Supplementary Table 1. Candidate photoreceptor and macular dystrophy genes.

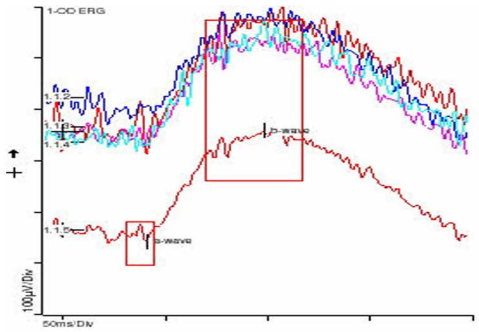
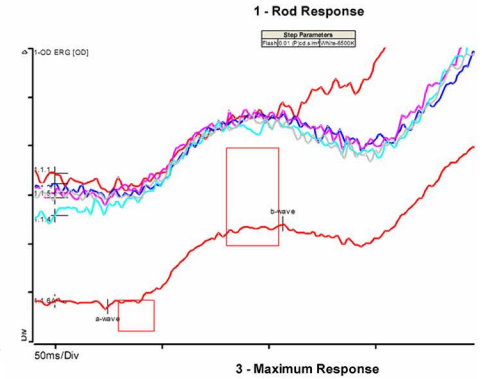
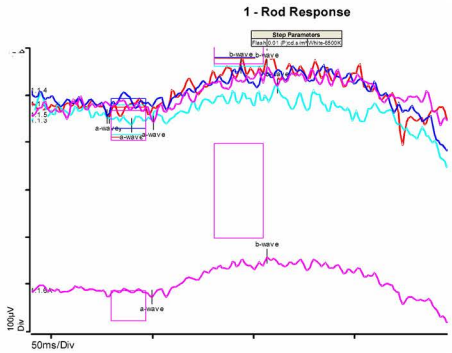
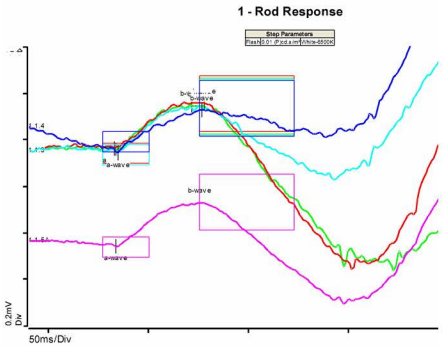
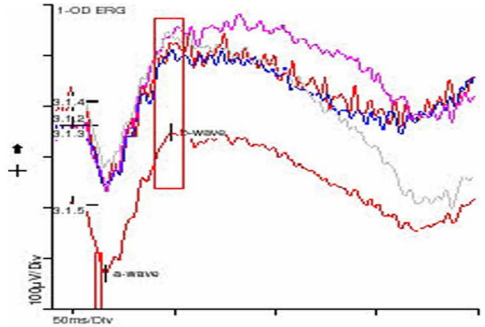
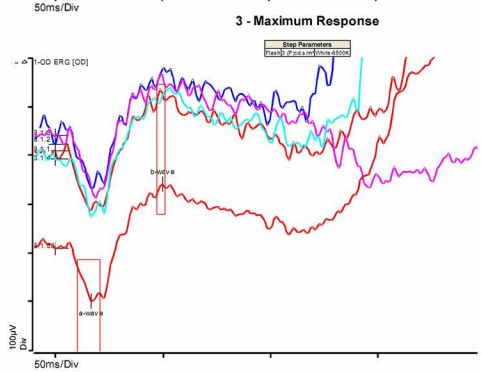
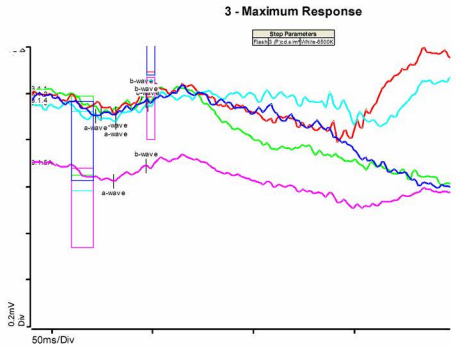
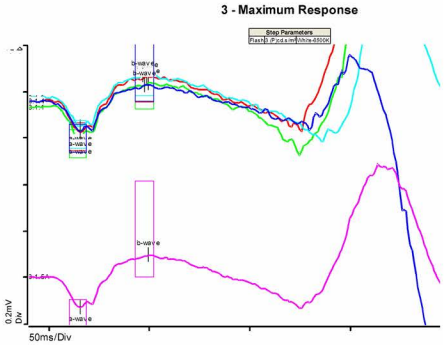
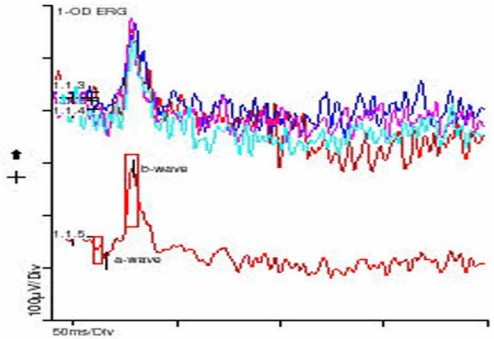
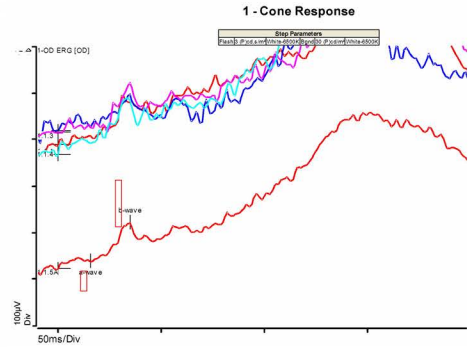
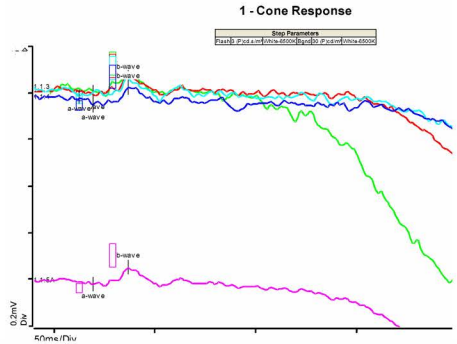
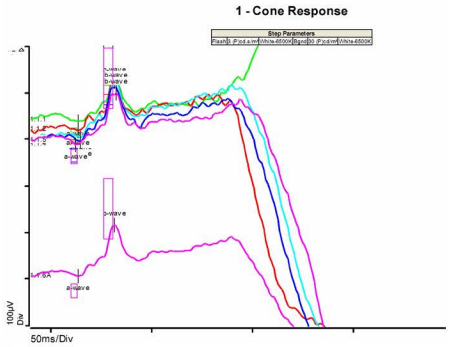
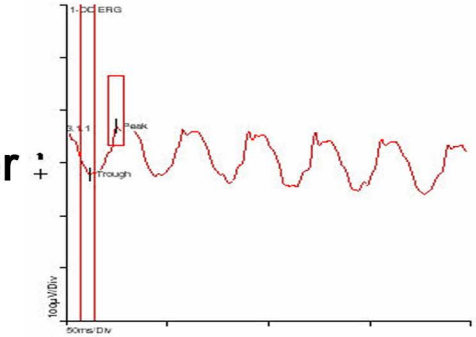
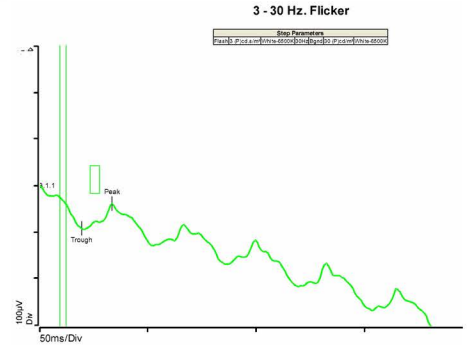
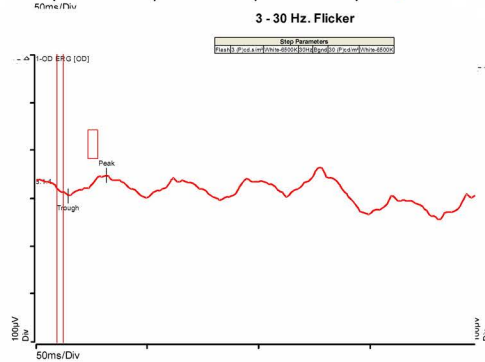
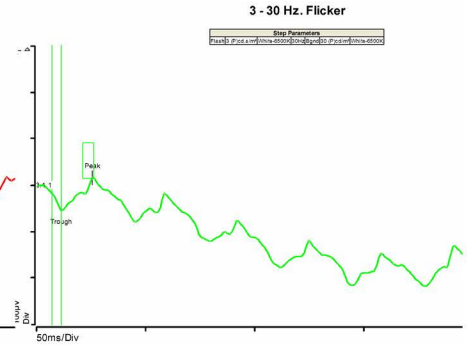
Pattern Dystrophy	Cone-Rod Dystrophy	Rod-Cone Dystrophy
ABCA4	*ADAM9*	*CA4*
ATXN7	*AIPL1*	*CERKL*
BEST1	*ALMS1*	*CRB1*
ELOVL4	*BBS10*	*RHO*
IMPG1	*C8orf37*	
IMPG2	*CACNA1F*	
RP1L1	*CDHR1*	
RPGR	*CNNM4*	**Cone Development, Function**
		BLIMP1
