Molecular analysis of Fanconi anemia: the experience of the Bone Marrow Failure Study Group of the Italian Association of Pediatric Onco-Hematology

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SUPPLEMENTARY MATERIAL

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Table 1S. Molecular data of 100 FA patients.

Family ID*	Exon/ Intron	FANCA cDNA** (NM_000135.2)	Mutation status	Inheritance***	FANCA protein (NP_000126.2)	Mutation classification	References****
FA1 C	38	c.3788_3790del	Homo	nd	p.Phe1263del	small deletion (in frame)	This paper/FA database
FA2 M CF	28	c.2738A>C	Homo	nd	p.His913Pro	missense	This paper/FA database
FA3 C	i5_i31	c.523-?_3066+?del	Homo	nd	p.Ser175_Gln1022del	large deletion (in frame)	This paper/Novel
FA4 C	28	c.2738A>C	Homo	P/nd	p.His913Pro	missense	This paper/FA database
FA5	i3_i20	c.284-?_1826+?del	Homo	P/M	p.Gly95Glufs*31	large deletion (frameshift)	This paper/Novel
FA6 C	37	c.3660del	Hetero	nd	p.Asn1221Thrfs*26	small deletion (frameshift)	This paper/Novel
	37	c.3692A>C	Hetero		p.His1231Pro	missense	This paper/Novel
FA7	i14_i15	c.1360-?_1470+?del	Hetero	M	r.1360_1470del p.Ala454_Gln490del	large deletion (in frame	This paper/FA database
	6	c.549G>A	Hetero	P	p.Trp183*	nonsense	This paper/Novel
FA8 C	29	c.2852G>A	Hetero	M	p.Arg951Gln	missense	This paper/FA database

	i42	c.4261-19_4261-12del	Hetero	P	nd	splicing (potential)	This paper/FA database
FA9 C	38	c.3788_3790del	Homo	nd	p.Phe1263del	small deletion (in frame)	This paper/FA database
FA10	32	c.3239G>A	Hetero	nd	p.Arg1080Gln	missense	This paper/FA database
	40	c.3971C>T	Hetero		p.Pro1324Leu	missense	This paper/FA database
FA11	1	c.11C>A	Hetero	M	p.Ser4*	nonsense	This paper/FA database
	i38	c.3829-9G>A	Hetero	P	nd	splicing (potential)	This paper/Novel
FA12 C	i14_i15	c.1360-?_1470+?del	Homo	P/M	r.1360_1470del p.Ala454_Gln490del	large deletion (in frame)	This paper/FA database
FA13 C	5	c.457C>T	Hetero	P	p.Gln153*	nonsense	This paper/Novel
	i7	c.709+1G>A	Hetero	M	r.[709+1a>g; 709_710ins709+1_709+30] (cryptic site in intron 7) p.250_251insGlyAlaPheMetThrArgCysGlyPheLeu	splicing (in frame)	This paper/FA database
FA14	i16	c.1567-20A>G	Homo	P/M	r.[1567-20a>g; 1566_1567ins1566+1_1567-1] (i16 retention) p.Val523Alafs*49	splicing (frameshift)	Savino et al., 2003 (EUFA265)
FA15 (2)	i7	c.710-5T>C	Hetero	P	r.710_792del (e8 skipping) p.Asp237Glyfs*4	splicing (frameshift)	This paper/FA database
	8	c.790C>T	Hetero	M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	This paper/FA database
FA16	38	c.3788_3790del	Homo	P/M	p.Phe1263del	small deletion (in frame)	This paper/FA database
FA17	29	c.2812_2830dup	Hetero	nd	p.Asp944Glyfs*5	small duplication (frameshift)	Savino et al., 2003 (FA19)
	40	c.3971C>T	Hetero	M	p.Pro1324Leu	missense	This paper/FA database
FA18	37	c.3715_3729del	Hetero	M	p.Glu1239_Arg1243del	small deletion (in frame)	This paper/FA database
	28	c.2738A>C	Hetero	P	p.His913Pro	missense	This paper/FA database

