Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Age-related Macular Degeneration (AMD)													
Early and intermediate AMD		RPE, Bruch's membrane	microscopic characteristics of regressing drusen, possibly representing different stages of drusen	N = 16 eyes, N = 12 patients	short report	rtx1, SLO, OCT			24290975	2014	Querques G,, Kamami-Levy C, Georges A, Pedinielli A, Souied EH	Ophthalmology	Appearance of regressing Drusen on adaptive optics in age-related macular degeneration.
Early and intermediate AMD		RPE, Bruch's membrane	AO allows differences in reflectivity between medium–large drusen and reticular pseudodrusen to be appreciated.	N = 8 eyes, N = 6 patients		rtx1, SLO, OCT			24985725	2014	Querques G, Kamami-Levy C, Blanco-Garavito R, Georges A, Pedinielli A, Capuano V, Poulon F, Souied EH.	Br J Ophthalmol	Appearance of medium-large drusen and reticular pseudodrusen on adaptive optics in age-related macular degeneration.
Early and intermediate AMD		Photoreceptor mosaic, large colloid drusen	The different AO features may suggest a different pathology and possible evolution between AMD drusen and this peculiar type of early onset drusen	N = 1		rtx1			21883987	2012	Querques G, Massamba N, Guigui B, Lea Q, Lamory B, Soubrane G, Souied EH.	Acta Ophthalmol	In vivo evaluation of photoreceptor mosaic in early onset large colloid drusen using adaptive optics.
Early and intermediate AMD		photoreceptors, subretinal drusenoid deposits	AOSLO revealed that photoreceptor reflectivity was qualitatively reduced by stage 1 subretinal drusenoid deposits and was greatly reduced by stage 2. AOSLO presented a distinct structure in stage 3, a hyporeflective annulus consisting of deflected, degenerated or absent photoreceptors	N = 53 AMD patients, N = 10 healthy		AOSLO			24907433	2014	Zhang Y, Wang X, Rivero EB, Clark ME, Witherspoon CD, Spaide RF, Girkin CA, Owsley C, Curcio CA.	Am J Ophthalmol	Photoreceptor perturbation around subretinal drusenoid deposits as revealed by adaptive optics scanning laser ophthalmoscopy.
Early and intermediate AMD		subretinal drusenoid deposits	AO-OCT suggested that the speckled appearance over the subretinal drusen deposits rendered by AO-SLO was the lesion material itself, rather than photoreceptors.	N = 3 AMD patients, N = 2 healthy		AOSLO, AOOCT			24688808	2014	Meadway A, Wang X, Curcio CA, Zhang Y.	Biomed Opt Express	Microstructure of subretinal drusenoid deposits revealed by adaptive optics imaging.
Intermediate age-related macular degeneration		dot subretinal drusenoid deposits	Adaptive optics scanning laser ophthalmoscopy reveals that dot SDD, like drusen, are dynamic. Dynamism, the absolute value of the areas affected by new and regressed lesions, ranged from 0.7% to 9.3%.	N = 6 eyes of 4 patients	Longitudinal study: observation time = 1 year	AO-SLO			28196054	2017	Zhang Y, Wang X, Godara P, Zhang T, Clark ME, Witherspoon CD, Spaide RF, Owsley C, Curcio CA	Retina	DYNAMISM OF DOT SUBRETINAL DRUSENOID DEPOSITS IN AGE-RELATED MACULAR DEGENERATION DEMONSTRATED WITH ADAPTIVE OPTICS IMAGING
Early and intermediate AMD		Cone Mosaic	The mean cone density was lower with subretinal drusenoid deposits compared to conventionl drusen. The difference in cone density reduction between the two lesion types was highly significant (P<0.001).	N = 11 (11 eyes) pseudodrusen, N = 6 (11 eyes) conventional drusen		AOSLO, OCT			24183341	2014	Mrejen S, Sato T, Curcio CA, Spaide RF.	Ophthalmology	Assessing the cone photoreceptor mosaic in eyes with pseudodrusen and soft Drusen in vivo using adaptive optics imaging.
Early and intermediate AMD		Cone Mosaic	This imaging approach and the image analysis metrics introduced may serve as the foundation for valuable imaging-based biomarkers for detecting the earliest stages of disease, tracking progression, and monitoring treatment response.	N = 1		AO Fundus Camera			21117594	2010	Godara P, Siebe C, Rha J, Michaelides M, Carroll J.	Ophthalmic Surg Lasers Imaging.	Assessing the photoreceptor mosaic over drusen using adaptive optics and SD-OCT.
Early and intermediate AMD		Microvasculature	AOSLO Offset Pinhole offers a non-invasive alternative to AOSLO FA without the need for any exogenous contrast agent.	N = 1	various diseases in this paper, method evaluation	AOSLO, Fundus Cam, OCT			24761299	2014	Chui TY, Dubow M, Pinhas A, Shah N, Gan A, Weitz R, Sulai YN, Dubra A, Rosen RB.	Biomed Opt Express	Comparison of adaptive optics scanning light ophthalmoscopic fluorescein angiography and offset pinhole imaging.
Early and intermediate AMD		Inner Retinal Reflectivity	Inner retinal phenotype: granular membrane; waxy membrane;	N = 2	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Early and intermediate AMD		Cone Structure	AO-SLO imaging reveals a decrease in photoreceptor density and increased spacing in patients with grade 1 to 3 fundi, as well as a spectrum of photoreceptor changes, ranging from variability in reflectivity to decreased density.	N = 40		AOSLO, OCT			25014365	2014	Land ME, Cooper RF, Young J, Berg E, Kitchner T, Xiang Q, Szabo A, Ivacic LC, Stepien KE, Page CD, Carroll J, Connor T Jr, Brilliant M.	Optom Vis Sci	Cone structure in subjects with known genetic relative risk for AMD.
Early and intermediate AMD		Cone Structure	Axial length and age were significantly correlated with parafoveal cone photoreceptor distribution. The results do not support that early AMD might influence cone photoreceptor density in the area without drusen or pigment abnormalities.	N = 60		rtx1, SD-OCT			24632778	2014	Obata R, Yanagi Y	PLoS One	Quantitative analysis of cone photoreceptor distribution and its relationship with axial length, age, and early age-related macular degeneration.
Early and intermediate AMD		RPE	Hypertransmission into the choroid, accompanied with subretinal drusenoid deposits (SDD) regression and thinning of choroid and photoreceptor layers, indicates RPE degeneration associated with advanced stages in the SDD life cycle.	N = 12 patients, N = 12 healthy		OCT, AO-SLO			27986424	2016	Xu X, Liu X, Wang X, Clark ME, McGwin G Jr, Owsley C, Curcio CA, Zhang Y.	Am J Ophthalmol	Retinal pigment epithelium degeneration associated with subretinal drusenoid deposits in age-related macular degeneration
Geographic Atrophy		Hyper Reflective Clumps	AO imaging revealed that a complex, dynamic process of redistribution of hyporeflective clumps throughout the posterior pole precedes and accompanies the emergence and progression of atrophy. Therefore, these clumps are probably also a biomarker of RPE damage.	N = 12 eyes, N = 9 GA patients, N = 7 control		rtx1			23620431	2013	Gocho K, Sarda V, Falah S, Sahel JA, Sennlaub F, Benchaboune M, Ullern M, Paques M.	Invest Ophthalmol Vis Sci	Adaptive optics imaging of geographic atrophy.
Geographic Atrophy		Fundus (whole retina)	While there was a strong correlation between altered retinal structure and reduction in visual function, there were a number of examples in which the photoreceptor inner/outer segment (IS/OS) junctions lost reflectivity at the margins of GA, while visual function was still demonstrated.	N = 5 eyes, N = 4 GA patients, N = 1 healthy		AO-OCT, FAF, CF	mfERG Function Testing		23696601	2013	Panorgias A, Zawadzki RJ, Capps AG, Hunter AA, Morse LS, Werner JS.	Invest Ophthalmol Vis Sci	Multimodal assessment of microscopic morphology and retinal function in patients with geographic atrophy.
Geographic Atrophy		Cones	Although cone spacing was often normal at baseline and remained normal over time, these regions showed focal areas of decreased cone reflectivity. These findings may provide insight into the pathophysiology of AMD progression.	N = 7 patients, N = 4 eyes GA, N = 4 eyes drusen		AOSLO, SD-OCT, FAF, CF, IF			24135755	2013	Zayit-Soudry S, Duncan JL, Syed R, Menghini M, Roorda AJ.	Invest Ophthalmol Vis Sci	Cone structure imaged with adaptive optics scanning laser ophthalmoscopy in eyes with nonneovascular age-related macular degeneration.
Geographic Atrophy		foveal structures	Preservation of functional cone photoreceptors was demonstrated on en face AO IR images in areas of foveal sparing detected by confocal SLO near-IR autofluorescence	N = 5 eyes, N = 4 patients		rtx1, OCT, SLO			26200512	2016	Querques G, Kamami-Levy C, Georges A, Pedinielli A, Capuano V, Blanco-Garavito R, Poulon F, Souied EH.	Retina	ADAPTIVE OPTICS IMAGING OF FOVEAL SPARING IN GEOGRAPHIC ATROPHY SECONDARY TO AGE-RELATED MACULAR DEGENERATION.
Geographic Atrophy		Photoreceptors	AO–SLO imaging revealed slight disruption in the photoreceptor mosaic in early stage AMD due to focal drusen formation and identified several small drusen deposits that were not observed with standard clinical im- aging techniques.	N = 4		AOSLO (Physical Sciences)			22930575	2012	Boretsky A, Khan F, Burnett G, Hammer DX, Ferguson RD, van Kuijk F, Motamedi M.	Lasers Surg Med	In vivo imaging of photoreceptor disruption associated with age-related macular degeneration: A pilot study.
Small hard macular drusen		Drusen, Cones	High lateral resolution imaging of small lobular hard retinal drusen suggests formation through the confluence of two or more smaller round lesions. The outline and size of these smaller lesions corresponds to 1–4 RPE cells.	N = 97 participants (21 affected)		SD-OCT, AO-SLO (Kongsberg), AO flood (Kongsberg)			29051326	2017	Pedersen HR, Gilson SJ, Dubra A, Munch IC, Larsen M, Baraas RC	Br J Ophthalmol.	Multimodal imaging of small hard retinal drusen in young healthy adults

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Retinal vascular diseases													
Diabetic retinopathy		Cone Mosaic	Cone density in the parafoveal retina is not reduced in adolescents with type 1 diabetes	N = 29 diabetic, N = 44 control		AOSLO (Physical Sciences)			26517403	2015	Tan W, Wright T, Rajendran D, Garcia-Sanchez Y, Finkelberg L, Kisilak M, Campbell M, Westall CA.	Invest Ophthalmol Vis Sci	Cone-Photoreceptor Density in Adolescents With Type 1 Diabetes.
Diabetic retinopathy		Cones	On average, cone density was 10% lower in the study than in the control group at each retinal eccentricity along the horizontal and vertical meridians.	N = 11 diabetic, N = 11 control		rtx1			23928676	2014	Lombardo M, Parravano M, Lombardo G, Varano M, Boccassini B, Stirpe M, Serrao S.	Retina	Adaptive optics imaging of parafoveal cones in type 1 diabetes.
Diabetic retinopathy		Retinal Capillaries	The parafoveal capillaries were narrower in patients with Type 1 diabetes and nonproliferative diabetic retinopathy than in healthy subjects, showing the potential capability of adaptive optics imaging to detect pathologic variations of the retinal microvascular structures in vaso-occlusive diseases.	N = 8 eyes	method introduction: measuring vessel lumen	rtx1			23492950	2013	Lombardo M, Parravano M, Serrao S, Ducoli P, Stirpe M, Lombardo G.	Retina	Analysis of retinal capillaries in patients with type 1 diabetes and nonproliferative diabetic retinopathy using adaptive optics imaging.
Diabetic retinopathy		Retinal parafoveal Capillaries	With a novel application of AOSLO imaging, it is possible to visualize parafoveal capillaries and identify AV channels noninva- sively. AV channels are disrupted in type 2 diabetes, even before the onset of diabetic retinopathy.	N = 12 patients, N = 11 control	method introduction: motion contrast	AOSLO			22039250	2011	Tam J, Dhamdhere KP, Tiruveedhula P, Manzanera S, Barez S, Bearse MA Jr, Adams AJ, Roorda A.	Invest Ophthalmol Vis Sci	Disruption of the retinal parafoveal capillary network in type 2 diabetes before the onset of diabetic retinopathy.
Diabetic retinopathy		Microvasculature, Mircoaneurysms	Retinal microaneuriysms can be classified in vivo into six different morphologic types, according to the geometry of their two-dimensional (2D) en face view. Imaging in a subject with CRVO before and after anti- VEGF injection shows regression of a mixed MA	N = 14 eyes		AOSLO (FA)		brief anti-VEGF treatment evaluation	24425852	2014	Dubow M, Pinhas A, Shah N, Cooper RF, Gan A, Gentile RC, Hendrix V, Sulai YN, Carroll J, Chui TY, Walsh JB, Weitz R, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Classification of human retinal microaneurysms using adaptive optics scanning light ophthalmoscope fluorescein angiography.
Diabetic retinopathy		Microvasculature	High-resolution serial AOSLO imaging enables in vivo observation of vasculopathic changes seen in diabetes mellitus.	N = 1 patient, N = 1 control		AOSLO, SD-OCT			26803289	2016	Chui TY, Pinhas A, Gan A, Razeen M, Shah N, Cheang E, Liu CL, Dubra A, Rosen RB.	Ophthalmic Physiol Opt	Longitudinal imaging of microvascular remodelling in proliferative diabetic retinopathy using adaptive optics scanning light ophthalmoscopy.
Diabetic retinopathy		Microvasculature, Hemodynamics	The preliminary data obtained to date by the authors suggest that the presence of DR correlates with changes in the hemodynamic environment of the parafoveal vasculature.	N = 2patients (3 eyes), N= 2control (3 eyes)	method evaluation	AOSLO			26738166	2015	Bernabeu MO, Yang Lu, Lammer J, Aiello LP, Coveney PV, Sun JK.	Conf Proc IEEE Eng Med Biol Soc	Characterization of parafoveal hemodynamics associated with diabetic retinopathy with adaptive optics scanning laser ophthalmoscopy and computational fluid dynamics.
Diabetic retinopathy		Parafoveal Cones	The extent of photoreceptor loss on AOimaging may correlate positively with severity of DR in patients with type II diabetes mellitus.	N = 25 patients (29 eyes), N = 10 control (20 eyes)		rtx1			27057752	2016	Soliman MK, Sadiq MA, Agarwal A, Sarwar S, Hassan M, Hanout M, Graf F, High R, Do DV, Nguyen QD, Sepah YJ.	PLoS One	High-Resolution Imaging of Parafoveal Cones in Different Stages of Diabetic Retinopathy Using Adaptive Optics Fundus Camera.
Diabetic retinopathy		Cones	The present set of AO imaging biomarkers identified reliably abnormalities in the spatial arrangement of the parafoveal cones in DM1 patients, even when no signs of diabetic reti- nopathy were seen on fundoscopy.	N = 16		rtx1			26963392	2016	Lombardo M, Parravano M, Serrao S, Ziccardi L, Giannini D, Lombardo G.	PLoS One	Investigation of Adaptive Optics Imaging Biomarkers for Detecting Pathological Changes of the Cone Mosaic in Patients with Type 1 Diabetes Mellitus.
Diabetic retinopathy		Fundus	red lesions on fundus photographs appeared on AO images as dark hyporeflective elements, but it could not be verified whether lesions represented haemorrhages or microaneurysms. The smallest of these lesions were circular with a size corresponding to that of blood cells. Hard exudates had irregular surfaces with buddings of various sizes protruding from the lesions.	N = 19		rtx1, OCT			24925100	2014	Bek T	Acta Ophthalmol	Fine structure in diabetic retinopathy lesions as observed by adaptive optics imaging. A qualitative study.
Diabetic retinopathy		Blood flow	Careful observation revealed that flow velocity fluctuations were found with higher frequency in diabetic patients than in normal subjects. Elongation rate differed significantly between the normal and NDR groups as well as the normal and NPDR groups.	N = 27 patients, N = 20 control		AOSLO (Canon)			25212778	2014	Arichika S, Uji A, Murakami T, Unoki N, Yoshitake S, Dodo Y, Ooto S, Miyamoto K, Yoshimura N.	Invest Ophthalmol Vis Sci	Retinal hemorheologic characterization of early-stage diabetic retinopathy using adaptive optics scanning laser ophthalmoscopy.
Diabetic retinopathy		Wavefront Aberrations	Diabetic retinopathy subjects had higher wavefront aberrations and less compact SH spots, likely attributable to pathological changes in the ocular optics. Wavefront aberrations were significantly reduced by AO, although AO performance was suboptimal in DR subjects as compared with control subjects	N = 19 patients, N = 10 control		AO-Cam (custom)			24748028	2014	Valeshabad AK, Wanek J, Grant P, Lim JI, Chau FY, Zelkha R, Camardo N, Shahidi M.	Optom Vis Sci	Wavefront error correction with adaptive optics in diabetic retinopathy.
Diabetic retinopathy		Microvasculature	Clinically undetected intraretinal vessel remodeling and varying blood flow patterns were found. Perifoveal capillary diameters were larger in the diabetic subjects, and small arteriolar walls were thickened, based on wall to lumen measurements.	N = 7		AOSLO, SD-OCT			24688827	2014	Burns SA, Elsner AE, Chui TY, Vannasdale DA Jr, Clark CA, Gast TJ, Malinovsky VE, Phan AD.	Biomed Opt Express	In vivo adaptive optics microvascular imaging in diabetic patients without clinically severe diabetic retinopathy.
Diabetic retinopathy		Microvasculature	AOSLO imaging can be used to longitudinally track capillaries, leukocytes, and photoreceptors in diabetic retinopathy. Capillary changes that can be detected include dropout of individual capillaries as well as formation and disappearance of microaneurysms	N = 1		AOSLO			22525131	2012	Tam J, Dhamdhere KP, Tiruveedhula P, Lujan BJ, Johnson RN, Bearse MA Jr, Adams AJ, Roorda A.	Optom Vis Sci	Subclinical capillary changes in non-proliferative diabetic retinopathy.
Diabetic retinopathy		Microvasculature, Hard exudates	The high resolution of the AOSLO allowed the detection of these early vascular changes induced by diabetes.	N = 1		AOSLO			17265801	2006	Roorda A, Garcia CA, Martin JA, Poonja S, Queener H, Romero-Borja F, Sepulveda R, Venkateswaran K, Zhang Y.	Bull Soc Belge Ophtalmol	What can adaptive optics do for a scanning laser ophthalmoscope ?
Diabetic retinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; vessel associated membrane;	N = 2	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Diabetic retinopathy		Microvasculature	AOSLO Offset Pinhole offers a non-invasive alternative to AOSLO FA without the need for any exogenous contrast agent.	N = 1	various diseases in this paper, method evaluation	AOSLO, Fundus Cam, OCT			24761299	2014	Chui TY, Dubow M, Pinhas A, Shah N, Gan A, Weitz R, Sulai YN, Dubra A, Rosen RB.	Biomed Opt Express	Comparison of adaptive optics scanning light ophthalmoscopic fluorescein angiography and offset pinhole imaging.
Diabetic retinopathy		Microvasculature	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA), SD-OCT			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Diabetic retinopathy		Hard Exudates	AO-SLO imaging enables morphological classification of retinal hard exudates (HE) into two types. The retinal thickness in regions with round HE was significantly increased compared to regions with irregular HE.	N = 22 patients	brief report	AOSLO (Canon), SD-OCT			27641223	2016	Yamaguchi M, Nakao S, Kaizu Y, Kobayashi Y, Nakama T, Arima M, Yoshida S, Oshima Y, Takeda A, Ikeda Y, Mukai S, Ishibashi T, Sonoda KH.	Sci Rep	High-Resolution Imaging by Adaptive Optics Scanning Laser Ophthalmoscopy Reveals Two Morphologically Distinct Types of Retinal Hard Exudates
Diabetic Retinopathy		Cones	This study shows an association between capillary non-perfusion of the deep capillary plexus and abnormalities in the photoreceptor layer in eyes with DR.	N = 11 patients		AO-SLO, OCT-A			28068435	2017	Nesper PL, Scarinci F, Fawzi AA.	PLoS One	Adaptive Optics Reveals Photoreceptor Abnormalities in Diabetic Macular Ischemia
Diabetic Retinopathy		retinal arterial wall	Retinal artery wall measurements can be potential surrogate markers of early diabetic microangiopathy	N = 28 patients, N = 31 healthy		AO-SLO, FCP			27913444	2017	Arichika S, Uji A, Murakami T, Suzuma K, Gotoh N, Yoshimura N.	Br J Ophthalmol	Correlation of retinal arterial wall thickness with atherosclerosis predictors in type 2 diabetes without clinical retinopathy
Diabetic Retinopathy		Cones	Absolute cone density and spacing don't appear to change substantially in DM. Decreased regularity of the cone arrangement is consistently associated with the presence of DM, increasing DR severity, and DME.	N = 53		AOSLO			27926754	2016	Lammer J, Prager SG, Cheney MC, Ahmed A, Radwan SH, Burns SA, Silva PS, Sun JK.	Invest Ophthalmol Vis Sci	Cone Photoreceptor Irregularity on Adaptive Optics Scanning Laser Ophthalmoscopy Correlates With Severity of Diabetic Retinopathy and Macular Edema
Diabetic Retinopathy		Microvasculature	Retinal vascular caliber measurement using adaptive optics is a highly sensitive method of visualization and monitoring of early signs of diabetic and hypertensive retinopathy.	N = 15 (diabetic and hypertensive)	various diseases in this paper	FA, AO-Fundus Cam			21721269	2011	Stepushina OA, Bol'shunov AV	Vestn Oftalmol	Combination of measurement of retinal vascular caliber, adaptive optics and fluorescent angiography in early diagnosis and monitoring of diabetic and hypertensive retinopathy
Diabetic retinopathy		Cones	The foveal cone mosaic can show localized areas of dark cones that persist over time, that apparently correspond to either missing or nonreflecting cones, and may be related to local retinal ischemia. All participants with these localized defects had alterations in the juxtafoveal capillary network.	N = 85 control, N = 54 diabetic		AO-SLO, SLO, OCT			28687853	2017	Sawides L, Sapoznik KA, de Castro A, Walker BR1, Gast TJ, Elsner AE, Burns SA	Invest Ophthalmol Vis Sci.	Alterations to the Foveal Cone Mosaic of Diabetic Patients
diabetic retinopathy (Nonproliferative)		Blood vessels	The relation between parent and daughter branch diameters changes in diabetes, but the branching angles do not.	N = 17 patients, N = 26 healthy		AO-SLO			28525557	2017	Luo T, Gast TJ, Vermeer TJ, Burns SA.	Invest Ophthalmol Vis Sci	Retinal Vascular Branching in Healthy and Diabetic Subjects
Prediabetes		Cones, retinal blood vessels	A multivariate regression analysis showed that the wall-to-lumen ratio was significantly correlated with BMI and total cholesterol. Abnormalities found, indicated early signs of arteriolar dysfunction, prior to impaired glucose tolerance progressing to diabetes.	N = 12 prediabetic patients, N = 22 healthy		rtx-1			29238728	2017	Zaleska-Żmijewska A, Piątkiewicz P, Śmigielska B, Sokołowska-Oracz A, Wawrzyniak ZM, Romaniuk D, Szaflik J, Szaflik JP	J Diabetes Res.	Retinal Photoreceptors and Microvascular Changes in Prediabetes Measured with Adaptive Optics (rtx1™): A Case-Control Study
Branch Retinal Vein Occlusion (BRVO)		Photoreceptors	After BRVO-associated retinal hemorrhage and macular edema resolved, affected parafoveal cone density decreases and the cone mosaic spatial arrangement is disrupted, becoming more irregular. These cone microstructural abnormalities may extend to parafovea in the BRVO-unaffected side.	N = 21		AOSLO (Canon), OCT			24531026	2014	Akagi-Kurashige Y, Tsujikawa A, Ooto S, Makiyama Y, Muraoka Y, Kumagai K, Uji A, Arichika S, Murakami T, Miyamoto K, Yoshimura N	Am J Ophthalmol	Retinal microstructural changes in eyes with resolved branch retinal vein occlusion: an adaptive optics scanning laser ophthalmoscopy study.
Branch Retinal Vein Occlusion (BRVO)		Microvasculature	AOSLO Offset Pinhole offers a non-invasive alternative to AOSLO FA without the need for any exogenous contrast agent.	N = 1	various diseases in this paper, method evaluation	AOSLO, Fundus Cam, OCT			24761299	2014	Chui TY, Dubow M, Pinhas A, Shah N, Gan A, Weitz R, Sulai YN, Dubra A, Rosen RB.	Biomed Opt Express	Comparison of adaptive optics scanning light ophthalmoscopic fluorescein angiography and offset pinhole imaging.
Branch Retinal Vein Occlusion (BRVO)		Microvasculature	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA)			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.
Branch Retinal Vein Occlusion (BRVO)		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane;	N = 3	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Branch Retinal Vein Occlusion (BRVO)		blood vessels	Affected venous segments showed a variable association of nicking, narrowing, deviation, and opacification. The degree of venous narrowing ranged from 40% to 77%, while at these sites, the width of the intervascular space ranged from 16µm to 42 µm.	N = 3 patients	various diseases in this paper	rtx1			25997175	2015	Paques M, Brolly A, Benesty J, Lermé N, Koch E, Rossant F, Bloch I, Girmens JF.	JAMA Ophthalmol	Venous Nicking Without Arteriovenous Contact: The Role of the Arteriolar Microenvironment in Arteriovenous Nickings.
Branch Retinal Vein Occlusion (BRVO)		Microvasculature	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA), SD-OCT			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.
Branch Retinal Vein Occlusion (BRVO)		Hard Exudates	AO-SLO imaging enables morphological classification of retinal hard exudates (HE) into two types. The retinal thickness in regions with round HE was significantly increased compared to regions with irregular HE.	N = 5 patients	brief report	AOSLO (Canon), SD-OCT			27641223	2016	Yamaguchi M, Nakao S, Kaizu Y, Kobayashi Y, Nakama T, Arima M, Yoshida S, Oshima Y, Takeda A, Ikeda Y, Mukai S, Ishibashi T, Sonoda KH.	Sci Rep	High-Resolution Imaging by Adaptive Optics Scanning Laser Ophthalmoscopy Reveals Two Morphologically Distinct Types of Retinal Hard Exudates
Branch Retinal Vein Occlusion (BRVO)		Microvasculature	Multimodal imaging illustrated a novel mechanism of branch retinal vein occlusion in which a primary retinal arteriolar macroaneurysm adjacent to the junction of two retinal veins led to obstruction of venous flow without evidence of direct compression.	N = 1		OCT, OCT-A, AO-Fundus, FA			28079651	2017	Chen Y, Chen SD, Chen FK.	Retin Cases Brief Rep	BRANCH RETINAL VEIN OCCLUSION SECONDARY TO A RETINAL ARTERIOLAR MACROANEURYSM: A NOVEL MECHANISM SUPPORTED BY MULTIMODAL IMAGING
Central Retinal Vein Occlusion (CRVO)		Microvasculature	AOSLO Offset Pinhole offers a non-invasive alternative to AOSLO FA without the need for any exogenous contrast agent.	N = 1	various diseases in this paper, method evaluation	AOSLO, Fundus Cam, OCT			24761299	2014	Chui TY, Dubow M, Pinhas A, Shah N, Gan A, Weitz R, Sulai YN, Dubra A, Rosen RB.	Biomed Opt Express	Comparison of adaptive optics scanning light ophthalmoscopic fluorescein angiography and offset pinhole imaging.
Central Retinal Vein Occlusion (CRVO)		Microvasculature	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA), SD-OCT			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.
Retinal Vein Occlusion (RVO)		Microvasculature	Quantitative evaluation of the parafoveal aggregated erythrocyte velocity.	N = 10 patients		OCT, AO-SLO		ranibizumab	28033234	2016	Iida Y, Muraoka Y, Uji A, Ooto S, Murakami T, Suzuma K, Tsujikawa A, Arichika S, Takahashi A, Miwa Y, Yoshimura N.	Retina	ASSOCIATIONS BETWEEN MACULAR EDEMA AND CIRCULATORY STATUS IN EYES WITH RETINAL VEIN OCCLUSION: An Adaptive Optics Scanning Laser Ophthalmoscopy Study
Retinal Vasculitis		Microvasculature	AO can be used as an additional investigative tool for diagnosis and to monitor the disease course during the treatment.	N = 6 patients		FCP, FFA, AO			28010147	2016	Mahendradas P, Vala R, Kawali A, Akkali MC, Shetty R.	Ocul Immunol Inflamm	Adaptive Optics Imaging in Retinal Vasculitis