	CRX	*NEUROD2*
	DFNB59	*NR2E3*
Cone Dystrophy	*GUCY2D*	*NRL*
CACNA2D4		
CNGA3	*PITPNM3*	*OPN1LW*
CNGB3	*POC1B*	*OPN1MW*
GUCA1A	*RAB28*	*OPN1SW*
KCNV2	*RAXL1*	*OTX2*
PDE6C	*RIMS1*	*RAX*
PDE6H	*RPGRIP1*	*RORB2*
PROM1	*SEMA4A*	*RXRG*
PRPH2	*TTLL5*	*THRB2*

Supplementary Table 2. Variants detected in candidate photoreceptor or macular dystrophy genes.

Position	Reference	Variant	Gene	OMIM Disease	Transcript IDDNA variant	Protein varian	Case 1	Case 2	Case 3	Grantham	PHRED/C-score	SIFT	SIFT Score	PolyPhen-2 Prediction	PhyloP p-value	dbSNP ID	1000 G MAF	EVS MAF	ExAC MAF
Chr1:94517254	C	G	**ABCA4**	**Stargardt disease, Cone-rod dystrophy 3 (recessive)**	NM_000350.2 c.2588G>C	p.G863A	**Het**			60	26.4	Damaging	0.002	Probably Damaging	1.25E-04	rs76157638	0.0012	0.0052	0.0051
Chr3:100962651	G	T	**IMPG2**	**Vitelliform macular dystrophy 5 (dominant)**	NM_016247.3 c.2524C>A	p.L842M	**Het**			15	23.1	Damaging	0	Probably Damaging		rs149896250	0.0006	0.0002	0.0004
