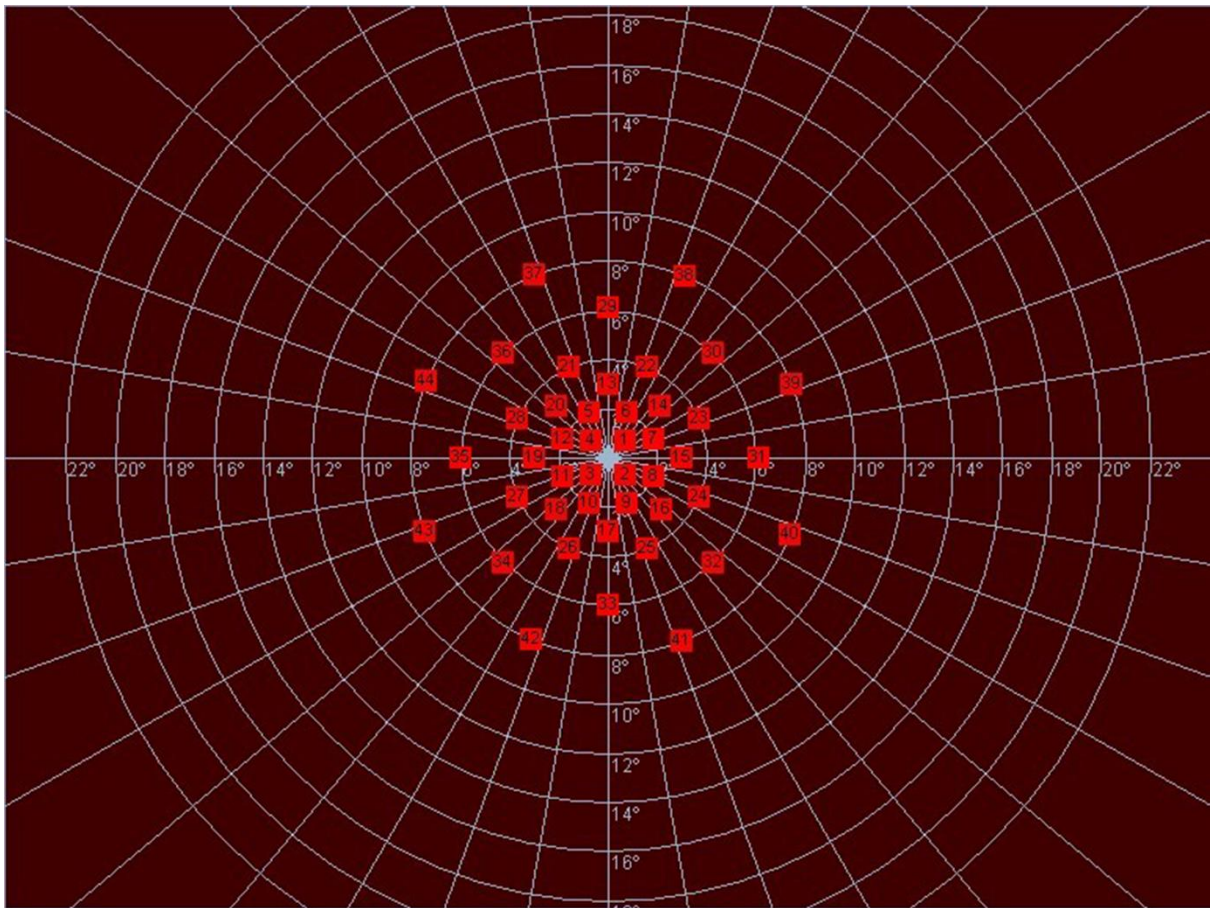


[Online-only material]

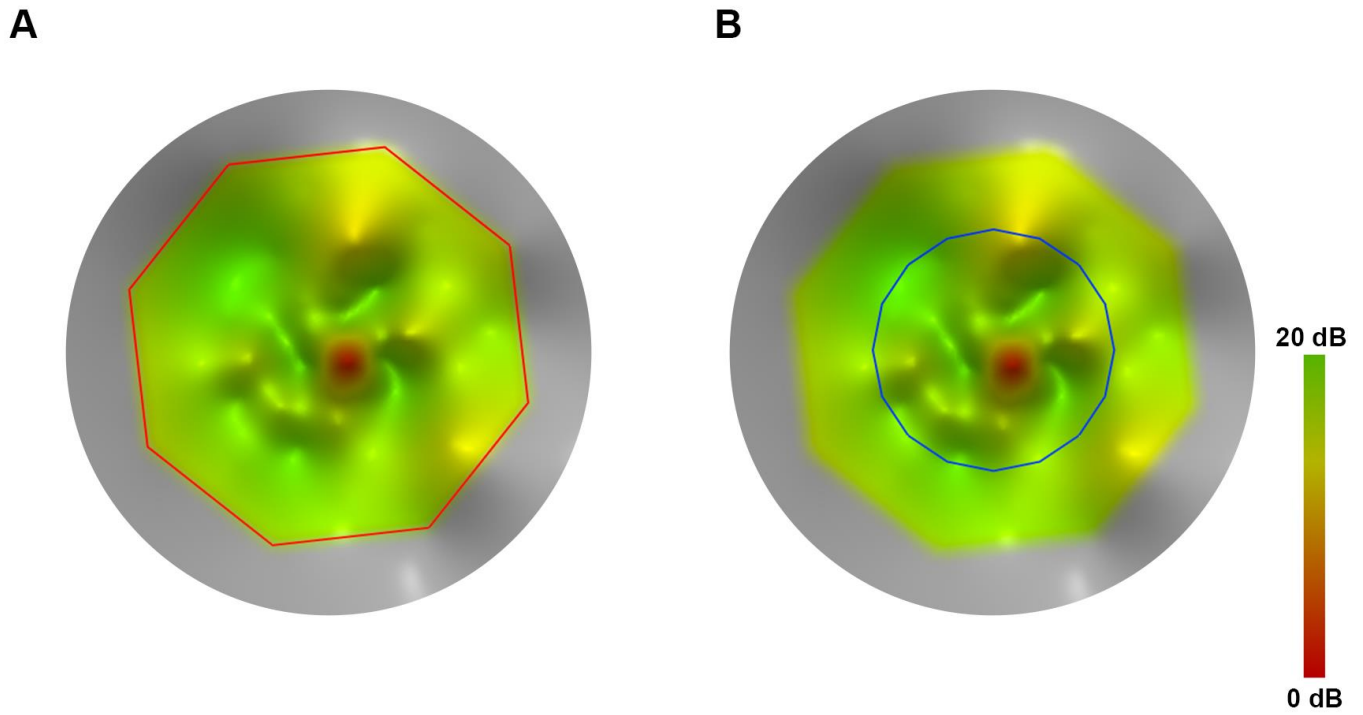
1. **eFigure 1: Microperimetry testing grid pattern**
2. **eFigure 2: V_{TOT} and V_{5° , volumetric indices of retinal function**
3. **eTable 1: Demographics and Genetics**



