YES
	NF1:NM 000267:c.4772+193T>C	rs7221954	55,43		Out of pre-NGS method covered region	
	NF1:NM 000267:c.5205+23T>C	rs9894648	48,12		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.5944-179A>C	rs10853138	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.6085-28T>A	rs7406039	50,06		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.6579+45T>A	rs17883614	48,28		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.7126+37C>G	rs7405740	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.8097+55T>C	rs143138479	47,06		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.2002-118 2002-117insACACACACACAC	10110100110	33.85		Out of pre-NGS method covered region	
	NF1:NM 000267:c.3974+33 3974+34insTG	·	67.97		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.6085-30 6085-29insA	rs66638017	37.03		Could not be detected by RNA-based analysis	
T10	101 1.100 000207.0.0000-00_0000-25IIISA	1300030017	37.03		Codid flot be detected by NNA-based affailysis	
110	MSH6:NM 000179:c.116G>A	rs1042821	100		Region not anlyzed by pre-NGS method	
	MSH6:NM 000179:c.255dupC	101012021	Not detecte	d PAT	Low coverage region – detected by Sanger sequencin	q NO, variant in low covered region
	MSH6:NM 000179:c.458-133A>G	rs3136316	100	41711	Out of pre-NGS method covered region	g res, ramanem non severea region
	MSH6:NM 000179:c.458-52G>T	rs1800934	100		Out of pre-NGS method covered region	
	MSH6:NM 000179:c.3173-101G>C	rs2072447	100		Out of pre-NGS method covered region	
	MSH6:NM 000179:c.3556+146G>A	rs7562048	100		Out of pre-NGS method covered region	
	MSH6:NM 000179:c.3336+146G>A MSH6:NM 000179:c.3646+91T>C		100		Out of pre-NGS method covered region	
		rs3136359				
	MSH6:NM_000179:c.3802-40C>G	rs3136367	100	+	Region not anlyzed by pre-NGS method	
	MSH6:NM_000179:c.3557-18_3557-17insT		37.5	+	Region not anlyzed by pre-NGS method	
	MSH6:NM_000179:c.3646+29_3646+32delCTAT	rs2234731	89.53	1	Region not anlyzed by pre-NGS method	
T44	MSH6:NM_000179:c.4002-27_4002-27delT		42.83		Region not anlyzed by pre-NGS method	
T11	NEA-NIM 000007:- 04 40005 A	0000055	100		Out of the NOO mathed assessed as its	
	NF1:NM_000267:c.61-123G>A	rs2269855	100	+	Out of pre-NGS method covered region	
	NF1:NM_000267:c.288+41G>A	rs2952976	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.480-90C>T	rs2905807	100	1	Out of pre-NGS method covered region	1/20
	NF1:NM_000267:c.702G>A	rs1801052	97,47	Known		YES
	NF1:NM_000267:c.730+85A>T		20,57	1	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.888+108C>T	rs2953000	100	1	Out of pre-NGS method covered region	
	NF1:NM_000267:c.888+118G>T	rs2952999	100	1	Out of pre-NGS method covered region	
	NF1:NM_000267:c.1393-130A>T	rs2905875	100	1	Out of pre-NGS method covered region	1

	NF1:NM 000267:c.1393-32T>C	rs2905876	100		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.1641+39T>C	rs2905880	100		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.1642-158A>G	rs17883230	70,59		Out of pre-NGS method covered region		
	NF1:NM 000267:c.2851-127G>A	rs181022226	62,86		Out of pre-NGS method covered region		
	NF1:NM 000267:c.5205+23T>C	rs9894648	51,64		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.5546+19T>A	rs2285894	47,42		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.5546+117G>A	rs3815154	41,22		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.6085-29T>A	rs7406038	42,05		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.6085-28T>A	rs7406039	47,78		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.7126+37C>G	rs7405740	50		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.7120+37C>G NF1:NM_000267:c.7395-29G>A	rs964288					
		15904200	41,51		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.730+15_730+16insT		29.59		Could not be detected by RNA-based analysis	+	
	NF1:NM_000267:c.1528-37_1528-36insT	rs67472948	94.59		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.2002-118_2002-117insACAC	rs141114572	28		Out of pre-NGS method covered region	,	(50
	NF1:NM_000267: exons 33-36 deletion			PAT)	/ES
T12							
	NF2:NM_000268:c.1-110G>C	rs1800540	100	<u> </u>	Out of pre-NGS method covered region		
	NF2:NM_000268:c.240+171T>A	rs2530662	100		Out of pre-NGS method covered region		
	NF2:NM_000268:c.241-58G>A	rs79901896	100	ļ	Could not be detected by RNA-based analysis		
	NF2:NM_000268:c.517-150C>T	rs6006220	46,78		Out of pre-NGS method covered region		
	NF2:NM_000268:c.600-154G>A	rs2071622	55		Out of pre-NGS method covered region		
	NF2:NM_000268:c.886-205C>T	rs2252472	45,95		Out of pre-NGS method covered region		
	NF2:NM_000268:c.1574+173G>A	rs56188151	46,15		Out of pre-NGS method covered region		
	NF2:NM_000268:c.1575-67G>A	rs140086	99,53		Could not be detected by RNA-based analysis		
	NF2:NM 000268:c.1737+1623T>C	rs7291645	60,29		Out of pre-NGS method covered region		
	NF2:NM 000268:c.599+41 599+41delT		28.98		Could not be detected by RNA-based analysis		
	NF2:NM 000268:c.885+173 885+173delT	rs35328234	57.89		Could not be detected by RNA-based analysis		
	NF2:NM 000268:c.1447-227 1447-226insTGAGGGA	rs3842713	31.71		Out of pre-NGS method covered region		
	NF2:NM 000268: exons 1-5 deletion		-	PAT		\	/ES
T13							
	NF1:NM 000267:c.61-300C>T	rs2269856	100		Out of pre-NGS method covered region		
	NF1:NM 000267:c.61-123G>A	rs2269855	100		Out of pre-NGS method covered region		
	NF1:NM 000267:c.288+41G>A	rs2952976	100		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.480-90C>T	rs2905807	100		Out of pre-NGS method covered region		
	NF1:NM 000267:c.702G>A	rs1801052	98,48	Known		\	/ES
	NF1:NM 000267:c.888+108C>T	rs2953000	100	KIIOWII	Out of pre-NGS method covered region		11111
	NF1:NM 000267:c.888+118G>T	rs2952999	100		Out of pre-NGS method covered region		
	NF1:NM 000267:c.1393-130A>T	rs2905875	98,66		Out of pre-NGS method covered region		
	NF1:NM 000267:c.1393-130A>1 NF1:NM 000267:c.1393-32T>C	rs2905876	99,78				
	-				Could not be detected by RNA-based analysis	+	
	NF1:NM_000267:c.1641+39T>C	rs2905880	100		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.4368-46G>C	rs17881285	50,72		Could not be detected by RNA-based analysis		
-	NF1:NM_000267:c.5205+23T>C	rs9894648	47,8	1	Could not be detected by RNA-based analysis		
-	NF1:NM_000267:c.5546+19T>A	rs2285894	49,56	_	Could not be detected by RNA-based analysis		
ļ	NF1:NM_000267:c.5546+117G>A	rs3815154	52,2	1	Out of pre-NGS method covered region		
	NF1:NM_000267:c.6085-29T>A	rs7406038	45,93	ļ	Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.6085-28T>A	rs7406039	48,88	ļ	Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.6365-2A>G		45,71	PAT		\	/ES
	NF1:NM_000267:c.7126+37C>G	rs7405740	43,85		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.7395-29G>A	rs964288	55,22		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.7907+166C>T	rs7350943	45,45		Out of pre-NGS method covered region		
1	NF1:NM_000267:c.8097+30G>T		27,54		Could not be detected by RNA-based analysis		
	NF1:NM_000267:c.8097+71A>T		30,61		Could not be detected by RNA-based analysis		
	NF1:NM 000267:c.8097+76C>A		26,19		Could not be detected by RNA-based analysis		
I				1			
			27.83		ICOUID HOLDE DELECTED DY KINA-DASED ANALYSIS		
	NF1:NM_000267:c.730+15_730+16insT	rs35966725	27.83 100		Could not be detected by RNA-based analysis Out of pre-NGS method covered region		
	NF1:NM_000267:c.730+15_730+16insT NF1:NM_000267:c.889-191_889-190insT	rs35966725	100		Out of pre-NGS method covered region		
	NF1:NM 000267:c.730+15 730+16insT NF1:NM 000267:c.889-191 889-190insT NF1:NM_000267:c.1528-37_1528-36insT	rs67472948	100 94.58		Out of pre-NGS method covered region Could not be detected by RNA-based analysis		
T14	NF1:NM_000267:c.730+15_730+16insT NF1:NM_000267:c.889-191_889-190insT		100		Out of pre-NGS method covered region		

	APC:NM_001127510:c.423-3T>A		44,24	PAT		YES
	APC:NM 001127510:c.645+173A>G	rs2289484	48,48	Known		YES
	APC:NM 001127510:c.1458T>C	rs2229992	49,55	Known		YES
	APC:NM 001127510:c.1635G>A	rs351771	46,6	Known		YES
	APC:NM 001127510:c.1743+193G>A	rs351772	47,62		Out of pre-NGS method covered region	
	APC:NM 001127510:c.4479G>A	rs41115	42,42	Known		YES
	APC:NM 001127510:c.5034G>A	rs42427	55,11	Known		YES
	APC:NM 001127510:c.5268T>G	rs866006	50,91	Known		YES
	APC:NM 001127510:c.5465T>A	rs459552	100	Known		YES
	APC:NM 001127510:c.5880G>A	rs465899	52,59	Known		YES
	APC:NM_001127510:c.934-120_934-119insG	rs138003874	26.76	Tanovin	Out of pre-NGS method covered region	1.20
	APC:NM 001127510:c.1959-145 1959-144insAGAA	rs3839284	47.69		Out of pre-NGS method covered region	
T15	AI C.IVIVI_001127310.C.1959-145_1959-144IIISAGAA	133033204	47.09		Out of pre-INGS method covered region	
110	MSH2:NM 000251:c.943-161C>T	rs17224276	48,65		Out of pre-NGS method covered region	
	MSH2:NM 000251:c.1077-80G>A	rs2347794	57,6		Out of pre-NGS method covered region	
	MSH2:NM_000251:c.1077-003>A MSH2:NM_000251:c.1277-118G>A	rs1981929	50,29		Out of pre-NGS method covered region	
	MSH2:NM 000251:c.1661+90T>C	rs10183143				
			49,43		Out of pre-NGS method covered region	
	MSH2:NM_000251:c.2006-127C>T	rs2059521	100		Out of pre-NGS method covered region	
	MSH2:NM_000251:c.2210+175G>A	rs4583514	32,14		Out of pre-NGS method covered region	
	MSH2:NM_000251:c.2635-111T>A	rs6737098	53,63	B 4 T	Out of pre-NGS method covered region	NO seeden Colores of the
	MSH2:NM_000251:c.597_598insT	•	47.18	PAT		NO, variant in homopolymer region
	MSH2:NM_000251:c.1276+117_1276+117delT		22.41		Out of pre-NGS method covered region	
T16						
	BRCA2:NM_000059:c.1-26G>A	rs1799943	49,36		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.425+246G>C	rs11571613	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.631+183T>A	rs3752451	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,29		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.3396A>G	rs1801406	99,68		Could not be detected by CSCE	
	BRCA2:NM_000059:c.4563A>G	rs206075	99,77		Could not be detected by CSCE	
	BRCA2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE	
	BRCA2:NM_000059:c.6841+80T>G	-	77,78		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.6841+191C>A	rs11571662	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.6938-120T>C	rs206080	99,6		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7242A>G	rs1799955	49,46		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.7397T>C	rs169547	100		Could not be detected by CSCE	
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	48,3		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.6841+78 6841+81delAATT	rs138193280	87.92		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.8942_8942delA		50.6	PAT		NO, variant in homopolymer region
T17		-				in a second and a second a second and a second a second and a second a second and a
	BRCA2:NM_000059:c.793+98G>A	rs206073	100		Could not be detected by CSCE	
	BRCA2:NM 000059:c.1114A>C	rs144848	100	Known	Codia not be detected by COCE	YES
	BRCA2:NM 000059:c.4563A>G	rs206075	99,66	TUIOWII	Could not be detected by CSCE	120
	BRCA2:NM_000059:c.6513G>C	rs206076	99,74		Could not be detected by CSCE	
	BRCA2:NM 000059:c.6938-120T>C	rs206080	99,07		Could not be detected by CSCE	
	BRCA2:NM_000059:c.7397T>C BRCA2:NM_000059:c.7617+190G>A	rs169547 rs206096	100	-	Could not be detected by CSCE Could not be detected by CSCE	
	BRCA2:NM_000059:c.7617+190G>A BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Could not be detected by CSCE Could not be detected by CSCE	
	BRCA2:NM 000059:c.2402 2420delACAATTATGAATCTGATGT	13200344		DAT	Could not be detected by CSCE	YES
		0470040	38,89	PAT	Out of man NOO menth and as a second section	IEO
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	44,23		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5333-153A>G	rs8176305	49,38		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5278-191A>T	rs8176297	52,89		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5152+66G>A	rs3092994	44,67		Out of pre-NGS method covered region	1.00
	BRCA1:NM_007294:c.5075-53C>T	rs8176258	45,16	Known		YES
	BRCA1:NM_007294:c.5075-238A>G	rs8176256	45,83		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4987-68A>G	rs8176234	51,65		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4987-92A>G	rs8176233	47,72		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4986+222A>G	rs3092987	73,68		Out of pre-NGS method covered region	

	BRCA1:NM 007294:c.4837A>G	rs1799966	48,05	Known		
	BRCA1:NM 007294:c.4485-63C>G	rs8176212	46,37	Known		YES
	BRCA1:NM 007294:c.4485-137T>A	rs2236762	45,12	1	Out of pre-NGS method covered region	1.20
	BRCA1:NM 007294:c.4358-2590T>G	rs8176194	52,1		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.4358-2885G>A	rs8176193	50,44		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.4308T>C	rs1060915	43,87	Known	i .	YES
	BRCA1:NM 007294:c.4097-141A>C	rs799916	54,68	KIIOWII	Out of pre-NGS method covered region	TEO
	BRCA1:NM 007294:c.3548A>G	rs16942	46,11	Known		YES
				Known		
	BRCA1:NM_007294:c.3113A>G	rs16941	47,65	Known	i	YES
	BRCA1:NM_007294:c.2612C>T	rs799917	52,96	Known		YES
	BRCA1:NM_007294:c.2311T>C	rs16940	49,12	Known	i	YES
	BRCA1:NM_007294:c.2082C>T	rs1799949	45,98	Known		YES
	BRCA1:NM_007294:c.2077G>A	rs4986850	45,48	Known		YES
	BRCA1:NM_007294:c.594-225G>A	rs8176147	44,74		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.593+167T>C	rs8176145	39,47	Known		YES
	BRCA1:NM_007294:c.547+146A>T	rs8176140	53,42		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.213-161A>G	rs799912	46,39		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.1-134T>C	rs3765640	35,29	Known		YES
	BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	40.58		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	49.24		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.441+52_441+63delCTTTTTTTTTT		52.52		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.441+36 441+38delCTT	rs147856441	44.87		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.134+224 134+224delT	rs35149296	60		Out of pre-NGS method covered region	
T18					out of profitor mounds covered region	
110	BRCA2:NM 000059:c.1-26G>A	rs1799943	45,98	Known		YES
	BRCA2:NM 000059:c.425+246G>C	rs11571613	50	Tallowin	Out of pre-NGS method covered region	1.23
	BRCA2:NM 000059:c.631+183T>A	rs3752451	34,21		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.793+98G>A	rs206073	99,41		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.1114A>C	rs144848	51,24	Known	i .	YES
	BRCA2:NM 000059:c.3396A>G	rs1801406	45,41	Known	i	YES
	BRCA2:NM 000059:c.4563A>G	rs206075	100	KIIOWII	Could not be detected by CSCE	TES
						+
	BRCA2:NM_000059:c.6513G>C	rs206076	100 52,69		Could not be detected by CSCE	
	BRCA2:NM_000059:c.6841+191C>A	rs11571662			Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.6937+239A>T	rs190434310	61,54		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.6938-120T>C	rs206080	98,45	1,	Out of pre-NGS method covered region	VEO
	BRCA2:NM_000059:c.7242A>G	rs1799955	47,78	Known		YES
	BRCA2:NM_000059:c.7397T>C	rs169547	100		Could not be detected by CSCE	
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7806-14T>C	rs9534262	49,88	Known	i	YES
	BRCA2:NM_000059:c.8754+183A>C	rs3764791	39,13		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.8754+187C>T	rs3764792	39,02		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.8755-66T>C	rs4942486	45,41		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9257-219A>G	rs7330025	70		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9257-16T>C	rs11571818	45,36	Known		YES
	BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9976A>T	rs11571833	49,17	Known		YES
	BRCA2:NM_000059:c.6275_6276delTT	rs11571658	43.6	PAT		YES
	BRCA2:NM 000059:c.6841+78 6841+81delAATT	rs138193280	47.75		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.8633-150 8633-149insA	rs200156944	44.83	Ì	Out of pre-NGS method covered region	
T19					, and a second second	
	MSH6:NM 000179:c.116G>A	rs1042821	52,11	Known		NO, variant in low covered regior
	MSH6:NM 000179:c.458-133A>G	rs3136316	50	Known		YES
	MSH6:NM 000179:c.458-52G>T	rs1800934	49,64	Known		YES
	MSH6:NM 000179:c.2633T>C	rs2020912	49,86	Known		YES
	MSH6:NM 000179:c.3148 3149insTGTAGAGTGTAT	192020312	33.72	PAT		YES
		•		PAI	Out of are NCC method	IES
T20	MSH6:NM_000179:c.4002-27_4002-27delT		36.45		Out of pre-NGS method covered region	
120	MSH6:NM 000179:c.186C>A	rs1042820	56,44	Known		NO, variant in low covered regior

