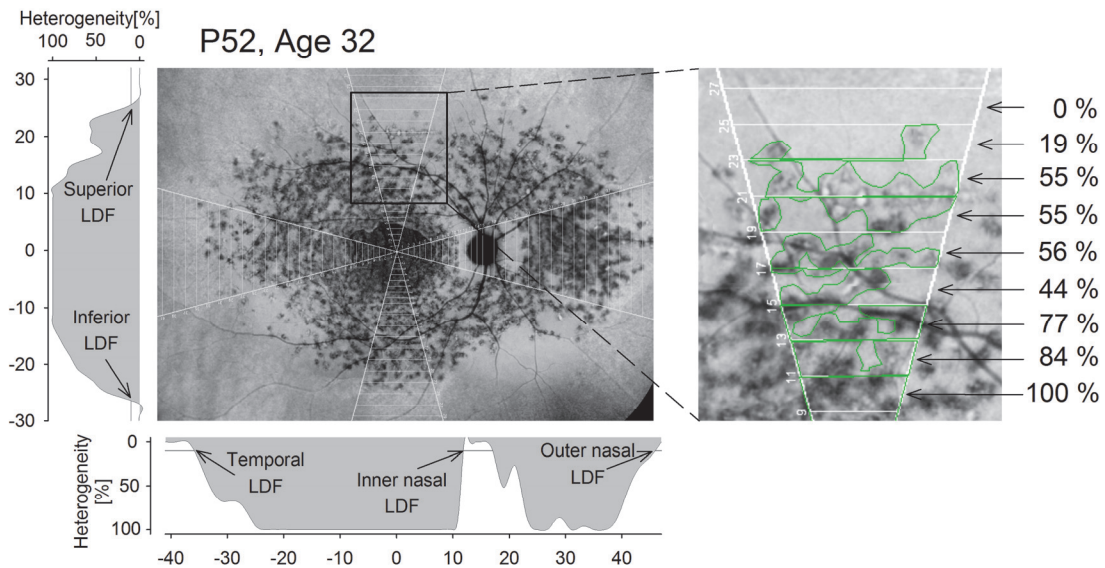


Supplementary Table 1. Molecular and LDF Characteristics of the ABCA4-RD Patients