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Hypertensive retinopathy		Retinal Arterioles	Retinal arteriolar remodeling comprised blood pressure and age-driven wall thickening as well as blood pressure-triggered lumen narrowing in younger individuals.	N = 1000	various diseases in this paper	rtx1			27065002	2016	Rosenbaum D, Mattina A, Koch E, Rossant F, Gallo A, Kachenoura N, Paques M, Redheuil A, Girerd X.	J Hypertens	Effects of age, blood pressure and antihypertensive treatments on retinal arterioles remodeling assessed by adaptive optics.
Hypertensive retinopathy		Microvasculature	Wall-to-Lumen-Ratio and Total Peripheral Resistance were significantly higher and aortic distensibility was significantly lower in hypertensives. Aortic dilation and arch elongation were found in uncontrolled hypertensives. The multivariate analysis indicated that WLR was associated with TPR (P=0.002) independent of age, BMI, gender, antihypertensive treatments, aortic diameter and central SBP.	N = 57 patients, N = 23 control		rtx1			27009576	2016	Rosenbaum D, Kachenoura N, Koch E, Paques M, Cluzel P, Redheuil A, Girerd X.	Hypertens Res	Relationships between retinal arteriole anatomy and aortic geometry and function and peripheral resistance in hypertensives.
Hypertensive retinopathy		Blood vessels	In the normal and hypertensive groups, Wall-To-Lumen-Ratio showed a strong correlation with systolic and diastolic blood pressure.	N = 22 patients, N = 51 control		AOSLO (Canon)			26192115	2015	Arichika S, Uji A, Ooto S, Muraoka Y, Yoshimura N	Sci Rep	Effects of age and blood pressure on the retinal arterial wall, analyzed using adaptive optics scanning laser ophthalmoscopy.
Hypertensive retinopathy		Blood vessel walls	The average wall thickness, with hypertension, was 18.7 µm, and the wall-to-lumen ratio was 0.44, both bigger than normal.	N = 1 patient, N = 1 control		AOSLO (Canon)			25336903	2014	Arichika S, Uji A, Yoshimura N.	Clin Ophthalmol	Adaptive optics assisted visualization of thickened retinal arterial wall in a patient with controlled malignant hypertension.
Hypertensive retinopathy		Blood vessel walls	High-resolution retinal imaging of subjects with essential hypertension showed a significant decrease in vessel inner diameter for a given outer diameter, and increases in wall to lumen ratio and wall cross-sectional areas over the entire range of vessel diameters and suggests that correcting for vessel size may improve the ability to identify significant vascular changes.	N = 23 hypertensive, N = 22 normal tensive, N = 10 hypotensive		AOSLO			27617182	2016	Hillard JG, Gast TJ, Chui TY, Sapir D, Burns SA.	Transl Vis Sci Technol	Retinal Arterioles in Hypo-, Normo-, and Hypertensive Subjects Measured Using Adaptive Optics.
Hypertensive retinopathy		Microvasculature	AOSLO Offset Pinhole offers a non-invasive alternative to AOSLO FA without the need for any exogenous contrast agent.	N = 1	various diseases in this paper, method evaluation	AOSLO, Fundus Cam, OCT			24761299	2014	Chui TY, Dubow M, Pinhas A, Shah N, Gan A, Weitz R, Sulai YN, Dubra A, Rosen RB.	Biomed Opt Express	Comparison of adaptive optics scanning light ophthalmoscopic fluorescein angiography and offset pinhole imaging.
Hypertensive retinopathy		Microvasculature	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA)			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.
Hypertensive retinopathy		Blood vessel walls	In the cohort of treatment-naive individuals, by multiple regression taking into account age, body mass index, mean, systolic, diastolic and pulse blood pressure, the Wall-to-Lumen-Ratio was found positively correlated to mean blood pressure and age which in combination accounted for 43% of the variability of WLR.	30 healthy, 19 hypertensive		rtx1			24406779	2014	Koch E, Rosenbaum D, Brolly A, Sahel JA, Chaumet-Riffaud P, Girerd X, Rossant F, Paques M.	J Hypertens	Morphometric analysis of small arteries in the human retina using adaptive optics imaging: relationship with blood pressure and focal vascular changes
Hypertensive retinopathy		blood vessels	Affected venous segments showed a variable association of nicking, narrowing, deviation, and opacification. The degree of venous narrowing ranged from 40% to 77%, while at these sites, the width of the intervascular space ranged from 16µm to 42 µm.	N = 4 patients	various diseases in this paper	rtx1			25997175	2015	Paques M, Brolly A, Benesty J, Lermé N, Koch E, Rossant F, Bloch I, Girmens JF.	JAMA Ophthalmol	Venous Nicking Without Arteriovenous Contact: The Role of the Arteriolar Microenvironment in Arteriovenous Nickings.
Hypertensive retinopathy		Hard Exudates	AO-SLO imaging enables morphological classification of retinal hard exudates (HE) into two types. The retinal thickness in regions with round HE was significantly increased compared to regions with irregular HE.	N = 1 patient	brief report	AOSLO (Canon), SD-OCT			27641223	2016	Yamaguchi M, Nakao S, Kaizu Y, Kobayashi Y, Nakama T, Arima M, Yoshida S, Oshima Y, Takeda A, Ikeda Y, Mukai S, Ishibashi T, Sonoda KH.	Sci Rep	High-Resolution Imaging by Adaptive Optics Scanning Laser Ophthalmoscopy Reveals Two Morphologically Distinct Types of Retinal Hard Exudates
Hypertensive retinopathy		Microvasculature	Retinal vascular caliber measurement using adaptive optics is a highly sensitive method of visualization and monitoring of early signs of diabetic and hypertensive retinopathy.	N = 15 (diabetic and hypertensive)	various diseases in this paper	FA, AO-Fundus Cam			21721269	2011	Stepushina OA, Bol'shunov AV.	Vestn Oftalmol	Combination of measurement of retinal vascular caliber, adaptive optics and fluorescent angiography in early diagnosis and monitoring of diabetic and hypertensive retinopathy
Hypertension		Microvasculature	Capillary density is increased only after treatment with lercanidipine + enalapril. In conclusion, lercanidipine both in monotherapy and in combination with enalapril but not with hydrochlorothiazide is able to improve microvascular structure; on the other hand, a decrease in central blood pressure is observed with both therapeutic combinations.	N = 30 patients		rtx-1		N = 15: lercanidipine + enalapril, N = 15: lercanidipine + hydrochlorothiazide	28647890	2017	De Ciuceis C, Salvetti M, Paini A, Rossini C, Muiesan ML, Duse S, Caletti S, Coschignano MA, Semeraro F, Trapletti V, Bertacchini F, Brami V, Petelca A, Agabiti Rosei E, Rizzoni D, Agabiti Rosei C	Intern Emerg Med.	Comparison of lercanidipine plus hydrochlorothiazide vs. lercanidipine plus enalapril on micro and macrocirculation in patients with mild essential hypertension
(resistant) Hypertension		Microvasculature	While a reverse eutrophic remodeling was observed in patients undergoing a standard-antihypertensive treatment, hypotrophic changes were found in RH patients undergoing baroreceptor activation therapy.	N = 5 RH-patients (treated), N = 21 hypertension patients (untreated)		rtx1		Baroreceptor activation therapy	28554698	2017	Gallo A, Rosenbaum D, Kanagasabapathy C, Girerd X	Ann Cardiol Angeiol (Paris)	Effects of carotid baroreceptor stimulation on retinal arteriole remodeling evaluated with adaptive optics camera in resistant hypertensive patients
Sickle cell retinopathy		Microvasculature, Blood vessel walls	Compared with healthy eyes, capillary nonperfusion in the vasculopathic eyes was more extensive. All six vasculopathic eyes had decreased microvascular densities.	N = 1	various diseases in this paper	AOSLO (FA)			25414179	2014	Pinhas A, Razeen M, Dubow M, Gan A, Chui TY, Shah N, Mehta M, Gentile RC, Weitz R, Walsh JB, Sulai YN, Carroll J, Dubra A, Rosen RB.	Invest Ophthalmol Vis Sci	Assessment of perfused foveal microvascular density and identification of nonperfused capillaries in healthy and vasculopathic eyes.
Retinopathy of prematurity		Photoreceptors	In the AOSLO images, cone density was lower and the packing pattern less regular in TROP, relative to control and untreated retinae. There was no evidence of cone loss in the TROP OCT images.	N = 5 treated, N = 5 untreated, N = 8 controls		AOSLO			26868749	2016	Ramamirtham R, Akula JD, Soni G, Swanson MJ, Bush JN, Moskowitz A, Swanson EA, Favazza TL, Tavormina JL, Mujat M, Ferguson RD, Hansen RM, Fulton AB.	Invest Ophthalmol Vis Sci	Extrafoveal Cone Packing in Eyes With a History of Retinopathy of Prematurity.
Retinopathy of prematurity		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Central Macular Arteriovenous Malformation		photoreceptors	New finding of photoreceptor damage associated with an anomalous macular vessel only detectable by new imaging techniques such as SD- OCT and AO imaging.	N = 1		custom AO fundus, OCT, FAF	mf ERG		20337274	2010	Telander DG, Choi SS, Zawadzki RJ, Berger N, Keltner JL, Werner JS.	Ophthalmic Surg Lasers Imaging.	Microstructural Abnormalities Revealed by High Resolution Imaging Systems in Central Macular Arteriovenous Malformation.
Renal retinopathy		Hard Exudates	AO-SLO imaging enables morphological classification of retinal hard exudates (HE) into two types. The retinal thickness in regions with round HE was significantly increased compared to regions with irregular HE.	N = 1 patient	brief report	AOSLO (Canon), SD-OCT			27641223	2016	Yamaguchi M, Nakao S, Kaizu Y, Kobayashi Y, Nakama T, Arima M, Yoshida S, Oshima Y, Takeda A, Ikeda Y, Mukai S, Ishibashi T, Sonoda KH.	Sci Rep	High-Resolution Imaging by Adaptive Optics Scanning Laser Ophthalmoscopy Reveals Two Morphologically Distinct Types of Retinal Hard Exudates

