

## Twenty years of the Italian Fanconi Anemia Registry: where we stand and what remains to be learned

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## SUPPLEMENTAL DATA

**Table S1. Genotypes of RIAF patients.**

Mutated gene	180 (%)
FANC-A	39 (21,5)
FANC-G	3 (2)
FANC-D2	2 (1)
FANC-C	1 (0.5)
other	3 (2)
unknown	132 (73)

**Table S2. Univariate analysis of RIAF patients.**

Variable	HR	95% CI (lower)	95% CI (upper)	p
Gender	1,177	0,776	1,808	0,458
Date of birth (quartiles)	-	-	-	0,452
Age at diagnosis	0,892	0,853	0,932	<0,001
Congenital abnormalities (yes/no)	1,504	0,831	2,722	0,177
Congenital abnormalities (n)	1,056	0,995	1,121	0,074
Congenital abnormalities (n, except skin)	1,081	1,014	1,152	0,016
Skin hyper/hypopigmentation	0,811	0,518	1,268	0,358
Skeletal abnormalities	1,315	0,82	2,11	0,256
CNS and/or eyes abnormalities	1,023	0,582	1,799	0,936
Renal and urinary tract abnormalities	1,38	0,879	2,168	0,162
Genital tract abnormalities	0,812	0,444	1,485	0,499
Gastrointestinal tract abnormalities	1,726	0,96	3,102	0,068
Endocrine abnormalities	0,474	0,228	0,987	0,46
Cardiovascular abnormalities	1,394	0,602	3,229	0,438
BMF at diagnosis	1,235	0,613	2,487	0,555
BMF anytime	1,877	0,739	4,772	0,186
Hematological malignancies	1,36	0,699	2,645	0,366
Solid tumours	0,743	0,418	1,32	0,311
Head/neck tumours	0,879	0,447	1,725	0,707
HSCT	0,769	0,5	1,183	0,232

**Table S3. Multivariate analysis of non-transplanted and transplanted RIAF patients.**

Variable	<i>Non-transplanted</i>				<i>Transplanted</i>			
	HR	95% CI (lower)	95% CI (upper)	p	HR	95% CI (lower)	95% CI (upper)	p
Gender	2,282	0,792	6,576	0,168	0,429	0,19	0,969	0,042
Date of birth (quartiles)	-	-	-	0,27	-	-	-	0,27
Age at diagnosis	0,784	0,705	0,872	<0,001	0,873	0,821	0,928	<0,001
Congenital abnormalities (n>10, except skin)	0,917	0,017	50,95	0,966	6,355	0,707	57,13	0,099
Skin hyper/hypopigmentation	0,367	0,134	1,007	0,052	1,302	0,483	3,512	0,602
Skeletal abnormalities	1,28	0,276	5,935	0,753	0,693	0,263	1,83	0,459
CNS and/or eyes abnormalities	1,877	0,277	12,72	0,519	0,677	0,196	2,34	0,538
Renal and urinary tract abnormalities	0,908	0,205	4,028	0,899	3,78	1,397	10,224	0,009
Genital tract abnormalities	3,984	0,837	18,955	0,082	0,541	0,175	1,676	0,287
Gastrointestinal tract abnormalities	6,844	1,34	34,948	0,021	0,708	0,143	3,51	0,672
Endocrine abnormalities	27,987	1,751	447,236	0,018	0,218	0,063	0,754	0,016
Cardiovascular abnormalities	0,448	0,089	2,246	0,329	0,059	0,006	0,593	0,016
BMF at diagnosis	1,834	0,474	7,106	0,38	1,732	0,126	23,732	0,681
BMF anytime	4,072	0,364	45,552	0,254	n.a.	n.a.	n.a.	n.a.
Hematological malignancies	4,367	0,782	24,377	0,093	1,77	0,605	5,181	0,297
Solid tumours	0,801	0,215	2,981	0,74	0,617	0,219	1,738	0,361