N N	MSH6:NM 000179:c.260+22C>G	rs55927047	44,44	Known		NO, variant in low covered region
N N	MSH6:NM_000179:c.276A>G	rs1800932	49,34	Known		YES
N N	MSH6:NM 000179:c.457+52T>A	rs3136282	52,34	Known		YES
	MSH6:NM 000179:c.458-133A>G	rs3136316	56,31	Known		YES
	MSH6:NM 000179:c.540T>C	rs1800935	44,8		Region not anlyzed by pre-NGS method	
	MSH6:NM_000179:c.3173-101G>C	rs2072447	55,2		Out of pre-NGS method covered region	
N	MSH6:NM 000179:c.3438+14A>T	rs2020911	53,03		Region not anlyzed by pre-NGS method	
	MSH6:NM 000179:c.3556+146G>A	rs7562048	56,45	Known	- J	YES
	MSH6:NM 000179:c.3646+91T>C	rs3136359	46,43		Out of pre-NGS method covered region	
	MSH6:NM 000179:c.3802-40C>G	rs3136367	47,03		Out of pre-NGS method covered region	
	MSH6:NM 000179:c.3252 3253insC		41.13	PAT		NO, variant in homopolymer region
	MSH6:NM 000179:c.3557-18 3557-17insT		43.88		Region not anlyzed by pre-NGS method	, , , , ,
	MSH6:NM 000179:c.3646+29 3646+32delCTAT	rs2234731	33.04	Known		YES
	MSH6:NM 000179:c.4002-27 4002-27delT		35.14		Region not anlyzed by pre-NGS method	1
T21						
	MLH1:NM 000249:c.453+79A>G	rs4234259	99,12		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.655A>G	rs1799977	99,59		Region not anlyzed by pre-NGS method	
	MLH1:NM 000249:c.791-79A>G	rs104894998	51,84		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1038+86T>C	rs2286939	100		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1039-78A>G	rs11129748	92,31		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1410-169C>T	rs2286940	100		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1668-19A>G	rs9876116	100		Region not anlyzed by pre-NGS method	
	MLH1:NM 000249:c.1990-121C>T	rs2241031	100		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.208-150 208-150delA	rs111908792	21.62		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.678-212 678-211insGGGA	rs138898092	70		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1039-28 1039-26delTTT	13130030032	37.5		Region not anlyzed by pre-NGS method	
	MLH1:NM_000249:c.1588_1589insCGTGGGCTG	<u> </u>	39.49	PAT	Tregion flot unity26d by pre 1466 metriod	YES
	MLH1:NM 000249:c.1897-167 1897-167delT	rs11290150	100	171	Out of pre-NGS method covered region	120
T22	WEITT.INM_000249.C.1097-107_1097-107 deit	1811290130	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.61-300C>T	rs2269856	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.61-123G>A	rs2269855	99,36		Out of pre-NGS method covered region	
	NF1:NM_000267:c.288+41G>A	rs2952976	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.480-90C>T	rs2905807	100		Out of pre-NGS method covered region	
	NF1:NM 000267:c.702G>A	rs1801052	98,56	Known	Out of pre-1400 method covered region	YES
	NF1:NM 000267:c.888+108C>T	rs2953000	100	TCHOWIT	Out of pre-NGS method covered region	120
	NF1:NM 000267:c.888+118G>T	rs2952999	100		Out of pre-NGS method covered region Out of pre-NGS method covered region	
	NF1:NM 000267:c.393-130A>T	rs2905875	100		Out of pre-NGS method covered region Out of pre-NGS method covered region	
	NF1:NM_000267:c.1393-130A>1	rs2905876	99,83			
	NF1:NM 000267:c.1393-321>C	rs2905880	100		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1041+391>C	rs17881285	43,74			
	NF1:NM_000267:c.5205+23T>C	rs9894648	100		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
N	NET INIVERSIGNATOR C 3203T2312C		1 100		,,,,	
N					Cauld not be detected by DNA becad analysis	
N N	NF1:NM_000267:c.5546+19T>A	rs2285894	99,19		Could not be detected by RNA-based analysis	
N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A	rs2285894 rs3815154	99,19 100		Out of pre-NGS method covered region	
N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A	rs2285894 rs3815154 rs7406038	99,19 100 88,82		Out of pre-NGS method covered region Could not be detected by RNA-based analysis	
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A	rs2285894 rs3815154	99,19 100 88,82 97,6	-	Out of pre-NGS method covered region	VEO
N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG	rs2285894 rs3815154 rs7406038 rs7406039	99,19 100 88,82 97,6 52,4	PAT	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	YES
N N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226deIG NF1:NM_000267:c.7126+37C>G	rs2285894 rs3815154 rs7406038 rs7406039	99,19 100 88,82 97,6 52,4 100		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	YES
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569	99,19 100 88,82 97,6 52,4 100 100		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region	YES
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288	99,19 100 88,82 97,6 52,4 100 100		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis	YES
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943	99,19 100 88,82 97,6 52,4 100 100 100 100		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Out of pre-NGS method covered region	YES
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288	99,19 100 88,82 97,6 52,4 100 100		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis	YES
N	NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.6226delG NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.7127-167T>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7907+166C>T NF1:NM 000267:c.8050+20G>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230	99,19 100 88,82 97,6 52,4 100 100 100 49,26		Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Out of pre-NGS method covered region	
N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T NF1:NM_000267:c.8050+20G>A BRCA2:NM_000059:c.1-26G>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230 rs1799943	99,19 100 88,82 97,6 52,4 100 100 100 49,26	Known	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Out of pre-NGS method covered region	YES
N N N N N N N N N N N N N N N N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T NF1:NM_000267:c.8050+20G>A BRCA2:NM_000059:c.1-26G>A BRCA2:NM_000059:c.425+67A>C	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230 rs1799943 rs11571610	99,19 100 88,82 97,6 52,4 100 100 100 49,26 45,37 51,53	Known	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Out of pre-NGS method covered region	YES YES
N N N N N N N N N N N N N N N N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T NF1:NM_000267:c.8050+20G>A BRCA2:NM_000059:c.1-26G>A BRCA2:NM_000059:c.425+67A>C BRCA2:NM_000059:c.426-89T>C	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230 rs1799943 rs11571610 rs3783265	99,19 100 88,82 97,6 52,4 100 100 100 49,26 45,37 51,53 52,3	Known	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis	YES
N N N N N N N N N N N N N N N N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T NF1:NM_000267:c.8050+20G>A NF1:NM_000267:c.8050+20G>A NF1:NM_000267:c.8050+20G>A NF1:NM_000059:c.425+67A>C NF1:NM_000059:c.426-89T>C NF1:NM_000059:c.426-89T>C NF1:NM_000059:c.631+183T>A	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230 rs1799943 rs11571610 rs3783265 rs3752451	99,19 100 88,82 97,6 52,4 100 100 100 100 49,26 45,37 51,53 52,3 100	Known	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region	YES YES
N N N N N N N N N N N N N N N N N N N	NF1:NM_000267:c.5546+19T>A NF1:NM_000267:c.5546+117G>A NF1:NM_000267:c.6085-29T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6085-28T>A NF1:NM_000267:c.6226delG NF1:NM_000267:c.7126+37C>G NF1:NM_000267:c.7127-167T>A NF1:NM_000267:c.7395-29G>A NF1:NM_000267:c.7907+166C>T NF1:NM_000267:c.8050+20G>A BRCA2:NM_000059:c.1-26G>A BRCA2:NM_000059:c.425+67A>C BRCA2:NM_000059:c.426-89T>C	rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs2525569 rs964288 rs7350943 rs55747230 rs1799943 rs11571610 rs3783265	99,19 100 88,82 97,6 52,4 100 100 100 49,26 45,37 51,53 52,3	Known	Out of pre-NGS method covered region Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis Out of pre-NGS method covered region Could not be detected by RNA-based analysis	YES YES

	BRCA2:NM_000059:c.1365A>G	rs1801439	43,9	Known		YES
	BRCA2:NM_000059:c.1910-51G>T	rs11571651	48,68		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.2229T>C	rs1801499	51,14	Known		YES
	BRCA2:NM_000059:c.2971A>G	rs1799944	50,49	Known		YES
	BRCA2:NM_000059:c.3396A>G	rs1801406	50,24	Known		YES
1	BRCA2:NM_000059:c.4563A>G	rs206075	99,77		Could not be detected by CSCE	
1	BRCA2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE	
1	BRCA2:NM_000059:c.6841+191C>A	rs11571662	45,3		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.6938-120T>C	rs206080	99,15		Out of pre-NGS method covered region	
1	BRCA2:NM 000059:c.7397T>C	rs169547	100		Could not be detected by CSCE	
1	BRCA2:NM 000059:c.7435+53C>T	rs11147489	48,33	Known		YES
1	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
1	BRCA2:NM 000059:c.7806-14T>C	rs9534262	49,39	Known		YES
1	BRCA2:NM 000059:c.8755-66T>C	rs4942486	46,5		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.8953+98T>C	rs81002901	51,75		Out of pre-NGS method covered region	
-	BRCA2:NM 000059:c.9257-219A>G	rs7330025	60		Out of pre-NGS method covered region	
-	BRCA2:NM_000059:c.6841+78_6841+81delAATT	rs138193280	41.81		Out of pre-NGS method covered region	
	BRCA1:NM_007300:c.5050-142_5050-142delA		24		Out of pre-NGS method covered region	
	BRCA1:NM 007300:c.1953 1956delGAAA	rs80357526	46.81	PAT	out or pro recommended covered region	YES
IDATION			.0.01			
V1						
	MUTYH:NM 012222:c.1477-40C>G	rs3219493	100	Known		
	MUTYH:NM 012222:c.1477-125C>A	rs3219492	37,74	Tallowin	Out of pre-NGS method covered region	
	MUTYH:NM 012222:c.1187G>A	rs36053993	51,76	PAT	Cut of pre 1400 metrod covered region	
	MUTYH:NM 012222:c.1014G>C	rs3219489	57,85	Known		
	MUTYH:NM 012222:c.504+35A>G	rs3219487	100	KIIOWII	Out of pre-NGS method covered region	
	MUTYH:NM_012222:c.304+33A>G	183219467	51,04	VUS	Out of pre-NGS method covered region	
	APC:NM 001127511:c.1-133C>G	rs79896135		VU3	Out of pre-NGS method covered region	
			43,64			
	APC:NM_001127511:c.1-31T>G	rs78429131	50		Out of pre-NGS method covered region	
	APC:NM_001127510:c.645+32C>T	rs2909961	55,04		Out of pre-NGS method covered region	
	APC:NM_001127510:c.645+173A>G	rs2289484	50		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-132G>T	rs12521276	100	14	Out of pre-NGS method covered region	
	APC:NM_001127510:c.1458T>C	rs2229992	99,51	Known		
	APC:NM_001127510:c.1635G>A	rs351771	50,97	Known	0.4.6. 1100 # 1	
	APC:NM_001127510:c.1743+193G>A	rs351772	91,67		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1744-158A>G	rs74915204	47,06		Out of pre-NGS method covered region	
	APC:NM_001127510:c.4479G>A	rs41115	48,48	Known		
	APC:NM_001127510:c.5034G>A	rs42427	57,46	Known		
	APC:NM_001127510:c.5268T>G	rs866006	43,49	Known		
	APC:NM_001127510:c.5465T>A	rs459552	99,61	Known		
	APC:NM_001127510:c.5880G>A	rs465899	49,35	Known		
	APC:NM_001127510:c.7201C>T	rs2229994	49,49	Known		
	APC:NM_001127510:c.934-132_934-132delG	rs141013063	45.9		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-120_934-119insG	rs138003874	24.71		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1959-145_1959-144insAGAA	rs3839284	100		Out of pre-NGS method covered region	
V2						
	APC:NM_001127510:c.645+173A>G	rs2289484	65,52	Known		
	APC:NM 001127510:c.934-132G>T	rs12521276	28,57		Out of pre-NGS method covered region	
	_			Known		
	APC:NM_001127510:c.1458T>C	rs2229992	46,25		!	
	APC:NM_001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C	rs2229992	47,74	PAT		
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A	rs2229992 rs351771		PAT Known		
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A APC:NM 001127510:c.1743+193G>A	rs2229992	47,74	PAT Known	Out of I2CHP ROI	
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A	rs2229992 rs351771	47,74 52,91	PAT Known	Out of I2CHP ROI	
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A APC:NM 001127510:c.1743+193G>A	rs2229992 rs351771	47,74 52,91 Not detected	PAT Known Known		
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A APC:NM 001127510:c.1743+193G>A APC:NM 001127510:c.1744-37A>G	rs2229992 rs351771 rs351772	47,74 52,91 Not detected 51,29	PAT Known Known Known		
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A APC:NM_001127510:c.1743+193G>A APC:NM_001127510:c.1744-37A>G APC:NM_001127510:c.4479G>A	rs2229992 rs351771 rs351772 rs41115	47,74 52,91 Not detected 51,29 51,63	PAT Known Known Known Known		
	APC:NM 001127510:c.1458T>C APC:NM_001127510:c.1548+1G>C APC:NM_001127510:c.1635G>A APC:NM 001127510:c.1743+193G>A APC:NM 001127510:c.1744-37A>G APC:NM_001127510:c.4479G>A APC:NM_001127510:c.5034G>A	rs2229992 rs351771 rs351772 rs41115 rs42427	47,74 52,91 Not detected 51,29 51,63 49,54	PAT Known Known Known Known Known		