Chr3:101039185	G	T	**IMPG2**	**Vitelliform macular dystrophy 5 (dominant)**	NM_016247.3 c.32C>A	p.S11Y		**Het**		144	22.6	Damaging	0	Possibly Damaging		rs137927107	0.0004	0.0003	0.0001
Chr8:10464692	A	T	**RP1L1**	**Occult macular dystrophy (dominant)**	NM_178857.5 c.6916T>A	p.W2306R		**Het**		101	8.732	Damaging	0.02	Benign		rs200403049	0.0026	0.003	0.0009
Chr8:10469738	C	T	**RP1L1**	**Occult macular dystrophy (dominant)**	NM_178857.5 c.1870G>A	p.A624T		**Het**		58	2.405	Tolerated	0.137	Benign		rs141846905	0.0024	0.003	0.0009
Chr8:38961159	G	T	**ADAM9**	**Cone-rod dystrpohy 9 (recessive)**	NM_003816.2 c.2400G>T	p.Q800H			**Het**	24	21.6	Damaging	0.004	Probably Damaging	4.49E-04	rs145276865	0	0.0002	0.00006
Chr17:7917269	G	A	**GUCY2D**	**Cone-rod dystrophy 6 (dominant)**	NM_000180.3 c.2335G>A	p.E779K		**Het**		56	29	Damaging	0.006	Probably Damaging	3.94E-06		0.0004	0	0.000008

Supplementary Table 3. Variants detected in 2 of 3 probands in genes with known retinal expression.