FA19	30	c.2932C>T	Hetero	nd	p.Gln978*	nonsense	This paper/Novel
	38	c.3788_3790del	Hetero	nd	p.Phe1263del	small deletion (in frame)	This paper/FA database
FA20	29	c.2812_2830dup	Hetero	P	p.Asp944Glyfs*5	small duplication (frameshift)	Savino et al., 2003 (FA32)
	i-1_i43	c.-42-?_5481+?del	Hetero	nd	no protein	large deletion (entire gene)	This paper/FA database
FA21 C	38	c.3788_3790del	Hetero	M	p.Phe1263del	small deletion (in frame)	This paper/FA database
	40	c.3971C>T	Hetero	P	p.Pro1324Leu	missense	This paper/FA database
FA22	29	c.2840C>G	Homo	P/M	p.Ser947*	nonsense	Savino et al., 2003 (EUFA341)
FA23 C	29	c.2852G>A	Hetero	P	p.Arg951Gln	missense	This paper/FA database
	28	c.2738A>C	Hetero	M	p.His913Pro	missense	This paper/FA database
FA24	25	c.2314C>T	Homo	P/M	p.Gln772*	nonsense	Savino et al., 2003 (EUFA268)
FA25 C	35	c.3490C>T	Homo	P/M	p.Pro1164Ser	missense	This paper/FA database
FA26	41	c.4075G>T	Hetero	nd	p.Asp1359Tyr	missense	Savino et al., 2003 (FA35)
	nd						
FA27	36	c.3558dup	Hetero	M	p.Arg1187Glufs*28	small duplication (frameshift)	This paper/FA database
	29	c.2852G>A	Hetero	P	p.Arg951Gln	missense	This paper/FA database
FA28	i10	c.893+5G>A	Hetero	P	nd	splicing (potential)	This paper/Novel
	i2_i3	c.190-?_283+?del	Hetero	nd	p.Val64Alafs*43	large deletion (frameshift)	This paper/FA database

FA29 C	36	c.3558dup	Hetero	M	p.Arg1187Glufs*28	small duplication (frameshift)	This paper/FA database
	i14_i28	c.1360-?_2778+?del	Hetero	nd	p.Ala454_His926del	large deletion (in frame)	This paper/Novel
FA30 (2)	2	c.123_124del	Hetero	P	p.Lys42Ilefs*21	small deletion (frameshift)	This paper/Novel
	32	c.3109_3137delinsG	Hetero	M	p.Pro1037Alafs*14	small indel (frameshift)	This paper/Novel
FA31	8	c.790C>T	Homo	P/M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	Savino et al., 2003 (FA63)
FA32	29	c.2840C>G	Hetero	P	p.Ser947*	nonsense	This paper/FA database
	nd						
FA33	28	c.2738A>C	Hetero	M	p.His913Pro	missense	This paper/FA database
	i10_i14	c.894-?_1359+?del	Hetero	nd	p.Phe299Profs*72	large deletion (frameshift)	This paper/Novel
FA34 C	11	c.987-990del	Homo	P/M	p.His330Alafs*4	small deletion (frameshift)	This paper/FA database
FA35 C	28	c.2770G>T	Homo	nd	p.Asp924Tyr	missense	This paper/Novel
	i-1_i6	c.-42-?_523+?del	Homo	nd	nd	large deletion (removal of initiation codon)	This paper/FA database
FA36	i7	c.710-5T>C	Hetero	P	r.710_792del (e8 skipping) p.Asp237Glyfs*4	splicing (frameshift)	This paper/FA database
	35	c.3490C>T	Hetero	M	p.Pro1164Ser	missense	This paper/FA database
FA37	32	c.3164G>A	Hetero	M	p.Arg1055Gln	missense	This paper/FA database
	28	c.2638C>T	Hetero	P	p.Arg880*	nonsense	This paper/FA database
FA38	i3	c.283+3A>C	Homo	P/M	r.190_283del (e3 skipping) p.Val64Alafs*43	splicing (frameshift)	Savino et al., 2003 (EUFA397)

FA39	i9	c.826+3del	Homo	P/M	nd	splicing (potential)	Savino et al., 2003 (EUFA330)
FA40	i14_i15	c.1360-?_1470+?del	Hetero	P	r.1360_1470del p.Ala454_Gln490del	large deletion (in frame)	This paper/FA database
	i10_i33	c.894-?_3348+?del	Hetero	M	p.Trp298*	large deletion (nonsense)	This paper/FA database
FA41	42	c.4258G>T	Hetero	P	p.Glu1420*	nonsense	This paper/Novel
	nd						
FA42 (2) C	37	c.3638_3639del	Hetero	M	p.Pro1213Argfs*64	small deletion (frameshift)	Savino et al., 2003 (FA58)
	40	c.3971C>T	Hetero	P	p.Pro1324Leu	missense	This paper/FA database
FA43	37	c.3761_3762dup	Hetero	P	p.Glu1255Argfs*12	small duplication (frameshift)	Savino et al., 2003 (EUFA232)
	27	c.2574C>G	Hetero	M	p.Ser858Arg	missense	Savino et al., 2003 (EUFA232)
FA44 C	25	c.2290 C>T	Hetero	P	p.Arg764Trp	missense	This paper/FA database
	41	c.4029T>G	Hetero	M	p.His1343Gln	missense	This paper/Novel
FA45 (2) C	18	c.1645C>T	Hetero	nd	r.1645c>u + r.1627_1715del (e18 skipping) Gln549* + p.Pro543Hisfs*26	nonsense + splicing (frameshift)	This paper/FA database
	33	c.3263C>T	Hetero	nd	p.Ser1088Phe	missense	This paper/FA database
	i20_i29	c.1827-?_2852+?del	Hetero	nd	r.1827_2852del p.Ala610_Arg951del	large deletion (in frame)	This paper/FA database
FA46	i28	c.2778+1G>A	Hetero	nd	r.2602_2778del (e28 skipping) p.Phe868_His926del	splicing (in frame)	This paper/FA database
	13	c.1115_1118del	Hetero	nd	p.Val372Alafs*42	small deletion (frameshift)	This paper/FA database
FA47 (2) C IT	37	c.3660del	Hetero	nd	p.Asn1221Thrfs*26	small deletion (frameshift)	This paper/Novel

