Table S3. Disease associated genotypes identified by exome sequencing in 136 out of 142 FA patients

Sample ID	Gene	DNA change	Type of mutation	cDNA change	Amino acid change	dbSNP ID	Zygosity	Varsome Results	EVE Results
FA-01	*FANCG*	NC_000009.11:g.35074215T>G	3' splice site mutation	NM_004629.2:c.1761-2A>C	-	rs765150956	Homozygous	Pathogenic	NA
FA-02	*FANCJ (BRIP1)*	NC_000017.10:g.59857679T>A	Missense	NM_032043.3:c.1878A>T	p.Glu626Asp	rs1567812484	Homozygous	Pathogenic	Pathogenic
FA-03	*FANCA*	NC_000016.9:g.89813237A>C	5' splice site mutation	NM_000135.4:c.3408+2T>G	-	Novel	Compound heterozygous	VUS	NA
		NC_000016.9:g.89828423T>G	Missense	NM_000135.4:c.2786A>C	p.Tyr929Ser	Novel		VUS	Pathogenic
FA-04	*FANCA*	NC_000016.9:g.89882396T>C	3' splice site mutation	NM_000135.4:c.80-2A>G	-	Novel	Compound heterozygous	VUS	NA
		NC_000016.9:g.89811367_89865640del	Deletion	NM_000135.4:c.827_3626del	-	Novel		Likely pathogenic	NA
FA-05	*FANCA*	NC_000016.9:g.89857866C>T	Missense	NM_000135.4:c.1304G>A	p.Arg435His	rs1060501879	Homozygous	Pathogenic	Pathogenic
FA-06	*FANCA*	NC_000016.9:g.89828423T>G	Missense	NM_000135.4:c.2786A>C	p.Tyr929Ser	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-07	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-08	*FANCA*	NC_000016.9:g.89805365dup	Frameshift insertion	NM_000135.4:c.4185dupG	p.Ile1396AspfsTer29	Novel	Homozygous	Pathogenic	NA
FA-09	*FANCA*	NC_000016.9:g.89877340_89877341del	5' splice site mutation	NM_000135.4:c.426_426+1delAG	K143Rfs*7/5' splice variant	rs763114336	Compound heterozygous	VUS	NA
		NC_000016.9:g.89877448del	Frameshift deletion	NM_000135.4:c.319delG	p.Val107PhefsTer31	rs1411237340		VUS	NA
FA-10	*FANCA*	NC_000016.9:g.89809218_89809219del	Frameshift deletion	NM_000135.4:c.3761_3762delAG	p.Glu1254GlyfsTer23	rs868273545	Homozygous	Pathogenic	NA
FA-11	*FANCA*	NC_000016.9:g.89811367_89816310del	Deletion	NM_000135.4:c.3067_3626del	-	Novel	Homozygous	Pathogenic	NA
FA-12	*FANCA*	NC_000016.9:g.89809286_89809290dup	Frameshift insertion	NM_000135.4:c.3690_3694dupGCACT	p.Phe1232CysfsTer17	Novel	Compound heterozygous	Likely pathogenic	NA
		NC_000016.9:g.89857867G>A	Missense	NM_000135.4:c.1303C>T	p.Arg435Cys	rs148473140		Likely pathogenic	Pathogenic
FA-13	*FANCG*	NC_000009.11:g.35077267_35077273del	Frameshift deletion	NM_004629.2:c.637_643delTACCGCC	p.Tyr213LysfsTer6	rs587776640	Homozygous	Pathogenic	NA
FA-15	*FANCA*	NC_000016.9:g.89851261C>T	Exonic splice donor variant	NM_000135.4:c.1470+1G>A	p.Lys369Lys	rs1555556175	Homozygous	Pathogenic	NA
FA-16	*FANCA*	NC_000016.9:g.89833551_89833647del	Deletion	NM_000135.4:c.2505_2601del	p.Lys835SerfsTer22	George et al. 2021	Homozygous	Pathogenic	NA