	APC:NM_001127510:c.934-120_934-119insG	rs138003874	28		Out of pre-NGS method covered region		
	APC:NM_001127510:c.1959-145_1959-144insAGAA	rs3839284	48.24		Out of pre-NGS method covered region		
V3							
	MSH2:NM_000251:c.1-118T>C	rs2303425	58,99		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.211+9C>G	rs2303426	98,51	Known			
	MSH2:NM_000251:c.211+98T>C	rs3815865	92,86		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.367-168C>T	rs17217758	60,38		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.518T>G	rs63750070	44,49	VUS			
	MSH2:NM 000251:c.646-224A>G	rs17217786	54,55		Out of pre-NGS method covered region		
	MSH2:NM 000251:c.1077-80G>A	rs2347794	100		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.1077-10T>C	rs17224360	54,4	Known			
	MSH2:NM 000251:c.1277-212T>A	rs1981928	100		Out of pre-NGS method covered region		
	MSH2:NM 000251:c.1277-118G>A	rs1981929	100		Out of pre-NGS method covered region		
	MSH2:NM 000251:c.1510+111T>C	rs3771278	99,23		Out of pre-NGS method covered region		
	MSH2:NM 000251:c.1510+115A>G	rs3771279	100		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.1510+118T>C	rs3771280	100		Out of pre-NGS method covered region		
	MSH2:NM 000251:c.1511-91G>T	rs3732182	100	Known	eat of profited method severed region		
	MSH2:NM 000251:c.1511-91621	rs12998837	57,84	Known			
	MSH2:NM 000251:c.1661+12G>A	rs3732183	100	Known		+	
	MSH2:NM 000251:c.1759+107A>G	rs3764959	100	Known		+	
	MSH2:NM 000251:c.1759+107A2G MSH2:NM 000251:c.1759+183G>A	rs3764960	98,31	Known		+	
	MSH2:NM 000251:c.2006-127C>T	rs2059521	100	_	Out of pre-NGS method covered region	+	
	MSH2:NM 000251:c.2006-127C>1 MSH2:NM 000251:c.2006-6T>C	rs2303428	48,79	Known	Tout of pre-1400 method covered region	+	
	MSH2:NM 000251:c.2210+175G>A		100	KIIOWII	Out of pro NCS method sovered region		
		rs4583514		+	Out of pre-NGS method covered region		
	MSH2:NM_000251:c.2635-214T>C	rs2042649	59,46		Out of pre-NGS method covered region		
V/4	MSH2:NM_000251:c.792+148_792+148delT		27.78		Out of pre-NGS method covered region		
V4	BRCA2:NM 000059:c.1-26G>A	4700040	40.0	1/			
		rs1799943	43,6	Known	0.1-1		
	BRCA2:NM_000059:c.631+183T>A	rs3752451	85,19		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,05	17	Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1114A>C	rs144848	51,94	Known			
	BRCA2:NM_000059:c.2803G>A	rs28897716	40,53	Known			
	BRCA2:NM_000059:c.3396A>G	rs1801406	49,7	Known			
	BRCA2:NM_000059:c.4563A>G	rs206075	99,73		Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.5351A>T		47,55	VUS	Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.6513G>C	rs206076	99,81		Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.6841+191C>A	rs11571662	51,2		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7008-62A>G	rs76584943	49,33	Known			
	BRCA2:NM_000059:c.7242A>G	rs1799955	47,43	Known			
	BRCA2:NM_000059:c.7397T>C	rs169547	99,62	1	Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7806-14T>C	rs9534262	47,76	Known			
	BRCA2:NM_000059:c.8754+183A>C	rs3764791	40		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.8754+187C>T	rs3764792	43,33		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.8755-66T>C	rs4942486	52,36		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.5345_5345delA		44.09	PAT			
	BRCA2:NM_000059:c.6841+78_6841+81delAATT	rs138193280	36.03		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	55		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4485-137T>A	rs2236762	49,44		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4097-141A>C	rs799916	50,73		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.2612C>T	rs799917	47,32		Out of pre-NGS method covered region		
-	BRCA1:NM_007294:c.213-161A>G	rs799912	59,09		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.441+64 441+64delT	rs368252296	53,33		Out of pre-NGS method covered region		
V5							
	MLH1:NM_000249:c.453+79A>G	rs4234259	100		Out of pre-NGS method covered region		
	MLH1:NM_000249:c.655A>G	rs1799977	55,38		Region not anlyzed by pre-NGS method		
	MLH1:NM_000249:c.1038+86T>C	rs2286939	100		Out of pre-NGS method covered region		
	<u></u> -:			_ i			1

MELITAM DIOCARD - 140 1695-7								
MILHTAM DOZAGE 1906-121C-T M7261031 97 677 Out of par MSS method covered region		MLH1:NM_000249:c.1410-169C>T	rs2286940	100		Out of pre-NGS method covered region		
MULHYMM, 000240c.1987-167-1687-1670eff 151200160 100 NUL of pre-NLSS method covered region MULHYMM, 000240c.2167, 247-2489 AB.77		MLH1:NM_000249:c.1668-19A>G	rs9876116	100		Region not anlyzed by pre-NGS method		
MILHTMM 000494e-2148 2149mAACA		MLH1:NM_000249:c.1990-121C>T	rs2241031	97,67		Out of pre-NGS method covered region		
PMS2-NM D005SS 2-257GS-C		MLH1:NM_000249:c.1897-167_1897-167delT	rs11290150	100		Out of pre-NGS method covered region		
PMS2-NM_000556_2570G+C		MLH1:NM 000249:c.2148 2149insAACA		48.71	PAT	-		
PMSS NM, 000055c, 267-1678-TC 98055490 41,42 Out of pre-NOS method covered region	V6							
PMSS NM, 000055c, 267-1678-TC 98055490 41,42 Out of pre-NOS method covered region		PMS2:NM 000535:c.2570G>C	rs1802683	42.07		Region not anlyzed by pre-NGS method		
MMS2 NM_000556_2074-SA_ 198568490			0					
PMSEX.NN 0005362 c 1001-AG		_	rs9655490					
PMSS_NM_0005SS_c160PGT n1895S1								
PMS2:NM 000536: 1495C-T s1905211 45.04 Region not anhized by pre-NSS method PMS2:NM 000536: 493C-T ra20980995 20.62 PMS2:NM 000536: 493C-T ra20980995 36.43 PAT Region not anhized by pre-NSS method PMS2:NM 000536: 20.61						Region not anlyzed by pre-NGS method		
PMS2:MM 000535:c.1145-5FC6A rs1995508965 20.62 PAT rs2096685 36.43 PAT rs2096685 36.43 PAT rs2096681 22.41 Region not anityzed by pre-NGS method rs1805319 99.66 Region not anityzed by pre-NGS method rs1805319 rs209681 23.41 Region not anityzed by pre-NGS method rs1805319 rs209681 23.43 Region not anityzed by pre-NGS method rs1805319 rs209681 23.43 Region not anityzed by pre-NGS method rs1805319 rs209681 rs20								
PMSZ.NM 000535: 094-014TP-0 sig2808081 22.41 Region not aniyzed by pre-NGS method								
PMSS_NM (000535: 204-1647-3C)					ΡΔΤ	eat of profited motified covered region		-
PMSZ-NM 000635c, 780C-PG rs 1806319 99,66 Region not anivated by pre-NGS method		_				Pegion not anlyzed by are NGS method		
PMS2NM 000635c 251-72A-C rsi2456162 43,37 Region not anived by pre-NGS method								
PMS2PM, 000536.291-72A-G rs17831773		_						+
PMS2-NM 000535: 904-88 904-86delCTG					-		+	+
PMS2 NM 000355 x; 705+118 r05+118 detT								+
V7 APCNM_001127511:c.1:133CPG								+
APC.MM .0011275110.c.133C-PG 678966135 38.78 Out of pre-NGS method APC.MM .0011275110.c.645+32C-T 62009961 48.44 Region not anized by pre-NGS method APC.MM .001127510.c.645+32C-T 62009961 48.44 Region not anized by pre-NGS method APC.MM .001127510.c.645+32C-T 612289484 60.71 Known APC.MM .001127510.c.645+32C-T 612221276 100 Known APC.MM .001127510.c.645+32C-T 612221276 100 Known APC.MM .001127510.c.645+32C-T 612222992 99.97 Known APC.MM .001127510.c.1435-3A-G 62229992 99.97 Known APC.MM .001127510.c.1435-13G-A 6351771 49.99 Known APC.MM .001127510.c.1435-13G-A 6351772 Not detected Known APC.MM .001127510.c.1474-158A-C 674915204 50 Out of pre-NGS method covered region APC.MM .001127510.c.1474-158A-C 674915204 50 Out of pre-NGS method APC.MM .001127510.c.4746-A 6411115 48.71 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456952 100 APC.MM .001127510.c.5465F-A 6456952 100 APC.MM .001127510.c.5465F-A 6456959 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456959 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456999 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456999 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456999 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456999 53.66 Region not anivzed by pre-NGS method APC.MM .001127510.c.5465F-A 6456999 641040	\/7		1511209444	03.33		Out of pre-1405 method covered region		
APC-NM 0011275110c.3512CF	V7	ADC:NM 004427544:a 4 422C> C	ro70000435	20.70		Out of pro NCS method sovered region		4
APC-NM 001127510:c.646+132A-5								+
APC-IMM 001127510c.934-132QFT rs12521275 100 Known					-			
APC.NM_001127510c.3434-32G-T Is12521276 50.0 Known APC.NM_001127510c.14581>C Is2229992 98.97 Known APC.NM_001127510c.14581>C Is2229992 98.97 Known APC.NM_001127510c.1743-1930>A Is351771 49.49 Known APC.NM_001127510c.1743-1930>A Is351772 Not detected Known APC.NM_001127510c.1743-1930>A Is351772 Not detected Known APC.NM_001127510c.1744-1580>G Is74915204 S0 Out of I2CHP RGI Out of I2CHP RGI APC.NM_001127510c.24790>A Is341115 45.71 Region not analyzed by pre-NGS method APC.NM_001127510c.52881>G Is341115 45.71 Region not analyzed by pre-NGS method APC.NM_001127510c.52881>G Is341115 A9.71 Region not analyzed by pre-NGS method APC.NM_001127510c.52881>G Is341000000000000000000000000000000000000					17	Region not aniyzed by pre-NGS method		+
APC:NM 001127510:c.183E3T-C								
APC.NM 001127510c.1458T>C			rs12521276					+
APC.NM 001127510c.1635G>A		_						
APC.NM 001127510: 1744-188A>G 8351772 Not detected Known Out of IZCHP ROI							_	
APC:NM 001127510c.1744-158A-G rs74915204 50 Cut of pre-NGS method covered region APC:NM 001127510c.52687-A rs41115 48,71 Region not anlyzed by pre-NGS method APC:NM 001127510c.52681-G rs866006 48,41 Region not anlyzed by pre-NGS method APC:NM 001127510c.52681-G rs866006 48,41 Region not anlyzed by pre-NGS method APC:NM 001127510c.52681-G rs866006 48,41 Region not anlyzed by pre-NGS method APC:NM 001127510c.5880G-A rs459552 100 Region not anlyzed by pre-NGS method APC:NM 001127510c.5880G-A rs465899 53,66 Region not anlyzed by pre-NGS method APC:NM 001127510c.534-132 934-132delG rs141013063 68.42 Out of pre-NGS method APC:NM 001127510c.934-132 934-132delG rs141013063 68.42 Out of pre-NGS method APC:NM 001127510c.934-120 934-119insG rs138003874 28.57 Out of pre-NGS method covered region APC:NM 001127510c.934-120 934-119insG rs138003874 28.57 Out of pre-NGS method covered region APC:NM 001027510c.934-120 934-119insG rs138003874 97.87 Out of pre-NGS method covered region APC:NM 000059c.5934-869-A rs206073 99.18 Out of pre-NGS method covered region APC:NM 000059c.5934-869-A rs206073 99.18 Out of pre-NGS method covered region APC:NM 000059c.5934-9G rs206075 99.75 Known BRCA2:NM 000059c.5633A-G rs206075 99.75 Known BRCA2:NM 000059c.5633G-C rs206076 100 Known BRCA2:NM 000059c.56337-C rs206076 rs206006 100 Known BRCA2:NM 000059c.57377-C rs206076 rs206076 100 Known BRCA2:NM 000059c.57377-C rs206076 rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059c.57377-C rs206076 rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059c.57357-BC rs206076 rs206096 100 Out of pre-NGS method covered region BRCA2:NM				-, -		0 / (100) ID DOI	_	
APC:NM 001127510:c.3034G>A rs41115		_						
APC:NM 001127510:c.5034G>A				_				
APC:NM 001127510c.5288T>G		_				, ,,		
APC:NM 001127510:c.54651>A								
APC:NM 001127510:c.5880G-A								
APC:NM 001127510:c.7201C>T						, ,,		
APC:NM_001127510:c.934-132_934-132delG rs141013063 68.42 Out of pre-NGS method covered region APC:NM_001127510:c.934-120_934-119insG rs138003874 28.57 Out of pre-NGS method covered region APC:NM_001127510:c.1959-145_1959-144insAGAA rs389284 97.87 Out of pre-NGS method covered region APC:NM_000059:c.1959-145_1959-144insAGAA rs389284 97.87 Out of pre-NGS method covered region APC:NM_000059:c.1934-96S-A rs206073 99.18 Out of pre-NGS method covered region APC:NM_000059:c.1114A>C rs144848 99.85 Known APC:NM_00059:c.1114A>C rs144848 99.85 Known APC:NM_00059:c.1599C>T rs206075 99.75 Known APC:NM_00059:c.5199C>T rs206075 99.75 Known APC:NM_00059:c.5199C>T rs206076 100 Known APC:NM_00059:c.6938-120T>C rs206076 100 Known APC:NM_00059:c.6938-120T>C rs206076 100 Known APC:NM_00059:c.6938-120T>C rs206080 98.48 Out of pre-NGS method covered region APC:NM_00059:c.6938-120T>C rs206080 98.48 Out of pre-NGS method covered region APC:NM_00059:c.7917-YC rs1069547 100 Known APC:NM_00059:c.7917-YC rs1069547 100 APC:NM_00059:c.7917-YC APC:NM_00059:c.7917-YC Rs1069								
APC:NM 001127510:c.934-120 934-119insG rs138003874 28.57 Out of pre-NGS method covered region APC:NM 001127510:c.1959-145 1959-144insAGAA rs3839284 97.87 Out of pre-NGS method covered region BRCA2:NM 000059:c.793+98G>A rs206073 99.18 Out of pre-NGS method covered region BRCA2:NM 000059:c.4563A>G rs206075 99.75 Known BRCA2:NM 000059:c.4563A>G rs206075 99.75 Known BRCA2:NM 000059:c.5199C>T rs28897734 48.22 Known BRCA2:NM 000059:c.6513G>C rs206076 100 Known BRCA2:NM 000059:c.6814191C>A rs11571662 34.78 Out of pre-NGS method covered region BRCA2:NM 000059:c.6831-20T>C rs206080 98.48 Out of pre-NGS method covered region BRCA2:NM 000059:c.7242A>G rs159647 100 Known BRCA2:NM 000059:c.7397T>C rs169547 100 Known BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.7596-14T>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+183A>C rs206096 100 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187684+181delAATT rs138193280 38,77 Out of pre-NGS method covered region Out of pre-NGS method covered region BRCA2:NM 0011								
APC:NM_001127510:c.1959-145_1959-144insAGAA rs3839284 97.87 Out of pre-NGS method covered region								
BRCA2:NM 000059:c.793+98G>A rs206073 99,18 Out of pre-NGS method covered region								
BRCA2:NM 000059:c.114A>C		APC:NM_001127510:c.1959-145_1959-144insAGAA	rs3839284	97.87		Out of pre-NGS method covered region		
BRCA2:NM 000059:c.1114A>C rs144848 99,85 Known BRCA2:NM 000059:c.4563A>G rs206075 99,75 Known BRCA2:NM 000059:c.5199C>T rs28897734 48,22 Known BRCA2:NM 000059:c.6513G>C rs206076 100 Known BRCA2:NM 000059:c.6841+191C>A rs11571662 34,78 Out of pre-NGS method covered region BRCA2:NM 000059:c.6938-120T>C rs206080 98,48 Out of pre-NGS method covered region BRCA2:NM 000059:c.7242A>G rs1799955 50 Known BRCA2:NM 000059:c.7397T>C rs169547 100 Known BRCA2:NM 000059:c.7397T>C rs169547 100 Known BRCA2:NM 000059:c.7606-14T>C rs9534262 54,05 Known BRCA2:NM 000059:c.7806-14T>C rs9534262 54,05 Known BRCA2:NM 000059:c.8754+183A>C rs3764791 50 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754+187C>T rs3764792 45,16 Out of pre-NGS method covered region BRCA2:NM 000059:c.8755-66T>C rs4942486 43,66 Out of pre-NGS method covered region BRCA2:NM 000059:c.8755-66T>C rs4942486 43,66 Out of pre-NGS method covered region BRCA2:NM 000059:c.8755-66T>C rs4942486 43,66 Out of pre-NGS method covered region BRCA2:NM 000059:c.8754-86841+81delAATT rs138193280 38,77 Out of pre-NGS method covered region TP53:NM 001126112:c.1101-221G>A rs6503048 39,58 Out of pre-NGS method covered region	V8							
BRCA2:NM 000059:c.4563A>G					1	Out of pre-NGS method covered region		
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TP53:NM_001126112:c.1101-221G>A rs6503048 39,58 Out of pre-NGS method covered region								
TP53:NM 001126112:c.743G>A rs11540652 43.57 PAT						Out of pre-NGS method covered region		
Page 12		TP53:NM_001126112:c.743G>A	rs11540652	43,57	PAT			

