

SUPPLEMENTARY MATERIAL

Supplementary Table S1. Subfoveal choroidal thickness (SFCT) associated with preserved, disrupted or lost ellipsoid zone (EZ) in CHM, versus normal controls matched for age and refractive error. 42 eyes of 23 CHM patients (mean age 37.5 yr, range 12-63) and 40 eyes of 20 normal controls (mean age 37.6 yr, range 13-62) were recruited. The mean SFCT was 162.5 μ m in CHM patients (range 21-336) and 302 μ m (range 249-354) in controls. All subjects had refractive error between -6.0 and +6.0 dioptres.

No.	Age	OD			OS			CHM mutation
		SFCT	EZ	Focus	SFCT	EZ	Focus	
CHOROIDEREMIA								
1	63	103	disrupted	+3.89	110	disrupted	+3.89	-
2	59	42	lost	+0.78	110	preserved	+1.17	-
3	59	34	lost	+1.32	37	lost	+1.32	c.877C>T
4	58	60	lost	+1.78	76	lost	+1.55	c.173G>A
5	53	151	disrupted	-2.58	171	disrupted	-2.58	c.562_655delTCAC
6	49	38	lost	-2.35	-	-	-	c.1300_1303delGTGG
7	49	137	preserved	-1.13	-	-	-	c.189+1 G>C
8	45	158	preserved	-3.84	114	preserved	-3.84	c.1762_1765del
9	43	110	disrupted	-4.69	120	disrupted	-4.69	c.529delG
10	42	45	lost	-1.74	69	lost	-1.74	c.877C>T
11	41	143	disrupted	-1.97	21	lost	-1.97	del exon 10-15
12	39	-	-	-	194	preserved	+0.97	c.1264C>T
13	37	230	disrupted	+0.17	-	-	-	c.492_493delGA
14	33	221	disrupted	+0.43	192	disrupted	+2.19	del exon 1-15
15	28	175	disrupted	+0.40	171	disrupted	+0.40	del exon 1-15
16	26	199	disrupted	-4.26	206	disrupted	-2.81	c.715C>T
17	25	218	disrupted	-2.40	205	disrupted	-0.90	c.1099_1100insTACC
18	24	191	disrupted	-5.25	200	disrupted	-5.25	c.715C>T
19	22	323	disrupted	-3.34	336	disrupted	-1.79	c.715C>T
20	21	232	preserved	+0.86	225	preserved	-0.82	c.179delA
21	19	279	disrupted	-1.51	254	disrupted	-1.50	c.492_493delGA
22	16	201	preserved	+0.94	246	preserved	+0.94	c.1780delC
23	12	245	disrupted	+0.63	237	disrupted	+0.65	c.940+2T>A
NORMAL CONTROLS								
1	62	316		+0.09	351		+0.09	
2	61	278		-1.05	271		-1.05	
3	56	294		-4.03	274		-2.39	
4	53	257		-1.28	320		-1.28	
5	52	305		+0.74	302		+0.74	
6	49	306		+1.64	314		+1.64	
7	47	320		+1.70	280		+1.70	
8	47	334		+4.72	337		+4.72	
9	41	249		+0.94	255		+0.94	
10	39	305		-3.73	312		-3.73	
11	37	260		+1.24	260		+1.24	
12	34	293		+0.10	323		+1.31	
13	31	353		+0.85	323		+0.85	
14	25	285		-3.19	309		-3.35	
15	25	341		+0.66	354		+0.66	
16	23	325		-1.02	292		-1.02	
17	22	281		-1.82	253		-1.82	
18	19	339		-4.03	328		-4.03	
19	17	328		+0.33	309		+0.33	
20	13	275		+0.10	268		+0.10	

Supplementary Figure S1. Fluctuations in the number of outer retinal cysts (ORCs) following subretinal gene therapy. The total number of ORCs seen on all B-scans of the standardized OCT for six treated eyes (A to F, corresponding to panels in Figure 5) was plotted against time post-gene therapy. While most eyes showed a slight reduction in the number of ORCs at 1 month (likely a result of mechanical disturbance from retinal detachment), linear regression analysis over 6 months showed no significant difference between the regression slopes for each eye ($F(5,6)=1.29$, $P=0.38$).

