Erratum to: Characterization and genotype-phenotype correlation of patients with Fanconi anemia in a multi-ethnic population

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Resequencing of one of the patients with *FANCD1* who was included in our manuscript (Table 4) July 2020 of Haematologica[1] revealed that he was not homozygous to the common variant in this gene, but rather compound heterozygous to the variants c.5946del (p.Ser1982Argfs*22) and c.7007G>C (p.Arg2336Pro).

This resulted in changes in the Results section (Genetics part, pages 1,829-1,831 in the original paper), that should now read:

Thirty-five different mutations were found, including 22 in *FANCA*, four in *FANCD1* and three each in *FANCC*, *FANCD1*, *FANCG* and *FANCJ* (Table 4); 75 patients were homozygous for mutations in Fanconi genes, and 13 patients were compound heterozygous; 33 different combinations of mutations were found (Table 4); 20 novel mutations were detected in the cohort, as detailed in the *Online Supplementary Table S1*; seven of these were previously reported by our group.[13-15]

The type of mutation varied, as detailed in Table 4. Twenty-five patients had frameshift mutations, 19 patients had splice site mutations, 15 patients had deletions, 13 patients had nonsense mutations, nine patients had missense mutations and seven patients had a combination of mutation types.

Corrected Table 4. Genetics of Fanconi anemia in Israel.

#	Gene	Mutation 1	Mutation 2	Effect 1	Effect 2	Ethnicities
11	FANCA	c.2172-2173 insG (p. S725Vfs*69)[13]	c.2172-2173 insG (p. S725Vfs*69)[13]	frame shift	frame shift	Jewish Sephardic
8	FANCA	c.4261-2A>C (IVS43-2a>c)[14]	c.4261-2A>C (IVS43-2a>c)[14]	splice site	splice site	Arab Muslim Arab Christian
8	FANCA	c.3788-3790delTCT[25]	c.3788-3790delTCT[25]	frame shift	frame shift	Arab Druze Jewish Sephardic
5	FANCA	ex6-13 del[14]	ex6-13 del[14]	large deletion	large deletion	Arab Muslim
5	FANCA	ex21 del[26, 27]	ex21 del[26, 27]	large deletion	large deletion	Arab Muslim
3	FANCA	c.4069-4070insT^	c.4069-4070insT^	frame shift	frame shift	Arab Muslim
3	FANCA	p.Gln1128Ter[15]	p.Gln1128T[15]	nonsense	nonsense	Arab Muslim
2	FANCA	ex31-37del[15]	4275delT (p.Asp1427Thr fsX6)[13]	large deletion	deletion	Jewish Sephardic
2	FANCA	p.Pro1164Ser^	p.Pro1164Ser^	missense	missense	Arab Muslim
1	FANCA	c.2172-2173 insG (p. S725Vfs*69)[13]	deletion exons 4-7[8]	frame shift	large deletion	Jewish mixed
1	FANCA	c.4275delT (p.Asp1427Thr fsX6)[13]	c.2172-2173 insG (p. S725Vfs*69)[13]	frame shift	frame shift	Jewish Sephardic
1	FANCA	c.4275delT (p.Asp1427Thr fsX6)[13]	4275delT (p.Asp1427Thr fsX6)[13]	frame shift	frame shift	Jewish Sephardic
1	FANCA	c.65G>A (p.Trp22Ter)[25]	c.65G>A (p.Trp22Ter)[25]	missense	missense	Jewish Ashkenazi
1	FANCA	c.65G>A (p.Trp22Ter)[25]	ex1-24del^	missense	large deletion	Jewish mixed
1	FANCA	c.891-893 delGCTG[13]	c.2172-2173 insG (p. S725Vfs*69)[13]	frame shift	frame shift	Jewish Sephardic
1	FANCA	c.2172-2173 insG (p. S725Vfs*69)[13]	ex1,2,4,5del^	frame shift	large deletion	Jewish Sephardic
1	FANCA	c.891-893 delGCTG[13]	c.65G>A (p.Trp22Ter)[25]	frame shift	nonsense	Jewish mixed
1	FANCA	ex15-21 del[28]	ex15-21 del[28]	large deletion	large deletion	Arab Muslim
1	FANCA	ex1-6 del[29]	ex1-6 del[29]	large deletion	large deletion	Arab Christian
1	FANCA	c.189+1G>A (IVS2+1 g>a)^	c.2778+2T>C (IVS28+2 T>C)^	splice site	splice site	Arab Muslim
1	FANCA	p.Arg880Ter[30]	p.Arg880Ter (30)	nonsense	nonsense	Jewish Ashkenazi
1	FANCA	c.3520-3522 delTGG[25]	c.1471-401_1626+395del^	deletion	large deletion	Jewish mixed
7	FANCC	c. 456+4a>t (IVS4+4 a>t)[5]	c. 456+4a>t (IVS4+4 a>t)[5]	splice site	splice site	Jewish Ashkenazi
2	FANCC	p. Gln3Ter^	p. Gln3Ter^	nonsense	nonsense	Arab Muslim
1	FANCC	c. 456+4a>t (IVS4+4 a>t)[5]	del 97116249-97124749[31]	splice site	deletion	Jewish mixed
1	FANCD1	c.5946del (p.Ser1982Argfs*22)[24]	c.7007G>C (p.Arg2336Pro)	frame shift	missense	Jewish mixed
1	FANCD1	c.7579delG (p.V2527X)^	c.9693delA (p.S3231fs16*)^	nonsense	frame shift	Jewish Ethiopian
6	FANCG	c.1742C<G (p.Ser581Ter)^	c.1742C<G (p.Ser581Ter)^	nonsense	nonsense	Arab Muslim
4	FANCG	c.212T>C (p. Leu71Pro)[32]	c.212T>C (p. Leu71Pro)[32]	missense	missense	Arab Muslim
3	FANCG	c.510+3A>G (IVS4+3 A>G)^	c.510+3A>G (IVS4+3 A>G)^	splice site	splice site	Arab Muslim
1	FANCJ	p.Arg251Cys[33]	p.Arg251Cys[33]	missense	missense	Arab Muslim
1	FANCJ	p.Arg848His[34]	p.Arg848His[34]	missense	missense	Arab Muslim
1	FANCJ	p.Gln376Ter^	p.Gln376Ter^	nonsense	nonsense	Arab Muslim
2	@ FANCC	c. 456+4a>t (IVS4+4 a>t)[5]	Unknown	splice site	Unknown	Jewish mixed
4	@	Unknown	Unknown			

#: number of patients; ^: mutations not previously described; @: not included in the analyses; ref: reference number.

References

1. Steinberg-Shemer O, Goldberg TA, Yacobovich J, et al. Characterization and genotype-phenotype correlation of patients with Fanconi anemia in a multi-ethnic population Haematologica 2020;105(7):1825-1834.