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Glacuoma and diseases of the ONH													
Glaucoma		Nerve fibres	As seen on AO-SLO, the pattern of abnormal RNF bundles near the border of the within normal limits and abnormal regions differed across eyes. However, in two of these eyes, a few bundles were seen within this region of otherwise missing bundles.	N = 7		AOSLO, OCT			25574048	2015	Chen MF, Chui TY, Alhadeff P, Rosen RB, Ritch R, Dubra A, Hood DC.	Invest Ophthalmol Vis Sci	Adaptive optics imaging of healthy and abnormal regions of retinal nerve fiber bundles of patients with glaucoma.
Glaucoma		lamina cribrosa	Average pore surface area was significantly different. In healthy subjects with at least one direct relative with POAG, 21% had pores with an appearance comparable to that of subjects in the glaucoma group.	N = 30 patients, N = 15 healthy control, N = 14 healthy but hereditary risk		rtx1			26987895	2016	Zwillinger S, Paques M, Safran B, Baudouin C	J Fr Ophtalmol	In vivo characterization of lamina cribrosa pore morphology in primary open-angle glaucoma.
Glaucoma		Nerve fibres	On AO-SLO images, three eyes showed small regions of preserved and/or missing RNFL bundles within the affected region.	N = 6		AOSLO, SS-OCT			26426403	2015	Hood DC, Fortune B, Mavrommatis MA, Reynaud J, Ramachandran R, Ritch R, Rosen RB, Muhammad H, Dubra A, Chui TY.	Invest Ophthalmol Vis Sci	Details of Glaucomatous Damage Are Better Seen on OCT En Face Images Than on OCT Retinal Nerve Fiber Layer Thickness Maps.
Glaucoma		Nerve fibres, Raphe	The raphe gap was larger in glaucomatous eyes than control eyes. The bundle index, GCC thickness, and RNFL thickness were on average reduced in glaucomatous eyes, with the first two showing statistically significant differences between the two groups.	N = 9 patients, N = 10 control		AOSLO			26047040	2015	Huang G, Luo T, Gast TJ, Burns SA, Malinovsky VE, Swanson WH.	Invest Ophthalmol Vis Sci	Imaging Glaucomatous Damage Across the Temporal Raphe.
Glaucoma		Nerve fibre bundles	Relatively similar 10-2 defects with similar fdOCT RNFL thickness profiles can have very different degrees of RNF bundle damage as seen on fdOCT and AO-SLO.	N = 6		AOSLO, fdOCT			25909035	2015	Hood DC, Chen MF, Lee D, Epstein B, Alhadeff P, Rosen RB, Ritch R, Dubra A, Chui TY.	Transl Vis Sci Technol	Confocal Adaptive Optics Imaging of Peripapillary Nerve Fiber Bundles: Implications for Glaucomatous Damage Seen on Circumpapillary OCT Scans.
Glaucoma		lamina cribrosa	The mean increase in pore area was larger following 3D transformation in glaucomatous eyes due to their more steeply curved laminar surfaces, while the change in pore elongation was comparable to that in normal eyes.	N = 4 patients, N = 11 control	method evaluation	AOSLO, OCT			23847739	2013	Sredar N, Ivers KM, Queener HM, Zouridakis G, Porter J.	Biomed Opt Express	3D modeling to characterize lamina cribrosa surface and pore geometries using in vivo images from normal and glaucomatous eyes.
Glaucoma		Cones	Both AO-SLO and SD-OCT showed cone integrity in eyes with glaucoma, even in areas with visual field and nerve fiber loss. In AO-SLO, microcystic lesions in the inner nuclear layer may influence images of the cone mosaic.	N = 35 patients, N = 21 control		AOSLO, SD-OCT			27565227	2016	Hasegawa T, Ooto S, Takayama K, Makiyama Y, Akagi T, Ikeda HO, Nakanishi H, Suda K, Yamada H, Uji A, Yoshimura N.	Am J Ophthalmol	Cone integrity in glaucoma: an adaptive-optics scanning laser ophthalmoscopy study.
Glaucoma		Inner and outer retinal morphology	The results demonstrate that nonglaucomatous and glaucomatous optic neuropathies are associated with outer retinal changes following long-term inner retinal pathology.	N = 10 patients, N = 6 control		AO Fundus Cam, AOOCT, FDOCT			21293495	2011	Werner JS, Keltner JL, Zawadzki RJ, Choi SS.	Eye (Lond)	Outer retinal abnormalities associated with inner retinal pathology in nonglaucomatous and glaucomatous optic neuropathies.
Glaucoma		lamina cribrosa	The pore area was significantly larger in glaucomatous subjects than in normal subjects, but elongation index was not.	N = 20 patients, N = 20 control		AOSLO			22669726	2012	Akagi T, Hangai M, Takayama K, Nonaka A, Ooto S, Yoshimura N.	Invest Ophthalmol Vis Sci	In vivo imaging of lamina cribrosa pores by adaptive optics scanning laser ophthalmoscopy.
Glaucoma		Cones	Identification of the exact location of structural changes within the cone photoreceptor layer. Images showing dark areas in the cone mosaic at the same retinal locations with reduced visual sensitivity.	N = 10		AO Fundus Cam, UHR-FD-OCT			20956277	2011	Choi SS, Zawadzki RJ, Lim MC, Brandt JD, Keltner JL, Doble N, Werner JS.	Br J Ophthalmol	Evidence of outer retinal changes in glaucoma patients as revealed by ultrahigh-resolution in vivo retinal imaging.
Glaucoma		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane; waxy membrane;	N = 11		AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Glaucoma (open angle)		Nerve fibres	Progressive changes in RNF bundles associated with deep defects on 10-2 VFs can be seen within about 1 year with AO-SLO imaging. Subtle thinning of regions with RNF bundles is not easy to see with current AO-SLO technology, and may be better followed with OCT.	N = 6 eyes (5 patients)		AO-SLO			28713646	2017	Hood DC, Lee D, Jarukasetphon R, Nunez J, Mavrommatis MA, Rosen RB, Ritch R, Dubra A, Chui TYP	Transl Vis Sci Technol.	Progression of Local Glaucomatous Damage Near Fixation as Seen with Adaptive Optics Imaging
Glaucoma (open angle)		Blood flow	The results reveal that tafluprost may not only lower IOP but may also improve retinal circulation in POAG eyes and AOSLO may be useful to evaluate retinal circulatory change after treatment.	N = 11 healthy, N = 11 patients		AO-SLO (canon)		topical tafluprost treatment	28694501	2017	Iida Y, Akagi T, Nakanishi H, Ohashi Ikeda H, Morooka S, Suda K, Hasegawa T, Yokota S, Yoshikawa M, Uji A, Yoshimura N	Sci Rep	Retinal Blood Flow Velocity Change in Parafoveal Capillary after Topical Tafluprost Treatment in Eyes with Primary Open-angle Glaucoma
Multiple sclerosis		Photoreceptor structure	Cone photoreceptors show structural changes when there is permanent damage to overlying inner retinal layers. There was a positive relation between the thickness of the three-layer inner retinal complex, visual sensitivity, and integrity of the cone mosaic.	N = 2	various diseases in this paper	AO Fundus Cam, AOOCT, FDOCT			18436843	2008	Choi SS, Zawadzki RJ, Keltner JL, Werner JS.	Invest Ophthalmol Vis Sci	Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies
Multiple sclerosis		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Optic nerve head drusen		Nerve fibres, Photoreceptors	Based on this study, changes occur not only in the RNFL but also in the photoreceptor layer in optic nerve drusen complicated by ischemic optic neuropathy.	N = 1	same as in: Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies	AO Fundus Cam, FDOCT			18562844	2008	Choi SS, Zawadzki RJ, Greiner MA, Werner JS, Keltner JL.	J Neuroophthalmol	Fourier-domain optical coherence tomography and adaptive optics reveal nerve fiber layer loss and photoreceptor changes in a patient with optic nerve drusen.
Optic nerve head drusen		Photoreceptor structure	Cone photoreceptors show structural changes when there is permanent damage to overlying inner retinal layers. There was a positive relation between the thickness of the three-layer inner retinal complex, visual sensitivity, and integrity of the cone mosaic.	N = 1	various diseases in this paper	AO Fundus Cam, AOOCT, FDOCT			18436843	2008	Choi SS, Zawadzki RJ, Keltner JL, Werner JS.	Invest Ophthalmol Vis Sci	Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies
Optic nerve head drusen		Inner Retinal Reflectivity	Inner retinal phenotype: nummular (disc-shaped) reflectivity;	N = 2	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Autosomal dominant optic atrophy (ADOA)		Microcystic Macular Edema				rtx1			24369534	2013	Gocho K, Kikuchi S, Kabuto T, Kameya S, Shinoda K, Mizota A, Yamaki K, Takahashi H.	Biomed Res Int	High-resolution en face images of microcystic macular edema in patients with autosomal dominant optic atrophy.
Autosomal dominant optic atrophy (ADOA)		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane; waxy membrane; vessel associated membrane; microcysts;	N = 5	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Optic neuritis		Inner Retinal Reflectivity	Inner retinal phenotype: microcysts;	N = 3	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Parkinson's disease		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane; waxy membrane; vessel associated membrane; striate reflectivity	N = 3	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Acquired optic disc pit		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; waxy membrane;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Idiopathic intracranial hypertension		Photoreceptor structure	Cone photoreceptors show structural changes when there is permanent damage to overlying inner retinal layers. There was a positive relation between the thickness of the three-layer inner retinal complex, visual sensitivity, and integrity of the cone mosaic.	N = 1	various diseases in this paper	AO Fundus Cam, AOOCT, FDOCT			18436843	2008	Choi SS, Zawadzki RJ, Keltner JL, Werner JS.	Invest Ophthalmol Vis Sci	Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies
Traumatic brain injury		Inner Retinal Reflectivity	microcystic spaces in the inner nuclear layer (INL) [has been previously described already with SD-OCT for all types of optic atrophy and glaucoma]	N = 1		AOSLO, SD-OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Nonarteritic anterior ischemic optic neuropathy (NAION)		Photoreceptor structure	Cone photoreceptors show structural changes when there is permanent damage to overlying inner retinal layers. There was a positive relation between the thickness of the three-layer inner retinal complex, visual sensitivity, and integrity of the cone mosaic.	N = 1	various diseases in this paper	AO Fundus Cam, AOOCT, FDOCT			18436843	2008	Choi SS, Zawadzki RJ, Keltner JL, Werner JS.	Invest Ophthalmol Vis Sci	Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies
Leber's hereditary optic neuropathy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Leber's hereditary optic neuropathy		Cones, Microcystic lesions	Manifestion of dark, partition-like areas in the cone mosaic on AO-SLO images. Microcystic lesions in the INL may affect the images of the cone mosaic.	N = 1	various diseases in this paper	AO-SLO, SD-OCT			28291071	2017	Hasegawa T, Ooto S, Makiyama Y, Hata M, Miyamoto K, Yoshimura N	Retin Cases Brief Rep	CIRCINATE PARTITION-LIKE FINDINGS ON CONE MOSAIC IMAGED BY ADAPTIVE OPTICS SCANNING LASER OPHTHALMOSCOPY IN EYES WITH INNER NUCLEAR LAYER MICROCYSTIC CHANGES

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
White dot syndroms, uveitis and inflammatory disorders													
Acute zonal occult outer retinopathy		Photoreceptors	Normal retina was observed in the areas with normal visual function. Discontinuity in the cone photoreceptor mosaic in the area of the relative scotoma was observed. Photoreceptor spacing in the area of the relative scotoma is consistent with the existence of rod photoreceptors.	N = 1	various diseases in this paper	AOSLO			21833357	2011	Merino D, Duncan JL, Tiruveedhula P, Roorda A.	Biomed Opt Express	Observation of cone and rod photoreceptors in normal subjects and patients using a new generation adaptive optics scanning laser ophthalmoscope.
Acute zonal occult outer retinopathy		Cones Mosaic	The cone mosaics were disrupted in the abnormal hyporeflective area of the IR image. However, the areas of abnormalities did not coincide with the hyporeflective areas in the IR images.	N = 12 eyes of 10 patients		rtx1	mfERG Function Testing		25923954	2015	Ueno S, Kawano K, Ito Y, Ra E, Nakanishi A, Nagaya M, Terasaki H.	Retina	NEAR-INFRARED REFLECTANCE IMAGING IN EYES WITH ACUTE ZONAL OCCULT OUTER RETINOPATHY.
Acute zonal occult outer retinopathy		Cones	This study might suggest reversible cone damage could occur in some cases of AZOOR with spontaneous remission.	N = 1		AOSLO (Canon), SD-OCT	mfERG Function Testing		25081027	2015	Nakao S, Kaizu Y, Yoshida S, Iida T, Ishibashi T.	Graefes Arch Clin Exp Ophthalmol	Spontaneous remission of acute zonal occult outer retinopathy: follow-up using adaptive optics scanning laser ophthalmoscopy.
Acute zonal occult outer retinopathy		Cones	In each patient, loss of retinal function correlated with structural changes in the outer retina. AOSLO showed focal cone loss in most patients, although in 1 patient with central vision loss such change was absent. In another patient, structural and functional analyses suggested that cones had degenerated but rods remained.	N = 4 patients, N = 27 control		AOSLO, SD-OCT	mfERG, ffERG and microperimetry Function Testing		22105799	2012	Mkrtchyan M, Lujan BJ, Merino D, Thirkill CE, Roorda A, Duncan JL.	Am J Ophthalmol	Outer retinal structure in patients with acute zonal occult outer retinopathy.
Multiple Evanescent White Dot Syndrome		choroid, RPE, Photoreceptorcells	Although changes in the choroid and RPE can be observed in MEWDS, adaptive optics imaging localized the visually significant changes seen in this disease at the level of the photoreceptors. These transient retinal changes specifically occur at the level of the inner segment ellipsoid and OS/RPE line.	N = 1	5-year observation	AOOCT, FAF, FD-OCT			26735319	2016	Labriola LT, Legarreta AD, Legarreta JE, Nadler Z, Gallagher D, Hammer DX, Ferguson RD, Iftimia N, Wollstein G, Schuman JS.	Retin Cases Brief Rep	IMAGING WITH MULTIMODAL ADAPTIVE-OPTICS OPTICAL COHERENCE TOMOGRAPHY IN MULTIPLE EVANESCENT WHITE DOT SYNDROME: THE STRUCTURE AND FUNCTIONAL RELATIONSHIP.
Multiple Evanescent White Dot Syndrome		Photoreceptors	microstructural changes may correlate with functional loss.	N = 19 eyes of 12 patients		rtx1, FAF, SD-OCT	Microperimetry Function Testing		26189087	2015	Agarwal A, Soliman MK, Hanout M, Sadiq MA, Sarwar S, Jack LS, Do DV, Nguyen QD, Sepah YJ.	Am J Ophthalmol	Adaptive Optics Imaging of Retinal Photoreceptors Overlying Lesions in White Dot Syndrome and its Functional Correlation.
Multiple Evanescent White Dot Syndrome		Photoreceptors	Photoreceptor disruption was apparent during the acute phase and recurrence	N = 1		rtx1, FAF			23676237	2013	Boretsky A, Mirza S, Khan F, Motamedi M, van Kuijk FJ.	Ophthalmic Surg Lasers Imaging Retina	High-resolution multimodal imaging of multiple evanescent white dot syndrome.
multiple evanescent white dot syndrome		Cones	Hyperreflective lesions (visible on AO-SLO), "Jampol dots," are the foveal corollaries of the same process associated with the classic "dot" lesions in MEWDS. The authors propose that material is accumulating at the level of the retinal pigment epithelium, based on the intact photoreceptor mosaic on AO-SLO .	N = 7 eyes of 6 patients (all female)		Ao-SLO, SD-OCT, CF			29190245	2017	Onishi AC, Roberts PK, Jampol LM, Nesper PL, Fawzi AA.	Retina	CHARACTERIZATION AND CORRELATION OF "JAMPOL DOTS" ON ADAPTIVE OPTICS WITH FOVEAL GRANULARITY ON CONVENTIONAL FUNDUS IMAGING
Acute middle macular neuroretinopathy		Photoreceptors	Both cases showed concomitant loss of integrity of the outer retinal structures on SD-OCT, and marked abnormalities on AO imaging with disruption of the visibility of the cone mosaic.	N = 4 eyes of 2 patients		rtx1, SD-OCT	mfERG Function Testing		26344727	2016	Audo I, Gocho K, Rossant F, Mohand-Said S, Loquin K, Bloch I, Sahel JA, Paques M.	Graefes Arch Clin Exp Ophthalmol	Functional and high-resolution retinal imaging monitoring photoreceptor damage in acute macular neuroretinopathy.
Acute middle macular neuroretinopathy		Photoreceptors	The cone photoreceptor density was decreased at the level of the AMN lesions. The cone mosaic disruption appeared heterogeneous and more widespread than the lesion detected in the IR-SLO and SD-OCT images. The areas of cone loss correlated with SD-OCT and microperimetry.	N = 4		rtx1, SD-OCT	Microperimetry Function Testing		25423637	2014	Mrejen S, Pang CE, Sarraf D, Goldberg NR, Gallego-Pinazo R, Klancnik JM, Sorenson JA, Yannuzzi LA, Freund KB	Ophthalmic Surg Lasers Imaging Retina	Adaptive optics imaging of cone mosaic abnormalities in acute macular neuroretinopathy.
Acute middle macular neuroretinopathy		Cones	AOSLO shows preferential disruption of cone photoreceptor structure within the region of decreased retinal sensitivity.	N = 1		AOSLO, SD-OCT	Microperimetry Function Testing		23615345	2013	Hansen SO, Cooper RF, Dubra A, Carroll J, Weinberg DV.	Retina	Selective cone photoreceptor injury in acute macular neuroretinopathy.
Acute middle macular neuroretinopathy									25659196	2015	Garnier MB, Castelbou M, Tumahai P, Montard M, Delbosc B, Saleh M.	J Fr Ophtalmol	[Acute macular neuroretinopathy and adaptive optics imaging. A case report].
Acute middle macular neuroretinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: striate reflectivity	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Acute idiopathic blind spot enlargement syndrome		Photoreceptors	This correspondence provided direct morphological evidence that damaged cones are capable, under some circumstances, of generating new outer segments.	N = 1		AOSLO			26213154	2015	Horton JC, Parker AB, Botelho JV, Duncan JL.	Sci Rep	Spontaneous Regeneration of Human Photoreceptor Outer Segments.
Acute Posterior Multifocal Placoid Pigment Epitheliopathy		Fundus	Acute posterior multifocal placoid pigment epitheliopathy is characterized by prominent RPE changes, but the permanent RPE damage (type 2 and 3 lesions) may be secondary to an acute transient choroidal inflammatory process (type 1 lesions)	N = 1	brief report	rtx1, FAF			23949236	2013	Mrejen S, Gallego-Pinazo R, Wald KJ, Freund KB.	JAMA Ophthalmol	Acute posterior multifocal placoid pigment epitheliopathy as a choroidopathy: what we learned from adaptive optics imaging.
Acute Posterior Multifocal Placoid Pigment Epitheliopathy		Cones	AO-SLO allowed for the direct observation of retinal disruptions and the ability of this technology to detect abnormalities in the left eye demonstrates a superior ability for in-depth retinal imaging.	N = 1		AOSLO			24392917	2014	Hong IH, Park SP, Chen CL, Kim HK, Tsang SH, Chang S.	Ophthalmic Surg Lasers Imaging Retina	Cone photoreceptor abnormalities correlate with vision loss in a case of acute posterior multifocal placoid pigment epitheliopathy.
Acute Posterior Multifocal Placoid Pigment Epitheliopathy		Fundus, Cones	the interdigitation zone could contribute substantially to the reflectance of the cone photoreceptormosaic.The absence of cones on adaptive optics images does not necessarily mean photoreceptor cell death.	N = 1	various diseases in this paper	rtx1			25284764	2015	Jacob J, Paques M, Krivosic V, Dupas B, Couturier A, Kulcsar C, Tadayoni R, Massin P, Gaudric A.	Am J Ophthalmol	Meaning of visualizing retinal cone mosaic on adaptive optics images.
Acute Posterior Multifocal Placoid Pigment Epitheliopathy		Photoreceptors	Irregularities in the reflectivity of the photoreceptor mosaic are visible on AO-SLO even in inactive APMPPE lesions, where the photoreceptor bands on SD-OCT have recovered.	N = 4 patients (N = 8 eyes)		CFP, SD-OCT, FAF, AO-SLO			28166161	2017	Roberts PK, Nesper PL, Onishi AC, Skondra D, Jampol LM, Fawzi AA.	Retina	CHARACTERIZING PHOTORECEPTOR CHANGES IN ACUTE POSTERIOR MULTIFOCAL PLACOID PIGMENT EPITHELIOPATHY USING ADAPTIVE OPTICS

