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Jane Morgan
An abnormality in the perifoveal retina is not usually detected in association with type 1 diabetes.
N = 20 diabetic, N = 18 control
GDS (physical examination)
2015

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Adaptive optics imaging of perfused foveal microvascular density in type 1 diabetes.

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Compiled by Niklas Domdei, Maximilian Paul and Wolf Horhing: Department of Ophthalmology, University Bonn, Germany. Last update: 19/11/2017
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<tr>
<td>Glaucoma</td>
<td>Human Name</td>
<td>See text for details</td>
<td>patients of abnormal lamina cribrosa (lacunar) hole</td>
<td>inferior foot of the inner nuclear layer and abnormal regions diffusion cone inner nuclear layer, in two of three eyes, a lacunar hole were the region of so intense arrowhead</td>
<td>5</td>
<td>2016</td>
<td>2008</td>
<td>Identification of the exact location of structural changes within the lamina cribrosa</td>
<td>AOSLO, OCT</td>
<td>N = 10 patients, N = 10</td>
<td>2016</td>
<td>Hasegawa T, Ooto S, Koyama Y, Hashimoto T, Hubbell A, Hashimoto K.</td>
<td>Transl Vis Sci Technol</td>
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</table>
Gene/mutation (if specified) AO-OCT, FAF, FD-OCT
Retinal structure Normal retina was observed in the areas with normal visual function.
Central finding
Number patients N = 1
Comments No visual loss in MD, MP.
Imaging modality OCT
Function testing No functional loss.
Treatment No treatment.
PMID 25659196
Year 2015
Author and year Massin P, Gaudric A.
Journal Retina
Title Adaptive optics imaging of cone mosaic abnormalities in acute multifocal placoid pigment epitheliopathy.

Gene/mutation (if specified) None
Retinal structure The cone photoreceptors were disrupted in the abnormal hyperreflective areas in the IR images. However, some areas of photoreceptor loss were observed. Photoreceptor spacing in the areas of relative scotoma is consistent with the presence of photoreceptor loss.
Central finding N = 10 eyes of 10 patients.
Comments No visual loss in MD, MP.
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PMID 23949236
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Microscopic inner retinal hyper-reflective phenotypes in retinal and cone photoreceptors show structural changes when there is retinal dysfunction and classic electronegative ERG in neuroretinopathy resulting in degeneration of photoreceptor cells, with a unique phenotype of acute progressive paravascular placoid maculopathy (AEPVM). This entity is being recognized in a variety of conditions, with severe photoreceptor damage can be induced by CMV retinitis, which can be monitored by OCT or AO modalities. AO imaging and OCT ranging can be used to monitor the seven photoreceptor layers damaged by Chlamydia trachomatis in HIV retinae.

Adaptation of retinal photoreceptor density, microvascular density in the foveal avascular zone (FAZ) sustained with tapered vascular calibers. HIV retinae also have increased microvascular perfusion density.

A spectrum of autoreactive anti-retinal antibodies is associated with various diseases in patients with autoimmune retinopathy. The use of broad-spectrum immunosuppressive agents, such as the use of rituximab, has been shown to be effective in controlling visual symptoms in foveal red spot syndrome. Monitoring disease progression and evaluating response to therapy.

Visual acuity was stable or improved in a majority of AIR patients treated with rituximab. OCT and ERG parameters, as well as AO-SLO cone densities, were stable during treatment.

Autoimmune Retinopathy Related Autoimmune Neurologic Disease

Visualization of Photoreceptors in Birdshot Chorioretinopathy Using Adaptive Optics Scanning Laser Ophthalmoscopy: A Pilot Study

N = 16 HIV+, N = 16 HIV-

N = 1

Autism spectrum disorder (acute paraneoplastic)

N = 1

18436843

Ward J, Yudofsky SC, Dean HC, Childs H, Kaderli T, Backlund M, Perlman JS. Multimodal imaging in foveal red spot syndrome. 2015

AJ Ophthalmol

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AJ Ophthalmol
Gene/mutation (if specified)

High-resolution imaging of resolved central serous chorioretinopathy
Jacob J, Paques M, Krivosic

Central serous diseases and injuries. Split-detector AOSLO revealed substantial
membrane; vessel associated membrane; striate

Disease
Other molecular descriptor

Central serous chorioretinopathy (CSCR)

Adaptive optics mfERG microperimetry and OCT images show preserved
photoreceptor cell death.