Patient†	Sex	----- ABCA4 Allele 1 -----		----- ABCA4 Allele 2 -----		Age [yrs]		FU [yrs]	LDF eccentricity along principal meridians [deg]										
		Nucleotide	Amino acid	Nucleotide	Amino acid	Seg.*	Visit 1		Visit 2	Superior		Inferior		Temporal		Inner Nasal		Outer Nasal	
										Visit 1	Visit 2	Visit 1	Visit 2	Visit 1	Visit 2	Visit 1	Visit 2	Visit 1	Visit 2
4	F	c.161 G>A	p.Cys54Tyr	c.2588 G>C	p.Gly863Ala	Y	37.2	42.6	5.5	25.2	30.2	32.0	52.3	27.7	30.9	13.8	15.0	44.9	54.3
7	M	c.161 G>A	p.Cys54Tyr	c.5882 G>A	p.Gly1961Glu	-	40.4			4.7		4.8		5.8		7.1			
11	M	c.454 C>T	p.Arg152Stop	c.5882 G>A	p.Gly1961Glu	Y	21.2	29.0	7.8	5.6	10.9	5.8	12.7	6.6	13.8	5.8	11.6		27.1
12	F	c.454 C>T	p.Arg152Stop	c.5882 G>A	p.Gly1961Glu	Y	19.5	24.4	4.9	3.9	4.8	3.7	3.7	5.8	6.9	4.9	4.9		
21	M	c.768 G>T	p.Val256Val	c.4577 C>T	p.Thr1526Met	Y	11.1	16.3	5.2									63.5	74.6
22	F	c.768 G>T	p.Val256Val	c.5882 G>A	p.Gly1961Glu	Y	45.6	53.9	8.3	5.8	7.5	5.8	7.6	7.7	10.5	12.0	12.7		33.4
23	F	c.768 G>T	p.Val256Val	c.5882 G>A	p.Gly1961Glu	Y	41.8	49.8	8.0	11.2	16.5	7.7	8.8	8.0	10.6	7.2	8.5	21.2	22.2
28	M	c.2041 C>T	p.Arg681Stop	c.6449 G>A	p.Cys2150Tyr	Y	9.4	14.6	5.2	7.6	30.6	5.1	20.3	11.9	27.8	6.9	15.3		33.1
29	M	c.2588 G>C	p.Gly863Ala	c.3322 C>T	p.Arg1108Cys	Y	31.4	35.3	4.0	5.8	5.8	5.7	6.0	6.6	6.8	5.8	7.1		
30	F	c.2588 G>C	p.Gly863Ala	c.4469 G>A	p.Cys1490Tyr	Y	36.6	44.3	7.7	22.4	29.3	17.6	28.8	20.9	29.6			41.1	60.1
32	F	c.2804 T>C	p.Val935Ala	c.5714+5 G>A	-	Y	32.2	37.4	5.3	7.3	15.9	6.9	10.4	10.6	13.0	9.5	11.1	22.9	27.8
34	M	c.3056 C>T	p.Thr1019Met	c.5882 G>A	p.Gly1961Glu	Y	29.6	38.7	9.1	4.8	6.8	9.4	11.8	8.1	14.7	8.7	10.2		25.7
36	F	c.1622 T>C;c.3113 C>T	p.Leu541Pro;p.Ala1038Val	c.2894 A>G	p.Asn965Ser	Y	19.3							52.8				72.1	
47	F	c.4139 C>T	p.Pro1380Leu	c.5882 G>A	p.Gly1961Glu	Y	52.8	59.9	6.0	23.9	27.6	16.2	20.6	24.2	38.7	13.4	13.6	36.7	51.0
49	F	c.4222 T>C	p.Trp1408Arg	c.5714+5 G>A	-	Y	14.1	20.1	6.0	26.8	51.0	16.2	40.5	26.7	47.6	9.8	11.8	37.8	56.1
52	M	c.4530_4531insC	p.Pro1511fs	c.5114 G>A	p.Arg1705Gln	Y	24.3	32.0	7.7	19.6	26.0	18.0	26.5	24.9	35.9	11.4	11.9	38.4	45.8
55	F	c.4577 C>T	p.Thr1526Met	c.6089 G>A	p.Arg2030Gln	Y	49.8	57.3	4.9	24.4	26.7	20.8	22.6	26.7	30.8	8.8	8.8	39.7	58.1
58	F	c.4919 G>A	p.Arg1640Gln	c.5882 G>A	p.Gly1961Glu	Y	54.6	61.9	7.3	22.8	24.6	19.6	20.4	24.6	25.4	13.7	13.7	38.9	44.4
64	F	c.3386 G>T	p.Arg1129Leu	c.5819 T>C	p.Leu1940Pro	Y	25.3	32.4	7.1	7.4	8.8	5.8	12.4	8.9	15.8	7.7	10.1		30.0
66	F	c.2005_2006delAT	p.Met669fs	c.5882 G>A	p.Gly1961Glu	Y	60.7			5.8		6.6		10.3		8.6			
67	F	c.203 C>G	p.Pro68Arg	c.634 C>T	p.Arg212Cys	Y	21.9			3.9		3.8		16.9		4.3			
68	M	c.658 C>T	p.Arg220Cys	c.3051-1 G>A	-	-	33.7			68.7				64.4				71.9	
69	F	c.1086 T>A	p.Tyr362Stop	c.5461-10 T>C	-	Y	17.9	20.1	2.2	54.4	59.8							62.6	63.6
70	M	c.1822 T>A	p.Phe608Ile	c.1022 A>G	p.Glu341Gly	Y	7.0	12.0	5.0	5.1	48.4	4.6		7.1	47.3	5.9	15.3		62.5
71	M	c.2461 T>A	p.Trp821Arg	c.2894 A>G	p.Asn965Ser	Y	12.3	17.5	5.3	10.2	29.8	9.2	24.5	16.4	32.2	8.4	12.2	31.8	50.5
72	M	c.2461 T>A	p.Trp821Arg	c.2894 A>G	p.Asn965Ser	Y	16.1	21.4	5.3	25.0	47.1	23.0	41.7	22.5	38.4	13.8	13.8	32.9	55.7
