

## Supplementary Information

### **Persistent Diffuse Photoreceptor Disorganization: Further evidence with potential associations, long-term clinical course and therapeutic responsiveness of a new clinical entity**

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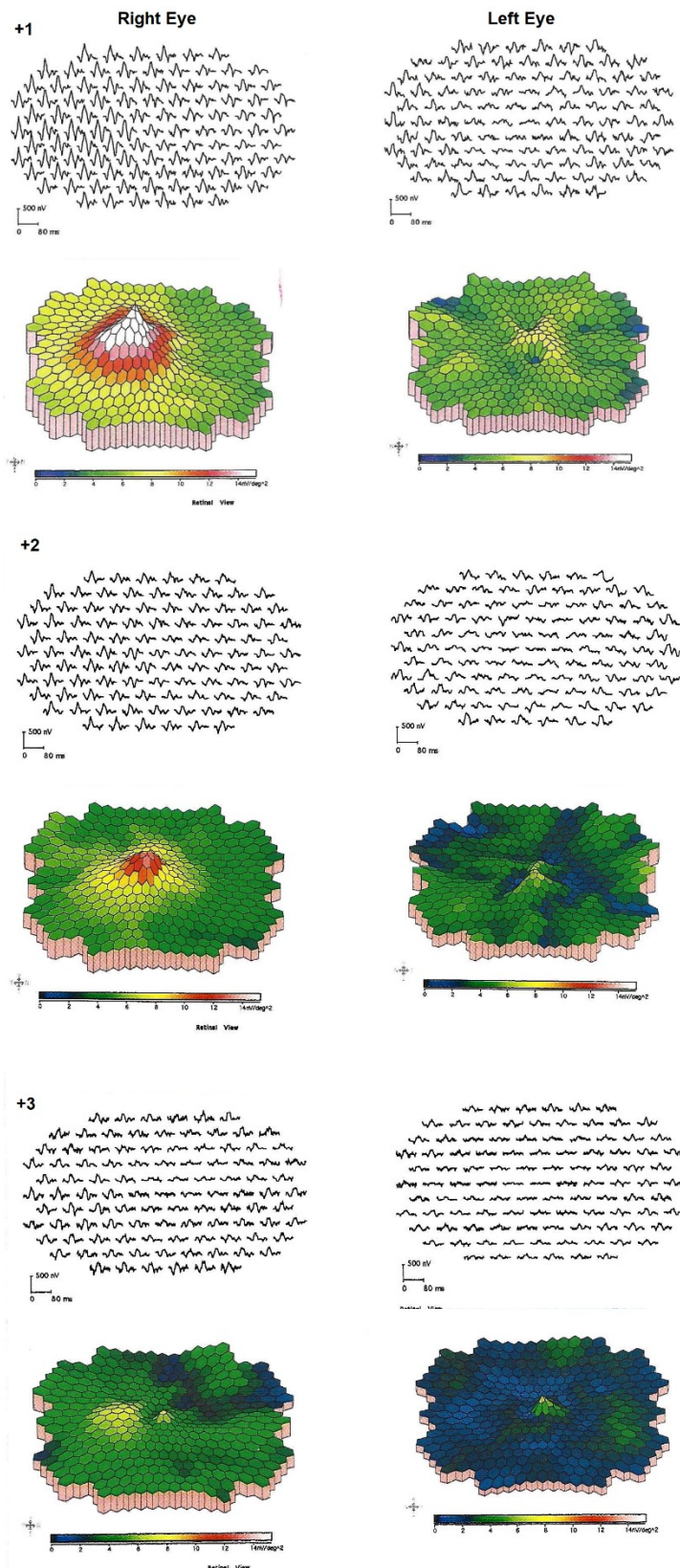
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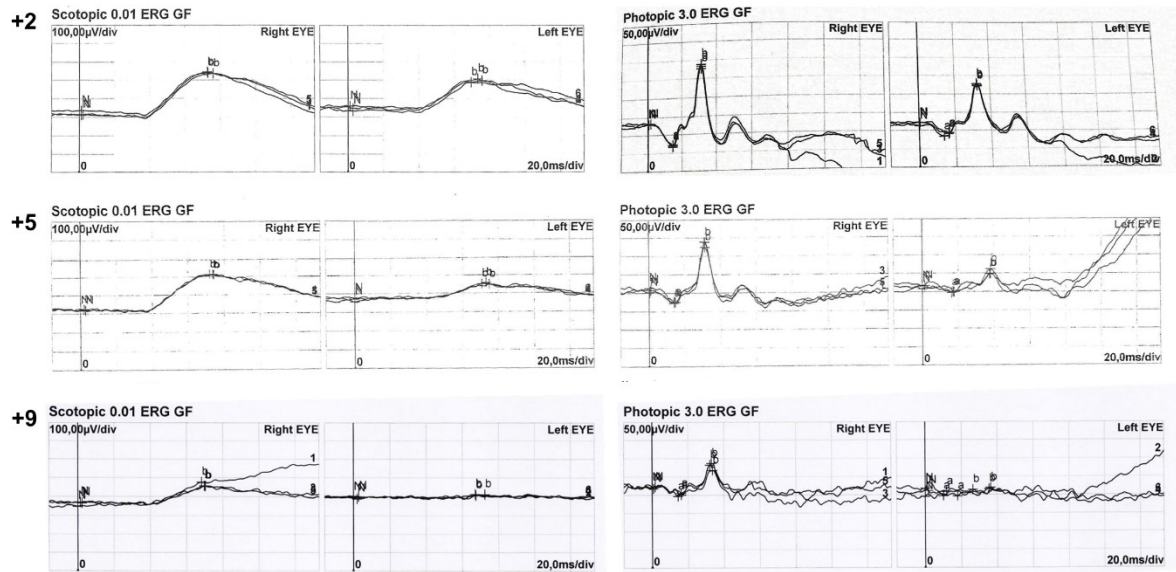
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**Supplemental Fig. 1** Patient 1. Multifocal electroretinogram (mfERG) traces 1, 2 and 3 years after onset of symptoms in the left (first) eye. Note the central onset and spread in the form of an increasing reduction in amplitudes from the center to outside first in the left, then in the right eye.