Null, 3 to 6 months post injection

Low-resolution imaging of resolved central serous chorioretinopathy in
patients with resolved central serous chorioretinopathy.

Null

Central serous chorioretinopathy (CSCR)

Case Missouri

Adaptive optics imaging revealed a gradual increase in the number
photoreceptor cell death.

Negative correlation between outer retinal layer thickness and cone density in
patients with resolved central serous chorioretinopathy.

Negative correlation between outer retinal layer thickness and cone density in
patients with resolved central serous chorioretinopathy.

Central serous chorioretinopathy (CSCR)

MacuTel study group with the findings of dermatologist and
ophthalmologist in macular telangiectasia type 2 suggests that keratoconus, cystoid macular edema and increased cone density in the inferior retina.

Central serous chorioretinopathy (CSCR)

Central serous chorioretinopathy (CSCR)

Adaptive optics imaging revealed a gradual increase in the number
photoreceptor cell death.

Central serous chorioretinopathy (CSCR)

Adaptive optics imaging demonstrated decreased cone density,
photoreceptor cell death.

Central serous chorioretinopathy (CSCR)

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Adaptive optics imaging demonstrated decreased cone density,
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Gene/mutation (if applicable)

Cones

Longitudinal study of cone photoreceptors during retinal degeneration

VA, visual field

Fundus-referenced vision testing is a useful tool to indicate the presence of cases that may be amenable to recovery or prevention in experimental therapies despite not being visible on standard testing tools or OCT images.

RHO

Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200)

27145477

Microscopic inner retinal hyper-reflective phenotypes in retinal and outer nuclear layer, cones

Journal


various diseases in retinal degeneration

A case report

Fundus-Ocular Phenotype of a Family with FAM161A-associated Retinal Degeneration

Outer retinal layers were significantly thicker in CNTF-treated eyes in this paper compared to sham-treated eyes. Neither the outer nuclear layer thickness nor the cone density in CNTF-treated eyes was significantly different from that in sham-treated eyes in this paper.

AOSLO, SD-OCT

Loss of outer retinal structures demonstrated with high-resolution AOSLO or SD-OCT images.

Stargardt's macular dystrophy and retinitis pigmentosa.

AOSLO, OCT

Changes in retinitis pigmentosa can be identified and reconciled with genetic findings.

AOSLO, SD-OCT

Northern blot

The development of the outer nuclear layer is disrupted in eyes with RP, even when visual sensitivity and foveal blood flow remain normal limits.

ff-ERG

Comparison between foveal cone structure and clinical measures of retinal degeneration.

ff-ERG

AOSLO, OCT

Sustained-release factor (CNTF)

AOSLO, OCT

AOSLO, SD-OCT

SLO

follow-up of acute bilateral foveolitis

fading

microperimetry

functionality

follow-up of acute bilateral foveolitis

AOSLO, OCT

SLO

microperimetry

functionality

follow-up of acute bilateral foveolitis

AOSLO, OCT

SLO

microperimetry

functionality

follow-up of acute bilateral foveolitis

AOSLO, OCT

SLO

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AOSLO, OCT

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AOSLO, OCT

SLO

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AOSLO, OCT

SLO

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functionality

follow-up of acute bilateral foveolitis

AOSLO, OCT

SLO
Gene/mutation (if applicable)/Retinal structure/Central finding/Number patients/Comments/Imaging modality/Functional testing/Treatment/PMID/Year/Author and year/Journal

