

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	Zygoty	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	FANCL (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 NC_000016.9:g.89828423T>G	5' splice site mutation Missense	NM_000135.4:c.3408+2T>G NM_000135.4:c.2786A>C	- p.Tyr929Ser	Novel Novel	Compound heterozygous	VUS VUS	NA Pathogenic
FA-04	FANCA	NC_000016.9:g.89823967C>T NC_000016.9:g.89811367_89865640del	3' splice site mutation Deletion	NM_000135.4:c.80-2A>G NM_000135.4:c.827_3626del	- -	Novel Novel	Compound heterozygous	VUS Likely pathogenic	NA 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 NC_000016.9:g.89877448del	5' splice site mutation Frameshift deletion	NM_000135.4:c.426_426+1delAG NM_000135.4:c.319delG	K143Rfs*7/5' splice variant p.Val107PhefsTer31	rs763114336 rs1411237340	Compound heterozygous	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 NC_000016.9:g.89857867G>A	Frameshift insertion Missense	NM_000135.4:c.3690_3694dupGCACT NM_000135.4:c.1303C>T	p.Phe1232CysfsTer17 p.Arg435Cys	Novel rs148473140	Compound heterozygous	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	rs155556175	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 NC_000016.9:g.89828358_89842224del	Nonsense Deletion	NM_000135.4:c.3671G>A NM_000135.4:c.1827_2852del	p.Trp1224Ter -	Novel Castella et al. 2011	Compound heterozygous	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.8982944C>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.8982944C>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 NC_000009.11:g.35076856_35076857del	Missense Frameshift deletion	NM_004629.2:c.425T>C NM_004629.2:c.792_793delAG	p.Leu142Pro p.Arg264SerfsTer24	Novel Novel	Compound heterozygous	VUS VUS	Pathogenic 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.8980929_8983055del	Deletion	NM_000135.4:c.21_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 NC_000016.9:g.89806402C>T	Frameshift deletion Missense	NM_000135.4:c.1164_1165delAG NM_000135.4:c.3934G>A	p.Arg388SerfsTer20 p.Asp1312Asn	Novel Novel	Compound heterozygous	VUS VUS	NA 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
FA-556	FANCA	NC_000016.9:g.89803957_89806507del	Deletion	NM_000135.4:c.3829_1052del	-	Novel	Compound heterozygous	Likely pathogenic	NA
FA-557	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.89833009C>T	Nonsense	NM_000135.4.c.158G>A	p.Trp35Ter	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	Homozygous	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	FANCL (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.119dup	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	Compound heterozygous	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.897C>A	p.Gln333Ter	rs776298788	Compound heterozygous	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.824-2A>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.3198delG	p.Val107PhefsTer31	rs1411237340	Compound heterozygous	Pathogenic	NA
		NC_000016.9.g.89883022A>T	Missense	NM_000135.4.c.217A>T	p.Met1Lys	rs769479800	Compound heterozygous	Pathogenic	NA
FA-580	FANCG	NC_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.Glu1254SerfsTer10	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	-	Savioia 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.3505G>C	p.Arg1117Thr	Novel	Homozygous	Likely pathogenic	Pathogenic
FA-12/19	FANCA	NC_000016.9.g.89877448del	Frameshift deletion	NM_000135.4.c.3198delG	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	Compound heterozygous	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	Compound heterozygous	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_89828945del	Deletion	NM_000135.4.c.79_177del	-	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	Compound heterozygous	Pathogenic	NA
FA-03/19	FANCI	NC_000015.9.g.89804822del	Frameshift deletion	NM_001113378.2.c.295delC	p.His99lefsTer10	rs759398314	Compound heterozygous	Pathogenic	NA
		NC_000015.9.g.89848828_89848832del	Intronic splice acceptor variant	NM_001113378.2.c.3256-8_3256-4delTCTTT	-	Novel	Compound heterozygous	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.3198del	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.Tyr551Trp	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	Compound heterozygous	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-849/18	BRCA2	NC_000013.10:g.32893238G>C	Missense	NM_000059.4:c.92G>C	p.Tip31Ser	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.