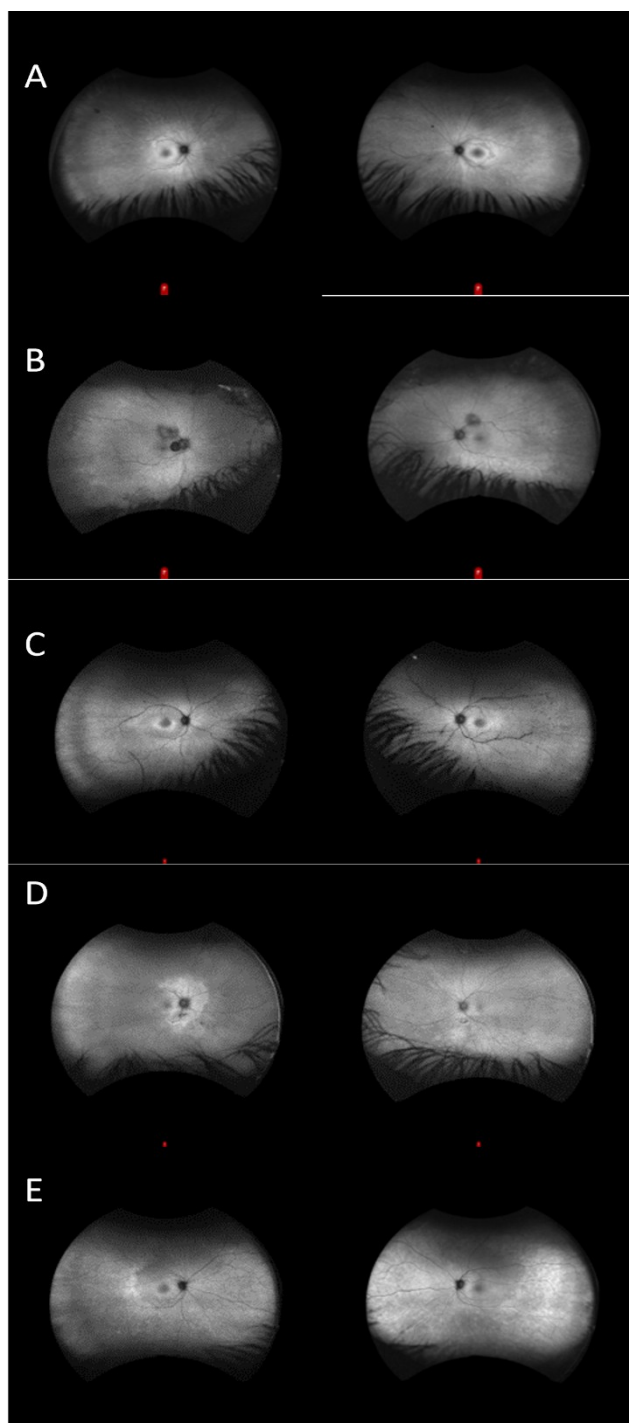


Norm-Values b-wave latency: 67-91 ms; b-wave amplitude: 95-305  $\mu\text{V}$

Norm-Values b-wave latency: 29-33 ms; b-wave amplitude: 103-250  $\mu\text{V}$

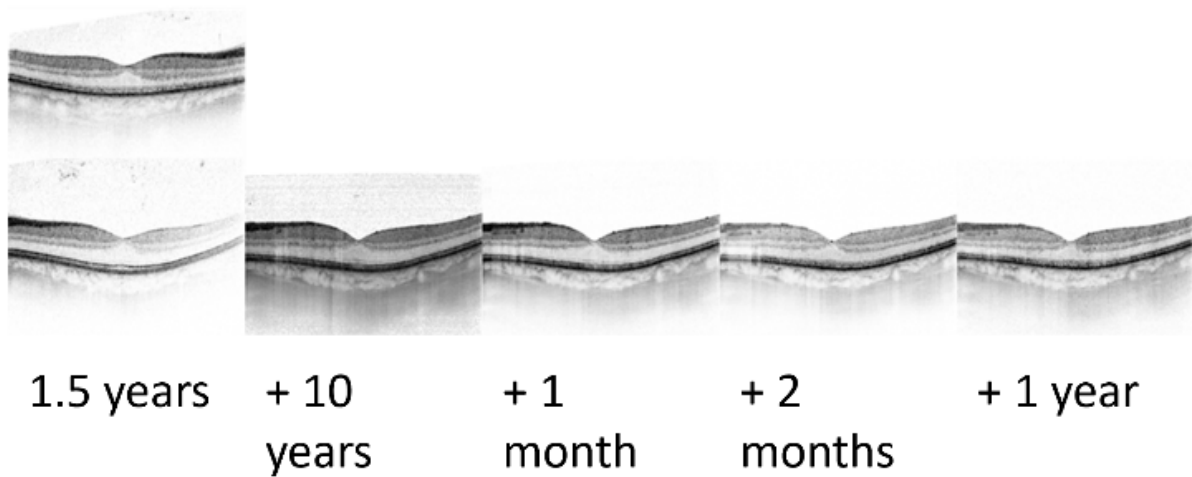
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25 **Supplemental Fig. 2** Patient 1. Full-field electroretinogram (ff-ERG) traces 2, 5 and 9 years after onset of  
 26 symptoms in the left (first) eye. Note the side-differential decrease in amplitudes in the photopic (on the right-  
 27 hand side) and scotopic (on the left-hand side) sum responses.



