

Appendix 5: dbSNP polymorphisms detected in African-American FECD cases

Gene	SNP rs #	SNP location within gene	Amino acid change	MAF AA (%)	MAF EA (%)
COL8A2	rs274754	Intron 1	n/a	39.5	21.5
	rs274753	Intron 1	n/a	--	--
	rs75864656	Exon 2	p.R155Q	0.16	0.04
	rs182708720	Exon 2	p.A441A	0.88	0.01
	rs35495320	Exon 2	p.G495G	0.37	6.32
	rs3738360	3' UTR	n/a	45.89	9.17
SLC4A11	rs3827076	5' UTR	n/a	45.53	37.37
	rs60867877	Intron 1	n/a	--	--
	rs41281866	Intron 1	n/a	0.50	0.63
	rs3803957	Intron 1	n/a	21.18	28.36
	rs6139040	Intron 3	n/a	--	--
	rs34460295	Exon 4	p.A135A	4.11	2.02
	rs34520315	Exon 4	p.N150S	4.24	0.02
	rs35262978	Exon 4	p.R158R	1.18	0
	rs3827075	Exon 4	p.R161R	35.59	39.73
	rs3803956	Exon 5	p.S213S	21.18	17.31
	rs3803955	Intron 6	n/a	5.29	23.19
	rs74174550	Intron 6	n/a	26.82	21.20
	rs3803954	Intron 8	n/a	--	--
	rs3803953	Intron 8	n/a	--	--
	rs41281862	Intron 10	n/a	4.22	13.14
	rs6084312	Exon 11	p.T463T	3.95	11.71
	rs3810561	Intron 11	n/a	--	--
	rs41281860	Exon 13	p.N553N	3.86	12.21
	rs115028271	Intron 16	n/a	--	--
	rs2281575	Intron 16	n/a	33.90	42.22
	rs10048856	Intron 16	n/a	16.37	0.56
	rs41281858	Intron 17	n/a	27.28	12.64
	rs58757394	Exon 18	p.T833T	16.23	0.56
	rs115581878	Intron 18	n/a	2.57	0.01
	rs114972331	Intron 18	n/a	--	--
	rs76962118	Exon 19	p.D886D	2.84	0
	rs6051657	3' UTR	n/a	20.99	0.36
	rs77978636	3' UTR	n/a	--	--
ZEB1	rs7918614	Exon 2	p.D64D	17.50	0.76
	rs80036666	Intron 3	n/a	2.18	0
	rs79134358	Exon 5	p.S201S	5.76	0
	rs220060	Intron 5	n/a	15.93	4.63
	rs143232269	Exon 6	p.S263S	0.48	0
	rs35753967	Exon 7	p.K553R	4.95	0
	rs151205909	Exon 7	p.N658K	0.48	0
	rs34846414	Exon 7	p.P687P	5.65	0.047
	rs139581793	Exon 7	p.Q854K	1.59	0

Minor allele frequencies (MAF) taken from the Exome Variant Server (EVS). All coding variants in this table are included in **Figures 1-3** (red font). UTR, untranslated region; n/a, not applicable; AA, African-American; EA, European-American; --, variant not found in EVS database.