Position	Reference	Sample	Gene	OMIM Function, Disease; Domain	Transcript ID	Transcript Variant	Protein Variant	Case 1	Case 2	Case 3	Grantham	PHRED/C-score	SIFT	SIFT Score	PolyPhen-2	PhyloP p-value	dbSNP ID	1000 G MAF	EVS MAF	ExAC MAF
Chr5:89981639	C	T	**GPR98**	**Usher syndrome 2C (recessive); Calx-beta domain**	NM_032119.3	c.6317C>T	p.A2106V	**Het**			64	27.5	Damaging	0.02	Probably Damaging	5.78E-07	186999408	0.18	0.0036	0.0016
Chr5:90016759	T	C	**GPR98**	**Usher syndrome 2C (recessive); No domain**	NM_032119.3	c.9631T>C	p.S3211P		**Het**		74	22.6	Tolerated	0.09	Probably Damaging			0	0	0.000033
Chr5:90106814	G	A	**GPR98**	**Usher syndrome 2C (recessive); Calx-beta domain**	NM_032119.3	c.15737G>A	p.R5246Q	**Het**			43	24.3	Tolerated	0.44	Possibly Damaging			0	0	0.0000082
Chr6:152720959	C	T	**SYNE1**	**Emery-Dreifuss muscular dystrophy 4 (dominant); Spinocerebellar ataxia 8 (recessive)**	NM_033071.3	c.7030-1G>A	Splice	**Het**			NA	18.06	NA	NA	NA	1.14E-06		0	0	0
Chr6:152847259	C	T	**SYNE1**	**Emery-Dreifuss muscular dystrophy 4 (dominant); Spinocerebellar ataxia 8 (recessive)**	NM_033071.3	c.181G>A	p.V61I		**Het**		29	17.8	Tolerated	0.14	Benign		151156420	0.0006	0.0005	0.00014