	1	c.50dup	Hetero	nd	p.Arg18Profs*19	small duplication (frameshift)	This paper/Novel
FA48 C	22	c.2005C>T	Hetero	M	p.Gln669*	nonsense	This paper/FA database
	23	c.2051T>C	Hetero	de novo (paternity confirmed)	p.Leu684Pro	missense	This paper/FA database
FA49 C IT	i5_i30	c.523-?_2981+?del	Hetero	nd	p.Ser175Leufs*5	large deletion (frameshift)	This paper/Novel
	6	c.548G>A	Hetero		p.Trp183*	nonsense	This paper/Novel
FA50 M	4	c.352G>C	Hetero	M	p.Ala118Pro	missense	This paper/Novel
	37	c.3715_3729del	Hetero	P	p.Glu1239_Arg1243del	small deletion (in frame)	This paper/FA database
FA51 M	8	c.790C>T	Hetero	M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	This paper/FA database
	29	c.2840C>G	Hetero	P	p.Ser947*	nonsense	This paper/FA database
FA52 C	i8	c.793-1G>C	Homo	nd	r.793_826del (e9 skipping) p.Val1265Leufs*20 r.[793-1g>c; 792_793ins793-57_793-1] (cryptic site in i8) p.Val265Lysfs*34	splicing (frameshift)	This paper/Novel
FA53 (2)	i26	c.2504+2T>C	Hetero	M	r.[2504+2u>c; 2504_2505ins2504+1_2504+129] (cryptic site in i26) p.Phe836*	splicing (nonsense)	Savino et al., 2003 (EUFA262-263)
	i10	c.893+1G>T	Hetero	P	r.[893+1g>u; 893_894ins893+1_893+215] (cryptic site in i10) p.Phe879Valfs*19	splicing (frameshift)	Savino et al., 2003 (EUFA262-263)
FA54	29	c.2840C>G	Homo	P/M	p.Ser947*	nonsense	Savino et al., 2003 (EUFA223)
FA55	i28	c.2778+83C>G	Hetero	P	nd	splicing (potential)	Savino et al., 2003 (EUFA578)
	nd						
FA56	21	c.1874G>C	Hetero	M	p.Cys625Ser	missense	This paper/FA database

	7	c.619G>T	Hetero	M	p.Gly207*	nonsense	This paper/Novel
	29	c.2840C>G	Hetero	P	p.Ser947*	nonsense	This paper/FA database
FA57 M CT	8	c.790C>T	Hetero	nd	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	This paper/FA database
	34	c.3367G>T	Hetero		p.Gly1123*	nonsense	This paper/Novel
FA58 M IT	38	c.3788_3790del	Hetero	P	p.Phe1263del	small deletion (in frame)	This paper/FA database
	i9	c.826+3del	Hetero	M	nd	splicing (potential)	This paper/FA database
FA59 M	1	c.66G>A	Hetero	nd	p.Trp22*	nonsense	This paper/FA database
	i11	c.1006+1G>T	Hetero	M	nd	splicing (potential)	This paper/Novel
FA60	27	c.2535_2536del	Hetero	P	p.Cys846Glnfs*20	small deletion (frameshift)	This paper/FA database
	i33	c.3348+1G>A	Hetero	M	r.3240_3348del (e33 skipping) p.Ile1081Glufs*10	splicing (frameshift)	This paper/FA database
FA61 M CT	i14_i20	c.1360-?_1826+?del	Hetero	nd	p.Ala454Serfs*3	large deletion (frameshift)	This paper/FA database
	nd						
FA62	i14	c.1359+1G>C	Homo	nd	r.1226_1359del (e14 skipping) p.Asp409Glyfs*31	splicing (frameshift)	Savino et al., 2003 (FA25)
FA63 C	42	c.4198C>T	Hetero	P	p.Arg1400Cys	missense	This paper/FA database
	i42	c.4261-2A>C	Hetero	M	nd	splicing (potential)	This paper/Novel
FA64	36	c.3558dup	Hetero	nd	p.Arg1187Glufs*28	small duplication (frameshift)	Savino et al., 2003 (FA37)
	nd						