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Rubella retinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; vessel associated membrane;	N = 1		AOSLO, SD-OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Rubella retinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; vessel associated membrane;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Lyme disease		Blood vessels	AO imaging revealed infiltrates in segments of the vessels with no or minimal changes detected otherwise.	N = 3	brief report	rtx1			24576889	2014	Errera MH, Coisy S, Fardeau C, Sahel JA, Kallel S, Westcott M, Bodaghi B, Paques M.	Ophthalmology	Retinal vasculitis imaging by adaptive optics.
CMV retinitis		Cone Mosaic	Severe photoreceptor damage can be induced by CMV retinitis, which can be monitored by OCT or AO modalities. AO imaging and OCT imaging can be used to monitor the severe photoreceptor damage induced by CMV retinitis.	N = 1	brief report	AOSLO (canon), rtx1			26148637	2016	Arichika S, Uji A, Yoshimura N.	Clin Exp Ophthalmol	Retinal structural features of cytomegalovirus retinitis with acquired immunodeficiency syndrome: an adaptive optics imaging and optical coherence tomography study.
HIV		Photoreceptors, retinal layers	Cone photoreceptor density is significantly reduced in HIV retinae compared with age-matched controls. HIV retinae also have increased macular retinal thickness.	N = 16 HIV+, N = 16 HIV-		rtx1, SD-OCT			26244973	2015	Arcinue CA, Bartsch DU, El-Emam SY, Ma F, Doede A, Sharpsten L, Gomez ML, Freeman WR.	PLoS One	Retinal Thickening and Photoreceptor Loss in HIV Eyes without Retinitis.
Birdshot choroidoretinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; waxy membrane;	N = 4	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Birdshot chorioretinopathy	HLA-A29+	Photoreceptors	Using AO-SLO, one instance of subclinical photoreceptor disruption not seen on SD-OCT was found.	N = 16 patients		AO-SLO (Apaeros), SD-OCT			28362542	2017	Khanna S, Nesper PL, Koreishi AF, Goldstein DA, Fawzi AA	Ocul Immunol Inflamm	Visualization of Photoreceptors in Birdshot Chorioretinopathy Using Adaptive Optics Scanning Laser Ophthalmoscopy: A Pilot Study
Cancer Associated and Related Autoimmune Retinopathy		Cone mosaic	This new imaging modality may be useful in establishing the diagnosis of this rare disease, monitoring disease progression and evaluating response to therapy.	N = 1	only case report	rtx1	Microperimetry Function Testing		26622144	2015	Dabir S, Mangalesh S, Govindraj I, Mallipatna A, Battu R, Shetty R.	Oman J Ophthalmol	Melanoma associated retinopathy: A new dimension using adaptive optics.
Autoimmune Retinopathy		Inner Retinal Reflectivity	Inner retinal phenotype: granular membrane;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2015	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Autoimmune retinopathy (acute paraneoplastic)		Cones	A spectrum of autoreactive anti-retinal antibodies is associated with a unique phenotype of acute progressive paravascular placoid neuroretinopathy resulting in degeneration of photoreceptor cells, inner retinal dysfunction and classic electronegative ERG in paraneoplastic retinopathy.	N = 1 patient		NIR, AF, ultra-widefield colour and green-light AF, F&IG-Angiography, SD-OCT, rtx1	ERG, microperimetry		28382556	2017	Chen FK, Chew AL, Zhang D, Chen SC, Chelva E, Chandrasekera E, Koay EM, Forrester J, McLenachan S	Doc Ophthalmol	Acute progressive paravascular placoid neuroretinopathy with negative-type electroretinography in paraneoplastic retinopathy
Autoimmune Retinopathy (AIR)		Photoreceptors	Visual acuity was stable or improved in a majority of AIR patients while they were being treated with rituximab. OCT and ERG parameters, as well as AO-SLO cone densities, were stable during treatment.	N = 16		SD-OCT, AO-SLO (Canon: RD102)	VA, ERG, VF	Rituximab	28483493	2017	Davoudi S, Ebrahimiadib N, Yasa C, Sevgi DD, Roohipoor R, Papavasilieou E, Comander J, Sobrin L	Am J Ophthalmol	Outcomes in Autoimmune Retinopathy Patients Treated With Rituximab.
Autoimmune Retinopathy		Cones	VA was stable or improved in a majority of AIR patients while they were being treated with rituximab. OCT and ERG parameters, as well as AO-SLO cone densities, were stable during treatment.	N = 16 patients		SD-OCT, AO-SLO	VA, ERG		28483493	2017	Davoudi S, Ebrahimiadib N, Yasa C, Sevgi DD, Roohipoor R, Papavasilieou E, Comander J, Sobrin L	Am J Ophthalmol	Outcomes in Autoimmune Retinopathy Patients Treated With Rituximab
Red Spot Syndrom		Fundus, Photoreceptors	When conventional clinical examination and imaging techniques fail to identify the presence of and visual symptoms in foveal red spot syndrome, advanced technologies may be used to confirm the diagnosis and explain the etiology of the abnormality.	N = 1		rtx1	Microperimetry Function Testing		25462132	2015	Yu S, Bellone D, Lee SE, Yannuzzi LA.	Retin Cases Brief Rep	Multimodal imaging in foveal red spot syndrome.
Unilateral acute idiopathic maculopathy		Photoreceptors	Images from AO-SLO revealed a remarkably restored cone mosaic, but with small, patchy, dark lesions in the fovea.	N = 1	brief report	AOSLO, SD-OCT	Microperimetry Function Testing		22159690	2011	Ooto S, Hangai M, Yoshimura N.	Arch Ophthalmol	Photoreceptor restoration in unilateral acute idiopathic maculopathy on adaptive optics scanning laser ophthalmoscopy.
Systemic lupus erythematosus		Photoreceptor structure	Cone photoreceptors show structural changes when there is permanent damage to overlying inner retinal layers. There was a positive relation between the thickness of the three-layer inner retinal complex, visual sensitivity, and integrity of the cone mosaic.	N = 1	various diseases in this paper	AO Fundus Cam, AOOCT, FDOCT			18436843	2008	Choi SS, Zawadzki RJ, Keltner JL, Werner JS.	Invest Ophthalmol Vis Sci	Changes in cellular structures revealed by ultra-high resolution retinal imaging in optic neuropathies
Vogt-Koyanagi-Harada (VKH) disease		Cones	Cone densities were gradually increased after the resolution of serous retinal detachment in the eyes of VKH disease patients. The presence of cystoid spaces might be a marker of severe damage to cone photoreceptors.	N = 8 patients (16 eyes), N = 30 healthy (control)	treatment study	rtx-1		high-dose corticosteroid treatment	29264653	2017	Nakamura T, Hayashi A, Oiwake T	Graefes Arch Clin Exp Ophthalmol.	Recovery of macular cone photoreceptors in Vogt-Koyanagi-Harada disease
Acute exudative polymorphous vitelliform maculopathy (AEPVM)		Photoreceptors	Optical coherence tomography angiography showed normal retinal and choroidal vasculature, whereas adaptive optics scanning laser ophthalmoscopy showed circular focal "target" lesions at the level of the photoreceptors in the area of foveal detachment.	N = 1		SD-OCT, AO-SLO, AF			28520626	2017	Skondra D, Nesper PL, Fawzi AA	Retin Cases Brief Rep	MULTIMODAL IMAGING OF ACUTE EXUDATIVE POLYMORPHOUS VITELLIFORM MACULOPATHY WITH OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY AND ADAPTIVE OPTICS SCANNING LASER OPHTHALMOSCOPY

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Other macular diseases													
Idiopathic central serous chorioretinopathy (CSCR)		Photoreceptors	Adaptive optics imaging revealed a gradual increase in the number of macular cone densities during 12 months in patients with resolved CSC, which was correlated with outer retinal layer thickness and visual acuity in a short term.	N = 12		rtx1, SD-OCT			27255458	2016	Nakamura T, Ueda-Consolvo T, Oiwake T, Hayashi A.	Graefes Arch Clin Exp Ophthalmol	Correlation between outer retinal layer thickness and cone density in patients with resolved central serous chorioretinopathy.
Idiopathic central serous chorioretinopathy (CSCR)		Cone Mosaic	Adaptive optics SLO images showed abnormal cone mosaic patterns and reduced cone densities in eyes with resolved CSC, and these abnormalities were associated with VA loss.	N = 45 eyes of 38 patients		AOSLO			20673590	2010	Ooto S, Hangai M, Sakamoto A, Tsujikawa A, Yamashiro K, Ojima Y, Yamada Y, Mukai H, Oshima S, Inoue T, Yoshimura N.	Ophthalmology	High-resolution imaging of resolved central serous chorioretinopathy using adaptive optics scanning laser ophthalmoscopy.
Idiopathic central serous chorioretinopathy (CSCR)		Inner Retinal Reflectivity	Inner retinal phenotype: Inner Retinal Reflectivity: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane; waxy membrane; vessel associated membrane; striate reflectivity.	N = 3	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Central serous chorioretinopathy		intraretinal hyperreflective clusters	AOSLO allows precise localization of intraretinal structures and detection of features that cannot be seen with SD-OCT alone	N = 5 patients		AO-SLO, SD-OCT			28055101	2017	Vogel RN, Langlo CS, Scoles D, Carroll J, Weinberg DV, Kim JE.	Invest Ophthalmol Vis Sci	High-Resolution Imaging of Intraretinal Structures in Active and Resolved Central Serous Chorioretinopathy
Central serous chorioretinopathy		Cones	Adaptive optics (AO) imaging demonstrated lower density, spacing, and changes in the photoreceptor mosaic pattern, suggesting that CSC may cause damage to cones after clinical recovery.	N = 1	Case Report	OCT, rtx-1			28832731	2017	Meirelles ALB, Rodrigues MW, Guirado AF, Jorge R	Arq Bras Oftalmol.	Photoreceptor assessment using adaptive optics in resolved central serous chorioretinopathy
central serous retinopathy		Photoreceptors	Pathologic alterations in photoreceptor microanatomy underlie residual visual acuity deficits. Observations of missing cones correlated well across all imaging modalities in the fovea and the temporal parafoveal region of missing cones.	N = 1		AO-SLO, D-OCT			29260123	2017	Sun LW, Carroll J, Lujan BJ	Am J Ophthalmol Case Rep.	Photoreceptor disruption and vision loss associated with central serous retinopathy
Chloroquine Retinopathy		Cones	Disrupted cone AO-SLO images were matched with visual field test results and functional deficits were associated with a precise location on the montage, which allowed correlation of histological findings with functional changes determined by HVF.	N = 1		AOSLO (Canon)	Humphrey visual field testing		24505207	2014	Bae EJ, Kim KR, Tsang SH, Park SP, Chang S.	Korean J Ophthalmol	Retinal damage in chloroquine maculopathy, revealed by high resolution imaging: a case report utilizing adaptive optics scanning laser ophthalmoscopy.
Chloroquine Retinopathy		Fundus, Cones	the interdigitation zone could contribute substantially to the reflectance of the cone photoreceptormosaic.The absence of cones on adaptive optics images does not necessarily mean photoreceptor cell death.	N = 1	various diseases in this paper	rtx1			25284764	2015	Jacob J, Paques M, Krivosic V, Dupas B, Couturier A, Kulcsar C, Tadayoni R, Massin P, Gaudric A.	Am J Ophthalmol	Meaning of visualizing retinal cone mosaic on adaptive optics images.
Chloroquine Retinopathy		Cones	In this pilot study, a moderate cone loss was observed as HCQ cumulative doses increased. The early detection of parafoveal cone metric changes may represent the earliest sign of HCQ macular toxicity during screening.	N = 40 eyes of 23 patients		rtx1, SD-OCT, FAF	mf-ERG		25989823	2015	Debellemanière G, Flores M, Tumahai P, Meillat M, Bidaut Garnier M, Delbosc B, Saleh M.	Acta Ophthalmol	Assessment of parafoveal cone density in patients taking hydroxychloroquine in the absence of clinically documented retinal toxicity.
Chloroquine Retinopathy		Cones	AO images showed disruption of the cone photoreceptor mosaic in areas corresponding to HVF 10-2 defects and SD-OCT IS/OS junction abnormalities. Additionally, irregularities in the cone photoreceptor density and mosaic were seen in areas with normal HVF 10-2 and SD-OCT findings.	N = 2		AO-Cam (custom), SD-OCT	Humphrey visual field testing		20126479	2009	Stepien KE, Han DP, Schell J, Godara P, Rha J, Carroll J.	Trans Am Ophthalmol Soc	Spectral-domain optical coherence tomography and adaptive optics may detect hydroxychloroquine retinal toxicity before symptomatic vision loss.
Chloroquine Retinopathy		Cones	Our study agrees with the findings of Debellemanière and associates (2015) and suggests that retinal toxicity could start with decreased cone density in the inferior retina.	N = 38	Letter to the editor	rtx-1, SD-OCT			27805308	2016	Babeau F, Busetto T, Hamel C, Villain M Daien V	Acta Ophthalmol.	Adaptive optics: a tool for screening hydroxychloroquine-induced maculopathy?
Chloroquine Retinopathy		bull's eye lesion, cones	Multimodal imaging illustrated pathology in the area surrounding the NIR bull's eye, characterized by reduced reflectance, wave-guiding cone density and retinal function.	N = 1	case report	OCT, rtx1	mfERG, FC microperimetry		29124422	2017	Chew AL, Sampson DM, Chelva E, Khan JC, Chen FK	Doc Ophthalmol.	Perifoveal interdigitation zone loss in hydroxychloroquine toxicity leads to subclinical bull's eye lesion appearance on near-infrared reflectance imaging

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Macular Telangiectasia		Photoreceptors	Idiopathic macular telangiectasia type- specific differences in the distribution of photoreceptor abnormalities were shown. For both (types 1 and 2), visual impairment was associated with cone damage.	N = 25 eyes, N = 10 normal eyes		AOSLO, SD-OCT, FAF			23465268	2013	Ooto S, Hangai M, Takayama K, Ueda-Arakawa N, Tsujikawa A, Yamashiro K, Oishi A, Hanebuchi M, Yoshimura N.	Am J Ophthalmol	Comparison of cone pathologic changes in idiopathic macular telangiectasia types 1 and 2 using adaptive optics scanning laser ophthalmoscopy.
Macular Telangiectasia		Cones	Adaptive optics showed that the macular cone density was lower than normal even outside the telangiectasia in MacTel 2 lacking intraretinal cavitation, although the ellipsoid zone remained intact on optical coherence tomography	N = 8 patients		rtx1, OCT			26418443	2016	Jacob J, Krivosic V, Paques M, Tadayoni R, Gaudric A.	Retina	CONE DENSITY LOSS ON ADAPTIVE OPTICS IN EARLY MACULAR TELANGIECTASIA TYPE 2.
Macular Telangiectasia		Cones	Visual sensitivity and recovery of cone visibility in areas of apparent focal cone loss suggests that MacTel type 2 lesions with a preserved ELM may contain functioning cones with abnormal scattering and/or waveguiding characteristics.	N = 3 patients		AOSLO, SD-OCT	Microperimetry Function Testing		25587056	2015	Wang Q, Tuten WS, Lujan BJ, Holland J, Bernstein PS, Schwartz SD, Duncan JL, Roorda A.	Invest Ophthalmol Vis Sci	Adaptive optics microperimetry and OCT images show preserved function and recovery of cone visibility in macular telangiectasia type 2 retinal lesions.
Macular Telangiectasia		Fundus, Cones	the interdigitation zone could contribute substantially to the reflectance of the cone photoreceptormosaic.The absence of cones on adaptive optics images does not necessarily mean photoreceptor cell death.	N = 4	various diseases in this paper	rtx1, OCT			25284764	2015	Jacob J, Paques M, Krivosic V, Dupas B, Couturier A, Kulcsar C, Tadayoni R, Massin P, Gaudric A.	Am J Ophthalmol	Meaning of visualizing retinal cone mosaic on adaptive optics images.
Macular Telangiectasia		Photoreceptors	In eyes with MacTel type 2, AO-SLO revealed unique dark regions in the cone mosaic and decreased cone density that was associated with decreased vision, even in areas with normal vasculature, which suggests that this feature represents early neuronal changes involved in the pathogenesis of MacTel type 2.	N = 13 eyes, N = 10 control eyes		AOSLO, SD-OCT, FAF, CBR	fundus-monitoring microperimetry		21642620	2011	Ooto S, Hangai M, Takayama K, Arakawa N, Tsujikawa A, Koizumi H, Oshima S, Yoshimura N.	Invest Ophthalmol Vis Sci	High-resolution photoreceptor imaging in idiopathic macular telangiectasia type 2 using adaptive optics scanning laser ophthalmoscopy.
Macular Telangiectasia		intraretinal crystalline deposits	Significant associations of crystalline deposits were found with a loss of retinal transparency, macular pigment optical density (MPOD) loss, fluorescein leakage, retinal thickness, and a break in the inner segment/outer segment junction line. Associations with environmental risk factors were not found.	N = 203 Crystals, N = 232 No Crystals		AOSLO, SD-OCT, CBR, CF			21839520	2011	Sallo FB, Leung I, Chung M, Wolf-Schnurrbusch UE, Dubra A, Williams DR, Clemons T, Pauleikhoff D, Bird AC, Peto T; MacTel Study Group..	Ophthalmology	Retinal crystals in type 2 idiopathic macular telangiectasia.
Macular Telangiectasia		Photoreceptor Mosaic	AO showed an overall rarefaction and disruption of the continuity of the photoreceptor mosaic within 5° to the fixation point. AO displayed also disappearance of macular cones.	N = 1	brief report	rtx1, OCT			20809908	2011	Massamba N, Querques G, Lamory B, Querques L, Souied E, Soubrane G.	Acta Ophthalmol	In vivo evaluation of photoreceptor mosaic in type 2 idiopathic macular telangiectasia using adaptive optics.
Macular Telangiectasia		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; vessel associated membrane; microcysts;	N = 7	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Macular Telangiectasia		Photoreceptor structure	Clinically available spectral domain OCT, viewed en face or as B-scan, may lead to misinterpretation of photoreceptor anatomy in a variety of diseases and injuries. Split-detector AOSLO revealed substantial populations of photoreceptors in areas of no, low, or ambiguous ellipsoid zone reflectivity with en face OCT and confocal AOSLO.	N = 1	various diseases in this paper	AOSLO, OCT			26166796	2016	Scoles D, Flatter JA, Cooper RF, Langlo CS, Robison S, Neitz M, Weinberg DV, Pennesi ME, Han DP, Dubra A, Carroll J.	Retina	ASSESSING PHOTORECEPTOR STRUCTURE ASSOCIATED WITH ELLIPSOID ZONE DISRUPTIONS VISUALIZED WITH OPTICAL COHERENCE TOMOGRAPHY.
Talc retinopathy		Crystals inside vessels	AO imaging allowed better elucidation of the clumps of the particles that form the talc microembolus with crystals clearly seen impacted inside the retinal vessels and within the surface of the retina. AO imaging also enabled detection of tiny talc particles.	N = 1 patient	case report	rtx1, SD-OCT			27847603	2015	Soliman MK, Sarwar S, Hanout M, Sadiq MA, Agarwal A, Gulati V, Nguyen QD, Sepah YJ.	Int J Retina Vitreous	High-resolution adaptive optics findings in talc retinopathy
acute bilateral foveolitis		Cones	Fundus-referenced vision testing is a useful tool to indicate the presence of cones that may be amenable to recovery or response to experimental therapies despite not being visible on confocal AOSLO or SD-OCT images.	N = 1	Case Report	SD-OCT, AO-SLO, T-SLO	AO-SLO based microperimetry		29057371	2017	Tu JH, Foote KG, Lujan BJ, Ratnam K, Qin J, Gorin MB, Cunningham ET Jr, Tuten WS, Duncan JL, Roorda A	Am J Ophthalmol Case Rep.	Dysflective cones: Visual function and cone reflectivity in long-term follow-up of acute bilateral foveolitis

