

Table S1. Number of patients in CNGA3 and PDE6C achromatopsia, grouped by structural changes obtained from OCT.

Features		OCT findings			Alternative hypothesis
		ISe change (-)	ISe change (+)	p-value	
Genes with variants, n	CNGA3	5	3	0.023 ^{*,a}	CNGA3-ACHM group has higher proportion of normal ISe
	Others	2	10		
	PDE6C	0	8	0.003 ^{*,b}	PDE6C-ACHM group has lower proportion of normal ISe
	Others	7	5		

^a Boschloo's one-tailed exact test comparing the proportion of patients with normal ISe. The alternative hypothesis of each test is described in the right-most column. OCT findings were derived from the date of the most recent OCT images. Abbreviations: OCT, optical coherence tomography; ISe, inner segment of ellipsoid zone.

Table S2. Results of pairwise comparison of one-sided, exact tests comparing features associated with structural changes.

Alternative Hypothesis			
The group of patients with the causative gene in the row has a higher proportion of normal ISe on the OCT than the groups of patients with the gene in the column.			
	CNGB3	PDE6C	GNAT2
CNGA3	0.094	0.005 *	1.0
CNGB3		N/A	1.0
PDE6C			1.0

^a Boschloo's one-tailed exact test comparing the proportion of patients with normal ISe. OCT findings were derived from the date of the most recent OCT images. Abbreviations: OCT, optical coherence tomography; ISe, inner segment of ellipsoid zone.

Table S3. Number of patients with structural changes obtained from OCT.

Genes with variants	OCT Findings		p-value
	ISe change (-)	ISe change (+)	
CNGA3	5	3	0.047 ^{*,a}
Others	2	10	
CNGB3	0	2	0.557 ^a
Others	7	11	
PDE6C	0	8	0.007 ^{*,a}
Others	7	5	
GNAT2	2	0	0.121 ^a
Others	5	13	

^a Boschloo's two-sided exact test, OCT findings, and age were derived from the date of the most recent OCT images. Abbreviations: OCT, optical coherence tomography; ISe, inner segment of ellipsoid zone.