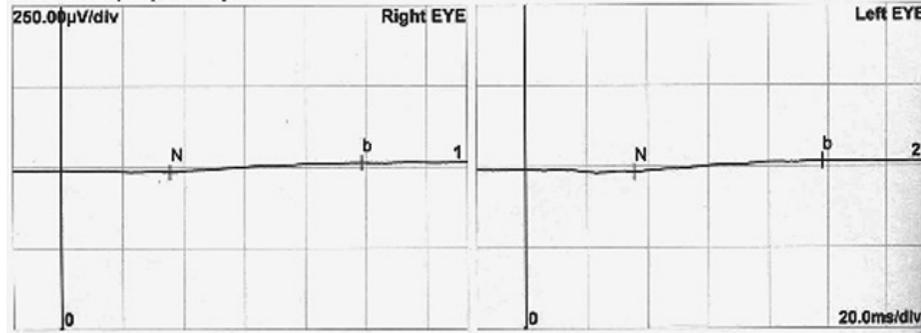


A

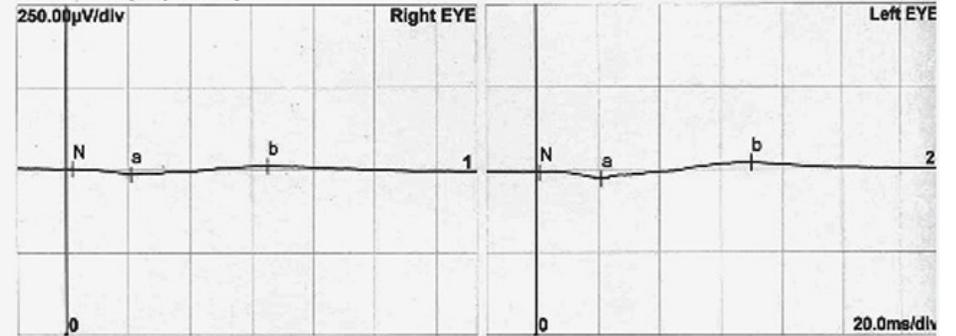
Diagnosis:

blue ERG (GF) scotopic



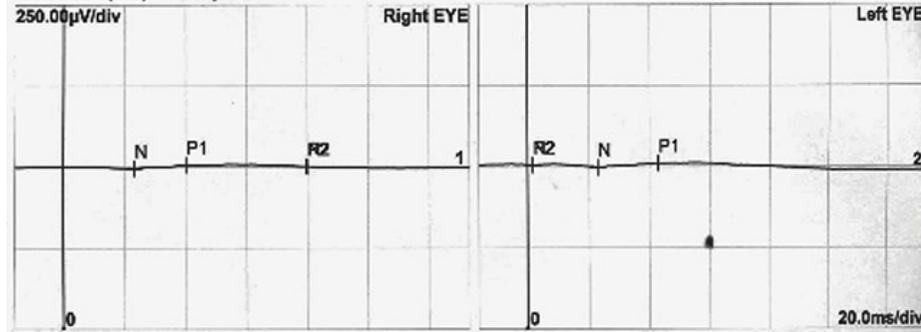
Normals	87-91	95µV-305µV
Channel	b [ms]	b-wave
1 R-1 scotopic	96.9 (!)	27.8µV (!)
2 L-2 scotopic	98.3 (!)	33.7µV (!)

white 10.0 (GF) scotopic



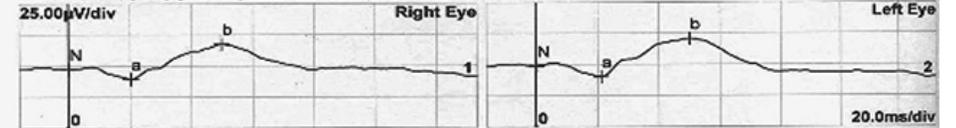
Channel	a [ms]	b [ms]	Amp. a	Amp. b
1 R-1 scotopic	21.1	65.5	16.1µV	25.9µV
2 L-2 scotopic	21.4	70.5	18.8µV	47.9µV

red ERG (GF) scotopic



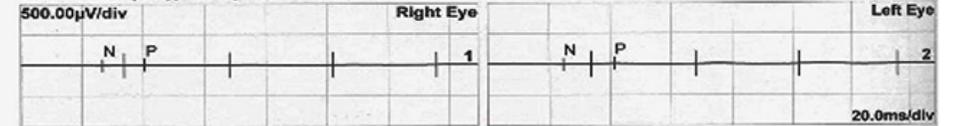
Normals	14-22	33-48	-	155µV-356µV	290µV-654µV
Channel	P1 [ms]	N2 [ms]	P2 [ms]	Amp. P1	Amp. P2
1 R-1 scotopic	40.5 (!)	80.4 (!)	80.1	11.9µV (!)	0V (!)
2 L-2 scotopic	43.2 (!)	1.8 (!)	1.8	12.7µV (!)	0V (!)