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Retinitis pigmentosa (RP)													
Retinitis Pigmentosa (RP)	RHO	Photoreceptors	Cone spacing values were significantly different from normal for patients with RP and demonstrated a statistically significant correlation with foveal threshold, BCVA, and mfERG amplitude. Little variation was observed in cone spacing measured during two sessions fewer than 8 days apart.	N = 5 patient eyes, N = 8 healthy eyes	various diseases in this paper	AOSLO			17591900	2007	Duncan JL, Zhang Y, Gandhi J, Nakanishi C, Othman M, Branham KE, Swaroop A, Roorda A.	Invest Ophthalmol Vis Sci	High-resolution imaging with adaptive optics in patients with inherited retinal degeneration.
Retinitis Pigmentosa (RP)	RHO	Cones	Outer retinal layers were significantly thicker in CNTF-treated eyes than in sham-treated eyes. Cone spacing increased in sham-treated eyes than in CNTF-treated eyes, and cone density decreased in sham-treated than in CNTF-treated eyes.	N = 2	various diseases in this paper	AOSLO, SD-OCT	VA, visual field sensitivity, ff-ERG	sustained-release ciliary neurotrophic factor (CNTF)	21087953	2011	Talcott KE, Ratnam K, Sundquist SM, Lucero AS, Lujan BJ, Tao W, Porco TC, Roorda A, Duncan JL.	Invest Ophthalmol Vis Sci	Longitudinal study of cone photoreceptors during retinal degeneration and in response to ciliary neurotrophic factor treatment.
Retinitis Pigmentosa (RP)	RHO	Photoreceptors	Foveal cone density can be decreased in RP and Usher syndrome before visible changes on OCT or a decline in visual function. Thus, AOSLO imaging may allow more sensitive monitoring of disease than current methods.	N = 11	various diseases in this paper	AOSLO, SD-OCT			27145477	2016	Sun LW, Johnson RD, Langlo CS, Cooper RF, Razeen MM, Russillo MC, Dubra A, Connor TB Jr, Han DP, Pennesi ME, Kay CN, Weinberg DV, Stepien KE, Carroll J.	Invest Ophthalmol Vis Sci	Assessing Photoreceptor Structure in Retinitis Pigmentosa and Usher Syndrome.
Retinitis Pigmentosa (RP)	RHO; RPGR; ABCA4	Outer Nuclear Layer, Cones	The ONL thickness and cone density were correlated in normal eyes and eyes with RP, but both were strongly correlated with retinal eccentricity, precluding estimation of cone density from ONL thickness.	N = 12 eyes of 7 patients, N = 20 eyes of 10 subjects		AOSLO, SD-OCT			25515570	2015	Menghini M, Lujan BJ, Zayit-Soudry S, Syed R, Porco TC, Bayabo K, Carroll J, Roorda A, Duncan JL.	Invest Ophthalmol Vis Sci	Correlation of outer nuclear layer thickness with cone density values in patients with retinitis pigmentosa and healthy subjects.
Retinitis Pigmentosa (RP)	RHO	Photoreceptors	AO-SLO imaging revealed noticeable spacing irregularities in the cone mosaic. AO-SLO allows researchers to characterize retinal structural abnormalities with precision so that early structural changes in retinitis pigmentosa can be identified and reconciled with genetic findings.	N = 2 (siblings)		AOSLO (Canon)			25215869	2014	Park SP, Lee W, Bae EJ, Greenstein V, Sin BH, Chang S, Tsang SH.	Ophthalmic Surg Lasers Imaging Retina	Early structural anomalies observed by high-resolution imaging in two related cases of autosomal-dominant retinitis pigmentosa.
Retinitis Pigmentosa (RP)	RHO	Macluar Cones	Cone density is decreased and the regularity of the cone mosaic spatial arrangement is disrupted in eyes with RP, even when visual acuity and foveal sensitivity are good.	N = 14 patients, N = 12 control		AOSLO (Canon)			24260224	2013	Makiyama Y, Ooto S, Hangai M, Takayama K, Uji A, Oishi A, Ogino K, Nakagawa S, Yoshimura N.	PLoS One	Macular cone abnormalities in retinitis pigmentosa with preserved central vision using adaptive optics scanning laser ophthalmoscopy.
Retinitis Pigmentosa (RP)	SNRNP200	Cone Mosaic	Mutations in SNRNP200 caused 1.6% of disease in this adRP cohort. Pathogenic mutations were found primarily in exons 16 and 25, but the novel p.Ala542Val mutation in exon 13 suggests that variation in other genetic regions is also responsible for causing dominant disease.	N = 251 families		AOSLO, OCT			24319334	2013	Bowne SJ, Sullivan LS, Avery CE, Sasser EM, Roorda A, Duncan JL, Wheaton DH, Birch DG, Branham KE, Heckenlively JR, Sieving PA, Daiger SP.	Mol Vis	Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200) cause 1.6% of autosomal dominant retinitis pigmentosa.
Retinitis Pigmentosa (RP)		Cones	Cone density was reduced by up to 62% below normal at or near the fovea in eyes with VA and sensitivity that remained within normal limits.	N = 16	various diseases in this paper	AOSLO			23908179	2013	Ratnam K, Carroll J, Porco TC, Duncan JL, Roorda A.	Invest Ophthalmol Vis Sci	Relationship between foveal cone structure and clinical measures of visual function in patients with inherited retinal degenerations.
Retinitis Pigmentosa (RP)		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane; waxy membrane; vessel associated membrane;	N = 4	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Retinitis Pigmentosa (RP)	FAM161A	Fundus	Loss of outer retinal structures demonstrated with high-resolution retinal imaging suggests FAM161A is important for normal photoreceptor structure and survival.	N = 6 family members (3 affected)		AOSLO, OCT	FF-ERG, Perimetry		25007332	2014	Duncan JL, Biswas P, Kozak I, Navani M, Syed R, Soudry S, Menghini M, Caruso RC, Jeffrey BG, Heckenlively JR, Reddy GB, Lee P, Roorda A, Ayyagari R.	Ophthalmic Genet	Ocular Phenotype of a Family with FAM161A-associated Retinal Degeneration.
Retinitis Pigmentosa (RP)		Cones, Microcystic lesions	Manifestion of dark, partition-like areas in the cone mosaic on AO-SLO images. Microcystic lesions in the INL may affect the images of the cone mosaic.	N = 1	various diseases in this paper	AO-SLO, SD-OCT			28291071	2017	Hasegawa T, Ooto S, Makiyama Y, Hata M, Miyamoto K, Yoshimura N	Retin Cases Brief Rep	CIRCINATE PARTITION-LIKE FINDINGS ON CONE MOSAIC IMAGED BY ADAPTIVE OPTICS SCANNING LASER OPHTHALMOSCOPY IN EYES WITH INNER NUCLEAR LAYER MICROCYSTIC CHANGES
Retinitis Pigmentosa (RP) and Stargardt (ST)		Cones	The developed and presented algorithms do not require spatial regularity in cone packing and are, therefore, useful for counting cones in diseased retinas, as demonstrated for eyes with Stargardt's macular dystrophy and retinitis pigmentosa.	N = 1 RP-patient, N = 1 ST-patient, N = 3 healthy	Testing of cone counting algorithm	AO flood-illum			17429482	2007	Xue B, Choi SS, Doble N, Werner JS.	J Opt Soc Am A Opt Image Sci Vis	Photoreceptor counting and montaging of en-face retinal images from an adaptive optics fundus camera
Usher syndrome type II	RHO	Cones	Outer retinal layers were significantly thicker in CNTF-treated eyes than in sham-treated eyes. Cone spacing increased in sham-treated eyes than in CNTF-treated eyes, and cone density decreased in sham-treated than in CNTF-treated eyes.	N = 1	various diseases in this paper	AOSLO, SD-OCT	VA, visual field sensitivity, ff-ERG	sustained-release ciliary neurotrophic factor (CNTF)	21087953	2011	Talcott KE, Ratnam K, Sundquist SM, Lucero AS, Lujan BJ, Tao W, Porco TC, Roorda A, Duncan JL.	Invest Ophthalmol Vis Sci	Longitudinal study of cone photoreceptors during retinal degeneration and in response to ciliary neurotrophic factor treatment.
Usher syndrome type II		Photoreceptors	Foveal cone density can be decreased in RP and Usher syndrome before visible changes on OCT or a decline in visual function. Thus, AOSLO imaging may allow more sensitive monitoring of disease than current methods.	N = 8	various diseases in this paper	AOSLO, SD-OCT			27145477	2016	Sun LW, Johnson RD, Langlo CS, Cooper RF, Razeen MM, Russillo MC, Dubra A, Connor TB Jr, Han DP, Pennesi ME, Kay CN, Weinberg DV, Stepien KE, Carroll J.	Invest Ophthalmol Vis Sci	Assessing Photoreceptor Structure in Retinitis Pigmentosa and Usher Syndrome.
Usher syndrome type II	CLRN1	Cones	Cones were observed centrally but not in regions with scotomas, and retinal pigment epithelial cells were visible in regions without cones in patients with CLRN1 mutations.	N = 3		AOSLO			22964989	2013	Ratnam K, Västinsalo H, Roorda A, Sankila EM, Duncan JL.	JAMA Ophthalmol	Cone structure in patients with usher syndrome type III and mutations in the Clarin 1 gene.
Usher syndrome type II		Cones	Cone density was reduced by up to 62% below normal at or near the fovea in eyes with VA and sensitivity that remained within normal limits.	N = 6	various diseases in this paper	AOSLO			23908179	2013	Ratnam K, Carroll J, Porco TC, Duncan JL, Roorda A.	Invest Ophthalmol Vis Sci	Relationship between foveal cone structure and clinical measures of visual function in patients with inherited retinal degenerations.
Neuropathy, ataxia, and retinitis pigmentosa	mtDNA m.8993T>C ATPase 6 mutation	Cones	High-resolution retinal and brain imaging in NARPsyndromerevealed analogous patterns of tissue injury characterized by heterogeneous areas of neuronal loss.	N = 5		AOSLO			20953793	2011	Gelfand JM, Duncan JL, Racine CA, Gillum LA, Chin CT, Zhang Y, Zhang Q, Wong LJ, Roorda A, Green AJ.	J Neurol	Heterogeneous patterns of tissue injury in NARP syndrome.
Neuropathy, ataxia, and retinitis pigmentosa	T8993C mutation	Cones	Visual function was better in patients with a contiguous and regular cone mosaic. Patients expressing high levels of the mtDNA T8993C mutation show abnormal cone structure, suggesting normal mitochondrial DNA is necessary for normal waveguiding by cones.	N = 5		AOSLO, OCT	Microperimetry		18997096	2009	Yoon MK, Roorda A, Zhang Y, Nakanishi C, Wong LJ, Zhang Q, Gillum L, Green A, Duncan JL.	Invest Ophthalmol Vis Sci	Adaptive optics scanning laser ophthalmoscopy images in a family with the mitochondrial DNA T8993C mutation.
X-linked Retinitis pigmentosa	RPGR and RP2	Cones	Qualitative and quantitative analyses by AO-SLO imaging revealed a mosaic pattern of cone disruption, even in the absence of visual symptoms, normal visual acuity and normal macular thickness on OCT.	N = 5 patients, N = 18 healthy		CFP, FAF, SD-OCT, AO-SLO (canon)	BCVA, FF-ERG		23443027	2013	Pyo Park S, Hwan Hong I, Tsang SH, Chang S.	Eur J Hum Genet	Cellular imaging demonstrates genetic mosaicism in heterozygous carriers of an X-linked ciliopathy gene

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
(Presumed) monogenic diseases (other than RP)													
Stargardt	ABCA4	Photoreceptors	cone spacing abnormalities were observed in regions of homogeneous AF,	N = 12 pat. / N = 27 controls		AOSLO	FF-ERG		21296825	2011	Chen Y, Ratnam K, Sundquist SM, Lujan B, Ayyagari R, Gudiseva VH, Roorda A, Duncan JL.	Invest Ophthalmol Vis Sci	Cone photoreceptor abnormalities correlate with vision loss in patients with Stargardt disease.
Stargardt	ABCA4	Photoreceptors	AOSLO, OCT, and microperimetry to create a method that conveys structure-function relationships	N = 14.pat. / N = 9		AOSLO, SD-OCT	Microperimetry		26981328	2016	Razeen MM, Cooper RF, Langlo CS, Goldberg MR, Wilk MA, Han DP, Connor TB Jr, Fishman GA, Collison FT, Sulai YN, Dubra A, Carroll J, Stepien KE.	Transl Vis Sci Technol	Correlating Photoreceptor Mosaic Structure to Clinical Findings in Stargardt Disease.
Stargardt	ABCA4	Photoreceptors	adaptive optics scanning light ophthalmoscopy reveals increased cone and rod spacing in areas that appear normal in conventional images	N = 2		AOSLO, SD-OCT			26247787	2015	Song H, Rossi EA, Latchney L, Bessette A, Stone E, Hunter JJ, Williams DR, Chung M.	JAMA Ophthalmol	Cone and rod loss in Stargardt disease revealed by adaptive optics scanning light ophthalmoscopy.
Stargardt	ABCA4	Photoreceptors	desctiption of multimodal imaging findings	N = 1 pat. / N = 2 eyes		AOSLO, SD-OCT			25707054	2015	Pang CE, Suqin Y, Sherman J, Freund KB	Ophthalmic Surg Lasers Imaging Retina	New insights into Stargardt disease with multimodal imaging.
Stargardt	ABCA4	Inner Retinal Reflectivity	Inner retinal phenotype: vessel associated membrane	N = 1	various diseases in this paper	AOSLO, SD-OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Stargardt-like	ELOVL4 or PROM1	whole fundus	cone density loss	N=2		rtx1, SD-OCT, FAF	FF-ERG		26110599	2016	Palejwala NV, Gale MJ, Clark RF, Schlechter C, Weleber RG, Pennesi ME.	Retina	INSIGHTS INTO AUTOSOMAL DOMINANT STARGARDT-LIKE MACULAR DYSTROPHY THROUGH MULTIMODALITY DIAGNOSTIC IMAGING.
Stargardt-like	Peripherin/RDS	Cones	peripherin/RDS mutations produced diffuse AF abnormalities, disruption of the photoreceptor/RPE junction, and increased cone spacing, consistent with cone loss in the macula.	N = 4 patients, N = 27 control		AOSLO, FAF, OCT	ffERG, mfERG, Mircoperimetry		21071739	2011	Duncan JL, Talcott KE, Ratnam K, Sundquist SM, Lucero AS, Day S, Zhang Y, Roorda A.	Invest Ophthalmol Vis Sci	Cone structure in retinal degeneration associated with mutations in the peripherin/RDS gene.
Retinopathy/ Macular degeneration	ABCA4	Cones	In childhood-onset ABCA4-associated retinopathy, the earliest stages of macular atrophy involve the parafovea and spare the foveola. In some cases, these changes are predated by tiny, foveal, yellow, hyperautofluorescent dots. Electroretinography suggests that the initial site of retinal dysfunction may occur after phototransduction.	N = 8 children		AO-SLO, OCT, FAF	VA, ERG		29310964	2018	Khan KN, Kasilian M, Mahroo OAR, Tanna P, Kalitzeos A, Robson AG, Tsunoda K, Iwata T, Moore AT, Fujinami K, Michaelides M	Ophthalmology	Early Patterns of Macular Degeneration in ABCA4-Associated Retinopathy
Pseudoxanthoma elasticum		Photoreceptors	Structural abnormalities at the level of Bruch's membrane, likely a result of calcification, correlate with the characteristic "orange peel" pattern known as peau d'orange.	N = 1		AO-SLO (Apaeros), OCT, FF			28499057	2017	Onishi AC, Nesper PL, Fawzi AA	Ophthalmic Surg Lasers Imaging Retina	Adaptive Optics Scanning Laser Ophthalmoscopy and Multimodal Imaging of Peau D'Orange in Pseudoxanthoma Elasticum
Bietti's crystalline Dystrophy	CYP4V2	Outer Retinal Tubules	reduction in the cone count in all eyes in the area outside the outer retinal tubules (ORT)	N = 5 pat. / N = 10 subj.		rtx1, SD-OCT, FAF	FF-ERG		26915747	2016	Battu R, Akkali MC, Bhanushali D, Srinivasan P, Shetty R, Berendschot TT, Schouten JS, Webers CA.	Eye (Lond)	Adaptive optics imaging of the outer retinal tubules in Bietti's crystalline dystrophy.
Bietti's crystalline Dystrophy	CYP4V2	Cone Mosaic	Reduction in mean cone density centrally	N = 7		AOSLO (Canon), SD-OCT, FAF	FF-ERG, HVF		26521715	2016	Miyata M, Ooto S, Ogino K, Gotoh N, Morooka S, Makiyama Y, Hasegawa T, Sugahara M, Hata M, Yamashiro K, Yoshimura N.	Am J Ophthalmol	Evaluation of Photoreceptors in Bietti Crystalline Dystrophy with CYP4V2 Mutations Using Adaptive Optics Scanning Laser Ophthalmoscopy.
Bietti's crystalline Dystrophy	CYP4V2	Fundus	The clusters of hyperreflective signals in the AO images corresponded to the crystals in the IR images. High-magnification AO images revealed that the clusters of hyperreflective signals consisted of circular spots that are similar to the signals of cone photoreceptors.	N = 3		rtx1, SD-OCT, FAF	MF-ERG, HVF		25276414	2014	Gocho K, Kameya S, Akeo K, Kikuchi S, Usui A, Yamaki K, Hayashi T, Tsuneoka H, Mizota A, Takahashi H.	J Ophthalmol	High-Resolution Imaging of Patients with Bietti Crystalline Dystrophy with CYP4V2 Mutation.
Enhanced S-cone syndrom	NR2E3	Cones	sparse distribution and multiple abnormal clusters within the cone mosaic in ESCS patients	N=3		AOSLO, SD-OCT, FAF	FF-ERG		23604511	2013	Park SP, Hong IH, Tsang SH, Lee W, Horowitz J, Yzer S, Allikmets R, Chang S.	Graefes Arch Clin Exp Ophthalmol	Disruption of the human cone photoreceptor mosaic from a defect in NR2E3 transcription factor function in young adults.
Fundus Albipunctatus	RDH5	subretinal yellow-white spots	decreased cone density revealed by FAOSLO in young patient with normal photopic ERG	N = 1		(F)AOSLO	FF-ERG		24922193	2014	Song H, Latchney L, Williams D, Chung M.	JAMA Ophthalmol	Fluorescence adaptive optics scanning laser ophthalmoscope for detection of reduced cones and hypoautofluorescent spots in fundus albipunctatus.
Fundus Albipunctatus	RDH5	Cones	Macular cone density is lower and the regularity of the macular cone mosaic spatial arrangement is disrupted in eyes with fundus albipunctatus.	N = 10 eyes / N = 11 control eyes		AOSLO (Canon)	FF-ERG, Microperimetry		24246574	2014	Makiyama, 2014	Am J Ophthalmol	Cone abnormalities in fundus albipunctatus associated with RDH5 mutations assessed using adaptive optics scanning laser ophthalmoscopy.
Retinitis punctata albescens	RLBP1	Photoreceptors, dot like deposits	Patients with RPA show variable degrees of foveal cone death, even at an early stage.	N = 11 healthy, N = 11 patients		OCT, FAF, rtx1	FF-ERG		23929416	2013	Dessalces, 2013	JAMA Ophthalmol	Early-onset foveal involvement in retinitis punctata albescens with mutations in RLBP1
Leber's congenital amaurosis		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; waxy membrane; vessel associated membrane;	N = 2	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Bilateral Progressive Maculopathy		RPE	The AOSLO image showed a very patchy cone mosaic, and RPE cells could be seen over much of the macular region but, unlike in the CRD case, they did not form an annular pattern.	N = 1		AOSLO, SD-OCT	Microperimetry		17460294	2007	Roorda A, Zhang Y, Duncan JL.	Invest Ophthalmol Vis Sci	High-resolution in vivo imaging of the RPE mosaic in eyes with retinal disease.
Bilateral Progressive Maculopathy		Cone Mosaic	The AO images revealed significant photoreceptor mosaic heterogeneity.	N = 1		AO-Cam (custom)	MF-ERG, HVF		20142554	2010	Godara P, Rha J, Tait DM, McAllister J, Dubis A, Carroll J, Weinberg DV.	Arch Ophthalmol	Unusual adaptive optics findings in a patient with bilateral maculopathy.