FA-18	*FANCA*	NC_000016.9:g.89828357C>G	Missense	NM_000135.4:c.2852G>C	p.Arg951Pro	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-20	*FANCG*	NC_000009.11:g.35074215T>G	3' splice site mutation	NM_004629.2:c.1761-2A>C	-	rs765150956	Homozygous	Pathogenic	NA
FA-21	*FANCA*	NC_000016.9:g.89807212_89816310del	Deletion	NM_000135.4:c.3067_3828del	-	Novel	Homozygous	Pathogenic	NA
FA-22	*FANCA*	NC_000016.9:g.89866011A>G	5' splice site mutation	NM_000135.4:c.826+2T>C	-	Novel	Homozygous	Pathogenic	NA
FA-23	*FANCA*	NC_000016.9:g.89818545C>A	5' splice site mutation	NM_000135.4:c.3066+1G>T	-	rs587783028	Homozygous	Pathogenic	NA
FA-24	*FANCA*	NC_000016.9:g.89809302C>T	Nonsense	NM_000135.4:c.3671G>A	p.Trp1224Ter	Novel	Compound heterozygous	Likely pathogenic	NA
		NC_000016.9:g.89828358_89842224del	Deletion	NM_000135.4:c.1827_2852del	-	Castella et al. 2011		Likely pathogenic	NA
FA-25	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-26	*FANCG*	NC_000009.11:g.35076427_35076431del	5' intronic splice donor variant (+2)	NM_004629.2:c.1076+3_1076+7delGAGGT	-	rs780410457	Homozygous	VUS	NA
FA-27	*FANCA*	NC_000016.9:g.89818545C>A	5' splice site mutation	NM_000135.4:c.3066+1G>T	-	rs587783028	Homozygous	Pathogenic	NA
FA-28	*FANCA*	NC_000016.9:g.89882944C>G	5' splice site mutation	NM_000135.4:c.79+1G>C	-	rs1483028018	Homozygous	Pathogenic	NA
FA-30	*FANCF*	NC_000011.9:g.22646233T>C	Stop loss	NM_022725.4:c.1124A>G	p.Ter375TrpextTer33	Novel	Homozygous	VUS	NA
FA-31	*FANCA*	NC_000016.9:g.89882944C>G	5' splice site mutation	NM_000135.4:c.79+1G>C	-	rs1483028018	Homozygous	Pathogenic	NA
FA-32	*FANCA*	NC_000016.9:g.89824984C>G	5' splice site mutation	NM_000135.4:c.2981+1G>C	-	Novel	Homozygous	Pathogenic	NA
FA-33	*FANCA*	NC_000016.9:g.89813247dup	Frameshift insertion	NM_000135.4:c.3401dupT	p.Phe1135LeufsTer80	Novel	Homozygous	Pathogenic	NA
FA-34	*FANCA*	NC_000016.9:g.89818545C>A	5' splice site mutation	NM_000135.4:c.3066+1G>T	-	rs587783028	Homozygous	Pathogenic	NA
FA-35	*FANCA*	NC_000016.9:g.89806416del	Frameshift deletion	NM_000135.4:c.3920delA	p.Gln1307ArgfsTer2	rs1228394297	Homozygous	Pathogenic	NA
FA-36	*FANCG*	NC_000009.11:g.35075650_35075651del	Frameshift deletion	NM_004629.2:c.1246_1247delCT	p.Leu416MetfsTer2	Novel	Homozygous	Pathogenic	NA
FA-38	*FANCG*	NC_000009.11:g.35078223A>G	Missense	NM_004629.2:c.425T>C	p.Leu142Pro	Novel	Compound heterozygous	VUS	Pathogenic
		NC_000009.11:g.35076856_35076857del	Frameshift deletion	NM_004629.2:c.792_793delAG	p.Arg264SerfsTer24	Novel		VUS	NA
FA-40	*FANCA*	NC_000016.9:g.89877448del	Frameshift deletion	NM_000135.4:c.319delG	p.Val107PhefsTer31	rs1411237340	Homozygous	Pathogenic	NA