FA65 C	5	c.457C>G	Hetero	M	p.Gln153Glu	missense	This paper/Novel
	i15_i17	c.1471-?_1626+?del	Hetero	nd	p.Val491_Glu542del	large deletion (in frame)	This paper/FA database
	35	c.3490C>T	Hetero	P	p.Pro1164Ser	missense	This paper/FA database
FA66 C IT	i20_i29	c.1827-?_2852+?del	Hetero	nd	r.1827_2852del p.Ala610_Arg951del	large deletion (in frame)	This paper/FA database
	i19	c.1776+7A>G	Hetero	M	r.1716_1776del (e19 skipping) p.Ile573Serfs*12	splicing (frameshift)	This paper/Novel
FA67 C	21	c.1850_1859del	Hetero	nd	p.Leu617Profs*20	small deletion (frameshift)	This paper/Novel
	i20_i28	c.1827-?_2778+?del	Hetero	M	p.Arg609Serfs*2	large deletion (frameshift)	This paper/FA database
FA68 M	i2	c.190-1G>T	Hetero	P	nd	splicing (potential)	Savino et al., 2003 (FA73)
	8	c.790C>T	Hetero	M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	Savino et al., 2003 (FA73)
FA69	8	c.790C>T	Homo	P/M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	Savino et al., 2003 (EUFA388)
FA70 M CF	15	c.1450G>C	Hetero	nd	p.Glu484Gln	missense	This paper/Novel
	i6	c.596+1G>T	Hetero	M	nd	splicing (potential)	This paper/Novel
FA71	30	c.2853dup	Homo	nd/M	p.Gln952Alafs*10	small duplication (frameshift)	This paper/Novel
FA72	i-1_i43	c.-42-?_5481+?del	Hetero	nd	no protein	large deletion (entire gene)	Centra et al., 1999; Savino et al., 2003 (EUFA337)
	i17_i21	c.1627-?_1900+?del	Hetero	M	p.Pro543Metfs*6	large deletion (frameshift)	Centra et al., 1999; Savino et al., 2003 (EUFA337)
FA73 M	29	c.2851C>T	Hetero	P	p.Arg951Trp	missense	This paper/FA database
	i19	c.1777-7_1779del	Hetero	M	nd	splicing (potential)	This paper/FA database

FA74 C	38	c.3788_3790del	Hetero	M	p.Phe1263del	small deletion (in frame)	This paper/FA database
	29	c.2840C>G	Hetero	P	p.Ser947*	nonsense	This paper/FA database
FA75	8	c.790C>T	Homo	P/M	r.790c>u + r.710_792del (e8 skipping) p.Gln264* + p.Asp237Glyfs*4	nonsense + splicing (frameshift)	This paper/FA database
FA76 C	i3_i20	c.284-?_1826+?del	Homo	P/M	p.Gly95Glufs*31	large deletion (frameshift)	This paper/Novel
FA77 C	i14	c.1359+1G>C	Homo	P/M	r.1226_1359del (e14 skipping) p.Asp409Glyfs*31	splicing (frameshift)	This paper/FA database
FA78 C	29	c.2852G>A	Hetero	M	p.Arg951Gln	missense	This paper/FA database
	i3_i4	c.284-?_426+?del	Hetero	nd	p.Gly95Glufs*38	large deletion (frameshift)	This paper/Novel
FA79 C	29	c.2852G>A	Hetero	M	p.Arg951Gln	missense	This paper/FA database
	38	c.3798G>A	Hetero	P	p.Met1266Ile	missense	This paper/Novel
	1	c.2T>A	Hetero	P	p.?	initiation codon	This paper/Novel
FA80 M	36	c.3558dup	Homo	P/M	p.Arg1187Glufs*28	small duplication (frameshift)	Savino et al., 2003 (EUFA393)
FA81	36	c.3558dup	Hetero	P	p.Arg1187Glufs*28	small duplication (frameshift)	Savino et al., 2003 (FA53)
	nd						
FA82	22	c.2005C>T	Hetero	P	p.Gln669*	nonsense	Savino et al., 2003 (FA38)
	nd						
FA83	37	c.3638_3639del	Homo	nd	p.Pro1213Argfs*64	small deletion (frameshift)	Savino et al., 2003 (FA41)
FA84 (2)	36	c.3558dup	Hetero	M	p.Arg1187Glufs*28	small duplication (frameshift)	This paper/FA database