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Albinism		Cone mosaic	Normal cone packing was observed in the absence of a foveal pit, suggesting a pit is not required for packing to occur.	N = 32		AOSLO, SD-OCT			24845642	2014	Wilk MA, McAllister JT, Cooper RF, Dubis AM, Patitucci TN, Summerfelt P, Anderson JL, Stepien KE, Costakos DM, Connor TB Jr, Wirostko WJ, Chiang PW, Dubra A, Curcio CA, Brilliant MH, Summers CG, Carroll J.	Invest Ophthalmol Vis Sci.	Relationship between foveal cone specialization and pit morphology in albinism.
Albinism		Cones	The quantitative analysis of cone density and outer segment elongation demonstrates, that foveal cone specialization is variable in albinism.	N = 6		AO-Cam (custom), SD-OCT			20149815	2010	McAllister JT, Dubis AM, Tait DM, Ostler S, Rha J, Stepien KE, Summers CG, Carroll J.	Vision Res	Arrested development: high-resolution imaging of foveal morphology in albinism.
Albinism		Cones	A foveal pit is not required for foveal cone specialization, anatomically or functionally.	N = 4		custom AO-Cam, OCT, SD-OCT	MF-ERG		18625935	2008	Marmor MF, Choi SS, Zawadzki RJ, Werner JS.	Arch Ophthalmol	Visual insignificance of the foveal pit: reassessment of foveal hypoplasia as fovea plana.
Stationary Night blindness	GRM6	Cone Mosaic	The selective thinning of the inner retinal layers in patients with GRM6 mutations suggests either reduced bipolar or ganglion cell numbers or altered synaptic structure in the inner retina.	N = 3		AOSLO, SD-OCT			22959359	2012	Godara P, Cooper RF, Sergouniotis PI, Diederichs MA, Streb MR, Genead MA, McAnany JJ, Webster AR, Moore AT, Dubis AM, Neitz M, Dubra A, Stone EM, Fishman GA, Han DP, Michaelides M, Carroll J.	Am J Ophthalmol	Assessing retinal structure in complete congenital stationary night blindness and Oguchi disease.
Oguchi's disease	GRK1	Cone Mosaic	The finding that rods, but not cones, change intensity after dark adaptation suggests that fundus changes in Oguchi disease are the result of changes within the rods as opposed to changes at a different retinal locus.	N = 2		AOSLO, SD-OCT			22959359	2012	Godara P, Cooper RF, Sergouniotis PI, Diederichs MA, Streb MR, Genead MA, McAnany JJ, Webster AR, Moore AT, Dubis AM, Neitz M, Dubra A, Stone EM, Fishman GA, Han DP, Michaelides M, Carroll J.	Am J Ophthalmol	Assessing retinal structure in complete congenital stationary night blindness and Oguchi disease.
X-linked juvenile retinoschisis		Cones	AOSLO data revealed that the resolution of cone cell images improves as the foveal schisis decreases in size.	N = 1		AOSLO, SD-OCT	ff-ERG	Acetazolamide (Diamox)	25796216	2015	Zhang L, Reyes R, Lee W, Chen CL, Chan L, Sujirakul T, Chang S, Tsang SH.	Doc Ophthalmol	Rapid resolution of retinoschisis with acetazolamide.
X-linked juvenile retinoschisis	RS1	Fundus	The AO images of the left eye showed spoke wheel retinal folds, and the folds were thinner than those in fundus photographs.	N = 1		rtx1	FF-ERG, MF-ERG		26356828	2015	Akeo K, Kameya S, Gocho K, Kubota D, Yamaki K, Takahashi H.	Case Rep Ophthalmol Med	Detailed Morphological Changes of Foveoschisis in Patient with X-Linked Retinoschisis Detected by SD-OCT and Adaptive Optics Fundus Camera.
X-linked juvenile retinoschisis	RS1	Cone Structures	XLRS revealed increased cone spacing and abnormal packing in the macula	N = 2		AOSLO, SD-OCT	FF-ERG, MF-ERG		22110067	2011	Duncan JL, Ratnam K, Birch DG, Sundquist SM, Lucero AS, Zhang Y, Meltzer M, Smaoui N, Roorda A.	Invest Ophthalmol Vis Sci	Abnormal cone structure in foveal schisis cavities in X-linked retinoschisis from mutations in exon 6 of the RS1 gene.
X-linked juvenile retinoschisis		Inner Retinal Reflectivity	Inner retinal phenotype: nummular (disc-shaped) reflectivity; microcysts; striate reflectivity.	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Choroideremia	REP1	RPE + blood vessels, later: Photorec.	AOSLO in combination with OCT, allows single cell analysis of disease in choroideremia	N = 57 patients / N = 18 carriers		OCT, SLO, AOSLO, SD-OCT, FAF			25190651	2014	Morgan JI, Han G, Klinman E, Maguire WM, Chung DC, Maguire AM, Bennett J.	Invest Ophthalmol Vis Sci	High-resolution adaptive optics retinal imaging of cellular structure in choroideremia.
Choroideremia	REP1	RPE + Photoreceptors	Patchy cone loss was present in two symptomatic carriers. In two affected males, cone mosaics were disrupted with increased cone spacing near the fovea but more normal cone spacing near the edge of atrophy.	N = 5 patients / N = 6 carriers		AOSLO, flood ill. AO, SD-OCT, FAF			23299470	2013	Syed R, Sundquist SM, Ratnam K, Zayit-Soudry S, Zhang Y, Crawford JB, MacDonald IM, Godara P, Rha J, Carroll J, Roorda A, Stepien KE, Duncan JL.	Invest Ophthalmol Vis Sci	High-resolution images of retinal structure in patients with choroideremia.
Choroideremia		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; waxy membrane; vessel associated membrane; striate reflectivity.	N = 8	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Choroideremia		Cones	Cone density was reduced by up to 62% below normal at or near the fovea in eyes with VA and sensitivity that remained within normal limits.	N = 1 patients / N = 3 carrier	various diseases in this paper	AOSLO, SD-OCT			23908179	2013	Ratnam K, Carroll J, Porco TC, Duncan JL, Roorda A.	Invest Ophthalmol Vis Sci	Relationship between foveal cone structure and clinical measures of visual function in patients with inherited retinal degenerations.
Choroideremia	CHM	RPE	The results support a model of choroideremia in which the RPE degenerates before photoreceptors.	N = 12 patients		SD-OCT, AO-SLO			27936069	2016	Sun LW, Johnson RD, Williams V, Summerfelt P, Dubra A, Weinberg DV, Stepien KE, Fishman GA, Carroll J.	PLoS One	Multimodal Imaging of Photoreceptor Structure in Choroideremia
Bornholm eye disease		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Bradyopsia		Photoreceptor mosaic and structure	Adaptive-optics imaging previously demonstrated a sparse mosaic of normal wave-guiding cones remaining at the fovea, with no visible structure outside the central fovea in oligocone trichromacy.	N = 3		AOSLO, SD-OCT			26343007	2015	Strauss RW, Dubis AM, Cooper RF, Ba-Abbad R, Moore AT, Webster AR, Dubra A, Carroll J, Michaelides M.	Am J Ophthalmol	Retinal Architecture in RGS9- and R9AP-Associated Retinal Dysfunction (Bradyopsia).

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Blue-cone monochromatism	OPN1MW/OPN1LW	Cones	Adaptive optics imaging confirmed the existence of inner segments at a spatial density greater than that expected for the residual blue cones.	N = 20		AOSLO, SD-OCT			24067079	2013	ideciyan AV, Hufnagel RB, Carroll J, Sumaroka A, Luo X, Schwartz SB, Dubra A, Land M, Michaelides M, Gardner JC, Hardcastle AJ, Moore AT, Sisk RA, Ahmed ZM, Kohl S, Wissinger B, Jacobson SG.	Hum Gene Ther	Human cone visual pigment deletions spare sufficient photoreceptors to warrant gene therapy.
Blue-cone monochromatism		Cones	Without adaptive optics correction, BCM carriers appeared to have normal visual function, with normal contrast sensitivity and visual resolution, but with AO-correction, visual resolution was significantly worse than normal.	N = 6 female carriers		AOSLO	Microperimetry Functiontesting		23469117	2013	Rossi EA, Achtman RL, Guidon A, Williams DR, Roorda A, Bavelier D, Carroll J.	PLoS One	Visual function and cortical organization in carriers of blue cone monochromacy.
Blue-cone monochromatism		Cones	Imaging of the cone mosaic in four females carrying an L/M array with deletion of the locus control region, resulting in an absence of L/M opsin gene expression (effectively acting as a cone opsin knockout). On average, they had cone mosaics with reduced density and disrupted organization compared to normal trichromats.	N = 4 female carriers		AOSLO, AO Fundus cam	FF-ERG		20638402	2010	Carroll J, Rossi EA, Porter J, Neitz J, Roorda A, Williams DR, Neitz M.	Vision Res	Deletion of the X-linked opsin gene array locus control region (LCR) results in disruption of the cone mosaic.
Blue-cone monochromatism	OPN1MW/OPN1LW	Photoreceptor structure	Clinically available spectral domain OCT, viewed en face or as B-scan, may lead to misinterpretation of photoreceptor anatomy in a variety of diseases and injuries. Split-detector AOSLO revealed substantial populations of photoreceptors in areas of no, low, or ambiguous ellipsoid zone reflectivity with en face OCT and confocal AOSLO.	N = 1	various diseases in this paper	AOSLO, SD-OCT	FF-ERG		26166796	2016	Scoles D, Flatter JA, Cooper RF, Langlo CS, Robison S, Neitz M, Weinberg DV, Pennesi ME, Han DP, Dubra A, Carroll J.	Retina	ASSESSING PHOTORECEPTOR STRUCTURE ASSOCIATED WITH ELLIPSOID ZONE DISRUPTIONS VISUALIZED WITH OPTICAL COHERENCE TOMOGRAPHY.
Complete or incomplete achromatopsia		Cones	The results of the linear mixed regression model analysis demonstrated a strong effect of observer in cone counting in images from patients with ACHM using two different imaging modalities.	N = 7		AOSLO			26427422	2016	Abozaid MA, Langlo CS, Dubis AM, Michaelides M, Tarima S, Carroll J.	Adv Exp Med Biol	Reliability and Repeatability of Cone Density Measurements in Patients with Congenital Achromatopsia.
Complete or incomplete achromatopsia	CNGB3	Photoreceptors	Peak foveal cone density ranged from 7,273 to 53,554 cones/mm2, significantly lower than normal (range, 84,733–234,391 cones/mm2), with the remnant cones being either contiguously or sparsely arranged.	N = 51		AOSLO, SD-OCT			27479814	2016	Langlo CS, Patterson EJ, Higgins BP, Summerfelt P, Razeen MM, Erker LR, Parker M, Collison FT, Fishman GA, Kay CN, Zhang J, Weleber RG, Yang P, Wilson DJ, Pennesi ME, Lam BL, Chiang J, Chulay JD, Dubra A, Hauswirth WW, Carroll J	Invest Ophthalmol Vis Sci	Residual Foveal Cone Structure in CNGB3-Associated Achromatopsia.
Complete or incomplete achromatopsia			Reduced cone density	N = 1		AOSLO, SD-OCT			21833357	2011	Merino D, Duncan JL, Tiruveedhula P, Roorda A.	Biomed Opt Express	Observation of cone and rod photoreceptors in normal subjects and patients using a new generation adaptive optics scanning laser ophthalmoscope.
Complete or incomplete achromatopsia	GNAT2, GNGA3, GNGB3	Cones	All subjects with ACHM had reduced numbers of cone photoreceptors, albeit to a variable degree. In addition, the relative cone reflectivity varied greatly.	N = 11 pat. / N = 7 controls		AOSLO, SD-OCT			25277229	2014	Dubis AM, Cooper RF, Aboshiha J, Langlo CS, Sundaram V, Liu B, Collison F, Fishman GA, Moore AT, Webster AR, Dubra A, Carroll J, Michaelides M.	Invest Ophthalmol Vis Sci	Genotype-dependent variability in residual cone structure in achromatopsia: toward developing metrics for assessing cone health.
Complete or incomplete achromatopsia	GNAT2 (c.730_743del)	Cones	Fundus shows no specific abnormalities, AO imaging shows a clearly defined but reduced (15-30%) mosaic.	N = 1 patient, N = 10 healthy	case report	rtx1, OCT	ERG		27718025	2016	Ueno S, Nakanishi A, Kominami T, Ito Y, Hayashi T, Yoshitake K, Kawamura Y, Tsunoda K, Iwata T, Terasaki H.	Jpn J Ophthalmol	In vivo imaging of a cone mosaic in a patient with achromatopsia associated with a GNAT2 variant
Complete or incomplete achromatopsia	congenital	Photoreceptors	In all cases, the mosaic was significantly disrupted from normal (Fig. 3).	N = 12		AOSLO, AO-Cam(custom), FF-ERG, SD-OCT	Microperimetry		21778272	2011	Genead MA, Fishman GA, Rha J, Dubis AM, Bonci DM, Dubra A, Stone EM, Neitz M, Carroll J.	Invest Ophthalmol Vis Sci	Photoreceptor structure and function in patients with congenital achromatopsia.
Complete or incomplete achromatopsia		Photoreceptors	A substantial number of foveal and parafoveal cone photoreceptors with apparently intact inner segments were identified in patients with the inherited disease achromatopsia.	N = 4 pat. / N = 2 controls	Method introduction: split detection	AOSLO, SD-OCT			24906859	2014	Scoles D, Sulai YN, Langlo CS, Fishman GA, Curcio CA, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	In vivo imaging of human cone photoreceptor inner segments.
Complete or incomplete achromatopsia		Cones	Retinal images revealed a severely disrupted photoreceptor mosaic in the fovea and parafovea, where the size and density of the visible photoreceptors.resembled that of normal rods.	N = 1 pat. / N = 1 carrier		AO Fundus-Cam			18499214	2008	Carroll J, Choi SS, Williams DR.	Vision Res	In vivo imaging of the photoreceptor mosaic of a rod monochromat.
Complete or incomplete achromatopsia		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane;	N = 5	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Complete or incomplete achromatopsia	RHO F45L, CNGA3	Cones	The RHO F45L allele is not pathogenic in this large family; hence, the two ACHM patients would unlikely develop RP in the future.	N = 2 ACHM patients / N = 24 familiy members		AOSLO			24049715	2013	Vincent AL, Carroll J, Fishman GA, Sauer A, Sharp D, Summerfelt P, Williams V, Dubis AM, Kohl S, Wong F.	Transl Vis Sci Technol	Rhodopsin F45L Allele Does Not Cause Autosomal Dominant Retinitis Pigmentosa in a Large Caucasian Family.
Complete or incomplete achromatopsia		Cones	Cone mosaics were present in the central fovea in the sibling with incomplete ACHM. This may explain the better visual acuity and color vision in this sibling.	N = 1 (complete), N = 1 (incomplete)	Siblings study	SD-OCT, rtx-1, FAF	microperimetry (MP-1)		28197754	2017	Ueno S, Nakanishi A, Sayo A, Kominami T, Ito Y, Hayashi T, Tsunoda K, Iwata T, Terasaki H.	Doc Ophthalmol	Differences in ocular findings in two siblings: one with complete and other with incomplete achromatopsia
Complete or incomplete achromatopsia	CNGB3	Foveal Cones	Foveal cone structure showed little or no change in subjects with CNGB3-associated achromatopsia. In the observed time interval, achromatopsia seems to be a structurally stable condition, although longer-term follow-up is needed.	N = 41 patients	Longitudinal Study: Observation Time = 6 to 26 months	OCT, AO-SLO			28145975	2017	Langlo CS, Erker LR, Parker M, Patterson EJ, Higgins BP, Summerfelt P, Razeen MM, Collison FT, Fishman GA, Kay CN, Zhang J, Weleber RG, Yang P, Pennesi ME, Lam BL, Chulay JD, Dubra A, Hauswirth WW, Wilson DJ, Carroll J	Retina	REPEATABILITY AND LONGITUDINAL ASSESSMENT OF FOVEAL CONE STRUCTURE IN CNGB3-ASSOCIATED ACHROMATOPSIA