FA-511	*FANCA*	NC_000016.9:g.89831327G>A	Nonsense	NM_000135.4:c.2749C>T	p.Arg917Ter	rs1060501880	Homozygous	Pathogenic	NA
FA-513	*FANCA*	NC_000016.9:g.89880929_89883055del	Deletion	NM_000135.4:c.-31_283del	-	Savoia et al. 1996	Homozygous	Pathogenic	NA
FA-516	*FANCA*	NC_000016.9:g.89825022del	Frameshift deletion	NM_000135.4:c.2944delA	p.Thr982ProfsTer7	Novel	Homozygous	Pathogenic	NA
FA-517	*FANCC*	NC_000009.11:g.97873912C>A	Nonsense	NM_000136.3:c.1162G>T	p.Gly388Ter	rs371897078	Homozygous	Pathogenic	NA
FA-521	*FANCA*	NC_000016.9:g.89877481T>G	3' splice site mutation	NM_000135.4:c.284-2A>C	-	rs756023006	Homozygous	Pathogenic	NA
FA-522	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-523	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-525	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-527	*FANCC*	NC_000009.11:g.97864081T>G	Missense	NM_000136.3:c.1585A>C	p.Thr529Pro	rs587778326	Homozygous	VUS	Pathogenic
FA-529	*FANCA*	NC_000016.9:g.89833551_89833647del	Deletion	NM_000135.4:c.2505_2601del	p.Lys835SerfsTer22	George et al. 2021	Homozygous	Pathogenic	NA
FA-531	*FANCG*	NC_000009.11:g.35076026C>G	3' splice site mutation	NM_004629.2:c.1077-1G>C	-	Novel	Homozygous	Pathogenic	NA
FA-532	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-533	*FANCA*	NC_000016.9:g.89858399_89858400del	Frameshift deletion	NM_000135.4:c.1164_1165delAG	p.Arg388SerfsTer20	Novel	Compound heterozygous	VUS	NA
		NC_000016.9:g.89806402C>T	Missense	NM_000135.4:c.3934G>A	p.Asp1312Asn	Novel		VUS	Pathogenic
FA-534	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-535	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-538	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-542	*FANCA*	NC_000016.9:g.89866011A>G	5' splice site mutation	NM_000135.4:c.826+2T>C	-	Novel	Homozygous	Pathogenic	NA
FA-543	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-544	*FANCA*	NC_000016.9:g.89858878C>T	5' splice site mutation	NM_000135.4:c.1083+1G>A	-	Novel	Homozygous	Pathogenic	NA
FA-547	*FANCA*	NC_000016.9:g.89845356G>A	Nonsense	NM_000135.4:c.1771C>T	p.Arg591Ter	rs753980264	Homozygous	Pathogenic	NA
FA-548	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-549	*FANCA*	NC_000016.9:g.89828423T>G	Missense	NM_000135.4:c.2786A>C	p.Tyr929Ser	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-554	*FANCA*	NC_000016.9:g.89857810C>G	5' Splice site mutation	NM_000135.4:c.1359+1G>C	-	rs1555561294	Compound heterozygous	Likely pathogenic	NA
		NC_000016.9:g.89803957_89806507del	Deletion	NM_000135.4:c.3829_*1052del	-	Novel		Likely pathogenic	NA
FA-556	*FANCA*	NC_000016.9:g.89806405del	Frameshift deletion	NM_000135.4:c.3931delA	p.Ser1311ValfsTer52	Novel	Homozygous	Pathogenic	NA