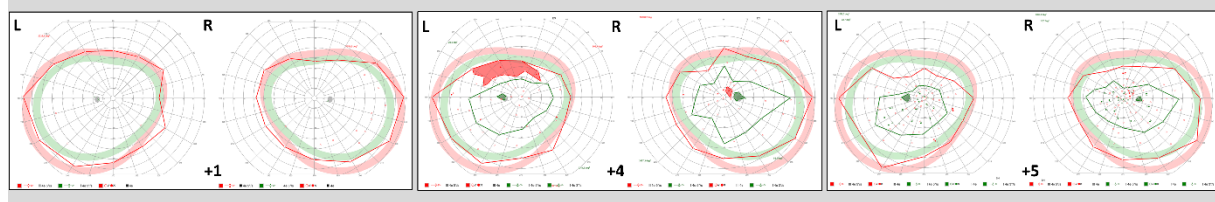
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30 **Supplemental Fig. 3** Fundus Autofluorescence in Patients No. 1-5 at last presentation. **A.** Patient 1. Note the  
 31 increased autofluorescence (IAF) at the posterior pole and the speckled pattern outside in both eyes and the  
 32 decreased autofluorescence (DAF) temporal to the macula in the left eye. **B.** Patient 2. Note the IAF in the mid-  
 33 periphery in both eyes. **C.** Patient 3. Note the linear perivascular DAF in the left eye more than in the right eye  
 34 and dotted DAF temporally in the left eye **D.** Patient 4. Note the IAF surrounding the optic disc in the right eye  
 35 and the IAF in the mid-periphery in the left eye. **E.** Patient 5. Note the speckled pattern outside the posterior  
 36 pole in the left eye more than in the right eye

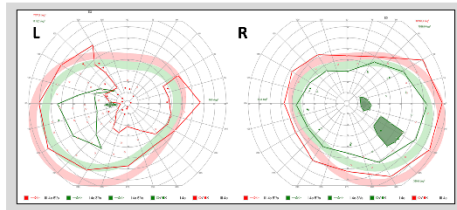


**Supplemental Fig. 4** Patient 4. Optical coherence tomography images of the right (upper panel) and left eye (lower panel), starting 1.5 years after symptom onset in the first (right) eye. Note the progressive disruption of the Ellipsoid zone (EZ) in the left eye over the course of 11 years starting in the foveola while EZ disruption had already occurred in the right eye