	i40	c.4011-1G>C	Hetero	P	r.4011_4033del (cryptic site recognition in e41) p.Ser1337Argfs*80	splicing (frameshift)	This paper/Novel
FA85	13	c.1126C>T	Hetero	nd	p.Gln376*	nonsense	This paper/Novel
	13	c.1115_1118del	Hetero	P	p.Val372Alafs*42	small deletion (frameshift)	This paper/FA database
Family ID*	Exon/Intron	FANCB cDNA (NM_152633.2)	Mutation status	Inheritance**	FANCB protein (NP_001018123.1)	Mutation classification	References
FA86 C	3	c.353T>C	Hemi	nd	p.Phe118Ser	missense	This paper/Novel
Family ID*	Exon/Intron	FANCC cDNA (NM_000136.2)	Mutation status		FANCC protein (NP_000127.2)	Mutation classification	References
FA87 C	15	c.1642C>T	Homo	nd	p.Arg548*	nonsense	This paper/FA database
FA88 IT	2	c.67del	Homo	nd	p.Asp23Ilefs*23	small deletion (frameshift)	This paper/FA database
FA89 IT	2	c.37C>T	Hetero	P	p.Gln13*	nonsense	This paper/FA database
	8	c.692_694del	Hetero	M	p.Lys231del	small deletion (in frame)	This paper/Novel
Family ID*	Exon/Intron	FANCD2 (NM_001018115.1)	Mutation status		FANCD2 protein (NP_001018125.1)	Mutation classification	References
FA90	12	c.904C>T	Hetero	nd	p.Arg302Trp	missense	Kalb et al., 2007
	13	c.1092G>A	Hetero		p.Trp364*	nonsense	
FA91	7	c.458T>C	Homo	nd	p.Leu153Ser	missense	Borriello et al., 2007
Family ID*	Exon/Intron	FANCG cDNA (NM_004629.1)	Mutation status		FANCG protein (NP_004620.1)	Mutation classification	References
FA92 C	10	c.1199_1204del	Homo	nd	p.Ala400_Ala401del	small deletion (in frame)	This paper/Novel
FA93 C	i9	c.1144-1G>T	Homo	P/M	r.1144_1168del (cryptic site in e10) p.Phe382Glyfs*13	splicing (frameshift)	This paper/Novel

FA94	13	c.1715G>A	Homo	nd	r.1715g>a + r.1637_1760del (e13 skipping) p.Trp572* + p.Asn547Leufs*6	nonsense + splicing (frameshift)	This paper/FA database
FA95	4	c.336del	Homo	nd	p.Arg113Glyfs*39	small deletion (frameshift)	This paper/Novel
FA96	13	c.1715G>A	Homo	nd	r.1715g>a + r.1637_1760del (e13 skipping) p.Trp572* + p.Asn547Leufs*6	nonsense + splicing (frameshift)	This paper/FA database
FA97 C	14	c.1788T>A	Homo	nd	p.Tyr596*	nonsense	This paper/Novel
FA98	10	c.1182_1192delinsC	Homo	nd	p.Glu395Trpfs*5	small indel (frameshift)	This paper/FA database
FA99 M CF	10	c.1223_1226dup	Hetero	nd	p.Ala410Glnfs*10	small duplication (frameshift)	This paper/Novel
	i7	c.924+1G>A	Hetero		r.778_924del (skipping exon 7) p.Gly260_Glu308del	splicing (in frame)	This paper/Novel
FA100 C	10	c.1344C>A	Homo	nd	p.Cys448*	nonsense	This paper/Novel

*In bracket number of FA patients if >1. Role of mutant gene supported by complementation analysis carried out in lymphoblastoid cell lines (C), primary fibroblasts (CF) or peripheral blood T lymphocytes (CT); M, revertant lymphoblastoid cell lines; IT, sequencing analysis performed by the Ion Torrent (Life Technology) next generation sequencing platform.

**Nucleotide numbering reflects the FANCA, FANCB, FANCC, FANCD2, and FANCG cDNAs with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence RefSeq NM_000135.2, NM_152633.2, NM_000136.2, NM_001018115.1, and NM_004629.1, respectively. Therefore, the initiation codon is codon 1.

*** Paternal (P) and maternal (M) transmission; nd, non determined.