Chr10:73464764	A	G	**CDH23**	**Usher syndrome 1D (recessive); Cadherin-like domain**	NM_022124.5	c.2830A>G	p.S944G			**Het**	56	24.3	Damaging	0.01	Probably Damaging	3.82E-04	188098974	0.0006	0.0006	0.0013
Chr10:73567371	G	A	**CDH23**	**Usher syndrome 1D (recessive); Cadherin-like domain**	NM_022124.5	c.8407G>A	p.V2803I		**Het**		29	23.9	Tolerated	0.41	Possibly Damaging	6.56E-05	369697366	0.0004	0.0001	0.00013
Chr10:73574991	C	T	**CDH23**	**Usher syndrome 1D (recessive); No domain**	NM_022124.5	c.10021C>T	p.R3341C			**Het**	180	14.85	Damaging	NA	Possibly Damaging		370074117	0	0.0001	0.00026
Chr20:368854	G	A	**TRIB3**	**Homolog of Drosophila *Tribbles* , modulator of AKT**	NM_021158.4	c.200G>A	p.R67H		**Het**		29	18.23	Tolerated	0.07	Benign		61746925	0.0032	0.0051	0.0012
Chr20:372180	C	T	**TRIB3**	**Homolog of Drosophila *Tribbles* , modulator of AKT**	NM_021158.4	c.541C>T	p.R181C			**Het**	180	33	Damaging	0.01	Probably Damaging		149447454	0.0004	0.0009	0.0006