73	F	c.2588 G>C	p.Gly863Ala	c.5018+2 T>C	-	Y	34.0			53.8		49.6		48.6				64.7	
74	M	c.2588 G>C	p.Gly863Ala	c.5018+2 T>C	-	Y	21.8	28.7	6.9	20.8	39.7	18.5	25.4	24.1	39.2	16.5		35.6	52.9
75	M	c.2588 G>C	p.Gly863Ala	c.4978 C>T	p.Pro1660Ser	-	27.5	32.1	4.6	22.2	24.6	20.2	24.1	22.5	23.8	9.5	10.6	36.0	43.7
76	F	c.2966 T>C	p.Val989Ala	c.4253+5 G>T	-	Y	16.1			6.7		7.3		10.4		8.6			
77	F	c.2966 T>C	p.Val989Ala	c.4253+5 G>T	-	Y	17.4			12.5		11.4		20.2		10.8		33.7	
78	F	c.2966 T>C	p.Val989Ala	c.4253+5 G>T	-	Y	18.3			5.6		8.2		8.0		6.7		39.9	
79	F	c.1622 T>C;c.3113 C>T	p.Leu541Pro;p.Ala1038Val	c.5882 G>A	p.Gly1961Glu	Y	27.9	30.2	2.3	6.9	8.0	9.5	10.9	14.0	16.6	10.0	10.0		
80	F	c.1622 T>C;c.3113 C>T	p.Leu541Pro;p.Ala1038Val	c.5773 A>G	p.Arg1925Gly	Y	34.3			18.2		18.4		21.0		11.0		29.2	
81	M	c.3292 C>T	p.Arg1098Cys	c.4352+5 G>A	-	Y	10.1			4.4		3.5		4.6		4.4			
82	M	c.3386 G>T	p.Arg1129Leu	c.5882 G>A	p.Gly1961Glu	-	18.3	21.8	3.5	5.7	9.5	6.2	12.4	7.5	12.6	9.8	10.6		
83	M	c.3190+21 A>G	-	c.4139 C>T	p.Pro1380Leu	-	8.7			26.2		24.9		33.4		12.6		39.8	
84	M	deletion of exon 7	-	c.4457 C>T	p.Pro1486Leu	Y	36.9			69.8				55.9				73.4	
85	F	deletion of exon 7	-	c.4457 C>T	p.Pro1486Leu	Y	51.4			68.5				-				71.9	
86	M	c.203 C>T;c.5882 G>A	p.Pro68Leu;p.Gly1961Glu	c.5882 G>A	p.Gly1961Glu	Y	20.5			16.9		11.7		18.9		9.6		18.9	
87	M	c.203 C>T;c.5882 G>A	p.Pro68Leu;p.Gly1961Glu	c.5882 G>A	p.Gly1961Glu	Y	23.1			11.2		10.6		18.8		10.8			
88	F	c.5312+1 G>A	-	c.5882 G>A	p.Gly1961Glu	-	56.0			25.3		14.8		14.7		12.1		46.2	
89	F	c.5461-10 T>C	-	c.6449 G>A	p.Cys2150Tyr	-	39.7			66.9		47.2		55.1					
90	F	c.6213 C>G	p.Tyr2071Stop	c.6319 C>T	p.Arg2107Cys	Y	23.7	27.1	3.4	28.1	32.2	18.1	19.0	26.7	30.0	11.6	12.8	34.9	43.6

†Patient numbers 4-66 correspond to those previously published in Cideciyan et al., 2009. Patients 22,23; 71,72; 73,74; 76,77,78; 84,85; 86,87 are siblings

\*Parental segregation of the reported alleles confirmed (Y, yes) or not available (-).



**SUPPLEMENTAL FIGURE 1.** Windmill analysis to determine the eccentricity of the LDF in *ABCA4*-RD along the four major meridians. Four 30° wedges starting at the anatomical fovea (determined using registered OCT image) and centered along each principle meridian are overlaid onto the wide-angle composite of NIR-RAFI. For the superior, temporal and inferior meridians, each wedge is divided into 1° segments up to eccentricities of 9° and 2° segments at further eccentricities. For the nasal meridian, 1° segments extend up to 15° eccentricity in order to obtain greater parapapillary detail. For each segment within the wedge, the fraction of the area showing local signal heterogeneity (Inset, green outline) is plotted as a function of eccentricity. Along each meridian, the furthest eccentricity showing a transition from less than 10% area of heterogeneity to greater than 10% is defined as the LDF (arrows on graphs). Along the nasal meridian, an inner nasal LDF is defined between the fovea and the optic nerve head and an outer nasal LDF at further eccentricities.