TPS3.NN 001128112.678-68A-G	
TPS3 NM_001126112c.276-91G>A rs2909430 57.98 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 56.63 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 56.65 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 56.65 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 56.65 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 56.65 Out of pre-NGS method covered region TPS3 NM_001126112c.37-52G>A 67.57-52D Out of pre-NGS method covered region Out of	
TP63.NM_001126112c.975-GD-A	
TP53.NM 001126112c.97-52G>A 56.65 Out of pre-NGS method covered region	
TP53:NM 001126112c.6972+194 672+19446IC	
TP53:NM 001126112c.96+41 97-54delACCTGGAGGCTGGGG rs146534833 44.64 Out of pre-NGS method covered region	
MSH6:NM 000179:c.276A>G	
MSH6:NM 000179:c.276A>G rs1800932 48.68 Known MSH6:NM 000179:c.457+52T>A rs3136282 52.54 Out of pre-NGS method covered region MSH6:NM 000179:c.458-133A>G rs3136282 52.54 Out of pre-NGS method covered region MSH6:NM 000179:c.358-133A>G rs1800935 46.62 Known MSH6:NM 000179:c.3517-101G>C rs20072447 72.5 Out of pre-NGS method covered region MSH6:NM 000179:c.3438+14A>T rs2020911 61.97 Out of pre-NGS method covered region MSH6:NM 00179:c.3565+146G>A rs7562048 35.20 Out of pre-NGS method covered region MSH6:NM 00179:c.3565+146G>A rs7562048 35.20 Out of pre-NGS method covered region MSH6:NM 000179:c.3566+146G>A rs7562048 35.20 Out of pre-NGS method covered region MSH6:NM 000179:c.3646+91T>C rs3136359 56.25 Known MSH6:NM 000179:c.3646+91T>C rs3136357 44.67 Known MSH6:NM 000179:c.3646+29 3464+32delCTAT rs2234731 42.86 Out of pre-NGS method covered region MSH6:NM 000179:c.3646+29 3464+32delCTAT rs2234731 42.86 Out of pre-NGS method covered region MSH6:NM 000179:c.3646+29 3464+32delCTAT rs2234731 42.86 Out of pre-NGS method covered region MSH6:NM 000079:c.4002-27 4002-	
MSH6:NM 000179:c.457+52T>A	
MSH6:NM 000179:c.458-133A>G	
MSH6:NM 000179:c.3407>C	
MSH6:NM 000179:c.3173-101G>C rs2072447 72,5 Out of pre-NGS method covered region MSH6:NM 000179:c.3438+14A>T rs2020911 61,97 Out of pre-NGS method covered region MSH6:NM 000179:c.3556+146G>A rs7562048 35,29 Out of pre-NGS method covered region MSH6:NM 000179:c.38046+91T>C rs3136359 56,25 Out of pre-NGS method covered region MSH6:NM 000179:c.3802-40C>G rs3136359 56,25 Out of pre-NGS method covered region MSH6:NM 000179:c.3802-40C>G rs3136367 44,67 Known MSH6:NM 000179:c.38046+92 3646+32delCTAT rs2234731 42,86 Out of pre-NGS method covered region MSH6:NM 000179:c.3646+29_3646+32delCTAT rs2234731 42,86 Out of pre-NGS method covered region MSH6:NM 000179:c.4002-27 4002-27delT	
MSH6:NM 000179:c.3438+14A>T	
MSH6:NM 000179:c.3556+146G>A	
MSH6:NM 000179:c.3646+91T>C	
MSH6:NM 000179:c.3802-40C>G	
MSH6:NM_000179:c.1614_1616delTCT	
MSH6:NM_000179:c.3646+29_3646+32delCTAT rs2234731 42.86 Out of pre-NGS method covered region	
MSH6:NM 000179:c.4002-27 4002-27delT	
No. No.	
BRCA2:NM_000059:c.793+98G>A rs206073 99,7 Out of pre-NGS method covered region	
BRCA2:NM 000059:c.1114A>C	
BRCA2:NM 000059:c.3807T>C rs543304 47,72 Known	
BRCA2:NM 000059:c.4563A>G rs206075 100 Could not be detected by CSCE	
BRCA2:NM 000059:c.6513G>C rs206076 99,6 Could not be detected by CSCE	
BRCA2:NM_000059:c.6938-120T>C rs206080 99,17 Out of pre-NGS method covered region	
BRCA2:NM_000059:c.6938-120T>C rs206080 99,17 Out of pre-NGS method covered region BRCA2:NM_000059:c.7397T>C rs169547 99,77 Could not be detected by CSCE BRCA2:NM_000059:c.7617+190G>A rs206096 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.9501+178A>G rs206343 66,67 Out of pre-NGS method covered region BRCA2:NM_000059:c.9648+244T>C rs206344 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.10257+105A>C rs15869 41,79 Out of pre-NGS method covered region BRCA1:NM_007294:c.5407-193A>G rs8176310 50 Out of pre-NGS method covered region BRCA1:NM_007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA2:NM_000059:c.7397T>C rs169547 99,77 Could not be detected by CSCE BRCA2:NM_000059:c.7617+190G>A rs206096 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.9501+178A>G rs206343 66,67 Out of pre-NGS method covered region BRCA2:NM_000059:c.9648+244T>C rs206344 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.10257+105A>C rs15869 41,79 Out of pre-NGS method covered region BRCA1:NM_007294:c.5407-193A>G rs8176310 50 Out of pre-NGS method covered region BRCA1:NM_007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA2:NM_000059:c.7617+190G>A rs206096 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.9501+178A>G rs206343 66,67 Out of pre-NGS method covered region BRCA2:NM_000059:c.9648+244T>C rs206344 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.10257+105A>C rs15869 41,79 Out of pre-NGS method covered region BRCA1:NM_007294:c.5407-193A>G rs8176310 50 Out of pre-NGS method covered region BRCA1:NM_007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA2:NM_000059:c.9501+178A>G rs206343 66,67 Out of pre-NGS method covered region BRCA2:NM_000059:c.9648+244T>C rs206344 100 Out of pre-NGS method covered region BRCA2:NM_000059:c.10257+105A>C rs15869 41,79 Out of pre-NGS method covered region BRCA1:NM_007294:c.5407-193A>G rs8176310 50 Out of pre-NGS method covered region BRCA1:NM_007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA2:NM 000059:c.9648+244T>C rs206344 100 Out of pre-NGS method covered region	
BRCA2:NM_000059:c.10257+105A>C rs15869 41,79 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.5407-193A>G rs8176310 50 Out of pre-NGS method covered region BRCA1:NM 007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.5278-191A>T rs8176297 45,65 Out of pre-NGS method covered region BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.5152+66G>A rs3092994 58,7 Out of pre-NGS method covered region	
	-
BRCA1:NM 007294:c.5074+65G>A rs8176235 55,49 Known	
BRCA1:NM 007294:c.4987-68A>G rs8176234 54,55 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.4987-92A>G rs8176233 48,15 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.4986+222A>G rs3092987 81,82 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.4837A>G	
BRCA1:NM_007294:c.4358-2590T>G rs8176194 51,53 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.4358-2885G>A rs8176193 48,27 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.4308T>C rs1060915 52,13 Known	
BRCA1:NM_007294:c.4097-141A>C rs799916 51,88 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.3548A>G rs16942 49,79 Known	
BRCA1:NM_007294:c.3113A>G rs16941 50,39 Known	
BRCA1:NM_007294:c.2612C>T rs799917 49,51 Known	
BRCA1:NM_007294:c.2311T>C rs16940 47,28 Known	
BRCA1:NM 007294:c.2082C>T rs1799949 52,33 Known	
BRCA1:NM_007294:c.594-225G>A rs8176147 43,75 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.593+167T>C rs8176145 50,94 Known	
BRCA1:NM_007294:c.547+146A>T rs8176140 63,64 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.213-161A>G rs799912 52,94 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.1-134T>C rs3765640 54,55 Out of pre-NGS method covered region	
BRCA1:NM_007294:c.5468-121_5468-121delA rs375078966 21.49 Out of pre-NGS method covered region	
BRCA1:NM 007294:c.4485-203 4485-199delAACCC rs34250703 40.74 Out of pre-NGS method covered region	

	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	51.58		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.441+52_441+63delCTTTTTTTTTT		53.66		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.441+36 441+38delCTT	rs147856441	39.26		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.68_69delAG	rs386833395	49.82	PAT			
V11							
	BRCA2:NM_000059:c.1-26G>A	rs1799943	47,96	Known			
	BRCA2:NM 000059:c.425+246G>C	rs11571613	55		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.631+183T>A	rs3752451	44,44		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.793+98G>A	rs206073	99,16		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.1114A>C	rs144848	45,68	Known	out or pro 1100 mother of torona region		
	BRCA2:NM 000059:c.3396A>G	rs1801406	51,67	Known			
	BRCA2:NM 000059:c.4563A>G	rs206075	100	Known			
	BRCA2:NM 000059:c.6513G>C	rs206076	99,83	Known			
	BRCA2:NM 000059:c.6841+191C>A	rs11571662	50		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.6938-120T>C	rs206080	99,71		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7242A>G	rs1799955	49,01	Known	eat of profited motified develor region		
	BRCA2:NM 000059:c.7397T>C	rs169547	99,24	Known			
	BRCA2:NM 000059:c.7617+190G>A	rs206096	97,22		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	99,63	Known	Cat of pre 1100 method toyered region		
	BRCA2:NM_000059:c.7500-1412C	rs3764791	49,06		Out of pre-NGS method covered region		
<u> </u>	BRCA2:NM_000059:c.8754+187C>T	rs3764791	51,02		Out of pre-NGS method covered region	+	
<u> </u>	BRCA2:NM_000059:c.8755-66T>C	rs4942486	99,28		Out of pre-NGS method covered region Out of pre-NGS method covered region	+	
<u> </u>	BRCA2:NM 000059:c.9257-219A>G	rs7330025	100		Out of pre-NGS method covered region Out of pre-NGS method covered region	+	
	BRCA2:NM 000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region		
	BRCA2:NW 000039:C:8048+2441>C BRCA2:NM 000059:c:6841+78 6841+81delAATT	rs138193280	43.32		Out of pre-NGS method covered region		
	BRCA2:NW 000059:C:0641+78 0641+81deiAA11 BRCA2:NM 000059:C:8633-150 8633-149insA	rs200156944	32.26		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.3119G>A	rs4986852	47,96	Known	Out of pre-1100 method covered region		
	BRCA1:NW 007294:C:5119G>A BRCA1:NM 007294:C:548-293G>A	rs117281398	57,14		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.442-34C>T	rs799923	53,02	Known	Out of pre-1103 method covered region		
	BRCA1:NM_007294:c.4987-142_4987-142delA	137 33323	22.22	KIIOWII	Out of pre-NGS method covered region		
V12	BRCAT.NW_007294.C.4907-142_4907-142delA		22.22		Out of pre-NGS method covered region		
V 12	BRCA2:NM_000059:c.793+98G>A	rs206073	97,27		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.3807T>C	rs543304	99,35		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.6513G>C	rs206076	100		Region not anlyzed by pre-NGS method		
		15200070					
	RPCA2:NM	re206080			Out of are NGS method covered region		
	BRCA2:NM_000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7397T>C	rs169547	100 99		Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.7397T>C BRCA2:NM_000059:c.7617+190G>A	rs169547 rs206096	100 99 100		Region not anlyzed by pre-NGS method Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C	rs169547	100 99 100 100		Region not anlyzed by pre-NGS method		
\/42	BRCA2:NM_000059:c.7397T>C BRCA2:NM_000059:c.7617+190G>A	rs169547 rs206096	100 99 100		Region not anlyzed by pre-NGS method Out of pre-NGS method covered region		
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG	rs169547 rs206096	100 99 100 100 40.95	PAT	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region	SANCED	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C)	rs169547 rs206096 rs15869	100 99 100 100 40.95	PAT	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region	SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA)	rs169547 rs206096 rs15869	100 99 100 100 40.95 Not detected Not detected	PAT	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region	SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C	rs169547 rs206096 rs15869 rs3214300 rs1802683	100 99 100 100 40.95 Not detected Not detected 36,5	PAT Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region		
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2466T>C	rs169547 rs206096 rs15869	100 99 100 100 40.95 Not detected Not detected 36,5 41,48	PAT Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region		
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2466T>C PMS2:NM_000535:c.2446-118G>A	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000	100 99 100 100 40.95 Not detected 36,5 41,48 33,33	PAT Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method	SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected	PAT Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region	SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.2334A>G	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected Not detected	PAT Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Complex region	SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-T>C PMS2:NM_000535:c.2446-T18G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.234A>G PMS2:NM_000535:c.2276-135T>C	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected Not detected	PAT Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.234A>G PMS2:NM_000535:c.2276-135T>C PMS2:NM_000535:c.2253T>C	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected Not detected 62,5 Not detected	PAT Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analyse	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.2344A>G PMS2:NM_000535:c.2276-135T>C PMS2:NM_000535:c.2253T>C PMS2:NM_000535:c.2174+103C>T	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325 rs2692553	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected Not detected 62,5 Not detected 29,09	PAT Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM_000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2446-T>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.2344A>G PMS2:NM_000535:c.2276-135T>C PMS2:NM_000535:c.2174+103C>T PMS2:NM_000535:c.2174+103C>T PMS2:NM_000535:c.207-7C>T	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325 rs2692553 rs55954143	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17	PAT Known Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analyse	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM 000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM_000535:c.2570G>C PMS2:NM_000535:c.2466T>C PMS2:NM_000535:c.2466T>C PMS2:NM_000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.2334A>G PMS2:NM_000535:c.2531>C PMS2:NM_000535:c.253T>C PMS2:NM_000535:c.253T>C PMS2:NM_000535:c.2174+103C>T PMS2:NM_000535:c.2174+103C>T PMS2:NM_000535:c.2007-7C>T PMS2:NM_000535:c.2007-7C>T PMS2:NM_000535:c.21621A>G	rs169547 rs206096 rs15869	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17 100	PAT Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analys Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM 000059:c.10257+105A>C BRCA2:NM 000059:c.1307_1308delAG PMS2:NM 000535:c.2606G>C (c.*17G>C) PMS2:NM 000535:c.2681_2682insA (c.*92dupA) PMS2:NM 000535:c.2570G>C PMS2:NM 000535:c.2466T>C PMS2:NM 000535:c.2466T>C PMS2:NM 000535:c.2446-118G>A PMS2:NM 000535:c.2340C>T PMS2:NM 000535:c.2334A>G PMS2:NM 000535:c.2551>C PMS2:NM 000535:c.2551>C PMS2:NM 000535:c.2531>C PMS2:NM 000535:c.2531>C PMS2:NM 000535:c.2531>C PMS2:NM 000535:c.2531>C	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 rs17420802 rs63750220 rs9655490 rs1805325 rs2692553 rs55954143 rs2228006 rs199550985	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17 100 20,42	PAT Known Known Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analyse	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM 000059:c.10257+105A>C BRCA2:NM 000059:c.1307_1308delAG PMS2:NM 000535:c.2606G>C (c.*17G>C) PMS2:NM 000535:c.2681_2682insA (c.*92dupA) PMS2:NM 000535:c.2570G>C PMS2:NM 000535:c.2466T>C PMS2:NM 000535:c.2446-118G>A PMS2:NM 000535:c.2340C>T PMS2:NM 000535:c.2334A>G PMS2:NM 000535:c.2575C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.25174+103C>T PMS2:NM 000535:c.2007-7C>T PMS2:NM 000535:c.2144-103C>T PMS2:NM 000535:c.21414-57G>A PMS2:NM 000535:c.1145-57G>A PMS2:NM 000535:c.1145-57G>A PMS2:NM 000535:c.780C>G	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325 rs2692553 rs55954143 rs2228006 rs199550985 rs1805319	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17 100 20,42 99,26	PAT Known Known Known Known Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analysed by pre-NGS method Region not anlyzed by pre-NGS method Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM 000059:c.10257+105A>C BRCA2:NM_000059:c.1307_1308delAG PMS2:NM_000535:c.2606G>C (c.*17G>C) PMS2:NM_000535:c.2681_2682insA (c.*92dupA) PMS2:NM 000535:c.2570G>C PMS2:NM 000535:c.2466T>C PMS2:NM 000535:c.2446-118G>A PMS2:NM_000535:c.2340C>T PMS2:NM_000535:c.2334A>G PMS2:NM 000535:c.2575C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.2531>C PMS2:NM 000535:c.2531>C PMS2:NM 000535:c.2537>C PMS2:NM 000535:c.2174+103C>T PMS2:NM 000535:c.2174+103C>T PMS2:NM 000535:c.1621A>G PMS2:NM 000535:c.1621A>G PMS2:NM 000535:c.1621A>G PMS2:NM 000535:c.780C>G PMS2:NM 000535:c.353+209G>A	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325 rs2692553 rs55954143 rs2228006 rs199550985 rs1805319 rs6463526	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17 100 20,42 99,26 61,9	PAT Known Known Known Known Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analysed by pre-NGS method Region not anlyzed by pre-NGS method Region not anlyzed by pre-NGS method Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	
V13	BRCA2:NM 000059:c.7397T>C BRCA2:NM 000059:c.7617+190G>A BRCA2:NM 000059:c.10257+105A>C BRCA2:NM 000059:c.1307_1308delAG PMS2:NM 000535:c.2606G>C (c.*17G>C) PMS2:NM 000535:c.2681_2682insA (c.*92dupA) PMS2:NM 000535:c.2570G>C PMS2:NM 000535:c.2466T>C PMS2:NM 000535:c.2446-118G>A PMS2:NM 000535:c.2340C>T PMS2:NM 000535:c.2334A>G PMS2:NM 000535:c.2575C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.2576-135T>C PMS2:NM 000535:c.25174+103C>T PMS2:NM 000535:c.2007-7C>T PMS2:NM 000535:c.2144-103C>T PMS2:NM 000535:c.21414-57G>A PMS2:NM 000535:c.1145-57G>A PMS2:NM 000535:c.1145-57G>A PMS2:NM 000535:c.780C>G	rs169547 rs206096 rs15869 rs3214300 rs1802683 rs10000 . rs17420802 rs63750220 rs9655490 rs1805325 rs2692553 rs55954143 rs2228006 rs199550985 rs1805319	100 99 100 100 40.95 Not detected 36,5 41,48 33,33 Not detected 62,5 Not detected 29,09 40,17 100 20,42 99,26	PAT Known Known Known Known Known Known Known Known Known	Region not anlyzed by pre-NGS method Out of pre-NGS method covered region Out of pre-NGS method covered region Out of pre-NGS method covered region Complex region Complex region Region not anlyzed by pre-NGS method Complex region Complex region Region not anlyzed by pre-NGS method Variant only detected by RNA-based analysed by pre-NGS method Region not anlyzed by pre-NGS method Region not anlyzed by pre-NGS method	SANGER SANGER SANGER	

