

Supplementary Table 1. Potentially pathogenic missense mutations in *PRF1* identified in the ASPREE cohort, and the predicted frequency of their co-inheritance with A91V or as homozygotes in 13,1313 participants of the ASPREE study.

Mutation RefSNP	A.A. substitution	Alleles,	A91V/mut,	mut/mut,
		observed, N=	predicted N=	predicted N=
rs141660796	R28C	15	0.290439	0.004284
rs377367451	P382S	11	0.212988	0.002304
rs145463632	G334S	10	0.193626	0.001904
rs144242324	A401V	10	0.193626	0.001904
rs528937278	P22L	6	0.116175	0.000685
rs147051158	A27T	6	0.116175	0.000685
rs139336186	R123H	5	0.096813	0.000476
rs779192397	E30A	4	0.077450	0.000305
rs142059134	G71S	4	0.077450	0.000305
rs140281371	R225Q	3	0.058088	0.000171
rs770354747	N205S	3	0.058088	0.000171
rs759235360	G198R	2	0.038725	0.000076
rs774503938	R177H	2	0.038725	0.000076
rs376289273	P389S	2	0.038725	0.000076
rs201468340	R177C	2	0.038725	0.000076
rs144892262	G548E	2	0.038725	0.000076
rs577367079	D469G	2	0.038725	0.000076
rs374588624	R373C	2	0.038725	0.000076
rs140787739	R357Q	2	0.038725	0.000076
rs749729753	G399V	2	0.038725	0.000076
rs751247865	H222Q	1	0.019363	0.000019
rs189650890	T450M	1	0.019363	0.000019
rs12263572	V135M	1	0.019363	0.000019
rs144024245	R96W	1	0.019363	0.000019
rs775966864	T250M	1	0.019363	0.000019
rs201909472	V38M	1	0.019363	0.000019
rs1031096686	T136S	1	0.019363	0.000019
rs760962796	V482F	1	0.019363	0.000019
rs765636779	H144R	1	0.019363	0.000019
rs902612105	E275K	1	0.019363	0.000019
rs776571416	G220S	1	0.019363	0.000019
rs754079962	D340N	1	0.019363	0.000019
rs150628656	T289M	1	0.019363	0.000019
rs776657932	C76Y	1	0.019363	0.000019
<i>rs753604724</i>	<i>G230D</i>	<i>1</i>	0.019363	0.000019
rs564234285	D134E	1	0.019363	0.000019
rs779956870	T404I	1	0.019363	0.000019
rs146414576	Q209P	1	0.019363	0.000019
rs745902829	K285del	1	0.019363	0.000019
rs142403457	A235V	1	0.019363	0.000019
rs776338709	V348L/M	2	0.019363	0.000019
rs557951406	R87H	1	0.019363	0.000019
rs902124045	R299C	1	0.019363	0.000019
rs748060067	R464Q	1	0.019363	0.000019
rs201235771	F316L	1	0.019363	0.000019
rs753226704	K285Q	1	0.019363	0.000019
rs202216158	F421L	1	0.019363	0.000019
rs774503938	R177H	1	0.019363	0.000019
rs144024245	R96W	1	0.019363	0.000019
rs200383720	H182R	1	0.019363	0.000019
rs781070813	R87C	1	0.019363	0.000019
<i>rs145513232</i>	<i>E447Q</i>	<i>1</i>	0.019363	0.000019
rs751467331	H35Q	1	0.019363	0.000019
rs141864315	V453M	1	0.019363	0.000019
rs201032696	R55C	1	0.019363	0.000019
rs763667216	I266T	1	0.019363	0.000019
rs755578676	R297W	1	0.019363	0.000019
rs200641752	P345S	1	0.019363	0.000019
rs370935432	R123C	1	0.019363	0.000019
rs756992644	L92F	1	0.019363	0.000019
rs766899179	R96Q	1	0.019363	0.000019
TOTAL EXPECTED NUMBER			2.691	0.80

Shown in *italics* are novel mutations in *PRF1*.