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Oligocone trichromacy		Cones	Peripherally the IS/OS layer decreased in intensity, and the RPE1 layer was no longer discernable, in keeping with the lack of cone structure observed on AO imaging outside the central fovea.	N = 4		AO-Cam (custom), SD-OCT, CFP			21436275	2011	Michaelides M, Rha J, Dees EW, Baraas RC, Wagner-Schuman ML, Mollon JD, Dubis AM, Andersen MK, Rosenberg T, Larsen M, Moore AT, Carroll J.	Invest Ophthalmol Vis Sci	Integrity of the cone photoreceptor mosaic in oligocone trichromacy.
Red/green color vision defect		Photoreceptors	Near the fovea, the cone mosaic was disrupted compared to normal, with only a sparse population of strongly waveguiding cones remaining (Figure 4A,B). In the parafoveal image (2 deg), normal-appearing rods were observed dispersed amongst a reduced number of cones with severely diminished wave guiding compared to normal (Figure 4C,D).	N = 1		AO-Cam (custom), SD-OCT	Color vision test		23337435	2013	McClements M, Davies WI, Michaelides M, Carroll J, Rha J, Mollon JD, Neitz M, MacLaren RE, Moore AT, Hunt DM.	Vision Res	X-linked cone dystrophy and colour vision deficiency arising from a missense mutation in a hybrid L/M cone opsin gene.
Red/green color vision defect		Cone Mosaic	While disruptions in retinal lamination and cone mosaic structure were observed in all subjects, genotype-specific differences were also observed.	N = 11		AOSLO, SD-OCT	FF-ERG		23139274	2012	Carroll J, Dubra A, Gardner JC, Mizrahi-Meissonnier L, Cooper RF, Dubis AM, Nordgren R, Genead M, Connor TB Jr, Stepien KE, Sharon D, Hunt DM, Banin E, Hardcastle AJ, Moore AT, Williams DR, Fishman G, Neitz J, Neitz M, Michaelides M.	Invest Ophthalmol Vis Sci	The effect of cone opsin mutations on retinal structure and the integrity of the photoreceptor mosaic.
Red/green color vision defect		Cone Mosaic	No significant difference in cone density between normal trichomats and multiple or single gene dichromats.	N = 10 deutans / N = 27 controls		AOSLO, SD-OCT	Color vision test		20854834	2010	Wagner-Schuman M, Neitz J, Rha J, Williams DR, Neitz M, Carroll J.	Vision Res	Color-deficient cone mosaics associated with Xq28 opsin mutations: a stop codon versus gene deletions.
Red/green color vision defect		Cones	Frequency-of-seeing curves were measured with 0.75′ and 7.5′ spots.	N = 1 protoanomalous trichromacy / N = 1 deuteranopia / N = 5 controls		AO-Fundus Cam	Microperimetry		16936137	2006	Makous W, Carroll J, Wolfing JI, Lin J, Christie N, Williams DR.	Invest Ophthalmol Vis Sci	Retinal microscotomas revealed with adaptive-optics microflashes.
Red/green color vision defect		Cones	The loss of one-third of the cones does not impair any aspect of vision other than color.	N = 2 dichromates / N = 1 trichromate		AO Fundus-Cam	FF-ERG		15148406	2004	Carroll J, Neitz M, Hofer H, Neitz J, Williams DR.	Proc Natl Acad Sci U S A	Functional photoreceptor loss revealed with adaptive optics: an alternate cause of color blindness.
Red/green color vision defect		Cones	Cone density was significantly reduced compared to normal and color-deficient controls, accompanying disruption in the cone mosaic in both individuals, and thinning of the outer nuclear layer.	N = 2		AO Fundus-Cam			19934058	2009	Carroll J, Baraas RC, Wagner-Schuman M, Rha J, Siebe CA, Sloan C, Tait DM, Thompson S, Morgan JI, Neitz J, Williams DR, Foster DH, Neitz M.	Proc Natl Acad Sci U S A	Cone photoreceptor mosaic disruption associated with Cys203Arg mutation in the M-cone opsin.
Red/green color vision defect	novel M-opsin sequence in transmembrane IV "LIAVA"	Cones	The static nature of the cone mosaic disruption combined with the normal lamination on SD-OCT suggests that the affected cones are likely still present.	N = 1 patient		SD-OCT, AO-SLO			20238030	2010	Rha J, Dubis AM, Wagner-Schuman M, Tait DM, Godara P, Schroeder B, Stepien K, Carroll J.	Adv Exp Med Biol	Spectral domain optical coherence tomography and adaptive optics: imaging photoreceptor layer morphology to interpret preclinical phenotypes
S-cone dystrophy		Cone Mosaic		N = 2 tritan		AO-Fundus Cam			17429491	2007	Baraas RC, Carroll J, Gunther KL, Chung M, Williams DR, Foster DH, Neitz M.	J Opt Soc Am A Opt Image Sci Vis	Adaptive optics retinal imaging reveals S-cone dystrophy in tritan color-vision deficiency.
Color vision defect, unspecified		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity;	N = 1	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Cone Rod Dystrophy	GUCY2D	Photoreceptor structure	Clinically available spectral domain OCT, viewed en face or as B-scan, may lead to misinterpretation of photoreceptor anatomy in a variety of diseases and injuries. Split-detector AOSLO revealed substantial populations of photoreceptors in areas of no, low, or ambiguous ellipsoid zone reflectivity with en face OCT and confocal AOSLO.	N = 1	various diseases in this paper	AOSLO, OCT			26166796	2016	Scoles D, Flatter JA, Cooper RF, Langlo CS, Robison S, Neitz M, Weinberg DV, Pennesi ME, Han DP, Dubra A, Carroll J.	Retina	ASSESSING PHOTORECEPTOR STRUCTURE ASSOCIATED WITH ELLIPSOID ZONE DISRUPTIONS VISUALIZED WITH OPTICAL COHERENCE TOMOGRAPHY.
Cone Rod Dystrophy		Cones	Central vision parameters progressively worsen in CDSR. Structural retinal and lipofuscin accumulation abnor- malities are commonly present. Macular cone photoreceptor mosaic is markedly disrupted early in the disease.	N = 7		AOSLO (Physical Sciences), SD-OCT, FAF			23221069	2013	Vincent A, Wright T, Garcia-Sanchez Y, Kisilak M, Campbell M, Westall C, Héon E.	Invest Ophthalmol Vis Sci	Phenotypic characteristics including in vivo cone photoreceptor mosaic in KCNV2-related "cone dystrophy with supernormal rod electroretinogram".
Cone Rod Dystrophy		Photoreceptors	Cone spacing values were significantly different from normal for patients with CRD and demonstrated a statistically significant correlation with foveal threshold, BCVA, and mfERG amplitude. Cone spacing increased in all CRD patients, even those with early disease. Little variation was observed in cone spacing measured during two sessions fewer than 8 days apart.	N = 3 patient eyes, N = 8 healthy eyes	various diseases in this paper	AOSLO	mfERG Function Testing		17591900	2007	Duncan JL, Zhang Y, Gandhi J, Nakanishi C, Othman M, Branham KE, Swaroop A, Roorda A.	Invest Ophthalmol Vis Sci	High-resolution imaging with adaptive optics in patients with inherited retinal degeneration.
Cone Rod Dystrophy			Large areas devoid of wave-guiding cones within atrophic regions. In these areas the cones were abnormally large, resulting in a 6.6-fold reduction from the normal peak cone density. Multifocal electroretinography confirmed a 5.5-fold reduction in amplitude of the central peak	N = 1		AO-flood illum, AOSLO, FAF, OCT	ERG, mfERG		16650474	2006	Wolfing JI, Chung M, Carroll J, Roorda A, Williams DR.	Ophthalmology	High-resolution retinal imaging of cone-rod dystrophy.
Cone Rod Dystrophy		Cones	A peripheral cone dystrophy, diagnosed by full-field ERGs and perimetry, is due to a reduction in the density of parafoveal and peripheral cones.	N = 1		rtx1, SS/SD-OCT	ffERG Function Testing		25708979	2015	Ito N, Kameya S, Gocho K, Hayashi T, Kikuchi S, Katagiri S, Gekka T, Yamaki K, Takahashi H, Tsuneoka H.	Doc Ophthalmol	Multimodal imaging of a case of peripheral cone dystrophy.

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Cone Rod Dystrophy	RPGR	RPE	The cone mosaic appeared patchy and nonuniform and RPE cell structure was visible in some small regions	N = 2		AOSLO			17460294	2007	Roorda A, Zhang Y, Duncan JL.	Invest Ophthalmol Vis Sci	High-resolution in vivo imaging of the RPE mosaic in eyes with retinal disease.
RPGR-associated retinopathy	RPGR	tapetal-like reflex areas	The photoreceptor mosaic in RPGR carriers with a TLR showed reduced cone densities, increased cone inner segment diameters, and increased rod outer segment reflectivity.	N = 9 patients, N = 3 healthy		AO-SLO, SD-OCT, FAF, NIR			29190250	2017	Kalitzeos A, Samra R, Kasilian M, Tee JJL, Strampe M, Langlo C, Webster AR, Dubra A, Carroll J, Michaelides M	Retina	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY
Cone Rod Dystrophy		Photoreceptors	In the eye with macular dystrophy, a relatively uniform photoreceptor mosaic was observed around the fi xation point, whereas presumed debris of photoreceptor degradation was observed in the other bull's eye retinal lesion.	N = 1 patient, N = 2		custom AO-Cam, SLO, CF			18991039	2008	Bessho K, Fujikado T, Mihashi T, Yamaguchi T, Nakazawa N, Tano Y.	Jpn J Ophthalmol	Photoreceptor images of normal eyes and of eyes with macular dystrophy obtained in vivo with an adaptive optics fundus camera.
Cone Rod Dystrophy		Cones	In all images of diseased retinas, there were extensive areas of dark space between groups of photoreceptors, where no cone photoreceptors were evident. These irregular features were not seen in healthy retinas, but were apparent in patients with retinal dystrophy. There were significant correlations between functional vision losses and the extent to which these irregularities, quantified by cone density, occurred in retinal images.	N = 5 patients, N = 3 control		AO Fundus	mfERG Function Testing		16639019	2006	Choi SS, Doble N, Hardy JL, Jones SM, Keltner JL, Olivier SS, Werner JS.	Invest Ophthalmol Vis Sci	In vivo imaging of the photoreceptor mosaic in retinal dystrophies and correlations with visual function.
Cone Rod Dystrophy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; granular membrane; waxy membrane; striate reflectivity.	N = 4		AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Cone Rod Dystrophy		Cone	Macular cone spacing measures were correlated between observers, visits, and fellow eyes of the same subject in normal eyes and in eyes with IRD.	N = 20 patients, N = 10 control		AOSLO			26416092	2015	Zayit-Soudry S, Sippl-Swezey N, Porco TC, Lynch SK, Syed R, Ratnam K, Menghini M, Roorda AJ, Duncan JL.	Invest Ophthalmol Vis Sci	Repeatability of Cone Spacing Measures in Eyes With Inherited Retinal Degenerations.
Cone Rod Dystrophy	CDHR1	Cones	High-resolution retinal imaging revealed outer retinal changes suggesting that CDHR1 is important for normal photoreceptor structure and survival.	N = 8 family members		AOSLO			23044944	2012	Duncan JL, Roorda A, Navani M, Vishweswaraiah S, Syed R, Soudry S, Ratnam K, Gudiseva HV, Lee P, Gaasterland T, Ayyagari R.	Arch Ophthalmol	Identification of a novel mutation in the CDHR1 gene in a family with recessive retinal degeneration.
cone-rod dystrophy (autosomal-dominant)	GUCA1A	Cones	A mutation in GUCA1A does not lead to the same degree of AD-CRD in all patients. Modifying factors may mitigate or augment disease severity, leading to different retinal cellular phenotypes.	N = 9 (affected), N = 3 (unaffectd)		SD-OCT,AO-SLO	VA, Goldman perimetry, ERG		29074494	2017	Song H, Rossi EA, Stone E, Latchney L, Williams D, Dubra A, Chung M	Br J Ophthalmol.	Phenotypic diversity in autosomal-dominant cone-rod dystrophy elucidated by adaptive optics retinal imaging
Best vitelliform dystrophy		Photoreceptors (Macular)	Combined confocal and nonconfocal split-detector AOSLO imaging reveals substantial variability within clinical lesions in all stages of BVMD.	N = 5		AOSLO			27467379	2016	Scoles D, Sulai YN, Cooper RF, Higgins BP, Johnson RD, Carroll J, Dubra A, Stepien KE.	Retina	PHOTORECEPTOR INNER SEGMENT MORPHOLOGY IN BEST VITELLIFORM MACULAR DYSTROPHY.
Best vitelliform dystrophy		Photoreceptor structure	Findings indicate that substantial photoreceptor structure persists within active lesions, accounting for good visual acuity in these patients. Despite previous reports of diffuse photoreceptor outer segment abnormalities in BVMD, this study reveals normal photoreceptor structure in areas adjacent to clinical lesions.	N = 4 (same family)		AOSLO, SD-OCT			23765342	2013	Kay DB, Land ME, Cooper RF, Dubis AM, Godara P, Dubra A, Carroll J, Stepien KE.	JAMA Ophthalmol	Outer retinal structure in best vitelliform macular dystrophy.
Best vitelliform dystrophy		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; microcysts; striate reflectivity.	N = 2	various diseases in this paper	AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Occult macular dystrophy	RP1L1	Cones	A sparse array of cone photoreceptors with significantly reduced density of the macula is one of the morphologic features of OMD.	N = 22 eyes of 11 patients		rtx1, SD-OCT			26544792	2015	Nakanishi A, Ueno S, Kawano K, Ito Y, Kominami T, Yasuda S, Kondo M, Tsunoda K, Iwata T, Terasaki H.	Invest Ophthalmol Vis Sci	Pathologic Changes of Cone Photoreceptors in Eyes With Occult Macular Dystrophy.
Occult macular dystrophy	RP1L1	Cones	The quantitative assessment of photoreceptor survival or loss, based on analysis of adaptive optics retinal images, was valuable to monitor disease progression at a cellular level.	N = 5 (same family)		rtx1, SD-OCT	mfERG Function Testing		25908487	2015	Ziccardi L, Giannini D, Lombardo G, Serrao S, Dell'Omo R, Nicoletti A, Bertelli M, Lombardo M.	Am J Ophthalmol	Multimodal Approach to Monitoring and Investigating Cone Structure and Function in an Inherited Macular Dystrophy.
Occult macular dystrophy		Cones	Cone densities in the macula of the OMD patient were greatly decreased.	N = 1		rtx1, SD-OCT	mfERG Function Testing		23696695	2013	Tojo N, Nakamura T, Ozaki H, Oka M, Oiwake T, Hayashi A.	Clin Ophthalmol	Analysis of macular cone photoreceptors in a case of occult macular dystrophy.
Occult macular dystrophy		Cones				AO fundus Cam, OCT			21468344	2011	Kitaguchi Y, Kusaka S, Yamaguchi T, Mihashi T, Fujikado T.	Clin Ophthalmol	Detection of photoreceptor disruption by adaptive optics fundus imaging and Fourier-domain optical coherence tomography in eyes with occult macular dystrophy.
Occult macular dystrophy		Photoreceptors	This here reported close topographical correspondence between the functional and structurally damaged retina is important because it might help to differentiate OMD from other macular dystrophies such as Stargardt disease, in which this correlation might be absent.	N = 1		SD-OCT, rtx1,	mfERG, ffERG, microperimetry		28591286	2017	Viana KÍ, Messias A, Siqueira RC, Rodrigues MW, Jorge R	Arq Bras Oftalmol	Structure-functional correlation using adaptive optics, OCT, and microperimetry in a case of occult macular dystrophy
Central Areolar Choroidal Dystrophy	PRPH2	Cones	Parafoveal cone photoreceptors can be affected even at the early stage of CACD.	N = 5					27977834	2016	Gocho K, Akeo K, Itoh N, Kameya S, Hayashi T, Katagiri S, Gekka T, Ohkuma Y, Tsuneoka H, Takahashi H.	Ophthalmic Surg Lasers Imaging Retina	High-Resolution Adaptive Optics Retinal Image Analysis at Early Stage Central Areolar Choroidal Dystrophy With PRPH2 Mutation
Cone Dystrophy	POC1B	Cones	The cone dystrophy associated with POC1B variants has features similar to achromatopsia, and genetic analyses is useful in discriminating these two diseases.	N = 1		rtx-1, SD-OCT, FAF	Ishihara Color Vision test, Standard Pseudoisochromatic Plates-2, Farnsworth dichromatic test, FF-ERG		29220607	2017	Kominami A, Ueno S, Kominami T, Nakanishi A, Ito Y, Fujinami K, Tsunoda K, Hayashi T, Kikuchi S, Kameya S, Iwata T, Terasaki H	Ophthalmic Genet.	Case of Cone Dystrophy with Normal Fundus Appearance Associated with Biallelic POC1B Variants
Hereditary retinal degeneration		parafoveal cones	Significant correlation between Contrast Sensitivity and parafoveal cone density	N = 15 healthy, N = 9 patients		AOSLO			28195612	2017	Hirota M, Morimoto T, Kanda H, Lohmann TK, Miyagawa S, Endo T, Miyoshi T, Fujikado T.	Ophthalmic Surg Lasers Imaging Retina	Relationships Between Spatial Contrast Sensitivity and Parafoveal Cone Density in Normal Subjects and Patients With Retinal Degeneration

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Miscellaneous													
Gunn's dots		Gunn's dots	Gunn's dpts_ The mean (±SD) diameter of Gunn's dots was 13.3 µm (±3.5). Their density peaked at ~120 per square millimeter and decreased with age to become barely detectable after 50 years.	N = 18		rtx1 + SLO			25077537	2015	Paques M, Miloudi C, Kulcsar C, Leseigneur A, Chaumette C, Koch E.	Retina	High-resolution imaging of gunn's dots.
Cone density in healthy eyes		Cones	Cone density at 2, 3, 5, 7° in line with histology data	N=10		rtx1, SD-OCT			24729030	2014	Muthiah MN, Gias C, Chen FK, Zhong J, McClelland Z, Sallo FB, Peto T, Coffey PJ, da Cruz L	Br J Ophthalmol	Cone photoreceptor definition on adaptive optics retinal imaging.
Lensing effect of microcysts		Photoreceptors	Lensing effect: cones underlying the microcyst appeared more tightly packed in the AOSLO images (average nearest neighbor spacing of 4.40 µm) than those immediately adjacent to the microcyst (average nearest neighbor spacing of 5.82 µm, p<0.0001)	N=1		AOSLO			23974999	2014	Langlo CS, Flatter JA, Dubra A, Wirostko WJ, Carroll J.	Retina	A lensing effect of inner retinal cysts on images of the photoreceptor mosaic.
Pathologic myopia		Inner Retinal Reflectivity	Inner retinal phenotype: waxy membrane	N = 1		AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Astrocytic hamartoma		Inner Retinal Reflectivity	Inner retinal phenotype: granular membrane; vessel associated membrane; striate reflectivity.	N = 1		AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Ocular Siderosis		Fundus	AO revealed an arterial tropism with a decrease in the amount of particles overtime, which may be consistent with macrophagic activity	N = 1		rtx1	ffERG Function Testing		24337723	2014	Faure C, Gocho K, Le Mer Y, Sahel JA, Paques M, Audo I.	Doc Ophthalmol	Functional and high resolution retinal imaging assessment in a case of ocular siderosis.
Laser injury		Photoreceptors	Very small, localized photoreceptor disruptions can be detected in patients with minimal titanium:sapphire laser injury by cross-sectional imaging using OCT, but their extent was delineated more precisely by en face AO imaging.	N = 2 pat. / N = 4 eyes		custom AO fundus, OCT			19327747	2009	Kitaguchi Y, Fujikado T, Kusaka S, Yamaguchi T, Mihashi T, Tano Y.	Am J Ophthalmol	Imaging of titanium:sapphire laser retinal injury by adaptive optics fundus imaging and Fourier-domain optical coherence tomography.
Solar retinopathy		Microvasculature, Hard exudates	The high resolution of the AOSLO allowed the detection of these early vascular changes induced by diabetes.	N = 1		AOSLO			17265801	2006	Roorda A, Garcia CA, Martin JA, Poonja S, Queener H, Romero-Borja F, Sepulveda R, Venkateswaran K, Zhang Y.	Bull Soc Belge Ophtalmol	What can adaptive optics do for a scanning laser ophthalmoscope ?
solar retinopathy		Cones	AO cone density map shows heterogeneous disruption of the cone mosaic with density reduction. 24 months later AO revealed incomplete recovery of the cone mosaic, with persistent loss at the level of the solar retinopathy lesion	N = 1		rtx1, OCT, CFP			27958216	2016	Lo Giudice G, Catania AG, Galan A.	Indian J Ophthalmol	Adaptive optics study of photoreceptors layer damage from presumed sun exposure: A case report
Acute solar retinopathy		Cones	The shape of the lesion on adaptive optics and en face OCT images of the left eye corresponded to the shape of the scotoma drawn by the patient on Amsler grid. Acute solar retinopathy can present with foveal cone photoreceptor mosaic disturbances on AO-SLO imaging. Corresponding reflectivity changes can be seen on en face OCT, especially in the middle and outer retina.	N = 1		AO-SLO, SD-OCT, CF	microperimetry		29222532	2018	Wu CY, Jansen ME, Andrade J, Chui TYP, Do AT, Rosen RB, Deobhakta A	JAMA Ophthalmol.	Acute Solar Retinopathy Imaged With Adaptive Optics, Optical Coherence Tomography Angiography, and En Face Optical Coherence Tomography
Idiopathic epiretinal membrane		Photoreceptors	The presence of microfolds was associated with metamorphopsia	N = 24 pat. / N = 25 eyes		AOSLO, SD-OCT			21074858	2011	Ooto S, Hangai M, Takayama K, Sakamoto A, Tsujikawa A, Oshima S, Inoue T, Yoshimura N.	Ophthalmology	High-resolution imaging of the photoreceptor layer in epiretinal membrane using adaptive optics scanning laser ophthalmoscopy.
Idiopathic epiretinal membrane		vitreomacular interface	After surgery, the morphology of the vitreomacular interface changed compared with the preoperative state.	N = 6		rtx1, SD-OCT			26110598	2016	Lombardo M, Scarinci F, Giannini D, Pileri M, Ripandelli G, Stirpe M, Lombardo G, Serrao S.	Retina	HIGH-RESOLUTION MULTIMODAL IMAGING AFTER IDIOPATHIC EPIRETINAL MEMBRANE SURGERY.
Idiopathic epiretinal membrane		ERM	Adaptive optics retinal images in eyes with ERM showed multiple abnormalities of the inner retinal interface. Various features were identified, including macrofolds, microfolds, and hyperreflective structures.	N = 6 / N = 6 controls		rtx1, SD-OCT			23823508	2013	Lombardo M, Scarinci F, Ripandelli G, Cupo G, Stirpe M, Serrao S	Ophthalmology	Adaptive optics imaging of idiopathic epiretinal membranes.
Microscotoma		Inner Retinal Reflectivity	Inner retinal phenotype: punctate reflectivity; nummular (disc-shaped) reflectivity; granular membrane;	N = 3		AOSLO, OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Macular hole		Photoreceptors	Post MH-repair: Photoreceptor disruption exists even after apparent MH closure.	N = 4		AOSLO, SD-OCT			25525907	2015	Hansen S, Batson S, Weinlander KM, Cooper RF, Scoles DH, Karth PA, Weinberg DV, Dubra A, Kim JE, Carroll J, Wirostko WJ.	Retin Cases Brief Rep	Assessing photoreceptor structure after macular hole closure.
Macular hole		Foveal Cone Loss	Macular hole (MH) after surgery: Cone loss ratio in the foveola correlated with postoperative visual acuity	N = 18 pat. / N = 19 eyes) and N = 10 normal		AOSLO, SD-OCT			23717484	2013	Yokota S, Ooto S, Hangai M, Takayama K, Ueda-Arakawa N, Yoshihara Y, Hanebuchi M, Yoshimura N.	PLoS One	Objective assessment of foveal cone loss ratio in surgically closed macular holes using adaptive optics scanning laser ophthalmoscopy.
Macular hole		Photoreceptors	Post MH-repair: Structural damage to the photoreceptor layer correlated with greater decreases in visual function in eyes with surgically closed MH.	N = 19 pat. / N = 21 eyes)		AOSLO, SD-OCT			22534108	2012	Ooto S, Hangai M, Takayama K, Ueda-Arakawa N, Hanebuchi M, Yoshimura N.	Am J Ophthalmol	Photoreceptor damage and foveal sensitivity in surgically closed macular holes: an adaptive optics scanning laser ophthalmoscopy study.
Macular hole					Couldn't access this paper				24703636	2014	Debellemanière G, Koehl A, Delbosc B, Saleh M.	J Fr Ophtalmol	[High-resolution retinal imaging using adaptive optics of a full thickness macular hole].
Retinal detachment surgery		Cone (loss)	Cone density post RD-surgery in comparision to partner eye: The parafoveal cone density was decreased in eyes operated for RD (mean ± SD 14,576 ± 4035/mm(2)) compared with fellow eyes (20,589 ± 2350/mm(2)) (p=0.0001)	N=21 patients (42 eyes)		rtx1, SD-OCT			25237163	2014	Saleh M, Debellemanière G, Meillat M, Tumahai P, Bidaut Garnier M, Flores M, Schwartz C, Delbosc B.	Br J Ophthalmol	Quantification of cone loss after surgery for retinal detachment involving the macula using adaptive optics.