white 10.0 (GF) photopic



Channel	a [ms]	b [ms]	Amp. a	Amp. b
1 R-1 photopic	20.5	50.2	7.6µV	28.1µV
2 L-2 photopic	21.4	50.5	8.7µV	30.3µV

white 10.0 (GF) photopic 30Hz Flicker



Channel	P [ms]	Amp. P
1 R-1 photopic	40.8	11µV
2 L-2 photopic	41.7	15.3µV

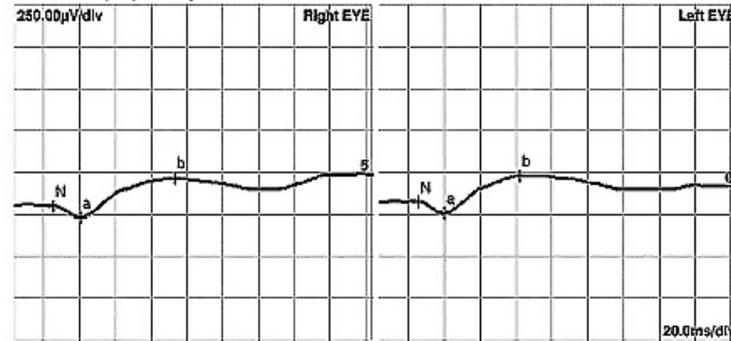
B

Diagnosis:
blue ERG (GF) scotopic



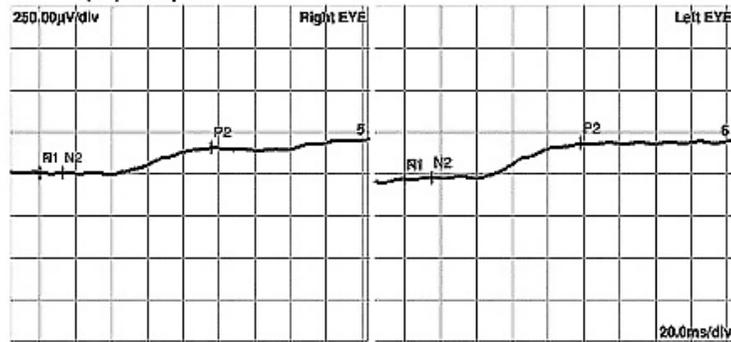
Normal	67-91	95µV-305µV
Channel	b [ms]	b-wave
1 R-1 scotopic		
3 R-1 scotopic	80.6	177.3µV
5 R-1 scotopic		
2 L-2 scotopic		
4 L-2 scotopic		
6 L-2 scotopic	91.6 (I)	205.6µV

white 10.0 (GF) scotopic



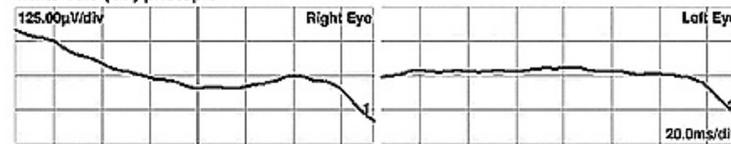
Channel	a [ms]	b [ms]	Amp. a	Amp. b
1 R-1 scotopic				
3 R-1 scotopic				
5 R-1 scotopic	21.1	79.6	64.2µV	231.1µV
2 L-2 scotopic				
4 L-2 scotopic				
6 L-2 scotopic	20.4	62.8	68.9µV	228.8µV

red ERG (GF) scotopic



Normal	14-22	33-46	-	155µV-358µV	290µV-654µV
Channel	P1 [ms]	N2 [ms]	P2 [ms]	Amp. P1	Amp. P2
1 R-1 scotopic					
3 R-1 scotopic					
5 R-1 scotopic	0.0 (I)	12.9 (I)	96.8	0V (I)	153.8µV (I)
2 L-2 scotopic					
4 L-2 scotopic					
6 L-2 scotopic	0.0 (I)	15.8 (I)	96.8	0V (I)	202.1µV (I)

white 10.0 (GF) photopic



Channel	a [ms]	b [ms]	Amp. a	Amp. b
1 R-1 photopic	0.0	0.0		
2 L-2 photopic	0.0	0.0		

white 10.0 (GF) photopic 30Hz Flicker



Channel	P [ms]	Amp. P
1 R-1 photopic	0.0	
2 L-2 photopic	0.0	

Figure S5 : Electroretinographic (ERG) recordings from the two patients with detectable ERGs in this study. **A**: ERGs recorded from patient 5B at the age of 12 months under chloral hydrate sedation using Burian-Allen electrodes. The scotopic responses were recorded after 20 minutes of dark adaptation. The scotopic responses to a blue flash showed severe amplitude reduction; the response to red flash under dark adaptation showed no discernable response. The dark-adapted bright flash response showed severe reduction of the a- and b-wave amplitudes. The printing scale precluded commenting on the waveform. The light-adapted single flash response showed amplitude reduction to approximately 25% of normal with severe b-wave delay. **B**: ERG recordings from patient 13B at the age of 15 years. The dark-adapted (DA) response to a blue flash is moderately reduced. Note the absence of the x-wave component, which originates from the dark-adapted cones, of the DA response to a red flash with relative preservation of the b-wave. The DA response to a bright flash showed a subnormal a-wave amplitude with moderate reduction of the b-wave amplitude. The light adapted responses were undetectable.