Table S1 Mutation details

Case No.	Gene	cDNA	#rs	MAF	SIFT	Polyphen-2	PROVEN	FATHMM	MutationTaster	MutationAssessor	ClinVar	LOVD	Comment
AA_018	BRCA2	c.5718_5719delCT/p.L1908Rfs*2	rs80359530	—	—	—	—	—	D	—	Pathogenic	Pathogenic	
AA_044	BRCA2	c.6879T>G/p.F2293L	rs1381512588	—	D	D	N	T	D	M	—	Pathogenic	
AA_049	BRCA2	c.572A>T/p.D191V	rs397507798	—	D	D	N	T	D	M	Uncertain significance	—	
AA_011	FANCA	c.530T>C/p.L177S	—	—		D	D	T	N		—	—	Novel
AA_008	FANCA	c.1874G>A/p.C625Y	rs139235751	0.0004	D	D	D	D	D	M	Uncertain significance	—	
AA_030	FANCA	c.1906G>C/p.A636P	rs1236146369	—	D	P	N	D	N	M	—	—	
AA_041	FANCA	c.205T>A/p.C69S	—	—	T	P	D	T	N	M	—	—	Novel
AA_056	FANCA	c.1328C>T/p.S443F	rs367733447	0	T	B	D	D	N	M	—	—	
AA_142	FANCA	c.2873C>T/p.A958V	rs1374262828	—	D	D	N	D	N	M	—	—	
AA_143	FANCA	c.2731T>C/p.W911R	—	—	D	D	D	D	D	M	—	—	Novel
AA_148	FANCA	c.83G>A/p.G28E	—	—	D	D	D	D	D	M	—	—	Novel
AA_233	FANCA	c.1682C>T/p.T561M	rs148154682	0	D	D	D	D	D	M	Uncertain significance	—	
AA_165	FANCC	c.1474C>T/p.L492F	—	—	D	D	N	T	N	M	—	—	Novel
AA_171	FANCC	c.436_438del/p.Y146del	rs761347179	0.0006	—	—	—	—	D	—	Uncertain significance	—	Recurrent
AA_199	FANCC	c.239T>C/p.I80T	rs4647419	0.0006	D	P	D	T	N	M	Uncertain significance	—	
AA_006	FANCD2	c.517C>T/p.P173S	—	—	D	D	D	T	D	M	—	—	Novel
AA_026	FANCD2	c.3973C>A/p.L1325M	rs555539811	0.0002	T	D	N	T	D	M	Uncertain	—	Recurrent

										significance		
AA_041	FANCD2	c.2885C>T/p.P962L	—	—	T	D	D	T	D	M	—	—	Novel
AA_222	FANCG	c.1144-1G>A	—	—	—	—	—	—	D	—	—	—	Novel
MDS_23	BRCA2	c.9898C>T/p.P3300S	rs770868371	0.0001	D	D	N	T	D	M	Uncertain significance	VUS	
MDS_16	BRCA2	c.1905T>A/p.D635E	—	—	D	P	N	T	N	M	Uncertain significance	—	
MDS_05	FANCA	c.1379G>A/p.R460Q	—	—	T	P	N	D	N	M	—	—	Novel
MDS_42	FANCA	c.1072C>G/p.L358V	rs748337423	0.0002	D	D	N	D	N	M	Uncertain significance	—	
MDS_55	FANCA	c.3653C>T/p.P1218L	—	—	D	B	D	D	N	M	—	—	Novel
MDS_14	FANCC	c.1182G>A/p.W394X	—	—	—	—	—	—	—	A	Likely pathogenic	—	
MDS_55	FANCD2	c.2351C>T/p.S784F	—	—	D	D	D	T	D	M	—	—	Novel
AML_037	BRCA2	c.8825C>T/p.A2942V	—	—	D	D	N	T	D	M	Uncertain significance	—	
AML_076	BRCA2	c.904dup/p.T302Nfs*3	—	—	—	—	—	.	.	.	—	Pathologic	
AML_120	BRCA2	c.5070A>C/p.K1690N	rs56087561	0	D	P	D	D	N	M	Benign	benign, VUS, likely benign	
AML_171	BRCA2	c.5218_5223del/p.1740_1741del	rs397507775	0.0002	.	.	.	.	D	.	—	Pathogenic	
AML_183	BRCA2	c.602C>T/p.P201L	—	—	D	D	D	T	D	M	Uncertain significance	—	
AML_019	FANCA	c.1874G>A/p.C625Y	rs139235751	0.0004	D	D	D	D	D	M	Uncertain significance	—	
AML_043	FANCA	c.2171C>T/p.T724M	rs777032467	0	D	B	D	D	N	L	—	—	
AML_054	FANCA	c.3046G>T/p.D1016Y	—	—	D	D	D	D	N	M	—	Pathologic	
AML_068	FANCA	c.1289C>T/p.A430V	rs772567344	0.0001	T	B	D	D	D	L	Uncertain		