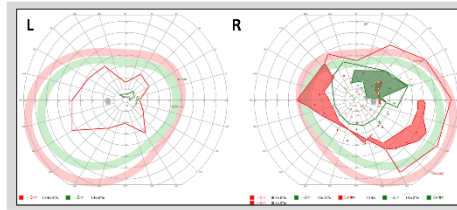
Pat. 2



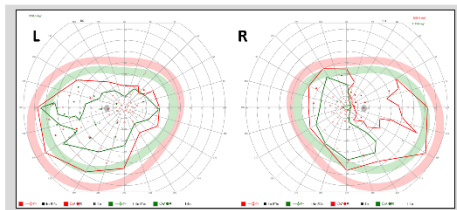
Pat. 1



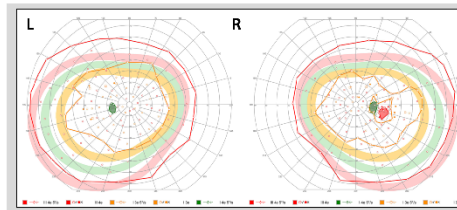
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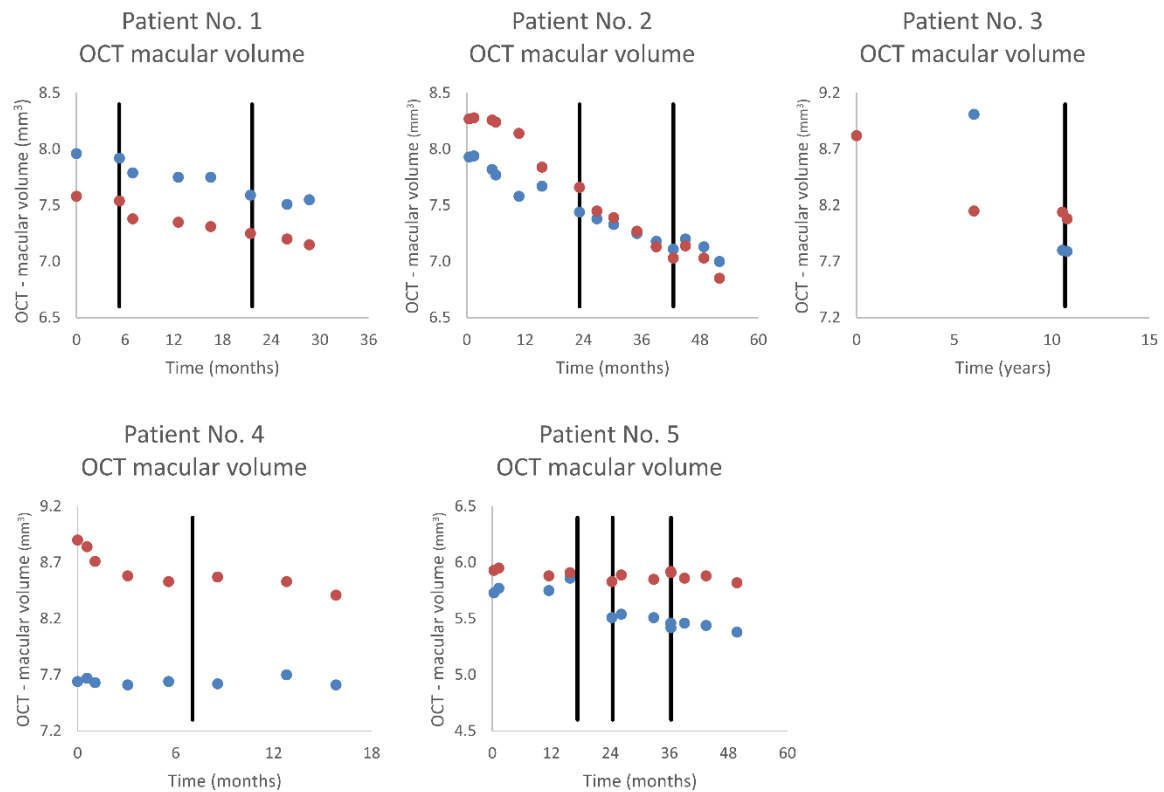
Pat. 4



Pat. 5



**Supplemental Fig. 5** Kinetic visual field (VF) exams in Patients 1-5 at last presentation (bottom two rows) as well as the longitudinal VF course of Patient 2 over a period of 5 years (top row, the numbers represent the time (years) since onset of symptoms in the first (right) eye). Goldmann target III4e is colored red, I4e green, and I3e yellow. Shaded areas represent normative data. Note the relatively preserved VF in all patients at last presentation. L = left eye, R = right eye.



**Supplemental Fig. 6** Longitudinal macular volumes for the right (blue) and left (red) eye of Patients 1-5. Treatments mentioned in the manuscript are indicated by vertical black lines and were the following:

- Patient 1: Start and stop of weight-adapted oral prednisolone (bars 1 and 2).
- Patient 2: Start and stop of weight-adapted oral prednisolone (bars 1 and 2).
- Patient 3: Start of weight-adapted oral prednisolone (bar 1).
- Patient 4: Start of weight-adapted oral prednisolone (bar 1).
- Patient 5: Start of first oral prednisolone (bar 1), start of second oral prednisolone (bar 2), start of oral prednisolone and subcutaneous adalimumab in combination (bar 3).