Supplementary Information

Table S1. Genotypic and demographic data of subjects with achromatopsia. The far right column indicates the best corrected visual acuity (BCVA) measured on an Early Treatment Diabetic Retinopathy Study (ETDRS) chart at 4 meters in the eye imaged with AOSLO (OD refers to the right eye).

Subject ID	Age	Gene	Allele 1	Allele 2	BCVA	Ocular axial length (mm)
JC_10069	18	*CNGA3*, heterozygous	c.847C>T-p.Arg283Trp	c.542A>G-p.Tyr181Cys	OD 20/125	23.11
JC_10028	13	*CNGB3*, homozygous	c.1148delC-p.Thr383fs	c.1148delC-p.Thr383fs	OD 20/100	21.78
KS_10088	64	*CNGA3*, heterozygous	c.450-1G>A	c.1557G>A-p.Met519Ile	OD 20/120	25.20
JC_10089	40	*CNGB3*, homozygous	c.1148delC-p.Thr383fs*	c.1148delC-p.Thr383fs	OS 20/125	21.67

*****A variation of unknown significance was also identified in PDE6C (p.Lys585 Asn:c.1755G>T).

Table S2. Ex vivo measurements of inner segment diameter between 0 and 12mm. Inner segment diameters measured between 0 and 0.39mm (2 retinas) were pooled across the superior, inferior, nasal and temporal meridians due to symmetry. Measurements from 1.0 to 11.94mm are averaged across 4 retinas.

		Inner segment diameter (mean ± SD, μm)	
Eccentricity (mm)	Eccentricity (degrees)	Nasal	Temporal
0.00	0.0	2.23 ± 0.11	2.23 ± 0.11
0.05	0.2	2.48 ± 0.04	2.48 ± 0.04
0.10	0.3	2.99 ± 0.03	2.99 ± 0.03
0.15	0.5	3.38 ± 0.14	3.38 ± 0.14
0.20	0.7	3.75 ± 0.25	3.75 ± 0.25
0.25	0.8	4.00 ± 0.23	4.00 ± 0.23
0.29	1.0	4.16 ± 0.22	4.16 ± 0.22
0.39	1.3	4.49 ± 0.19	4.49 ± 0.19
1.00	3.4	6.27 ± 0.71	6.84 ± 0.51
1.99	6.8	6.84 ± 0.85	7.33 ± 0.49
2.99	10.3	7.61 ± 0.45	7.54 ± 0.6
3.98	13.7	*	7.88 ± 0.69
4.98	17.1	7.84 ± 0.65	8.10 ± 0.73
5.97	20.5	7.81 ± 0.72	8.26 ± 0.61
6.97	23.9	7.78 ± 0.56	8.38 ± 0.55
7.96	27.4	7.86 ± 0.44	8.30 ± 0.58
8.96	30.8	7.98 ± 0.33	8.42 ± 0.57
9.95	34.2	8.09 ± 0.24	8.51 ± 0.59
10.95	37.6	8.21 ± 0.15	8.57 ± 0.63
11.94	41.0	8.32 ± 0.11	8.55 ± 0.57

*****The optic nerve head precluded measurement along the nasal meridian at this eccentricity.

Table S3. Rod photoreceptor size estimate using nearest neighbor analysis in achromatopsia subject JC_10069 compared to minimum foveal cone size estimate resolvable with split-detector in normal volunteer AD_1225. In achromat JC_10069, rod photoreceptors across the retina are significantly enlarged compared to both normal rods, as well as normal foveal cones (Kruskal-Wallis test, Dunn's multiple comparisons test of each group to normal cones at the fovea, see p-values on right column). This substantial difference in size (~1 μm or 40%) makes rods resolvable in the achromatopsia subjects, and not in normal subjects in split-detection.

Retinal location	Subject	NN*± SD (μm)	# cells	p
Cones – fovea	Normal	2.63 ± 0.40	123	-
Rods – 10° eccentricity	Normal	2.31 ± 0.32	70	$p < 0.001$
Rods – fovea	Achromat	3.34 ± 0.45	145	$p < 0.001$
Rods – 5° eccentricity	Achromat	3.19 ± 0.51	253	$p < 0.001$

*Photoreceptor nearest neighbor distance**.**