		T	T	1			T
	PMS2:NM_000535:c.23+72C>T	rs3735295	34,21	Known			
	PMS2:NM_000535:c.705+118_705+118delT	rs11289444	53.7		Region not anlyzed by pre-NGS method		
V14							
	BRCA2:NM_000059:c.793+98G>A	rs206073	98,49		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1114A>C	rs144848	99,69		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.6513G>C	rs206076	99,43		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.6938-120T>C	rs206080	97,83		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7397T>C	rs169547	99,52		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	47,44		Region not anlyzed by pre-NGS method		
	BRCA2:NM_000059:c.8755-66T>C	rs4942486	49,81		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.3934_3952delAATTACAAGAGAAATACTG	134342400	35.53	PAT	Cut of pre-1400 method covered region		
V15	BRCAZ:NIM_0000039.C:3934_393206IAATTACAAGAGAAATACTG	•	33.33	FAI			
v io	ADC:NM 004407540:2 402 255C> A	re 202470	100		Out of are NCC method sourced region		
	APC:NM_001127510:c.423-256G>A	rs392179	100	17	Out of pre-NGS method covered region		
	APC:NM_001127510:c.645+173A>G	rs2289484	100	Known			
	APC:NM_001127510:c.730-22G>C	rs115634618	47,09	Known			
	APC:NM_001127510:c.934-132G>T	rs12521276	100	1	Out of pre-NGS method covered region		
	APC:NM_001127510:c.1458T>C	rs2229992	100	Known			
	APC:NM_001127510:c.1635G>A	rs351771	99,73	Known			
	APC:NM_001127510:c.1743+193G>A	rs351772	100	Known			
	APC:NM_001127510:c.2344A>T		50,63	PAT			
	APC:NM 001127510:c.4479G>A	rs41115	99,4	Known			
	APC:NM 001127510:c.5034G>A	rs42427	99,47	Known			
	APC:NM 001127510:c.5268T>G	rs866006	100	Known			
	APC:NM 001127510:c.5465T>A	rs459552	99,42	Known			
	APC:NM 001127510:c.5880G>A	rs465899	100	Known			
	APC:NM 001127510:c.934-133 934-132delTG	rs369189394	20	KIIOWII	Out of pre-NGS method covered region		
	APC:NM 001127510:c.934-132 934-132delG	rs141013063	65.31	+	Out of pre-NGS method covered region		
					, ,		
	APC:NM_001127510:c.934-120_934-119insG	rs138003874	77.42	+	Out of pre-NGS method covered region		
1/40	APC:NM_001127510:c.1959-145_1959-144insAGAA	rs3839284	93.58		Out of pre-NGS method covered region		
V16	NUTT (1111 0 10000 1177 100 0	0040400	00.51		D : / / // NOO // /		
	MUTYH:NM_012222:c.1477-40C>G	rs3219493	99,54	+	Region not anlyzed by pre-NGS method		
	MUTYH:NM_012222:c.1187G>A	rs36053993	100	PAT			
	MUTYH:NM_012222:c.504+35A>G	rs3219487	100		Region not anlyzed by pre-NGS method		
V17							
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,35		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1114A>C	rs144848	48,33	Known			
	BRCA2:NM_000059:c.3807T>C	rs543304	47,13	Known			
	BRCA2:NM_000059:c.4563A>G	rs206075	100		Could not be detected by CSCE		
	BRCA2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE		
	BRCA2:NM 000059:c.6938-120T>C	rs206080	99,12		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7397T>C	rs169547	99,5	1	Could not be detected by CSCE		
	BRCA2:NM 000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.9501+178A>G	rs206343	34,62	1	Out of pre-NGS method covered region		+
	BRCA2:NW 000059:c.10257+105A>C	rs15869	63,75		Out of pre-NGS method covered region		
		rs80359530		PAT	Out of pre-1400 method covered region		+
	BRCA2:NM_000059:c.5718_5719delCT	1200303030	49.73	PAI	Out of one NOO continue to the	+	
//10	BRCA1:NM_007294:c.4987-142_4987-142delA		23.68		Out of pre-NGS method covered region		
V18							
	BRCA2:NM_000059:c.1-26G>A	rs1799943	49,85	Known			
	BRCA2:NM_000059:c.631+183T>A	rs3752451	47,06		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,27		Out of pre-NGS method covered region		
	DD 0.4.0.11.4.00.0000	rs144848	47,34	Known			
	BRCA2:NM_000059:c.1114A>C			1.2	<u> </u>	1	<u> </u>
	BRCA2:NM_000059:c.1114A>C BRCA2:NM_000059:c.3396A>G	rs1801406	43,85	Known			
			43,85 100	Known			
	BRCA2:NM_000059:c.3396A>G BRCA2:NM_000059:c.4563A>G	rs1801406 rs206075	100	Known			
	BRCA2:NM_000059:c.3396A>G	rs1801406			Out of pre-NGS method covered region		

	BRCA2:NM_000059:c.7242A>G	rs1799955	48,96	Known			
	BRCA2:NM 000059:c.7397T>C	rs169547	99,61	Known			
	BRCA2:NM 000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		1
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	48,26	Known			
	BRCA2:NM 000059:c.8754+183A>C	rs3764791	38,64		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.8754+187C>T	rs3764792	37,84		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.8755-66T>C	rs4942486	47,33		Out of pre-NGS method covered region		-
	BRCA2:NM_000059:c.9501+178A>G	rs206343	48,28		Out of pre-NGS method covered region		_
	BRCA2:NM 000059:c.10257+105A>C	rs15869	64,29		Out of pre-NGS method covered region		_
	BRCA2:NM 000059:c.6841+78 6841+81delAATT	rs138193280	45,78		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.2584A>G	rs80356927	47,47	Known	Dut of pre-1400 method covered region		+
	BRCA1:NM 007294:C:2564A2G BRCA1:NM 007294:C:442-34C>T	rs799923	50,78	Known			+
	BRCA1:NM_007294:c.4987-142_4987-142delA	18799923		KIIOWII	Out of are NCC method accord region		-
		************************************	26.58	DAT	Out of pre-NGS method covered region		+
1/40	BRCA1:NM_007294:c.3869_3870delAA	rs80357918	51.32	PAT			
V19							4
	MSH2:NM_000251:c.211+9C>G	rs2303426	48,72		Region not anlyzed by pre-NGS method		
	MSH2:NM_000251:c.367-86A>C	rs17217765	46,15	+	Out of pre-NGS method covered region		
	MSH2:NM_000251:c.691G>T		46,99		Region not anlyzed by pre-NGS method	1	
	MSH2:NM_000251:c.1277-118G>A	rs1981929	46,95		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.1661+90T>C	rs10183143	68,57		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.2006-127C>T	rs2059521	100		Out of pre-NGS method covered region		
	MSH2:NM_000251:c.689_689delC		45.2	PAT			
	MSH2:NM_000251:c.2005+43_2005+43delT	rs201305711	22.78		Region not anlyzed by pre-NGS method		
	MLH1:NM 000249:c.453+79A>G	rs4234259	51,39		Out of pre-NGS method covered region		
	MLH1:NM 000249:c.655A>G	rs1799977	55,83	Known			
	MLH1:NM 000249:c.1038+86T>C	rs2286939	65,62		Out of pre-NGS method covered region		
	MLH1:NM 000249:c.1039-78A>G	rs11129748	60		Out of pre-NGS method covered region		
	MLH1:NM 000249:c.1668-19A>G	rs9876116	42,14		Region not anlyzed by pre-NGS method		
	MLH1:NM 000249:c.1990-121C>T	rs2241031	55,36		Out of pre-NGS method covered region		
	MLH1:NM_000249:c.1897-167_1897-167delT	rs11290150	85,81		Out of pre-NGS method covered region		
V20			33,37				
720	PMS2:NM 000535:c.2446-118G>A		27,59		Out of pre-NGS method covered region		
	PMS2:NM_000535:c.2276-135T>C	rs9655490	39,23		Out of pre-NGS method covered region		-
	PMS2:NM 000535:c.1621A>G	rs2228006	99,79	Known	eat of pre 1400 method dovered region		_
	PMS2:NM 000535:c.1408C>T	rs1805321	56,54	Known			_
	PMS2:NM 000535:c.804-164T>G	rs2286681	61,54	TUIOWII	Out of pre-NGS method covered region		
	PMS2:NM 000535:c.780C>G	rs1805319	99,11	Known	Out of pre-NGS method covered region		
	PMS2:NM 000535:c.760C/G	rs62456182	50	Known			+
	PMS2:NM 000535:c.538-223A>G	rs2345060	75	_	Out of pre-NGS method covered region		+
	PMS2:NM 000535:c.163+138A>G				Out of pre-NGS method covered region		+
		rs62456183	70	+		+	+
	PMS2:NM_000535:c.989-105_989-104insA	rs149249785	36	DAT	Out of pre-NGS method covered region	+	+
	PMS2:NM_000535:c.780_780delC		49.53	PAT	Out of one NOO mathe days and a size	+	+
1/24	PMS2:NM_000535:c.705+118_705+118delT	rs11289444	48.84		Out of pre-NGS method covered region		+
V21		100					
	MLH1:NM_000249:c.1-93G>A	rs1800734	100	Known			
	MLH1:NM_000249:c.116+199C>T		40,91		Out of pre-NGS method covered region		
	MLH1:NM_000249:c.1039-31A>T	rs59684491	43,75		Region not anlyzed by pre-NGS method	1	
	MLH1:NM_000249:c.1039-29A>T	rs6771325	73,68		Region not anlyzed by pre-NGS method		
	MLH1:NM_000249:c.1039-31_1039-29delATA	-	26.92		Region not anlyzed by pre-NGS method		
V22							
	BRCA2:NM_000059:c.793+98G>A	rs206073	99,61		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1114A>C	rs144848	99,42	Known			
	BRCA2:NM_000059:c.4563A>G	rs206075	100	Known			
	BRCA2:NM_000059:c.6513G>C	rs206076	99,66	Known			
	BRCA2:NM_000059:c.6938-120T>C	rs206080	98,37		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7397T>C	rs169547	100	Known			
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100	_	Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9648+84G>A	rs81002832	40,86		Out of pre-NGS method covered region		
	1	1.00.002	Page 16	-1	1 p		

	BRCA1:NM_007294:c.5407-193A>G	rs8176310	38,71		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5278-191A>T	rs8176297	59,57		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5152+66G>A	rs3092994	42,2		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5075-237C>A	rs8176257	53,85		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5074+65G>A	rs8176235	50,44		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4987-68A>G	rs8176234	52,94		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4987-92A>G	rs8176233	50		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4986+222A>G	rs3092987	77,78		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4837A>G	rs1799966	51,23	Known			
	BRCA1:NM_007294:c.4485-63C>G	rs8176212	50,72		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4485-137T>A	rs2236762	53,25		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4358-2590T>G	rs8176194	47,86		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4358-2885G>A	rs8176193	48,88		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4357+117G>A	rs3737559	41,79		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4308T>C	rs1060915	46,19	Known			
	BRCA1:NM 007294:c.4097-141A>C	rs799916					
			45,81		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.3548A>G	rs16942	47,99	Known		+	
	BRCA1:NM_007294:c.3113A>G	rs16941	47,08	Known			
	BRCA1:NM_007294:c.2612C>T	rs799917	47,71	Known			
	BRCA1:NM_007294:c.2311T>C	rs16940	49,37	Known		-	
	BRCA1:NM_007294:c.2082C>T	rs1799949	47,7	Known			
	BRCA1:NM_007294:c.594-225G>A	rs8176147	69,23		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.593+167T>C	rs8176145	46,81		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.547+146A>T	rs8176140	56,52		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.213-161A>G	rs799912	43,24		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.1-134T>C	rs3765640	60		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	53.85		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.3770_3771delAG	rs80357993	50.44	PAT			
	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	46.24		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.441+52 441+63delCTTTTTTTTTT		37.04		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.441+36_441+38delCTT	rs147856441	46.15		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.134+224_134+224delT	rs35149296	46.15		Out of pre-NGS method covered region		
V23							
	BRCA2:NM 000059:c.793+98G>A	rs206073	100		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.1114A>C	rs144848	54,95		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.3807T>C	rs543304	42,21		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.6513G>C	rs206076	100		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.7397T>C	rs169547	99,43		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.7617+190G>A	rs206096	100	1	Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7806-14T>C	rs9534262	49,52		Region not anlyzed by pre-NGS method		
	BRCA2:NM 000059:c.8755-66T>C	rs4942486	50		Out of pre-NGS method covered region		
	BRCA2:NW 000059:c.10257+105A>C	rs15869	59,09	-	Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.1126_1126delT	1313003	43,55	PAT	Tout or pre-1400 method tovered region		
1/04	DRGAZ.NW_000033.C.1120_11200011		43,55	FAI			
V24	ADC:NIM 004497540:e 493 9560: A	re200470	100		Out of are NCC method account and		
	APC:NM_001127510:c.423-256G>A	rs392179	100		Out of pre-NGS method covered region		
	APC:NM_001127510:c.645+173A>G	rs2289484	100	-	Out of pre-NGS method covered region		
	APC:NM_001127510:c.933+261G>T	rs12656359	100		Out of pre-NGS method covered region	-	
	APC:NM_001127510:c.934-132G>T	rs12521276	100		Out of pre-NGS method covered region	-	
	APC:NM_001127510:c.1458T>C	rs2229992	100	-	Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.1635G>A	rs351771	99,68		Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.1743+193G>A	rs351772	100		Out of pre-NGS method covered region		
	APC:NM_001127510:c.3249T>G	rs201629780	45,35		Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.4479G>A	rs41115	100		Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.5034G>A	rs42427	100		Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.5268T>G	rs866006	99,65		Region not anlyzed by pre-NGS method		
	APC:NM_001127510:c.5465T>A	rs459552	100		Region not anlyzed by pre-NGS method		