											significance		
AML_091	FANCA	c.1196G>A/p.C399Y	—	—	D	P	D	T	D	M	—	—	Novel
AML_099	FANCA	c.1237C>T/p.R413C	rs780135578	0.0002	D	B	D	D	N	L	—	—	
AML_135	FANCA	c.3538G>T/p.V1180L	rs372706571	—	T	B	N	D	D	M	—	—	
AML_198	FANCA	c.2471G>A/p.C824Y	—	—	T	B	D	D	N	M	—	—	Novel
AML_205	FANCA	c.2582C>T/p.S861F	—	—	D	P	D	D	N	M	Uncertain significance	VUS	
AML_132	FANCC	c.1424C>T/p.T475M	rs41281200	0	B	.	.	.	N	.	Benign	—	
AML_177	FANCC	c.329T>C/p.L110P	rs77773156+D360	0	T	D	N	T	D	M	Uncertain significance	—	
AML_040	FANCD2	c.3973C>A/p.L1325M	rs555539811	0.02%	T	D	N	T	D	M	Uncertain significance	—	Recurrent
AML_100	FANCD2	c.418C>G/p.L140V	—	—	D	D	N	T	D	M	—	—	Novel
AML_139	FANCG	c.366G>C/p.W122C	rs546023787	0	D	D	D	D	D	M	Likely benign	—	
ALL_031	BRCA2	c.8351G>A/p.R2784Q	rs80359076	0	D	D	N	D	D	M	Conflict	—	
ALL_033	BRCA2	c.5848_5851delGTTA/p.S1951WfsX11	rs80359543	—	.	.	.	.	D	.	Pathogenic	—	
ALL_034	BRCA2	c.247G>A/p.E83K	—	—	D	B	N	T	D	M	—	—	Novel
ALL_093	BRCA2	c.7544C>T/p.T2515I	rs28897744	0	T	P	N	D	N	M	Benign	—	
ALL_159	BRCA2	c.8525G>A/p.R2842H	rs80359105	0.0006	D	D	N	D	D	M	Benign	—	
ALL_175	BRCA2	c.6092C>T/p.T2031I	—	—	D	P	D	T	N	M	—	—	Novel
ALL_179	BRCA2	c.1588_1591delinsTTT/p.K530FfsX28	—	—	.	.	.	.	D	.	—	—	Novel
ALL_190	BRCA2	c.4258G>T/p.D1420Y	rs28897727	0.01%	D	B	D	T	N	M	Benign	—	
ALL_209	BRCA2	c.8356G>A/p.A2786T	rs80359077	0.08%	D	P	N	D	D	M	Conflict	—	
ALL_253	BRCA2	c.2990T>G/p.L997X	—	—	.	.	.	.	A	.	Pathogenic	—	
ALL_002	FANCA	c.1634G>T/p.S545I	rs200922390	0.0007	D	D	D	D	D	M	—	—	
ALL_074	FANCA	c.1675G>A/p.E559K	rs201323171	0.0005	T	P	N	D	D	M	Uncertain	—	

										significance		
ALL_080	FANCA	c.596+5C>T	rs758345423	0.0005	.	.	.	.	.	Uncertain significance	—	
ALL_090	FANCA	c.3328C>T/p.H1110Y	—	—	T	B	D	D	N	M	—	—	Novel
ALL_100	FANCA	c.2882A>G/p.E961G	—	—	D	P	N	D	N	M	—	—	Novel
ALL_198	FANCA	c.2760G>T/p.L920F	—	—	D	D	D	D	N	M	—	—	Novel
ALL_216	FANCA	c.1827-1G>C	—	—	.	.	.	.	D	.	—	—	
ALL_240	FANCA	c.457C>T/p.Q153X	—	—	.	.	.	.	A	.	—	—	Novel
ALL_185	FANCC	c.436_438del/p.Y146del	rs761347179	0.06%	.	.	.	.	D	Uncertain significance	—	Recurrent
ALL_178	FANCD2	c.2777G>A/p.R926Q	rs532250395	0.02%	D	D	D	T	D	M	—	—	
ALL_181	FANCD2	c.3973C>A/p.L1325M	rs555539811	0.02%	T	D	N	T	D	M	Uncertain significance	—	Recurrent
ALL_252	FANCD2	c.3670C>T/p.P1224S	—	—	T	D	D	T	D	M	—	—	Novel
ALL_169	FANCG	c.420C>A/p.H140Q	rs764566651	0.09%	D	D	D	T	D	M	—	—	