

Supplementary Table 1: Molecular characterization by WES of 68 FA patients

Sample ID	Sex	Mosaic	Subtyping	DNA	Gene	Notes	Mutation 1					Mutation 2						
							Exon/Intron	cDNA	ACMG classification	ACMG Rules	Protein	Reported/Novel	Exon/Intron2	cDNA2	ACMG classification2	ACMG Rules2	Protein2	Reported/Novel2
FA029	F	Yes	No A,B,C,G,E,F,D2	PF	FANCD2 (NM_014176.4)		12	c.110-15_110-5del	Pathogenic	PM2	p.Glu377Argfs*499	Novel	Ex2-6	Deletion Ex2-6	–	–	–	–
FA049	M	Yes	A	LCL	FANCA (NM_000135.4)		112	c.1083-2T>C	Pathogenic	PM2; PM2; PP3	–	Novel	Ex21-29	Deletion Ex21-29	–	–	–	–
FA050	M	Yes	A	LCL	FANCA (NM_000135.4)	Sibling FA049	112	c.1083-2T>C	Pathogenic	PM2; PM2; PP4	–	Novel	Ex21-29	Deletion Ex21-29	–	–	–	–
FA090	F	Yes	A	LCL	FANCA (NM_000135.4)		E6	c.3558delG	Pathogenic	PM2; PM2; PP5	p.Arg1186Glnfs*28	PMID: 9399890	Ex5	c.472dupC	Likely Pathogenic	PM2; PM2; PP5	p.Hist158Profs*23	Novel
FA104	F	No	No A,B,C,G,E,F,D2,J	PB	FANCA (NM_005236.2)		Ex11	c.2065C>A	Uncertain Significance	PM2; PP3; PP5	p.Arg698Ser	PMID: 23623386	Ex8	c.1484_1488delCTCAA	Pathogenic	PM2; PM2; PP5	p.Theo95Asnfs*5	PMID: 23623386
FA121	M	Yes	E	PB	FANCD2 (NM_021922.2)		Ex4	c.929dupC	Likely Pathogenic	PM2	p.Val311Serfs*1	Novel	–	–	–	–	–	–
FA124	F	Yes	No A, C, G	PB	FANCD2 (NM_03084.4)		Ex13	c.1068T>A	Pathogenic	PM2; PM2; PP3	p.Tyr356*	Novel	–	–	–	–	–	–
FA158	M	Yes	–	PB	FANCD1 (NM_000099.3)		119	c.8488-1G>A	Pathogenic	PM2; PM2; PP3; PP5	p.The2831_Leu2834del	PMID: 16115142	119	c.8488-1G>A	Pathogenic	PM2; PM2; PP3; PP5	r.The2831_Leu2834del	PMID: 16115142
FA166	F	No	A	LCL	FANCA (NM_000135.4)		Ex36	c.3558dupG	Pathogenic	PM2; PM2; PP6	p.Arg1186Glnfs*28	PMID: 9399890	–	–	–	–	–	–
FA255	F	No	–	PB	FANCD2 (NM_03084.4)		Ex26	c.2444G>A	Uncertain Significance	PP3; PP5	p.Arg815Gln	PMID: 17436244	Ex38	c.3817C>T	Pathogenic	PM2; PM2; PP3; BP7	p.Arg1273*	Novel
FA287	F	No	No A,B,C,G,E,F,D2,J	PB	FANCA (NM_032043.2)		Ex7	c.751C>T	Likely Benign	PM2; PP3; BS2; BP1	p.Arg251Cys	PMID: 23613520	Ex7	–	Pathogenic	PM2; PM2; PP3	p.Arg279Glyfs*10	Novel
FA310	M	No	–	PB	FANCA (NM_000135.4)		Ex32	c.3164G>T	Uncertain Significance	PM2; PP3; PP5	p.Arg105Leu	PMID: 12444997	Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PM2; PP3	p.Phe1263del	PMID: 19464302
FA326	F	No	No A,C,G,E,F,D2	PF	FANCA (NM_004629.1)		Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564	Ex3	c.566delA	Likely Pathogenic	PM2; PM2; PP3	p.Glu1063Glyfs*45	Novel
FA331	M	Yes	No A,C,G	PF	–		–	–	–	–	–	–	–	–	–	–	–	–
FA357	F	No	No A,B,C,G,E,F,D2	PB	FANCD2 (NM_03084.4)		Ex3	c.98dupA	Likely Pathogenic	PM2	p.Lys34Glnfs*6	Novel	Ex3	c.98dupA	Likely Pathogenic	PM2; PM2; PP3	p.Lys34Glnfs*6	Novel
FA383	F	No	D2	PF	FANCD2 (NM_03084.4)		Ex26	c.2444G>A	Uncertain Significance	PP3; PP5	p.Arg815Gln	PMID: 17436244	137	c.3777-1G>A	Pathogenic	PM2; PM2; PP3	–	Novel
FA388	F	No	A	PB	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	–	–	–	–	–	–
FA460	F	No	A	PB	FANCA (NM_000135.4)		Ex37	c.3763G>T	Pathogenic	PM2; PM2; PP3	p.Glu1255*	Novel	17	c.709+5G>A	Uncertain Significance	PM2; PP5	–	PMID: 10004191