	APC:NM_001127510:c.5880G>A	rs465899	100		Region not anlyzed by pre-NGS method	
	APC:NM_001127510:c.934-133_934-132delTG	rs369189394	28.05		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-132_934-132delG	rs141013063	67.8		Out of pre-NGS method covered region	
	APC:NM_001127510:c.934-120_934-119insG	rs138003874	89.52		Out of pre-NGS method covered region	
	APC:NM_001127510:c.1959-145_1959-144insAGAA	rs3839284	97.56		Out of pre-NGS method covered region	
	APC:NM_001127510:c.3180_3184delAAAAC		53.1	PAT		
V25						
	MSH2:NM 000251:c.1661+90T>C	rs10183143	37,93		Out of pre-NGS method covered region	
	MSH2:NM 000251:c.2006-127C>T	rs2059521	100		Out of pre-NGS method covered region	
	MSH2:NM 000251:c.388 389delCA	rs63750704	45.72	PAT		
	MSH2:NM_000251:c.2005+43_2005+43delT	rs201305711	21.5		Out of pre-NGS method covered region	
V26						
	BRCA2:NM 000059:c.793+98G>A	rs206073	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.1114A>C	rs144848	50,84		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.3807T>C	rs543304	47,4		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.6513G>C	rs206076	100		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.6938-120T>C	rs206080	99,6	1	Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7397T>C	rs169547	100	1	Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.75971>C	rs206096	100	1	Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	49,51		Region not anlyzed by pre-NGS method	
	BRCA2:NM 000059:c.8755-66T>C	rs4942486	51,45		Out of pre-NGS method covered region	
					, v	
	BRCA2:NM_000059:c.9257-235G>A	rs7327867	63,64		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9257-219A>G	rs7330025	50		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9501+178A>G	rs206343	50		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.10257+105A>C	rs15869	47,93		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7227_7227delT		42,31	PAT		
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	75,93		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5333-153A>G	rs8176305	45,71		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5278-191A>T	rs8176297	53,27		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5152+66G>A	rs3092994	48,16		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.5075-238A>G	rs8176256	66,67		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4987-68A>G	rs8176234	46,86		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4987-92A>G	rs8176233	46,34		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4986+222A>G	rs3092987	54,17		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4837A>G	rs1799966	47,99		Region not anlyzed by pre-NGS method	
	BRCA1:NM_007294:c.4485-63C>G	rs8176212	51,8		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4485-137T>A	rs2236762	51,2		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4358-2590T>G	rs8176194	50,92		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4358-2885G>A	rs8176193	50		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.4308T>C	rs1060915	48,09		Region not anlyzed by pre-NGS method	
	BRCA1:NM_007294:c.4097-141A>C	rs799916	42,68		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.3548A>G	rs16942	50,19		Region not anlyzed by pre-NGS method	
	BRCA1:NM 007294:c.3113A>G	rs16941	47,86		Region not anlyzed by pre-NGS method	
	BRCA1:NM_007294:c.2612C>T	rs799917	45,07		Region not anlyzed by pre-NGS method	
	BRCA1:NM 007294:c.2311T>C	rs16940	48,22		Region not anlyzed by pre-NGS method	
	BRCA1:NM 007294:c.2082C>T	rs1799949	52,39		Region not anlyzed by pre-NGS method	
	BRCA1:NM_007294:c.2077G>A	rs4986850	52,9	1	Region not anlyzed by pre-NGS method	
	BRCA1:NM 007294:c.594-225G>A	rs8176147	62,07		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.593+167T>C	rs8176145	48,18		Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.547+146A>T	rs8176140	66,67	1	Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.442-34C>T	rs799923	43,84	1	Region not anlyzed by pre-NGS method	
	BRCA1:NM_007294:c.213-161A>G	rs799912	37,08	1	Out of pre-NGS method covered region	
	BRCA1:NM 007294:c.1-134T>C	rs3765640	57,78	1	Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.1-1341>C BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	27.27	+		
	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	45.99	1	Out of pre-NGS method covered region Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.346-56_546-56delT BRCA1:NM_007294:c.441+52_441+63delCTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTT	150170144	45.99	1	Out of pre-NGS method covered region Out of pre-NGS method covered region	
	DNGA1.NW_007294.0.441+52_441+630elG11111111111		44.23	1	Tout of pre-ingo method covered region	

			upprementary 1				
	BRCA1:NM 007294:c.441+36 441+38delCTT	rs147856441	44.89		Region not anlyzed by pre-NGS method		
	BRCA1:NM_007294:c.134+224_134+224delT	rs35149296	59.09		Out of pre-NGS method covered region		
V27							
	MSH6:NM_000179:c.186C>A	rs1042820	58,18	Known			
	MSH6:NM_000179:c.260-22C>G		Not detecte	d Known	Low covered region	SANGER	
	MSH6:NM_000179:c.276A>G	rs1800932	51,93	Known			
	MSH6:NM 000179:c.457+52T>A	rs3136282	55,36	Known			
	MSH6:NM 000179:c.458-133A>G	rs3136316	58,65	Known			
	MSH6:NM 000179:c.540T>C	rs1800935	51,34	Known			
	MSH6:NM 000179:c.628-56C>T	rs1800936	45,11	Known			
	MSH6:NM_000179:c.3173-101G>C	rs2072447	49,04		Out of pre-NGS method covered region		
	MSH6:NM 000179:c.3438+14A>T	rs2020911	49,63	Known			
	MSH6:NM_000179:c.3556+146G>A	rs7562048	42,55	Known			
	MSH6:NM 000179:c.3557-144G>A	rs7562367	66,67		Out of pre-NGS method covered region		
	MSH6:NM_000179:c.3646+91T>C	rs3136359	48,98		Out of pre-NGS method covered region		
	MSH6:NM_000179:c.3802-40C>G	rs3136367	49,02	Known	-		
	MSH6:NM_000179:c.2905_2906delTA		49.4	PAT			
	MSH6:NM 000179:c.3646+29 3646+32delCTAT	rs2234731	41.11	Known			
	MSH6:NM_000179:c.4002-27_4002-27delT		33.27				
V28							
	BRCA2:NM_000059:c.425+246G>C	rs11571613	57,89		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.631+183T>A	rs3752451	63,64		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.793+98G>A	rs206073	99,55		Out of pre-NGS method covered region		
	BRCA2:NM 000059:c.3396A>G	rs1801406	46,92	Known			
	BRCA2:NM 000059:c.3807T>C	rs543304	49,13	Known			
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Could not be detected by CSCE		
	BRCA2:NM_000059:c.6513G>C	rs206076	100		Could not be detected by CSCE		
	BRCA2:NM 000059:c.6841+191C>A	rs11571662	51,47		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7242A>G	rs1799955	48,74	Known	-		
	BRCA2:NM_000059:c.7397T>C	rs169547	100		Could not be detected by CSCE		
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.7806-14T>C	rs9534262	52,33	Known			
	BRCA2:NM_000059:c.8754+183A>C	rs3764791	36,36		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.8754+187C>T	rs3764792	36,67		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.8755-66T>C	rs4942486	47,32		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9257-219A>G	rs7330025	45		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9501+178A>G	rs206343	61,9		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.10257+105A>C	rs15869	55,81		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.260_261delCT		44.9	PAT			
	BRCA2:NM_000059:c.6841+78_6841+81delAATT	rs138193280	46.06		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.8633-150_8633-149insA	rs200156944	36.11		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5407-193A>G	rs8176310	49,38		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5333-153A>G	rs8176305	54,98		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5278-191A>T	rs8176297	48,85		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5152+66G>A	rs3092994	52,53		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.5075-238A>G	rs8176256	51,43		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4987-68A>G	rs8176234	43,29		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4987-92A>G	rs8176233	42,49		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4986+222A>G	rs3092987	60	1,	Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4837A>G	rs1799966	55,39	Known			
	BRCA1:NM_007294:c.4485-63C>G	rs8176212	46,72		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4485-137T>A	rs2236762	50	1	Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4358-2590T>G	rs8176194	50	1	Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4358-2885G>A	rs8176193	49,27	IZ:	Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.4308T>C	rs1060915	45,37	Known			
	BRCA1:NM_007294:c.4097-141A>C	rs799916	53,38		Out of pre-NGS method covered region		

		-				
	BRCA1:NM 007294:c.3548A>G	rs16942	44,53	Known		
	BRCA1:NM 007294:c.3113A>G	rs16941	51,03	Known		
	BRCA1:NM 007294:c.2612C>T	rs799917	47,68	Known		
	BRCA1:NM 007294:c.2311T>C	rs16940	50,74	Known		+
	BRCA1:NM 007294:c.2082C>T	rs1799949	50,65	Known		
	BRCA1:NM_007294:0.2002C>1 BRCA1:NM_007294:0.2077G>A	rs4986850	50,59	Known		
	BRCA1:NM 007294:c.671-178G>A	rs8176151	48,84		Out of pre-NGS method covered region	
	_	rs7503154				+
	BRCA1:NM_007294:c.670+259T>G		50		Out of pre-NGS method covered region	+
	BRCA1:NM_007294:c.594-225G>A	rs8176147	56,25	I/o access	Out of pre-NGS method covered region	+
	BRCA1:NM_007294:c.593+167T>C	rs8176145	53,01	Known	0.1.6. 1100 # 1 1	
	BRCA1:NM_007294:c.547+146A>T	rs8176140	51,06		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.213-161A>G	rs799912	41,76		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.1-134T>C	rs3765640	44,12	Known		
	BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	32.65		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	44.24		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.441+52_441+63delCTTTTTTTTTTT	0	48.33		Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.441+36_441+38delCTT	rs147856441	42.96	1	Out of pre-NGS method covered region	
	BRCA1:NM_007294:c.134+224_134+224delT	rs35149296	46.15		Out of pre-NGS method covered region	
V29						
	BRCA2:NM_000059:c.1-26G>A	rs1799943	100		Could not be detected by CSCE	
	BRCA2:NM_000059:c.67+82C>G	rs189026060	50,34		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.631+183T>A	rs3752451	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.793+98G>A	rs206073	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.3396A>G	rs1801406	100		Could not be detected by CSCE	
	BRCA2:NM 000059:c.4563A>G	rs206075	100		Could not be detected by CSCE	
	BRCA2:NM 000059:c.6513G>C	rs206076	99,17		Could not be detected by CSCE	
	BRCA2:NM 000059:c.6841+191C>A	rs11571662	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.6938-120T>C	rs206080	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7242A>G	rs1799955	100		Could not be detected by CSCE	1
	BRCA2:NM 000059:c.7397T>C	rs169547	100		Could not be detected by CSCE	1
	BRCA2:NM 000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	100		Could not be detected by CSCE	1
	BRCA2:NM 000059:c.8754+183A>C	rs3764791	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.8754+187C>T	rs3764792	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.8755-66T>C	rs4942486	96,3		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.9257-235G>A	rs7327867	100		Out of pre-NGS method covered region	+
	-	rs7330025	100			+
	BRCA2:NM_000059:c.9257-219A>G				Out of pre-NGS method covered region	+
	BRCA2:NM_000059:c.6841+78_6841+81delAATT	rs138193280	87.25		Out of pre-NGS method covered region	+
<u> </u>	BRCA2:NM_000059:c.8633-150_8633-149insA	rs200156944	84.62	+	Out of pre-NGS method covered region	+
	BRCA1:NM_007294:c.5468-121_5468-121delA	rs375078966	24.62	DAT	Out of pre-NGS method covered region	 +
1 (0.0	BRCA1:NM_007294:c.4110_4111insATCT		47.21	PAT		
V30	DD040 NW 0000F0 4 000 A	47000:5	5 0.00	1,4		4
	BRCA2:NM_000059:c.1-26G>A	rs1799943	52,65	Known	0 (1 100 1 1 1 1 1 1 1 1	
	BRCA2:NM_000059:c.425+246G>C	rs11571613	70		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.631+183T>A	rs3752451	53,19		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.793+98G>A	rs206073	100		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.1114A>C	rs144848	47,2	Known		
	BRCA2:NM_000059:c.3396A>G	rs1801406	45,99	Known		
	BRCA2:NM_000059:c.4563A>G	rs206075	100	Known		
	BRCA2:NM_000059:c.6513G>C	rs206076	99,8	Known		
	BRCA2:NM_000059:c.6841+191C>A	rs11571662	53,79		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.6938-120T>C	rs206080	99,55		Out of pre-NGS method covered region	
	BRCA2:NM_000059:c.7242A>G	rs1799955	46,14	Known		
1	BRCA2:NM_000059:c.7397T>C	rs169547	100	Known		
	BRCA2:NM_000059:c.7617+190G>A	rs206096	100		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.7806-14T>C	rs9534262	51,41	Known	X ·	
	BRCA2:NM 000059:c.8754+183A>C	rs3764791	44,83		Out of pre-NGS method covered region	
	BRCA2:NM 000059:c.8754+187C>T	rs3764792	42,86		Out of pre-NGS method covered region	
			,			_ •