eFigure 1: Microperimetry testing grid pattern.

The customized testing grid consisted of 44 testing locations and 8° radius to cover the macular and paramacular region.

The grid pattern was of radial design with centrally-condensed spacing and covered the macular and para-macular region.



eFigure 2: V_{TOT} and V_{5° volumetric indices of retinal function

Topographical Model of MP1 microperimetry testing of the right eye, in a subject with *CNGA3*-ACHM (MM_0446). (A) Red outlines the total volume (V_{TOT}), which represented the entire tested field. (B) Blue outlines the central five degrees of the field volume (V_{5°). The volume represents the total sensitivity across the solid angle of the base of the test grid for V_{TOT} and the solid angle of a 5° radius selection for V_{5° , and is reported in units of decibel-steradians (dB-sr). The calibration scale in the figures, reflects the topographic display in sensitivity (dB) from a low of 0 to a ceiling of 20dB.

eTable 1: Demographics, Genetics and OCT Findings

Patient ID*	Sex	Age (years)	Follow-up Time (years)	Hypoplasia	OCT Group [§]	Gene	cDNA	Protein	cDNA	Protein
MM_0014†	F	29.08	7.58	YES	2	<i>CNGA3</i>	848A>G	Arg283Gln	667T>C	Arg223Trp
MM_0015†	F	22.42	6.83	YES	2	<i>CNGA3</i>	848A>G	Arg283Gln	667T>C	Arg223Trp
MM_0165	M	7.00	8.08	YES	2	<i>CNGA3</i>	1641A>C	Phe547Leu	1641A>C	Phe547Leu
MM_0167	F	22.42	6.58	NO	1	<i>CNGA3</i>	847T>C	Arg283Trp	1279T>C	Arg427Cys
MM_0168	M	35.33	7.83	NO	1	<i>CNGA3</i>	661T>C	Arg221X	848A>G	Arg283Gln
MM_0169	M	11.08	6.83	NO	1	<i>CNGA3</i>	485A>T	Asp162Val	485A>T	Asp162Val
MM_0170	M	10.17	7.50	YES	1	<i>CNGA3</i>	1642A>G	Gly548Arg	67T>C	Arg23X
MM_0171	M	17.50	7.25	YES	2	<i>CNGA3</i>	1001T>C	Ser334Phe	1360T>A	Lys454X
MM_0446	F	49.67	7.25	YES	2	<i>CNGA3</i>	67T>C	Arg23X	67T>C	Arg23X
MM_0480	F	29.33	7.00	NO	1	<i>CNGA3</i>	661T>C	Arg221X	848A>G	Arg283Gln
MM_0004	M	23.75	7.25	YES	2	<i>CNGB3</i>	1148delC	Thr383Ile fs * 13	1148delC	Thr383Ile fs * 13
MM_0022	F	33.17	6.67	YES	2	<i>CNGB3</i>	1148delC	Thr383Ile fs * 13	1148delC	Thr383Ile fs * 13
MM_0067	M	29.83	5.25	YES	1	<i>CNGB3</i>	1148delC	Thr383Ile fs * 13	1148delC	Thr383Ile fs * 13
MM_0123	M	47.75	7.42	NO	2	<i>CNGB3</i>	1148delC	Thr383Ile fs * 13	1148delC	Thr383Ile fs * 13
MM_0124	M	11.75	6.92	YES	2	<i>CNGB3</i>	1148delC	Thr383Ile fs * 13	1148delC	Thr383Ile fs * 13
MM_0147‡	F	19.08	7.83	YES	2	<i>ATF6</i>	c.1187+5G>C	p.Asn366His fs*12	c.1187+5G>C	p.Asn366His fs*12
MM_0152‡	F	23.08	7.92	YES	2	<i>ATF6</i>	c.1187+5G>C	p.Asn366His fs*12	c.1187+5G>C	p.Asn366His fs*12
MM_0106	M	43.83	7.58	NO	1	<i>GNAT2</i>	c.843-844insAGTC	p.His282Ser fs*11	c.843-844insAGTC	p.His282Ser fs*11

*All patients were previously reported^{15, 19, 21, 22, 25, 27}

[§] OCT Group: Group 1: presence of foveal ellipsoid zone, ii) Group 2: absence of foveal ellipsoid zone.

‡ Siblings