- **Bilateral Progression Fundus Albipunctatus**
  - Disease:
  - Gene:
  - Mutation:
  - Retinal structure:
  - Central finding:
  - Number patients:
  - Comments:
  - Imaging modality:
  - Functional testing:
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  - Author and year:
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- **Unilateral abnormal photoreceptor mosaic in a patient with Stargardt-like macular disease:**
  - Disease:
  - Gene:
  - Mutation:
  - Retinal structure:
  - Central finding:
  - Number patients:
  - Comments:
  - Imaging modality:
  - Functional testing:
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  - Author and year:
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Ophthamology
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<td>Macular degeneration</td>
<td>GUCY2D</td>
<td>Inner retina</td>
<td>There were significant differences in density, distribution, and vitality of cone photoreceptors in affected eyes compared to normals.</td>
<td>10 patients</td>
<td>Localised</td>
<td>AOSLO, OCT</td>
<td>Mirlin et al.</td>
<td>Correlation</td>
<td>17460294</td>
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<td>CMOX1</td>
<td>Inner retina</td>
<td>Photoreceptor layer morphology to interpret preclinical signs of disease.</td>
<td>20 patients</td>
<td>Localised</td>
<td>AOSLO, OCT</td>
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**Notes:**
- **AOSLO** indicates adaptive optics scanning laser ophthalmoscopy.
- **mfERG** indicates multifocal electroretinography.
- **FF-ERG** indicates full-field electroretinography.
- **SD-OCT** indicates spectral domain optical coherence tomography.
- **AO-Cam** indicates adaptive optics fundus camera.
- **Berne JJ** indicates Berne et al.
- **SD-OCT** indicates spectral domain optical coherence tomography.
- **AO-Cam** indicates adaptive optics fundus camera.
- **AO-SLO** indicates adaptive optics scanning laser ophthalmoscopy.
- **Bessho K** indicates Bessho et al.
- **AO-Fundus-Cam** indicates adaptive optics fundus camera.
- **AOSLO** indicates adaptive optics scanning laser ophthalmoscopy.
Gene/mutation (if specified)

Central finding

Retinal structure

Number patients

Comments

Imaging modality

Functional testing

Treatment

PMID

Year

Author and year

Journal

Title

Vitelliform macular dystrophy

Gene/mutation (if specified)

Vitriform inner retinal hyper-reflective phenotypes in retinal and vitreous

12 patients, 6 - 10 control

RTX1, SD-OCT

 mfERG, ffERG

Light sensitivity

KD84, 2013


Retinal dysplasia

Gene/mutation (if specified)

Punctate inner retinal lesions

50 patients

RTX1, SD-OCT

 mfERG, ffERG

Light sensitivity

KD85, 2013


Best vitelliform dystrophy

Gene/mutation (if specified)

Pathologic changes of cone photoreceptors in eyes with occult macular dystrophy

N = 15 healthy, N = 9 affected, N = 3 unaffected

Retinal structure

mfERG, ffERG, perimetry, ERG

Light sensitivity

KD86, 2015


Inner retinal reflectivity

Gene/mutation (if specified)

Inner retinal reflectivity

28 patients

Retinal structure

AOSLO, OCT

Light sensitivity

KD87, 2017


Retinal dysplasia

Gene/mutation (if specified)

Cone inner retinal reflectivity

5 patients

Retinal structure

AOSLO, OCT

Light sensitivity

KD90, 2000

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<th>Number patients</th>
<th>Comments</th>
<th>Imaging modality</th>
<th>Functional testing</th>
<th>Treatment</th>
<th>PMID</th>
<th>Year</th>
<th>Author and year</th>
<th>Journal</th>
<th>Manuscript title</th>
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<td>Retina</td>
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Compiled by Niklas Domdei, Maximilian Pfau and Wolf Harmoning; Department of Ophthalmology, University Bonn, Germany. Last update: 19/11/2017

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Recovery of cone packing density measured by AO was associated with functional recovery. Later imaging studies revealed that the lesion remained overlying the choroidal lesion and adjacent retina.

Although retinal vascular densities are reduced and cone spacing is increased in advanced disease, central foveal structure is maintained until late stages of disease, which may contribute to maintenance of foveal vision in eyes with MAK-related retinal degeneration.
**Retinal structure**

**Central finding**

**Number patients**

**Comments**

**Imaging modality**

**Functional testing**

**Treatment**

**PMID**

**Year**

**Author and year**

**Journal**

**Manuscript title**

**Department of Ophthalmology**, University Bonn, Germany. Last update: 19/11/2017

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**Gene/mutation (if specified)**

**Central finding**

**Number patients**

**Comments**

**Imaging modality**

**Functional testing**

**Treatment**

**PMID**

**Year**

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**Journal**

**Manuscript title**

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