	BRCA2:NM_000059:c.8755-66T>C	rs4942486	46,47		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9257-219A>G	rs7330025	60		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.9648+244T>C	rs206344	100		Out of pre-NGS method covered region		
	BRCA2:NM_000059:c.6841+78_6841+81delAATT	rs138193280	41,61		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5407-193A>G	rs8176310	48,57		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5333-153A>G	rs8176305	44,14		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5278-191A>T	rs8176297	57,14		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5152+66G>A	rs3092994	57		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.5075-238A>G	rs8176256	64,29		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4987-68A>G	rs8176234	60		Out of pre-NGS method covered region		1
	BRCA1:NM 007294:c.4987-92A>G	rs8176233	63,64		Out of pre-NGS method covered region		+
	BRCA1:NM 007294:c.4986+222A>G	rs3092987	66,67		Out of pre-NGS method covered region		+
	BRCA1:NM 007294:c.4837A>G	rs1799966	46,25	Known	eat of profited method develor region		-
	BRCA1:NM 007294:c.4485-63C>G	rs8176212	57,06		Out of pre-NGS method covered region		-
	BRCA1:NM 007294:c.4485-137T>A	rs2236762	57,35		Out of pre-NGS method covered region		+
	BRCA1:NM 007294:c.4358-2590T>G	rs8176194	48,96		Out of pre-NGS method covered region		
	BRCA1:NM 007294:c.4358-2885G>A	rs8176193	46,13		Out of pre-NGS method covered region		
-	BRCA1:NM 007294:c.4308T>C	rs1060915	45,86		Out of pre-1400 method covered region	+	+
	BRCA1:NM_007294:c.43081>C BRCA1:NM_007294:c.4097-141A>C		· · · · · ·	Known	Out of pro NCS method sourced region		+
		rs799916	44,07		Out of pre-NGS method covered region		+
	BRCA1:NM_007294:c.3548A>G	rs16942	51,05	Known		-	+
	BRCA1:NM_007294:c.3113A>G	rs16941	46,46	Known		-	+
	BRCA1:NM_007294:c.2612C>T	rs799917	45,34	Known			
	BRCA1:NM_007294:c.2311T>C	rs16940	46,85	Known			
	BRCA1:NM_007294:c.2082C>T	rs1799949	47,46	Known			
	BRCA1:NM_007294:c.2077G>A	rs4986850	48,35	Known			
	BRCA1:NM_007294:c.671-178G>A	rs8176151	60,71		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.593+167T>C	rs8176145	52,88		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.547+146A>T	rs8176140	41,03		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.213-161A>G	rs799912	50		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.1-134T>C	rs3765640	55,88	Known			
	BRCA1:NM_007294:c.4485-203_4485-199delAACCC	rs34250703	46.34		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.548-58_548-58delT	rs8176144	41.23		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.441+52_441+63delCTTTTTTTTTT		54.23		Out of pre-NGS method covered region		
	BRCA1:NM_007294:c.441+36_441+38delCTT	rs147856441	49.57	Known			
V31							
	APC:NM_001127510:c.1-176G>A		65		Out of pre-NGS method covered region		
	APC:NM_001127510:c.136-53T>C	rs2304793	50,67	Known			
	APC:NM_001127510:c.423-256G>A	rs392179	100		Out of pre-NGS method covered region		
	APC:NM_001127510:c.645+129A>C	rs2289485	42,86	Known			
	APC:NM_001127510:c.645+173A>G	rs2289484	57,14	Known			
	APC:NM_001127510:c.934-132G>T	rs12521276	100		Out of pre-NGS method covered region		
	APC:NM_001127510:c.934-92G>C	rs80307314	64,56	Known			
	APC:NM_001127510:c.1458T>C	rs2229992	47,67	Known			
	APC:NM_001127510:c.1635G>A	rs351771	50,22	Known			
	APC:NM_001127510:c.1660C>T	rs137854573	48,4	PAT			
	APC:NM_001127510:c.1743+193G>A	rs351772	47,37	Known			
	APC:NM 001127510:c.3264G>A	rs114774495	50,59	VUS			
	APC:NM 001127510:c.4479G>A	rs41115	47,86	Known			+
	APC:NM 001127510:c.5034G>A	rs42427	49,41	Known			+
	APC:NM 001127510:c.5268T>G	rs866006	50,4	Known			
	APC:NM_001127510:c.5465T>A	rs459552	100		Could not be detected by CSCE		+
	APC:NM 001127510:c.5880G>A	rs465899	47,14	Known	COLUMN TO COLORO DE COLORO		
	APC:NM_001127510:c.8532+86C>A	rs1804197	41,3	Known			+
	APC:NM_001127510:c.6532+66C>A APC:NM_001127511:c.1-134_1-133insG	181700181	46.67		Out of pre-NGS method covered region		+
	APC:NM 001127511:C:1-134_1-133IIISG APC:NM 001127510:c:934-133 934-132delTG	rs369189394	29.17		Out of pre-NGS method covered region		+
	APC:NM 001127510:C:934-133_934-132delG	rs141013063	64.71		Out of pre-NGS method covered region Out of pre-NGS method covered region	+	+
 	APC:NM 001127510:C:934-132_934-132detG APC:NM 001127510:c:934-120_934-119insG	rs138003874	25		Out of pre-NGS method covered region	+	+
-	APC:NM 001127510:c:934-120_934-119InsG APC:NM 001127510:c:1959-145 1959-144insAGAA	rs3839284	47.73		Out of pre-NGS method covered region Out of pre-NGS method covered region	+	+
I	JAF C.NIVI_UU I 1270 TU.C. 1909-140_1909-144IIISAGAA	183039204	47.73 Page 21	1	Out of pre-1405 method covered region		

V32						
V32	MILLIA-NIM 000040 4 000 A	ro 1000724	60.64		Out of are NCC method sovered region	
	MLH1:NM_000249:c.1-93G>A	rs1800734	60,64		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.453+79A>G	rs4234259	43,08	17	Out of pre-NGS method covered region	
	MLH1:NM_000249:c.655A>G	rs1799977	66,67	Known		
	MLH1:NM_000249:c.678-211T>A		75		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.1038+86T>C	rs2286939	50		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.1039-78A>G	rs11129748	47,06		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.1039-29A>T	rs6771325	46,15		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.1410-169C>T	rs2286940	70		Out of pre-NGS method covered region	
	MLH1:NM_000249:c.1668-19A>G	rs9876116	41,51		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.1990-121C>T	rs2241031	53,09		Out of pre-NGS method covered region	
	MLH1:NM 000249:c.541 541delG		46.22	PAT		
	MLH1:NM 000249:c.1897-167 1897-167delT	rs11290150	88.24		Out of pre-NGS method covered region	
V33		1011200100	00.21		eat of profited motified covered region	
V 33	NF1:NM 000267:c.889-192G>T		100		Could not be detected by RNA-based analysis	
	_			I/n au m	Could flot be detected by RNA-based analysis	
	NF1:NM_000267:c.2034G>A	rs2285892	100	Known	O II	<u> </u>
	NF1:NM_000267:c.3197+139T>C	rs2071009	98,33		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3315-130G>C	rs2072131	99,45		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3496+33C>A	rs2066736	100	1	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3974+46T>G	rs72813695	83,97		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7126+37C>G	rs7405740	99,48		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.8097+71A>T		23,53		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.2002-118_2002-117insACACACACAC		28.57		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.2956 2957insT		48.22	PAT	-	
	NF1:NM 000267:c.3974+33 3974+34insTG		72.76		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.6085-29 6085-28insA	rs33925668	78.53		Could not be detected by RNA-based analysis	
V34	141 1.1411 000201.0.0000 20_0000 20110/4	1300020000	70.00		Codia not be detected by 1414/1 based unarysis	
VOT	NF1:NM 000267:c.61-300C>T	rs2269856	66,67		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.61-123G>A	rs2269855	50,61	+	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.288+41G>A	rs2952976	47,83	+	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.480-90C>T	rs2905807	46,91		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.702G>A	rs1801052	49,55	Known		
	NF1:NM_000267:c.731-1G>C		52,86	PAT		
	NF1:NM_000267:c.731-1G>C NF1:NM_000267:c.888+108C>T	rs2953000	52,86 53,49	PAT	Could not be detected by RNA-based analysis	
		rs2953000 rs2952999		PAT	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.888+108C>T		53,49	PAT		
	NF1:NM_000267:c.888+108C>T NF1:NM_000267:c.888+118G>T	rs2952999	53,49 54,67	PAT	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C	rs2952999 rs2905875 rs2905876	53,49 54,67 48,99 43,77	PAT	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C	rs2952999 rs2905875 rs2905876 rs2905880	53,49 54,67 48,99 43,77 54,49		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892	53,49 54,67 48,99 43,77 54,49 46,09	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009	53,49 54,67 48,99 43,77 54,49 46,09 45,65		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.5546+29T>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.5546+29T>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.6085-28T>A	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.7126+37C>G	rs2952999 rs2905875 rs2905876 rs2905880 rs285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7405740	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3946+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.6085-28T>A NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7907+166C>T	rs2952999 rs2905875 rs2905876 rs2905880 rs285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs964288	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3946+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.6085-29T>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7907+166C>T NF1:NM 000267:c.8097+30G>T	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs964288 rs7350943	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3944+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+119T>A NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5085-29T>A NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.71907+166C>T NF1:NM 000267:c.8997+30G>T NF1:NM 000267:c.889-191_889-190insT	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7406039 rs7405740 rs964288 rs7350943 .	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69 35,29	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3944+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+17G>A NF1:NM 000267:c.5085-29T>A NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7907+166C>T NF1:NM 000267:c.889-191_889-190insT NF1:NM 000267:c.889-191_889-190insT	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7405740 rs964288 rs7350943	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69 35.29 44.86	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.33496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.56085-29T>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.8997+30G>T NF1:NM 000267:c.8997+30G>T NF1:NM 000267:c.889-191_889-190insT NF1:NM 000267:c.1528-37 1528-36insT NF1:NM 000267:c.2002-118 2002-117insACACACACACACACACACACACACACACACACACACAC	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7406039 rs7405740 rs964288 rs7350943 .	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69 35.29 44.86 20.45	Known	Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.3944+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+17G>A NF1:NM 000267:c.5085-29T>A NF1:NM 000267:c.7126+37C>G NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7907+166C>T NF1:NM 000267:c.889-191_889-190insT NF1:NM 000267:c.889-191_889-190insT	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7406039 rs7405740 rs964288 rs7350943 .	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69 35.29 44.86	Known	Could not be detected by RNA-based analysis	
V35	NF1:NM 000267:c.888+108C>T NF1:NM 000267:c.888+118G>T NF1:NM 000267:c.1393-130A>T NF1:NM 000267:c.1393-32T>C NF1:NM 000267:c.1641+39T>C NF1:NM 000267:c.2034G>A NF1:NM 000267:c.3197+139T>C NF1:NM 000267:c.3315-130G>C NF1:NM 000267:c.33496+33C>A NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.3974+46T>G NF1:NM 000267:c.5205+23T>C NF1:NM 000267:c.5546+19T>A NF1:NM 000267:c.5546+117G>A NF1:NM 000267:c.56085-29T>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.7395-29G>A NF1:NM 000267:c.8997+30G>T NF1:NM 000267:c.8997+30G>T NF1:NM 000267:c.889-191_889-190insT NF1:NM 000267:c.1528-37 1528-36insT NF1:NM 000267:c.2002-118 2002-117insACACACACACACACACACACACACACACACACACACAC	rs2952999 rs2905875 rs2905876 rs2905880 rs2285892 rs2071009 rs2072131 rs2066736 rs72813695 rs9894648 rs2285894 rs3815154 rs7406038 rs7406039 rs7406039 rs7405740 rs964288 rs7350943 .	53,49 54,67 48,99 43,77 54,49 46,09 45,65 44,98 49,69 40,44 47,1 47,2 51,36 49,11 58,56 100 44,59 53,33 20,69 35.29 44.86 20.45	Known	Could not be detected by RNA-based analysis	

	NF1:NM 000267:c.61-123G>A	rs2269855	100		Could not be detected by RNA-based analysis	T
	NF1:NM_000267:c.288+41G>A	rs2952976	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.480-90C>T	rs2905807	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.702G>A	rs1801052	97,9	Known	Codid not be detected by MA-based analysis	
	NF1:NM 000267:c.730+85A>T	131001032	20,48		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.888+108C>T	rs2953000	100		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.888+118G>T	rs2953000	100		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1393-130A>T					+
		rs2905875	100		Could not be detected by RNA-based analysis	+
	NF1:NM_000267:c.1393-32T>C	rs2905876	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1641+39T>C	rs2905880	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1642-158A>G	rs17883230	81,03		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5205+23T>C	rs9894648	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5546+19T>A	rs2285894	99,25		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5546+117G>A	rs3815154	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-29T>A	rs7406038	94,88		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-28T>A	rs7406039	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7126+37C>G	rs7405740	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7395-29G>A	rs964288	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.7907+166C>T	rs7350943	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.8050+20G>A	rs55747230	22,4		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.730+15_730+16insT		33.89		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.889-191_889-190insT	rs35966725	87.5		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.1528-37 1528-36insT	rs67472948	92.45		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.2002-118 2002-117insAC	rs141114572	43.4		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.3314+154 3314+154delT	rs17881842	84.21		Could not be detected by RNA-based analysis	
	NF1:NM_000267: whole gene deletion	Detected by I2HCP				
V36	J					
	NF1:NM 000267:c.61-300C>T	rs2269856	100		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.61-123G>A	rs2269855	99,38		Could not be detected by RNA-based analysis	1
	NF1:NM 000267:c.288+41G>A	rs2952976	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.480-90C>T	rs2905807	98,53		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.655-37A>C	rs375655707	46,49		Could not be detected by RNA-based analysis	
	NF1:NM 000267:c.702G>A	rs1801052	97,55	Known	Could not be detected by 14474 based analysis	
	NF1:NM 000267:c.888+108C>T	rs2953000	100		Could not be detected by RNA-based analysis	
	NF1:NM_000207:c.888+118G>T	rs2952999	100		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1381C>T	152902999	49,23	PAT	Codid not be detected by KNA-based analysis	
		rs2905875	100		Could not be detected by DNA based analysis	+
	NF1:NM_000267:c.1393-130A>T				Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1393-32T>C	rs2905876	99,6		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1641+39T>C	rs2905880	99,55		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5205+23T>C	rs9894648	49,25		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5546+19T>A	rs2285894	46,97		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.5546+117G>A	rs3815154	52,46		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-29T>A	rs7406038	46,63		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-28T>A	rs7406039	54,27	ļ	Could not be detected by RNA-based analysis	1
	NF1:NM_000267:c.7126+37C>G	rs7405740	54,17		Could not be detected by RNA-based analysis	_
	NF1:NM_000267:c.7395-29G>A	rs964288	37,62		Could not be detected by RNA-based analysis	_
	NF1:NM_000267:c.7970+166C>T;NF1:NM_000267:c.7907+166C>T	rs7350943	59,09		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.730+15_730+16insT		33.75		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.889-191_889-190insT	rs35966725	100		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.1528-37_1528-36insT	rs67472948	93.47		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.2002-118_2002-117insAC	rs141114572	46.94		Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3198-29_3198-29delT		20.49		Could not be detected by RNA-based analysis	
V37						1
	NF1:NM_00267:c.61-123G>A	rs2269855	52,49		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.278G>A	rs199474728	59,06	PAT		
	NF1:NM 00267:c.288+41G>A	rs2952976	63,44		Could not be detected by RNA-based analysis	1
	NF1:NM 00267:c.480-90C>T	rs2905807	45,05		Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.587-140A>G		55,17		Could not be detected by RNA-based analysis	
		•	,			