FA486	F	No	G	PB	FANCA (NM_004629.1)		Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564	Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564
FA522	M	No	G	PB	FANCA (NM_004629.1)		Ex4	c.450G>A	Pathogenic	PM2; PM2; PP3	p.Tyr150*	Novel	Ex4	c.450G>A	Pathogenic	PM2; PM2; PP3	p.Tyr150*	Novel
FA530	F	No	–	PB	FANCA (NM_000135.4)		Ex36	c.3558dupG	Pathogenic	PM2; PM2; PP5	p.Arg1186Glnfs*28	PMID: 9399890	Ex36	c.3558dupG	Pathogenic	PM2; PM2; PP5	p.Arg1186Glnfs*28	PMID: 9399890
FA531	F	No	No A,C,G,E,F,D2	PB	FANCD2 (NM_03084.4)		Ex26	c.2444G>A	Uncertain Significance	PP3; PP5	p.Arg815Gln	PMID: 17436244	Chr:3 1000519-10183337 (HG19)	Microdeletion	–	–	–	–
FA535	M	No	A	PB	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	Ex13	c.1100G>T	Uncertain Significance	PM2	p.Ser367Ile	Novel
FA537	F	No	A	PB	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302
FA550	F	Yes	A	PF	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex41	c.4130C>G	Likely Pathogenic	PM2; PM2; BP4	p.Ser1377*	PMID: 17924555
FA551	M	No	A	PF	FANCA (NM_000135.4)	Sibling FA550	Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex41	c.4130C>G	Likely Pathogenic	PM2; PM2; BP5	p.Ser1377*	PMID: 17924555
FA553	M	Yes	A	PF	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	Ex32-33	Deletion Ex32-33	–	–	–	–
FA568	F	Yes	A	PB	FANCA (NM_000135.4)		Ex39	c.3913C>T	Uncertain Significance	PM2; PP3; PP5	p.Leu1305Phe	PMID: 8655138	Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138
FA569	F	Yes	No A, B, C	PB	FANCA (NM_000135.4)		Ex8	c.790G>T	Pathogenic	PM2; PM2; PP3	p.Glu264*	PMID: 9399890	Ex36	c.3558dupG	Pathogenic	PM2; PM2; PP5	p.Arg1186Glnfs*28	PMID: 9399890
FA572	F	No	No A,C,D,E,E,F	PB	FANCA (NM_032043.2)		Ex7	c.751C>T	Likely Benign	PM2; PP3; BS2; BP2	p.Arg251Cys	PMID: 23613520	Ex7	c.751C>T	Likely Benign	PM2; PP3; BS2; BP2	p.Arg251Cys	PMID: 23613520
FA573	F	No	G	PB	FANCA (NM_004629.1)		Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564	Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564
FA574	M	Yes	No A,C,G	PB	FANCA (NM_032043.2)		Ex3	c.786G>T	Uncertain Significance	PM2; PP3	p.Lys262Asn	Novel	–	–	–	–	–	–
FA577	F	No	A	PB	FANCA (NM_000135.4)		Ex3	Deletion Ex3	–	–	–	–	–	–	–	–	–	–
FA596	M	No	A	PB	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	Ex3	c.233_236delTTGA	Likely Pathogenic	PM2; PM2	p.Thr781Ilefs*16	PMID: 21273304
FA609	M	No	A	PB	FANCA (NM_000135.4)		Ex1	c.65G>A	Pathogenic	PM2; PM2; PP3; PP5	p.Tyr22*	Novel	Ex1	c.65G>A	Pathogenic	PM2; PM2; PP3; PP5	p.Tyr22*	Novel
FA616	M	No	A	PB	FANCA (NM_000135.4)		Ex2	c.163C>T	Pathogenic	PM2; PM2; PP5	p.Glu55*	PMID: 10004191	Ex2	c.163C>T	Pathogenic	PM2; PM2; PP5	p.Glu55*	PMID: 10004191
FA625	M	No	A	PB	FANCA (NM_000135.4)		Ex12	c.1033G>T	Pathogenic	PM2; PM2; PP3	p.Glu345*	Novel	Ex4	c.295C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Glu69*	PMID: 15522956
FA638	F	No	A	PF	FANCA (NM_000135.4)		Ex28	c.2641C>T	Likely Pathogenic	PM2	p.Glu881*	PMID: 21273304	Ex35	c.3448dupC	Likely Pathogenic	PM2; PM2; PP3	p.Leu1305Profs*65	Novel