FA-557	*FANCA*	NC_000016.9:g.89882943A>G	5' splice site mutation	NM_000135.4:c.79+2T>C	-	rs1319795682	Homozygous	Pathogenic	NA

FA-561	*FANCA*	NC_000016.9:g.89833551_89833647del	Deletion	NM_000135.4:c.2505_2601del	p.Lys835SerfsTer22	George et al. 2021	Homozygous	Pathogenic	NA
FA-564	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-565	*FANCA*	NC_000016.9:g.89883009C>T	Nonsense	NM_000135.4:c.15G>A	p.Trp5Ter	Novel	Homozygous	Pathogenic	NA
FA-568	*FANCA*	NC_000016.9:g.89828423T>G	Missense	NM_000135.4:c.2786A>C	p.Tyr929Ser	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-629/18	*FANCG*	NC_000009.11:g.35075954C>G	5' intronic splice donor variant (+5)*	NM_004629.2:c.1143+5G>C	-	rs778328620	Homozygous	Pathogenic	NA
FA-631/18	*FANCA*	NC_000016.9:g.89831438G>A	Nonsense	NM_000135.4:c.2638C>T	p.Arg880Ter	rs762804216	Compound heterozygous	Pathogenic	NA
		NC_000016.9:g.89811367_89816310del	Deletion	NM_000135.4:c.3067_3626del	-	Novel		Pathogenic	NA
FA-636/18	*UBE2T/FANCT*	NC_000001.10:g.202300964_202301088del	Deletion	NM_014176.4:c.470_594del	p.Ala157GlyfsTer18	Novel	Homozygous	Pathogenic	NA
FA-637/18	*FANCJ (BRIP1)*	NC_000017.10:g.59885995G>A	Missense	NM_032043.3:c.751C>T	p.Arg251Cys	rs752309409	Homozygous	Likely pathogenic	Pathogenic
FA-638/18	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-641/18	*FANCF*	NC_000011.9:g.22647316A>C	Missense	NM_022725.4:c.41T>G	p.Leu14Arg	Novel	Homozygous	VUS	VUS
FA-646/18	*FANCA*	NC_000016.9:g.89828357C>T	Missense	NM_000135.4:c.2852G>A	p.Arg951Gln	rs755922289	Homozygous	Likely pathogenic	Pathogenic
FA-650/18	*FANCG*	NC_000009.11:g.35079204dup	Frameshift insertion	NM_004629.2:c.119dupA	p.Gln41AlafsTer16	Novel	Homozygous	Pathogenic	NA
FA-652/18	*FANCA*	NC_000016.9:g.89816214G>A	Missense	NM_000135.4:c.3163C>T	p.Arg1055Trp	rs753063086	Homozygous	Likely pathogenic	Pathogenic
FA-659/18	*FANCA*	NC_000016.9:g.89809284A>G	Missense	NM_000135.4:c.3689T>C	p.Leu1230Pro	Novel	Homozygous	VUS	Pathogenic
FA-660/18	*FANCA*	NC_000016.9:g.89865641C>T	3' splice site mutation	NM_000135.4:c.827-1G>A	-	rs753728435	Homozygous	Pathogenic	NA
FA-665/18	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-672/18	*FANCA*	NC_000016.9:g.89811367_89816310del	Deletion	NM_000135.4:c.3067_3626del	-	Novel	Homozygous	Pathogenic	NA
FA-674/18	*FANCA*	NC_000016.9:g.89816214G>A	Missense	NM_000135.4:c.3163C>T	p.Arg1055Trp	rs753063086	Compound heterozygous	Pathogenic	Pathogenic
		NC_000016.9:g.89811367_89865640del	Deletion	NM_000135.4:c.827_3626del	-	Novel		Pathogenic	NA
FA-675/18	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Compound heterozygous	Pathogenic	NA