	NF1:NM 00267:c.702G>A	rs1801052	58,9	Known		
	NF1:NM 00267:c.1393-130A>T	rs2905875	47,92		Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.1393-32T>C	rs2905876	50,36		Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.1641+39T>C	rs2905880	34,88		Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.2034G>A	rs2285892	41,1	Known		
	NF1:NM_00267:c.3315-130G>C	rs2072131	48,21		Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.3496+33C>A	rs2066736	53,33		Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
	NF1:NM 00267:c.3974+46T>G	rs72813695	· · · · · · · · · · · · · · · · · · ·		i	
			38,46		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.5268+23T>C;NF1:NM_000267:c.5205+23T>C	rs9894648	36,73		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.5609+19T>A;NF1:NM_000267:c.5546+19T>A	rs2285894	44,9		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.5609+117G>A;NF1:NM_000267:c.5546+117G>A	rs3815154	43,22		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.6148-29T>A;NF1:NM_000267:c.6085-29T>A	rs7406038	35,86		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.7189+37C>G;NF1:NM_000267:c.7126+37C>G	rs7405740	100		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.7458-29G>A;NF1:NM_000267:c.7395-29G>A	rs964288	54,29		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.8160+30G>T;NF1:NM_000267:c.8097+30G>T		33,33		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.730+15_730+16insT	·	25.41		Could not be detected by RNA-based analysis	
	NF1:NM_00267:c.1528-37_1528-36insT	rs67472948	47.26	<u> </u>	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.2002-118_2002-117insACACACACACAC		35	1	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.3974+33_3974+34insTG		32.61	1	Could not be detected by RNA-based analysis	
	NF1:NM_000267:c.6085-29_6085-28insA	rs33925668	54.05		Could not be detected by RNA-based analysis	
V38						
	NF2:NM_000268:c.241-85G>T	rs5763378	51,43		Could not be detected by RNA-based analysis	
	NF2:NM_000268:c.364-39A>C	rs2530664	46,58		Could not be detected by RNA-based analysis	
	NF2:NM_000268:c.600-154G>A	rs2071622	59,04		Could not be detected by RNA-based analysis	
	NF2:NM 000268:c.886-205C>T	rs2252472	72,73		Could not be detected by RNA-based analysis	
	NF2:NM 000268:c.1122+129A>T	rs2527336	52,73		Could not be detected by RNA-based analysis	
	NF2:NM 000268:c.1123-51C>T	rs13055076	43,08		Could not be detected by RNA-based analysis	
	NF2:NM 000268:c.1575-67G>A	rs140086	99,18		Could not be detected by RNA-based analysis	
	NF2:NM 000268:c.1737+1623T>C	rs7291645	100		Could not be detected by RNA-based analysis	
	NF2:NM 000268: exon 15 deletion	Detected by I2HCF	copy numbe	rl PAT		
V39	NF2:NM_000268: exon 15 deletion	Detected by I2HCI	copy numbe	r PAT		
V39				r PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM_003073:c.1-117C>T	rs11090285	69,23	r PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM_003073:c.1-117C>T SMARCB1:NM_003073:c.363-154G>C	rs11090285 rs9608185	69,23 100	r PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM_003073:c.363-153C>T	rs11090285 rs9608185 rs9608186	69,23 100 98,33	r PAT	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G	rs11090285 rs9608185 rs9608186 rs738797	69,23 100 98,33 100	r PAT	Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T	rs11090285 rs9608185 rs9608186 rs738797 rs11090286	69,23 100 98,33 100 62,96	r PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030	69,23 100 98,33 100 62,96 100	r PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738	69,23 100 98,33 100 62,96 100 99,45	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032	69,23 100 98,33 100 62,96 100 99,45 50,79	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4	PAT	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65		Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+121G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89		Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+153A>C SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89		Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+18C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.987-176A>C	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9608192 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-139G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9608192 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267045 rs2267046 rs17003998	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs207393 rs2070458 rs2070459 rs2267045 rs2267045 rs2267045 rs2267045 rs2267045 rs2267045 rs2267045	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.897G>A SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.987-139G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267046 rs17003998 rs9612483 rs5030613	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-41G>A SMARCB1:NM 003073:c.629-130 629-130delC	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267045 rs17003998 rs9612483 rs5030613 rs34737764	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55 51,32	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+198T>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-164G>A	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267046 rs17003998 rs9612483 rs5030613	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-41G>A SMARCB1:NM 003073:c.629-130 629-130delC	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267045 rs17003998 rs9612483 rs5030613 rs34737764	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55 51,32	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-62A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.987-176A>C SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-41G>A SMARCB1:NM 003073:c.629-130 629-130delC SMARCB1:NM 003073:c.629-130 629-130delC SMARCB1:NM 003073:c.986+54_986+55insAA	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2073393 rs2070458 rs2070459 rs2229354 rs2267045 rs2267046 rs17003998 rs9612483 rs5030613 rs34737764 rs5844569	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55 51,32 44,27	Known	Could not be detected by RNA-based analysis	
V39	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.937-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-41G>A SMARCB1:NM 003073:c.986+54_986+55insAA SMARCB1:NM 003073:c.986+143 986+145delTCC	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2070459 rs2267045 rs2267045 rs2267045 rs17003998 rs9612483 rs5030613 rs34737764 rs5844569 rs34624937	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55 51.32 44,27 75	Known	Could not be detected by RNA-based analysis	
	SMARCB1:NM 003073:c.1-117C>T SMARCB1:NM 003073:c.363-154G>C SMARCB1:NM 003073:c.363-153C>T SMARCB1:NM 003073:c.500+234C>G SMARCB1:NM 003073:c.501-164C>T SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.501-87A>G SMARCB1:NM 003073:c.628+66G>C SMARCB1:NM 003073:c.628+118C>T SMARCB1:NM 003073:c.628+200T>C SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.629-248A>G SMARCB1:NM 003073:c.795+121G>A SMARCB1:NM 003073:c.795+153A>G SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.795+184A>T SMARCB1:NM 003073:c.937-176A>C SMARCB1:NM 003073:c.987-139G>A SMARCB1:NM 003073:c.1118+163G>T SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-164G>A SMARCB1:NM 003073:c.1119-41G>A SMARCB1:NM 003073:c.986+54_986+55insAA SMARCB1:NM 003073:c.986+143 986+145delTCC	rs11090285 rs9608185 rs9608186 rs738797 rs11090286 rs5760030 rs5751738 rs5760032 rs17003912 rs2073392 rs9608192 rs9612452 rs2070459 rs2267045 rs2267045 rs2267045 rs17003998 rs9612483 rs5030613 rs34737764 rs5844569 rs34624937	69,23 100 98,33 100 62,96 100 99,45 50,79 54,55 100 48,29 44,14 43,4 65 38,89 49,89 62,16 53,57 53,06 60 55 51.32 44,27 75	Known	Could not be detected by RNA-based analysis	

NF2:NM_000268:c.240+171T>A	rs2530662	42,11		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.241-85G>T	rs5763378	50,28		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.241-13T>A		50	PAT		
NF2:NM_000268:c.600-154G>A	rs2071622	49,44		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.886-205C>T	rs2252472	62,5		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.1123-51C>T	rs13055076	50,76		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.1575-67G>A	rs140086	99,6		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.1737+1623T>C	rs7291645	29,27		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.599+41_599+41delT	-	21.8		Could not be detected by RNA-based analysis	
NF2:NM_000268:c.1447-227_1447-226insTGAGGGA	rs3842713	27.03		Could not be detected by RNA-based analysis	

Table S5: Results from Validation set (n=40)

Sample	HC syndrome	Genes	Pathogenic Variant	Detected
V1	FAP	MUTYH	NM_012222.2:c.1187G>A; <u>c.481G>C</u>	YES
V2	FAP	APC	NM_001127510.2:c.1548+1G>C	YES
V3	HNPPC	MSH2	NM_000251.2: <u>c.518T>G</u>	YES
V4	НВОС	BRCA2	NM_000059.3:c.5350_5351delAAinsT	YES
V5	HNPPC	MLH1	NM_000249.3:c.2150_2153dup	YES
V6	HNPPC	PMS2	NM_000535:.5:c.943C>T	YES
V7	FAP	APC	NM_001127510.2:c.1312+3A>G	YES
V8	LI-FRAUMENI	TP53	NM_001126112.2:c.743G>A	YES
V9	HNPPC	MSH6	NM_000179.2:c.1618_1620delCTT	YES
V10	НВОС	BRCA1	NM_007294.2:c.3770_3771delAG	YES
V11	НВОС	BRCA1	Unknown disease-causing mutation	
V12	НВОС	BRCA2	NM_000059.3:c.1308_1309delGA	YES
V13	HNPPC	PMS2	NM_000535.5:c.59G>A	YES
V14	НВОС	BRCA2	NM_000059.3:c.3936_3954del(19bp)	YES
V15	FAP	APC	NM_001127510.2:c.2344A>T	YES
V16	FAP	MUTYH	NM_012222.2:c.1187G>A	YES
V17	НВОС	BRCA2	NM_000059.3:c.262_263delCT	YES
V18	НВОС	BRCA1	NM_007294.2:c.3869_3870delAA	YES
V19	HNPPC	MSH2	NM_000251.2:c.689_691delCTGinsTT	YES
V20	HNPPC	PMS2	NM_000535.5:c.780delC	YES
V21	HNPPC	MLH1	Unknown disease-causing mutation	
V22	НВОС	BRCA1	NM_007294.2:c.68_69delAG	YES
V23	НВОС	BRCA2	NM_000059.3:c1128delT	YES
V24	FAP	APC	NM_001127510.2:c.3183_3187delACAAA	YES
V25	HNPPC	MSH2	NM_000251.2:c.388_389delCA	YES
V26	НВОС	BRCA2	NM_000059.3:c.7230delT	YES

V27	HNPPC	MSH6	NM_000179.2:c.2906_2907del	YES
V28	НВОС	BRCA2	NM_000059.3:c.5722_5723delCT	YES
V29	НВОС	BRCA1	NM_007294.2:c.4107_4110dupATCT	YES
V30	НВОС	BRCA1	Unknown disease-causing mutation	
V31	FAP	APC	NM_001127510.2:c.1660C>T; <u>c.3264C>A</u>	YES
V32	HNPPC	MLH1	NM_000249.3:c.542delG	YES
V33	NF1	NF1	NM_000267.3: c.2957_2958insT	YES
V34	NF1	NF1	NM_000267.3:c.731-1G>C	YES
V35	NF1	NF1	NM_000267.3: whole gene deletion	YES
V36	NF1	NF1	NM_000267.3: c.1381C>T	YES
V37	NF1	NF1	NM_000267.3: c.278G>A	YES
V38	NF2	NF2	NM_000268.3: exon 15 deletion	YES
V39	SCHWANNOMATOSIS	SMARCB1	Unknown disease-causing mutation	
V40	NF2	NF2	NM_000268.3: c.241-13T>A	YES

Sheet1

			Sheeti		
Sample	HBOC FAP		HNPCC NF1	NF	-2
T1	3	0	2	2	2
T2	2	0	1	1	3
T3	2	0	1	1	3
T4	2	0	1	1	2
T5	2	0	1	1	2
T6	7	0	4	2	4
T7	2	0	1	1	1
T8	7	0	4	2	4
T9	1	0	0	1	1
T10	4	0	2	1	3
T11	10	1	6	4	4
T12	5	0	3	2	4
T13	1	0	0	1	1
T14	6	0	3	1	4
T15	3	0	2	1	2
T16	3	0	2	1	3
T17	18	5	8	6	9
T18	7	0	3	1	3
T19	3	0	2	1	3
T20	2	0	1	1	2
T21	10	0	4	2	4
T22	9	0	4	3	4
T23	5	0	3	1	3
V1	4	0	3	1	3
V2	8	1	5	3	4
V3	1	0	0	1	1
V4	4	0	2	1	3
V5	47	7	27	10	4
V6	2	0	1	1	2
V7	7	1	4	2	4
V8	17	5	11	2	5
V9	1	0	0	1	1
V10	2	0	1	1	1
V11	2	0	1	1	3
V12	2	0	1	1	3
V13	3	0	2	1	3
V14	2	0	2 1	1	2
V15	7	2	5	1	4
V16	7	1	4	1	4
V17	3	0		1	2
V18	3	0	2 2	1	2
V19	2	0	1	1	2

			Shee	et1	
V20	3	0	2	1	3
V21	2	0	1	1	3
V22	2	0	1	2	3
V23	3	0	2	1	2
V24	3	0	2	1	2
V25	11	1	5	3	4
V26	6	1	4	1	4
V27	5	0	3	1	3
V28	2	0	1	1	3
V29	5	0	3	1	2
V30	4	0	3	2	3
V31	5	0	2	2	3
V32	15	1	7	5	7
V33	31	7	17	6	10
V34	2	0	1	1	2
V35	2	0	1	1	3
V36	2	0	1	1	2
V37	2	0	1	1	2
V38	1	0	0	1	2
V39	8	1	4	1	3
V40	14	2	8	5	5

Resum_CNA_Panell_Abril2016

Gene	Samples	MLPA analysis (-)	I2HCP CNA analysis (-)	MLPA analysis (+)	12HCP CNA analysis	1
NF1	46	43	43	3	3	(whole gene dele
CDKN2A	6	1	1	5	5	
BRCA1	16	16	15 (1 undetermined)	0	0	
BRCA2	16	16	15 (1 undetermined)	0	0	
TP53	16	16	15 (1 undetermined)	0	0	
MUTYH	1	1	1	0	0	
MSH2	25	25	25	0	0	
MSH6	12	12	12	0	0	
MLH1	25	25	25	0	0	
PMS2	1	1	1	0	0	
CDH1	4	3	3	1 (exons 1-2 deleted)	1	(exon 2 deleted, є
TSC1	1	1	1	0	0	
TSC2	1	1	1	0	0	
APC	1	1	1	0	0	
TOTAL	171	162	159	9	9	

SUPPLEMENTARY TABLES AND FIGURES

<u>Table. S1</u>: List of tested genes. The table includes the genes tested for the five conditions with pre-defined groups. Red dots indicate genes tested in the pre-NGS workflows and black dots indicate genes included in the new gene sets. The list below the table includes the remaining genes present in the I2HCP panel.

<u>Table S2</u>: Training set results. Training set contains 23 samples previously genetically characterized. For each sample, gene analyzed and pathogenic mutation detected in each approach is indicated. In addition, for non-identified information on variant context is provided. Pathogenic mutations are colored depending on mutation type: <u>blue</u>: substitution, <u>light green</u>: small deletion or insertion; <u>dark green</u>: medium-sized deletion or insertion; <u>red</u>: big deletions. <u>HomoP</u> indicates variants are within a homopolymer region. <u>Low coverage</u> indicates coverage <30x.

<u>Table S3:</u> Sequencing and coverage statistics. Sequencing and coverage statistics of samples in the training (T) and validation (V) sets. Coverage statistics were calculated over the ROIs. Cx indicates the percentage of ROI covered by more than x reads. Ux indicates the uniformity, that is, the percentage of reads within +/- x% of the mean coverage.

<u>Table S4:</u> Concordance with previously detected variants. Variants detected in the training (T) and validation (V) sets by I2HCP second approach (V2) in genes previously analyzed by pre-NGS tests. For each variant, dbSNP identifier (rs), frequency, classification (previously known (Known), pathogenic (PAT) or variant of unknown significance (VUS)) are indicated. A short explanation for variants detected by only one method (pre-NGS analysis or I2HCP) is included. In addition, variants also detected by I2HCP first approach (V1) are indicated.

<u>Table S5:</u> Validation set results. Validation set contains 40 samples previously genetically characterized. For each sample, gene analyzed and pathogenic mutation detected are indicated. Variants are colored depending on mutation type: <u>blue</u>: substitution, <u>light green</u>: small deletion or insertion; <u>dark green</u>: medium-sized deletion or insertion; <u>red</u>: big deletions. <u>VUS</u> are underlined.

<u>Table S6</u>: Complementary Sanger sequencing tests required. The table shows the number of ROIs to be Sanger sequenced due to low coverage for each sample of the training (T) and validation (V) sets if tested for one of the five predefined groups of genes.

<u>Table S7</u>: Comparison of CNA analyses. One hundred seventy-one copy number alteration (CNA) analyses in 100 patients comprising 14 different genes were performed by MLPA and I2HCP exon-level copy-number calling algorithm. One sample, comprising 3 MLPAs, was non-assessable by I2HCP due to quality criteria.

<u>Figure S1</u>: Bait redesign. Example of one exon of *TP53* where bait redesign and rebalancing were applied. Y-axis indicates the depth of coverage of a region of the *TP53* gene (X-axis, reference genome) for different samples. Grey lines: samples using the first approach with V1 baits. Colored lines: samples from the second approach after bait redesign (V2 baits). Yellow and blue lines represent bait density in V1 and V2 (respectively) over the X-axis. A green doted line denotes the recurrent low coverage region of the *TP53* (grey lines going under the 30x depth coverage, denoted by a pale yellow line) that was clearly improved after bait redesign.

Figure S2: Examples of coverage per sample. Global coverage as in Figure 3c for 12 representative samples of the

validation set.

<u>Figure S3</u>: Regions with recurrent low coverage. A) Plot showing coverage and %GC content as in Figure 3b. Exons with coverage below 30x in at least 1bp in ≥75% of samples are highlighted in red. Detailed coverage of the two highlighted exons with highest mean coverage are shown. B) List of all highlighted exons in A.

Figure S4: Analysis pipeline. Diagram of the I2HCP data analysis pipeline.

<u>Figure S5:</u> New genetic testing workflow solved uncertain clinical diagnosis. A patient with clinical suspicion of Neurofibromatosis Type 1 (3 neurofibromas, 1 glioblastoma multiforme, 1 malignant fibrous histiocytoma) (a) was analyzed with the I2HCP. A deletion in *CDKN2A* gene was detected by the exon-level copy-number calling algorithm. (b). Grey dots represent the normalized mean depth of coverage of 27 ROIs close to *CDKN2A* in chromosome 9; black dots represent normalized patient's sample coverage of the same ROIs and red dots indicate *CDKN2A* deleted exons. (c). MLPA analysis validated the loss of *CDKN2A*. Deletion of the whole *CDKN2B* gene was also detected, a locus that was not included in I2HCP. Red dots indicate the deleted region evidenced by MLPA analysis).

<u>Figure S6:</u> Frequency of pathogenic variants. The graph depicts the frequency of all pathogenic variants present in the training and validation sets. SNV: single nucleotide variant.