Chr22:38109357	C	G	**TRIOBP**	**Deafness 28 (recessive)**	NM_001039141.2	c.395C>G	p.S132C			**Het**	112	6.772	Damaging	NA	Possibly Damaging			0	0	0.000041
Chr22:38130889	C	G	**TRIOBP**	**Deafness 28 (recessive)**	NM_001039141.2	c.4546C>G	p.P1516A	**Het**			27	5.953	Tolerated	NA	Benign			0	0	0
Chr22:38134708	C	G	**TRIOBP**	**Deafness 28 (recessive)**	NM_001039141.2	c.5166C>G	p.D1722E		**Het**		0	14.01	Tolerated	0.19	Benign		183189469	0.0004	0.0003	0.00015
Chr22:38153637	A	C	**TRIOBP**	**Deafness 28 (recessive)**	NM_001039141.2	c.5705A>C	p.K1902T			**Het**	78	12.78	Damaging	0.02	Probably Damaging	2.61E-03	138804394	0.0002	0.0005	0.0013

	Normal	Case 1	Case 2	Case 3
Rod				
Bright White				
Cone				
30 Hz Flicker				

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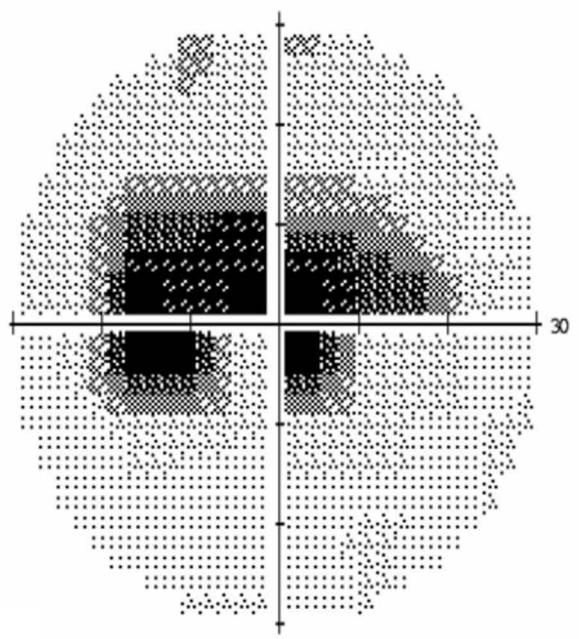
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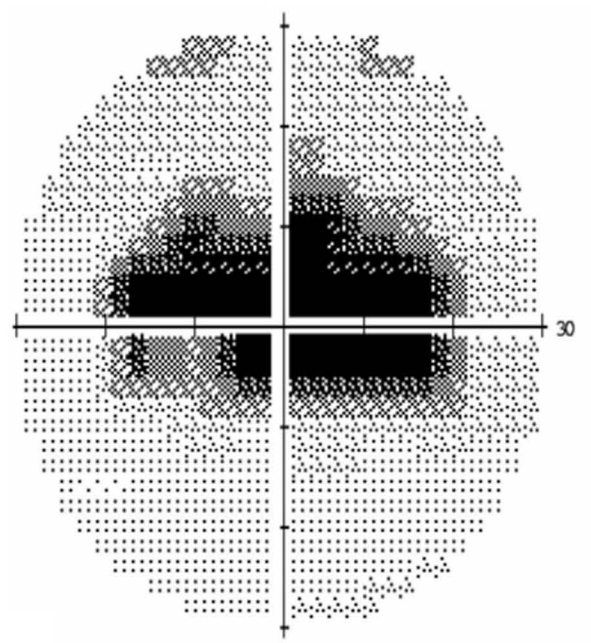
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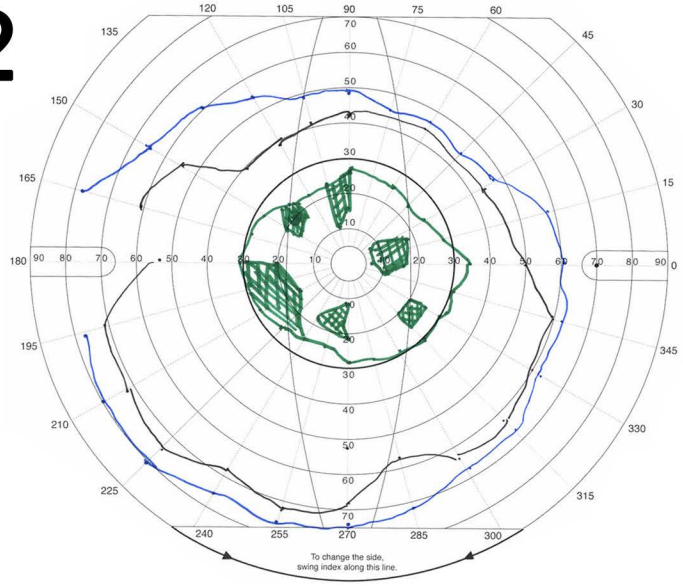
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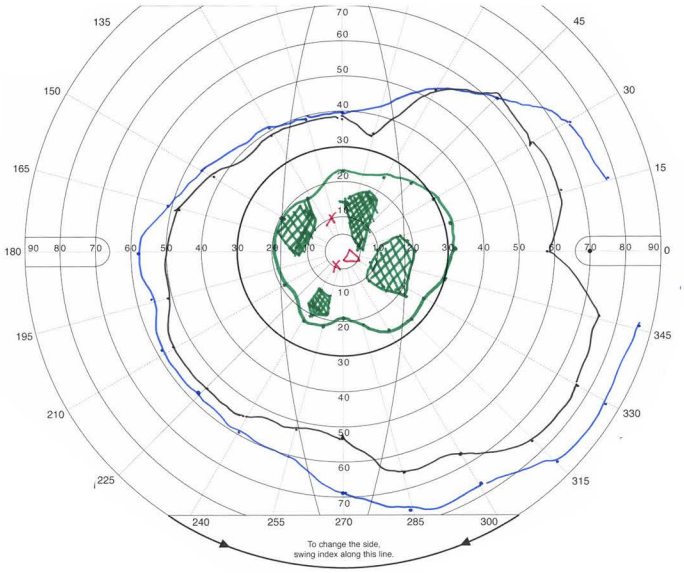