		NC_000002.11:g.58388695G>A	Nonsense	NM_001114636.1:c.997C>T	p.Gln333Ter	rs776298788		Pathogenic	NA
FA18/20	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA01/19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FP-23-P-11	*FANCA*	NC_000016.9:g.89828423T>G	Missense	NM_000135.4:c.2786A>C	p.Tyr929Ser	Novel	Homozygous	VUS	Pathogenic
FA-17/19	*FANCA*	NC_000016.9:g.89871689_89871801del	Deletion	NM_000135.4:c.597_709del	p.Ser199GlyfsTer24	Esmail nia et al. 2016	Homozygous	Pathogenic	NA
FA-18/19	*FANCA*	NC_000016.9:g.89871689_89871801del	Deletion	NM_000135.4:c.597_709del	p.Ser199GlyfsTer24	Esmail nia et al. 2016	Homozygous	Pathogenic	NA
FA-20/19	*FANCA*	NC_000016.9:g.89877481T>G	3' splice site mutation	NM_000135.4:c.284-2A>C	-	rs756023006	Homozygous	Pathogenic	NA
FA-27/19	*FANCA*	NC_000016.9:g.89866011A>G	5' splice site mutation	NM_000135.4:c.826+2T>C	-	Novel	Homozygous	Pathogenic	NA
FA-30/19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Likely pathogenic	NA
FA-36/19	*FANCG*	NC_000009.11:g.35074215T>G	3' splice site mutation	NM_004629.2:c.1761-2A>C	-	rs765150956	Homozygous	Pathogenic	NA
FA-37/19	*FANCG*	NC_000009.11:g.35074215T>G	3' splice site mutation	NM_004629.2:c.1761-2A>C	-	rs765150956	Homozygous	Pathogenic	NA
FA-49/19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-573	*FANCA*	NC_000016.9:g.89877448del	Frameshift deletion	NM_000135.4:c.319delG	p.Val107PhefsTer31	rs1411237340	Compound heterozygous	Pathogenic	NA
		NC_000016.9:g.89883022A>T	Missense	NM_000135.4:c.2T>A	p.Met1Lys	rs769479800		Pathogenic	NA
FA-580	*FANCG*	\C_000009.11:g.35078222_35078223delinsAGCAGT	Frameshift insertion	NM_004629.2:c.425_426delTGinsAACTGCT	p.Leu142GlnfsTer12	Novel	Homozygous	Pathogenic	NA
FA-581	*FANCA*	NC_000016.9:g.89809216_89809219del	Frameshift deletion	NM_000135.4:c.3759_3762del	p.Glu1254SerfsTer11	Novel	Homozygous	Pathogenic	NA
FA-584	*FANCA*	NC_000016.9:g.89877327_89877344del	Frameshift deletion	NM_000135.4:c.423_426+14del	-	Novel	Homozygous	Pathogenic	NA
FA-588	*FANCG*	NC_000009.11:g.35074215T>G	3' splice site mutation	NM_004629.2:c.1761-2A>C	-	rs765150956	Homozygous	Pathogenic	NA
FA-591	*FANCA*	NC_000016.9:g.89866011A>G	5' splice site mutation	NM_000135.4:c.826+2T>C	-	Novel	Homozygous	Pathogenic	NA
FA-595	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-598	*FANCG*	NC_000009.11:g.35075059G>A	Nonsense	NM_004629.2:c.1501C>T	p.Gln501Ter	Novel	Homozygous	Pathogenic	NA
FA-599	*FANCA*	NC_000016.9:g.89877327_89877344del	Frameshift deletion	NM_000135.4:c.423_426+14del	-	Novel	Homozygous	Pathogenic	NA
FA-601	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-614	*FANCA*	NC_000016.9:g.89849270_89849510del	Deletion	NM_000135.4:c.1472_1624del	-	Neil V morgan et al. 1999	Homozygous	Pathogenic	NA
