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Supplemental Table 1. Exonic rare variants within *PRRX1* region

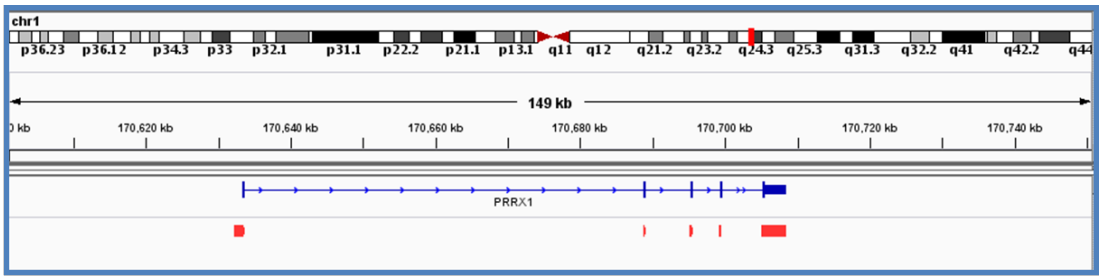
Chromosome	Position*	dbSNP	Reference allele	Alternative allele	Effect	Allele change	Residue change	Damaging Potential[+]
1	170,688,889		A	G	Synonymous	GAA=>GAG	E88E	
1	170,688,894	rs79567938	A	G	Missense	AAG=>AGG	K90R	
1	170,688,957		G	A	Missense	CGT=>CAT	R111H	D
1	170,689,011		G	A	Missense	CGC=>CAC	R129H	D
1	170,689,021		C	T	Synonymous	AAC=>AAT	N132N	
1	170,689,027		C	T	Synonymous	ACC=>ACT	T134T	
1	170,689,033		G	C	Synonymous	GCG=>GCC	A136A	
1	170,695,377		G	A	Missense	CGA=>CAA	R145Q	D
1	170,695,435		C	G	Synonymous	CTC=>CTG	L164L	
1	170,695,456		C	T	Synonymous	GAC=>GAT	D171D	
1	170,695,466	rs201365132	G	T	Missense	GTG=>TTG	V175L	
1	170,695,472		C	G	Missense	CAG=>GAG	Q177E	D
1	170,695,522		G	A	Nonsense	TGG=>TGA	W193*	D
1	170,699,429		T	A	Missense	CTC=>CAC	L204H	
1	170,705,236		A	C	Missense	CAG=>CCG	Q216P	D
1	170,705,313	rs201359953	C	T	Missense	CCA=>TCA	P242S	D

* The position is based on NCBI Genome Build 37 (hg19).

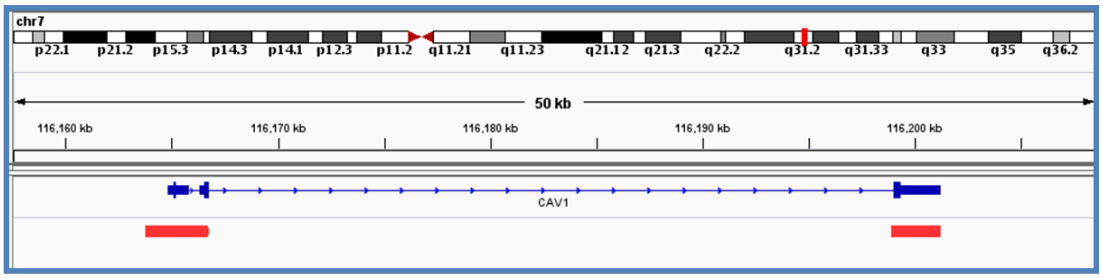
[+] "D" indicates that nonsense variants or variants located in the splicing sites or missense variants predicted to causing damaging effects to the encoded proteins based on PolyPhen (http://genetics.bwh.harvard.edu/pph2/).

Supplemental Figure 1. Genomic location of targeted regions for the 4 AF candidate genes. The blue bars represent the exonic regions, whereas the red bars indicate the overlap with the genomic regions actually covered by our targeted sequencing approach.

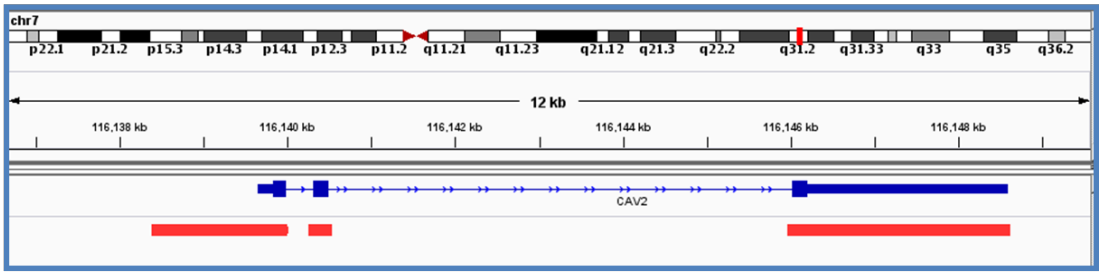
PRRX1



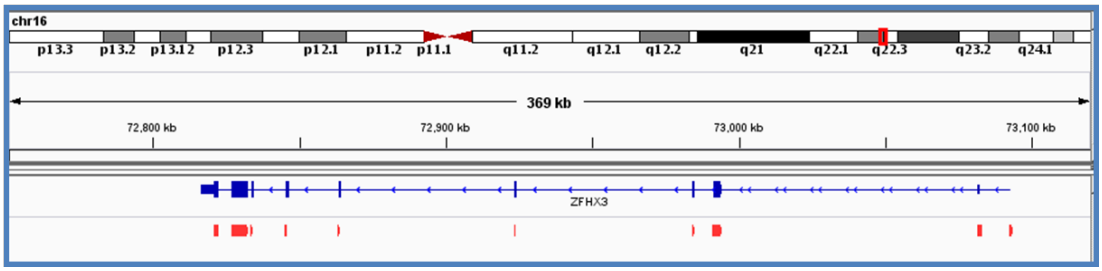
CAV1



CAV2



ZFHX3



Supplemental Figure 2. QQ plot of common variants by unweighted analysis. Each point represents one common variant. The x-axis represents the distribution of -$\log_{10}$ p-values if no association were present, whereas y-axis represents the distribution of -$\log_{10}$ p-values in our study. Only one SNP (rs11265611, P=1.7x10^{-6}) significantly deviated from the diagonal line, indicating a strong association between the SNP and AF.