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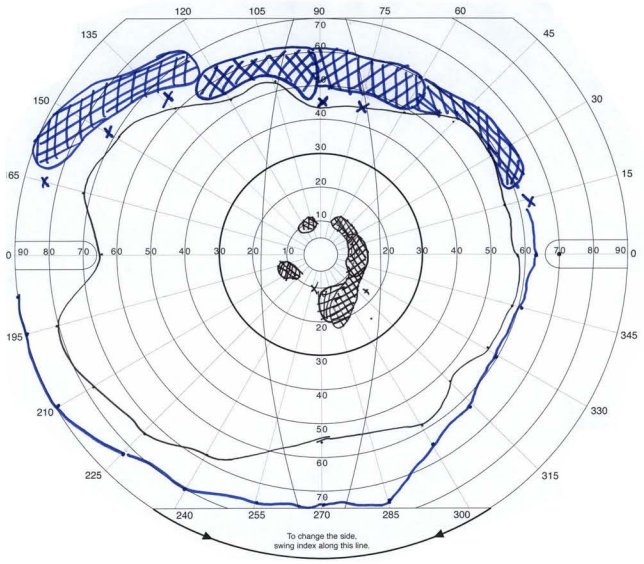
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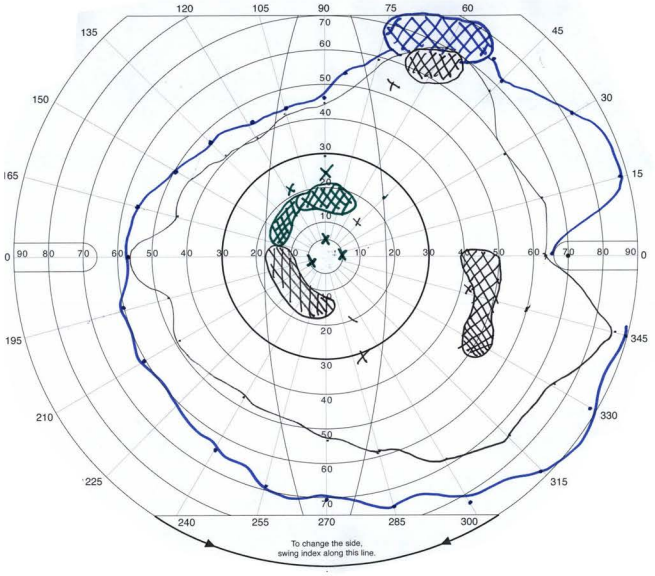
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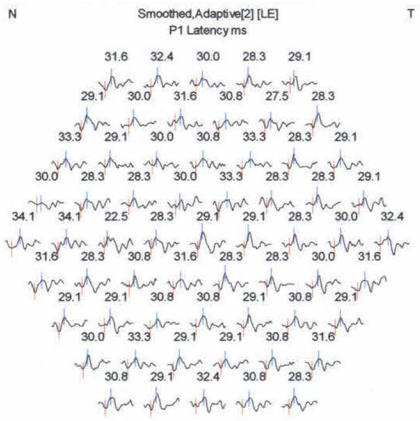
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Smoothed,Adaptive[2] [LE]
P1 Latency ms
T
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33.3 29.1 30.0 30.8 33.3 28.3 29.1
30.0 28.3 28.3 30.0 33.3 28.3 28.3 29.1
34.1 34.1 22.5 28.3 29.1 29.1 28.3 30.0 32.4
31.6 28.3 30.8 31.6 28.3 28.3 30.0 31.6
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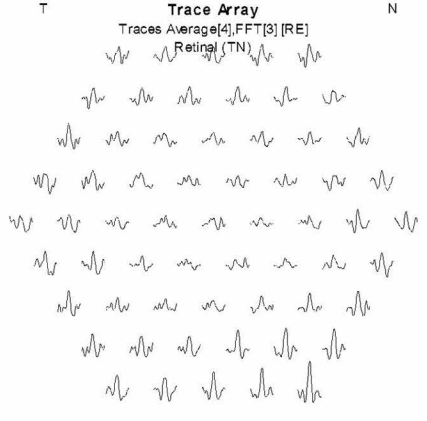
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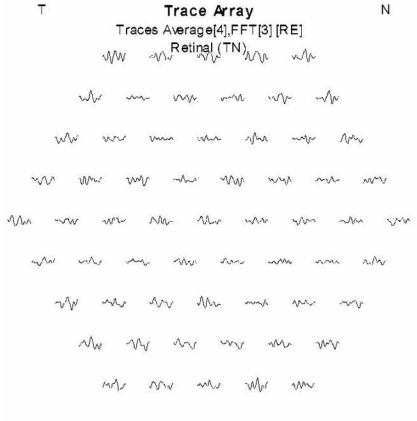
Trace Array
Traces Average[4],FFT[3] [RE]
Retinal (TN)
N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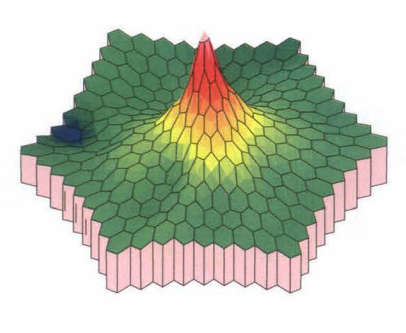

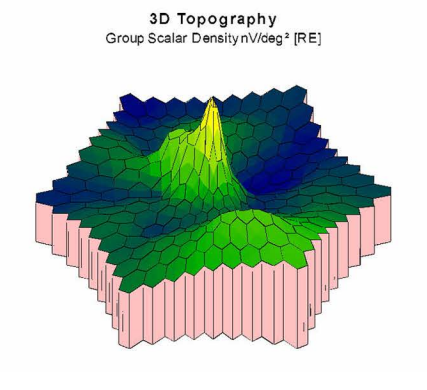
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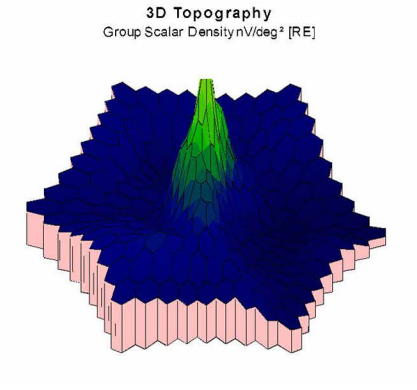
Group Scalar Density nV/deg² [RE]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