O-117	*FANCA*	NC_000016.9:g.89874703_89877479del	Deletion	NM_000135.4:c.285_596del	-	Savoia et al. 1996	Homozygous	Pathogenic	NA
O-123	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
O-126	*FANCA*	NC_000016.9:g.89862316_89862426del	Deletion	NM_000135.4:c.894_1004del	p.Trp298_Lys335delinsGly	Solanki et al. 2016	Homozygous	Pathogenic	NA
P-177	*FANCA*	NC_000016.9:g.89813297C>G	Missense	NM_000135.4:c.3350G>C	p.Arg1117Thr	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-12/19	*FANCA*	NC_000016.9:g.89877448del	Frameshift deletion	NM_000135.4:c.319delG	p.Val107PhefsTer31	rs1411237340	Compound heterozygous	Pathogenic	NA
		NC_000016.9:g.89882396T>C	3' splice site mutation	NM_000135.4:c.80-2A>G	-	Novel		Pathogenic	NA
FA-622	*FANCA*	NC_000016.9:g.89818552_89825113del	Deletion	NM_000135.4:c.2853_3060del	-	Novel	Compound heterozygous	Pathogenic	NA
		NC_000016.9:g.89816231_89816232del	Frameshift deletion	NM_000135.4:c.3146_3147delTT	p.Phe1049Ter	Novel		Pathogenic	NA
FA-593	*FANCA*	NC_000016.9:g.89828358G>A	Missense	NM_000135.4:c.2851C>T	p.Arg951Trp	rs755546887	Homozygous	Likely pathogenic	Pathogenic
FA-592	*FANCA*	NC_000016.9:g.89845258_89882945del	Deletion	NM_000135.4:c.79_1777del	-	Castella et al. 2011	Compound heterozygous	Pathogenic	NA
		NC_000016.9:g.89833551_89833647del	Deletion	NM_000135.4:c.2505_2601del	p.Lys835SerfsTer22	George et al. 2021		Pathogenic	NA
FA-03/19	*FANCI*	NC_000015.9:g.89804822del	Frameshift deletion	NM_001113378.2:c.295delC	p.His99IlefsTer10	rs759398314	Compound heterozygous	Pathogenic	NA
		NC_000015.9:g.89848828_89848832del	Intronic splice acceptor variant	NM_001113378.2:c.3256-8_3256-4delTCTTT	-	Novel		VUS	NA
P-603	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-04/20	*FANCA*	NC_000016.9:g.89877448del	Frameshift deletion	NM_000135.4:c.319del	p.Val107PhefsTer31	rs1411237340	Homozygous	Pathogenic	NA
FA-06/20	*FANCA*	NC_000016.9:g.89805352G>A	Missense	NM_000135.4:c.4198C>T	p.Arg1400Cys	rs745882980	Homozygous	Pathogenic	Pathogenic
FA-10/20	*FANCA*	NC_000016.9:g.89805352G>A	Missense	NM_000135.4:c.4198C>T	p.Arg1400Cys	rs745882980	Homozygous	Pathogenic	Pathogenic
FA-10/21	*FANCA*	NC_000016.9:g.89809216_89809219del	Frameshift deletion	NM_000135.4:c.3759_3762del	p.Glu1254SerfsTer11	Novel	Homozygous	Pathogenic	NA
FA-11/20	*FANCA*	NC_000016.9:g.89806411_89806427dup	Frameshift insertion	NM_000135.4:c.3909_3925dup	p.Thr1309ArgfsTer6	Novel	Homozygous	Pathogenic	NA
FA-15/20	*FANCG*	NC_000009.11:g.35077289del	Frameshift deletion	NM_004629.2:c.619del	p.Leu207SerfsTer2	Novel	Homozygous	Pathogenic	NA