FA649	M	No	A	PB	FANCA (NM_000135.4)		Ex37	c.3763G>T	Pathogenic	PM2; PM2; PP3	p.Glu1255*	Novel	Ex33	c.3335T>G	Uncertain Significance	PM2; PM2; PP3	p.Val1112Gly	PMID: 21273304
FA652	M	No	A	PB	FANCA (NM_000135.4)		Ex21	c.1860dupC	Likely Pathogenic	PM2	p.Tyr521Leufs*9	Novel	110	c.893-2T>C	Pathogenic	PM2; PM2; PP3	p.Val1112Gly	Novel
FA654	M	No	A	PB	FANCA (NM_000135.4)		Ex39	c.3913C>T	Uncertain Significance	PM2; PP3; PP5	p.Leu1305Phe	PMID: 8655138	Ex41	c.4124_4125delCA	Likely Pathogenic	PM2; PM2; PP3	p.Phe1375Sfs*49	PMID: 8655138
FA664	M	Yes	D2 (WBlo)	PF	FANCD2 (NM_03084.4)		Ex4	c.227delA	Likely Pathogenic	PM2; PM2; PP3	p.Lys778Ile*	Novel	–	–	–	–	–	–
FA681	M	Yes	D2 (WBlo)	PF	FANCD2 (NM_03084.4)	Sibling FA664	Ex4	c.227delA	Likely Pathogenic	PM2; PM2; PP3	p.Lys778Ile*	Novel	–	–	–	–	–	–
FA705	M	No	–	PB	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	Ex27-43	Deletion Ex27-43	–	–	–	–
FA707	F	No	–	PB	FANCA (NM_000135.4)		Ex13	Deletion Ex13-17	–	–	–	–	–	–	–	–	–	–
FA712	M	No	A	PB	FANCA (NM_000135.4)		Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138	Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138
FA718	F	No	–	PB	FANCA (NM_000135.4)		Ex23	c.2128A>T	Likely Pathogenic	PM2; PM2; BP4	p.Arg710*	Novel	138	c.3828-1G>T	Pathogenic	PM2; PM2; PP3	–	PMID: 9399890
FA746	F	No	–	PB	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	110	c.893-1G>T	Pathogenic	PM2; PM2; PP3	–	PMID: 9399890
FA766	F	No	A	PB	FANCA (NM_000135.4)		Ex15	Deletion Ex15	–	–	–	–	–	–	–	–	–	–
FA796	M	No	–	PB	FANCA (NM_021922.2)		Ex5	c.1111C>T	Uncertain Significance	PP3; BP1	p.Arg371Trp	PMID: 17924555	Ex5	c.1111C>T	Uncertain Significance	PP3; BP1	p.Arg371Trp	PMID: 17924555
FA829	M	Yes	–	PB	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex39	c.3913C>T	Uncertain Significance	PM2; PP3; PP5	p.Leu1305Phe	PMID: 8655138
FA830	F	No	–	PB	FANCA (NM_000135.4)		Ex41	c.4130C>G	Likely Pathogenic	PM2; PM2; BP4	p.Ser1377*	PMID: 17924555	Ex13	c.1115_1118delTTGG	Pathogenic	PM2; PM2; PP5	p.Val372Alafs*42	PMID: 8655138
FA842	F	Yes	–	PB	FANCA (NM_000135.4)		Ex26	c.2398G>T	Pathogenic	PM2; PM2; PP5	p.Glu800*	Novel	Ex15	Deletion Ex15	–	–	–	–
FA846	F	No	–	PB	FANCA (NM_004629.1)		Ex13	c.1642C>T	Pathogenic	PM2; PM2; PP3; PP5	p.Arg548*	PMID: 12552564	Ex7	c.908T>C	Uncertain Significance	PM2; PM2; BP1	p.Leu303Pro	PMID: 12552564
FA847	M	No	–	PB	FANCA (NM_000135.4)		Ex37	c.3763G>T	Pathogenic	PM2; PM2; PP3	p.Glu1255*	Novel	Ex37	c.3763G>T	Pathogenic	PM2; PM2; PP3	p.Glu1255*	Novel
FA848	M	No	–	PB	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex29	c.2814C>G	Uncertain Significance	PM2; PM2; PP3	p.Arg951Gly	Novel
FA850	F	Yes	A	PB	FANCA (NM_000135.4)		Ex38	c.3788_3790delTCT	Uncertain Significance	PM2; PP5	p.Phe1263del	PMID: 19464302	Ex19-20	Deletion Ex19-20	–	–	–	–
FA860	F	No	No A,C,G	PB	FANCA (NM_000135.4)		Ex1-30	Deletion Ex1-30	–	–	–	–	–	–	–	–	–	–
FA875	F	Yes	A	PB	FANCA (NM_000135.4)		Ex30	c.2870G>A	Pathogenic	PM2; PM2; PP3; PP5	p.Tyr957*	PMID: 17924555	Ex8					