FA-16/20	*FANCA*	NC_000016.9:g.89818545C>A	5' splice site mutation	NM_000135.4:c.3066+1G>T	-	rs587783028	Homozygous	Pathogenic	NA
FA-21/20	*FANCA*	NC_000016.9:g.89805660C>A	Nonsense	NM_000135.4:c.4048G>T	p.Glu1350Ter	Novel	Homozygous	Pathogenic	NA
FA-5/21	*UBE2T/FANCT*	NC_000001.10:g.202302631T>G	Missense	NM_014176.4:c.232A>C	p.Asn78His	rs776219033	Homozygous	VUS	Pathogenic
FA-01/20	*FANCG*	NC_000009.11:g.35074476dup	Nonsense	NM_004629.2:c.1652dup	p.Tyr551Ter	Novel	Homozygous	Pathogenic	Pathogenic
FA-02/21	*FANCA*	NC_000016.9:g.89807252A>G	Missense	NM_000135.4:c.3788T>C	p.Phe1263Ser	George et al. 2021	Compound heterozygous	Likely pathogenic	VUS
		NC_000016.9:g.89849441C>T	Missense	NM_000135.4:c.1540G>A	p.Ala514Thr	Novel		VUS	Pathogenic

FA-08/19	*FANCA*	NC_000016.9:g.89833551_89833647del	Deletion	NM_000135.4:c.2505_2601del	p.Lys835SerfsTer22	George et al. 2021	Homozygous	Pathogenic	NA
FA-13/19	*FANCA*	NC_000016.9:g.89831446G>C	Nonsense	NM_000135.4:c.2630C>G	p.Ser877Ter	Solanki et al. 2016	Homozygous	Likely pathogenic	NA
FA-18/21	*FANCA*	NC_000016.9:g.89836600G>A	Missense	NM_000135.4:c.2290C>T	p.Arg764Trp	De Rocco et al. 2014	Homozygous	Pathogenic	Pathogenic
FA-21/21	*FANCA*	NC_000016.9:g.89816138C>T	Missense	NM_000135.4:c.3239G>A	p.Arg1080Gln	rs1555538571	Homozygous	Likely pathogenic	Pathogenic
FA-23/19	*FANCA*	NC_000016.9:g.89836973T>G	Exonic splice donor variant	NM_000135.4:c.2221A>C	p.Arg741=	Novel	Homozygous	Likely pathogenic	NA
FA-26/21	*FANCA*	NC_000016.9:g.89809211_89809212insCT	Frameshift insertion	NM_000135.4:c.3761_3762insAG	p.Glu1255GlyfsTer12	rs868273545	Homozygous	Pathogenic	NA
FA-30/21	*UBE2T/FANCT*	NC_000001.10:g.202300964_202301088del	Deletion	NM_014176.4:c.470_594del	p.Ala157GlyfsTer18	Novel	Homozygous	Pathogenic	NA
FA-31/21	*FANCA*	NC_000016.9:g.89851302A>G	Missense	NM_000135.4:c.1430T>C	p.Leu477Ser	Novel	Homozygous	Pathogenic	Pathogenic
FA-33/19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-33/21	*FANCA*	NC_000016.9:g.89809211_89809212insCT	Frameshift insertion	NM_000135.4:c.3761_3762insAG	p.Glu1255GlyfsTer12	rs868273545	Homozygous	Pathogenic	NA
FA-34/19	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA
FA-35/21	*FANCA*	NC_000016.9:g.89805365dup	Frameshift insertion	NM_000135.4:c.4185dup	p.Ile1396AspfsTer29	Novel	Homozygous	Pathogenic	NA
FA-649/18	*BRCA2*	NC_000013.10:g.32893238G>C	Missense	NM_000059.4:c.92G>C	p.Trp31Ser	Caleca et al. 2018	Homozygous	Likely pathogenic	NA
FA-12/22	*FANCL*	NC_000002.11:g.58387243C>T	Exonic splice donor variant	NM_001114636.1:c.1107G>A	p.Lys369(=)	rs577063114	Homozygous	Pathogenic	NA

NA: Not applicable as EVE is available only for missense mutations

* likely pathogenic by ACMG guidelines but pathogenic by Varsome.