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Scleral Buckling Surgery for Rhegmatogenous Retinal Detachment		Photoreceptors	Recovery of cone packing density measured by AO was associated with structural recovery of the outer retina observed in OCT, suggesting regeneration of the photoreceptor outer segment after surgery.	N = 21 patients	Photoreceptor regeneration, Observation Time = 12 months	AO-Fundus, OCT		Treatment study: Scleral Buckling Surgery	28189482	2017	Ra E, Ito Y, Kawano K, Iwase T, Kaneko H, Ueno S, Yasuda S, Kataoka K, Terasaki H.	Am J Ophthalmol	Regeneration of Photoreceptor Outer Segments after Scleral Buckling Surgery for Rhegmatogenous Retinal Detachment
Macular Microholes		Photoreceptors	Macular microholes: Cone disruption occurs in eyes with macular microholes and a larger cone disruption area translates into a poorer visual acuity.	N = 12 pat. / N = 14 eyes		AOSLO			25146990	2014	Ooto S, Hangai M, Takayama K, Ueda-Arakawa N, Makiyama Y, Hanebuchi M, Yoshimura N.	Invest Ophthalmol Vis Sci	High-resolution imaging of photoreceptors in macular microholes.
Macular Microholes		Photoreceptors	AO images indicated the absence of the cone mosaic in the foveal zone in all 3 cases	N = 3		custom AO fundus, OCT			18486223	2008	Kitaguchi Y, Fujikado T, Bessho K, Sakaguchi H, Gomi F, Yamaguchi T, Nakazawa N, Mihashi T, Tano Y.	Ophthalmology	Adaptive optics fundus camera to examine localized changes in the photoreceptor layer of the fovea.
Macular lesions		RPE	AO showed a well-preserved cone mosaic, suggesting that the abnormality was localized under the photoreceptor layers. Macular RPE hypopigmentation should be considered in case of an isolated macular lesion without functional visual impairment or anatomical defect on SD-OCT	N = 2 (unilateral), N = 1 (bilateral)	brief report (no AO images shown)	SD-OCT, NIR & SW AF, FA, OCT-A, rtx1	VA, microperimetry		28593392	2017	Boulanger-Scemama E, Akesbi J, Tick S, Mohand-Said S, Sahel JA, Audo I	Doc Ophthalmol	Multimodal imaging and functional correlations identify unusual cases of macular retinal pigment epithelium hypopigmentation occurring without functional loss
Blunt ocular trauma		Inner Retinal Reflectivity	Phenotype of the inner retina post trauma: Punctate reflectivity; nummular (disc-shaped) reflectivity; microcysts; striate reflectivity.	N = 5		AOSLO, SD-OCT			24894394	2014	Scoles D, Higgins BP, Cooper RF, Dubis AM, Summerfelt P, Weinberg DV, Kim JE, Stepien KE, Carroll J, Dubra A.	Invest Ophthalmol Vis Sci	Microscopic inner retinal hyper-reflective phenotypes in retinal and neurologic disease.
Blunt ocular trauma		Photoreceptors	Partial recovery of damaged cone photoreceptors following closed globe blunt ocular trauma can be documented using AO-SLO longitudinal tracking.	N = 1		prototype Canon AOSLO, SD-OCT			27391597	2016	Kaizu Y, Nakao S, Yamaguchi M, Murakami Y, Salehi-Had H, Ishibashi T.	BMC Ophthalmol	Detection of airbag impact-induced cone photoreceptor damage by adaptive optics scanning laser ophthalmoscopy: a case report.
Blunt ocular trauma		Cones	disruptions of the photoreceptor mosaic were seen in all subjects with AOSLO imaging, AOSLO in some cases more sensitive than SD-OCT	N = 9		AOSLO, SD-OCT			24752010	2014	Flatter JA, Cooper RF, Dubow MJ, Pinhas A, Singh RS, Kapur R, Shah N, Walsh RD, Hong SH, Weinberg DV, Stepien KE, Wirostko WJ, Robison S, Dubra A, Rosen RB, Connor TB Jr, Carroll J.	Retina	Outer retinal structure after closed-globe blunt ocular trauma.
Blunt ocular trauma		Cones	AOSO detected photoreceptor disruption resulting from head trauma not apparent clinically or by other standard imaging modalities, including SD-OCT.	N = 1		AOSLO, OCT, FLA	HFV 10-2		22411676	2012	Stepien KE, Martinez WM, Dubis AM, Cooper RF, Dubra A, Carroll J.	Arch Ophthalmol	Subclinical photoreceptor disruption in response to severe head trauma.
Blunt ocular trauma		Photoreceptor structure	Clinically available spectral domain OCT, viewed en face or as B-scan, may lead to misinterpretation of photoreceptor anatomy in a variety of diseases and injuries. Split-detector AOSLO revealed substantial populations of photoreceptors in areas of no, low, or ambiguous ellipsoid zone reflectivity with en face OCT and confocal AOSLO.	N = 2	various diseases in this paper	AOSLO, OCT			26166796	2016	Scoles D, Flatter JA, Cooper RF, Langlo CS, Robison S, Neitz M, Weinberg DV, Pennesi ME, Han DP, Dubra A, Carroll J.	Retina	ASSESSING PHOTORECEPTOR STRUCTURE ASSOCIATED WITH ELLIPSOID ZONE DISRUPTIONS VISUALIZED WITH OPTICAL COHERENCE TOMOGRAPHY.
Blunt ocular trauma		Cones, Microcystic lesions	Manifestion of dark, partition-like areas in the cone mosaic on AO-SLO images. Microcystic lesions in the INL may affect the images of the cone mosaic.	N = 1	various diseases in this paper	AO-SLO, SD-OCT			28291071	2017	Hasegawa T, Ooto S, Makiyama Y, Hata M, Miyamoto K, Yoshimura N	Retin Cases Brief Rep	CIRCINATE PARTITION-LIKE FINDINGS ON CONE MOSAIC IMAGED BY ADAPTIVE OPTICS SCANNING LASER OPHTHALMOSCOPY IN EYES WITH INNER NUCLEAR LAYER MICROCYSTIC CHANGES
Idiopathic Central Ring Scotoma		Photoreceptors	AOSLO-findings revealed a parafoveal circular abnormality of the cone mosaic approximately 3° in diameter that corresponded to the ring of visual disturbance.	N = 1		AOSLO, SD-OCT			18413527	2008	Joeres S, Jones SM, Chen DC, Silva D, Olivier S, Fawzi A, Castellarin A, Sadda SR.	Arch Ophthalmol	Retinal imaging with adaptive optics scanning laser ophthalmoscopy in unexplained central ring scotoma.
Subthreshold laser therapy		Photoreceptors, RPE	Subthreshold-Laser in single eye: SLO-AO and SD-OCT imaging of subthreshold laser therapy in human retina showed no cone cell or RPE damage at all time points during a 9-month period using the 25% threshold power 577-nm laser in the human retina	N = 1		AOSLO, SD-OCT			26985801	2016	Wood EH, Leng T, Schachar IH, Karth PA.	Ophthalmic Surg Lasers Imaging Retina	Multi-Modal Longitudinal Evaluation of Subthreshold Laser Lesions in Human Retina, Including Scanning Laser Ophthalmoscope-Adaptive Optics Imaging.
Photocoagulation		Photoreceptors	Sub 1 Hemi-CRVO -> Treatment grid laser, Sub 2 PDR -> treated with PRP // AO images show a normal appearing mosaic around the lesion	N=2		AO fundus, AOSLO, SD-OCT			22491923	2012	Han DP, Croskrey JA, Dubis AM, Schroeder B, Rha J, Carroll J.	Arch Ophthalmol	Adaptive optics and spectral- domain optical coherence tomography of human photoreceptor structure after short-duration [corrected] pascal macular grid and panretinal laser photocoagulation.
Healthy eyes, perifoveal achromatic, L- and M-cone acuity			A significant correlation was observed between thicker retinal pigment epithelium (RPE) complex, higher cone density and better L-cone logMAR at 5 deg eccentricity, but not for achromatic or M-cone logMAR.	N = 32		KAOO II			27353223	2016	Baraas RC, Gjelle JV, Finstad EB, Jacobsen SB, Gilson SJ.	Vision Res	The relationship between perifoveal achromatic, L- and M-cone acuity and retinal structure as assessed with multimodal high resolution imaging.
Choroidal Nevus		Photoreceptors	Detection of photoreceptor abnormalities in the retina overlying the choroidal nevi. These abnormalities may correlate and possibly predict functional visual loss.	N = 3 patients		CF, EDI-SD-OCT, SLO, rtx1	Microperimetry (MAIA)		27711926	2016	Rodrigues MW, Correa ZM, Say EA, Borges FP, Siqueira RC, Cardillo JA, Jorge R.	JAMA Ophthalmol	Photoreceptor Arrangement Changes Secondary to Choroidal Nevus
Outer retinal tubulation		outer retinal tubs, Cones	ORTs demonstrate surviving photoreceptors in tubular structures found within otherwise nonsupportive atrophic areas that lack retinal pigment epithelium and choriocapillaris.	N = 47 patients, N = 29 (no ORT)		SD-OCT, AO-SLO			27984506	2016	King BJ, Sapoznik KA, Elsner AE, Gast TJ, Papay JA, Clark CA, Burns SA.	Optom Vis Sci	SD-OCT and Adaptive Optics Imaging of Outer Retinal Tubulation
Small choroidal melanoma		Photoreceptors	Detection of potential photoreceptor abnormalities in the retina overlying the choroidal lesion and adjacent retina.	N = 1 asymptomatic patient		AO-...?			28419403	2017	Rodrigues MW, Say EA, Shields CL, Jorge R	Ophthalmic Surg Lasers Imaging Retina	Adaptive Optics of Small Choroidal Melanoma
Posterior polar annular choroidal dystrophy		Cones, Paravascular system (FAF)	Longitudinal follow-up of PPACD showed progression of the paravascular atrophy of the pigment epithelium. Foveal cone photoreceptors can be reduced even in the presence of preserved visual acuity.	N = 2	longitudinal study: 3 years				27579567	2018	Forte R, Aptel F, Feldmann A,	Retin Cases Brief Rep.	MULTIMODAL IMAGING OF POSTERIOR POLAR ANNULAR CHOROIDAL DYSTROPHY
(MAK)-related Retinal Degeneration		Cones, blood vessels	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 preservation of foveal vision in eyes with MAK-related retinal degeneration	N = 6 patients with rod-cone degeneration and disease-causing mutations in MAK, N = 5 healthy		OCT-A, AO-SLO, SD-OCT			29103961	2017	Lew YJ, Rinella N, Qin J, Chiang J, Moore AT, Porco TC, Roorda A, Duncan JL	Am J Ophthalmol.	High Resolution Imaging in male germ cell associated kinase (MAK)-related Retinal Degeneration

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Reviews													
Age-related Macular Degeneration(AMD), Diabetes, Glaucoma		Cones	Adaptive optics is opening a new frontier for clinical research in ophthalmology, providing new information on the early pathological changes of the retinal microstructures in various retinal diseases.	REVIEW	REVIEW, various diseases	rtx1			23271600	2013	Lombardo M, Serrao S, Devaney N, Parravano M, Lombardo G.	Sensors (Basel)	Adaptive optics technology for high-resolution retinal imaging.
Age-related Macular Degeneration(AMD), Diabetes, Glaucoma, Macular Telangiectasia		Nerve fibres, microvasculature, cones, RPE	showcase of the latest capabilities of AO systems for imaging the human retina and by an extensive review of the literature on clinical uses of AO.	REVIEW	REVIEW	AO-SLO			26973867	2015	Roorda A, Duncan JL	Annu Rev Vis Sci.	Adaptive optics ophthalmoscopy
Diabetic retinopathy		Microvasculature	Some of these instruments allow a more detailed in vivo examination of the retinal vasculature than fluorescein angiography with- out its potentially serious side effects, thus better allowing us to further study retinal vascular homeostasis in healthy sub- jects and to identify preclinical changes in early disease stages.	REVIEW	REVIEW	AO-SLO, AO-OCT, DOCT			23733525	2013	Deák GG, Schmidt-Erfurth U.	Curr Diab Rep	Imaging of the parafoveal capillary network in diabetes.
Glaucoma		Nerve fibre layer, blood vessels, vitreous	This review is focused on new ocular imaging modalities used for glaucoma diagnosis.	REVIEW	REVIEW	OCT, AO-Fundus, AOOCT			27087829	2015	Kostanyan T, Wollstein G, Schuman JS.	Expert Rev Ophthalmol	Evaluating glaucoma damage: emerging imaging technologies.
Glaucoma			The incorporation of AO into ophthalmic imaging modalities has enhanced OCT by improving image resolution and quality, particularly in the posterior segment of the eye.	REVIEW	REVIEW	AO-OCT			27916682	2016	Dong ZM, Wollstein G, Wang B, Schuman JS.	Prog Retin Eye Res	Adaptive optics optical coherence tomography in glaucoma
Albinism		Cones	REVIEW	REVIEW	REVIEW	AO-SLO			21057346	2010	Godara P, Dubis AM, Roorda A, Duncan JL, Carroll J.	Optom Vis Sci	Adaptive optics retinal imaging: emerging clinical applications.
Several Diseases (mainly non hereditary)		Nerve fibres, microvasculature, cones, RPE	Review about studying retinal diseases with adaptive optics ophthalmoscopy	REVIEW	REVIEW	AO-SLO, AO-OCT, AO-Fundus, rtx1			27995325	2016	Domdei N, Reiniger JL, Pfau M, Charbel Issa P, Holz FG, Harmening WM.	Ophthalmologe	Histology of the living eye : Noninvasive microscopic structure and functional analysis of the retina with adaptive optics
Several Diseases (hereditary)		Nerve fibres, microvasculature, cones, RPE	Review about studying retinal diseases with adaptive optics ophthalmoscopy with a focus on heriditary diseases	REVIEW	REVIEW	AO-SLO, AO-OCT, AO-Fundus, rtx1			28355660	2017	Domdei, Reiniger, 2017	Klinische Monatsblätter	Potential of Adaptive Optics for the Diagnostic Evaluation of Hereditary Retinal Diseases
Diabetic retinopathy		Microvasculature	The potential of newly developed techniques for assessing retinal blood flow and metabolism, such as Doppler techniques, adaptive optics, and retinal oximetry, is promising and may potentially contribute to significant advances in our understanding of diabetic retinopathy.	REVIEW	REVIEW				28791532	2017	Bek T	Curr Diab Rep	Diameter Changes of Retinal Vessels in Diabetic Retinopathy
photoreceptorbased metrics as candidate biomarkers		Photoreceptors	Ongoing and future clinical trials for inherited retinal diseases will benefit from the improved resolution and sensitivity that multimodal AO retinal imaging affords to evaluate safety and efficacy of emerging therapies	REVIEW	REVIEW	AO-SLO			28873135	2017	Litts KM, Cooper RF, Duncan JL, Carroll J	Invest Ophthalmol Vis Sci.	Photoreceptor-Based Biomarkers in AOSLO Retinal Imaging
Inherited retinal diseases		Cones	AO ophthalmoscopy offers invaluable identification of structural detail on a cellular level, with several studies described herein exploring correlation between structure and function. Evolving AO-guided retinal sensitivity assessments ('nanoperimerty') will better allow correlation between cellular imaging and functional testing with exquisite retinotopic precision.	REVIEW	REVIEW	AO-SLO, OCT			29141905	2017	Georgiou M, Kalitzeos A, Patterson EJ, Dubra A, Carroll J, Michaelides M	Br J Ophthalmol.	Adaptive optics imaging of inherited retinal diseases
Fundus imaging with AO-SLO		photoreceptors (cones and rods), fundus vessels, RPE, retinal nerve fiber layer, GC, lamina cribrosa	Comparison with conventional imaging methods and other AO rechniques. Current research situation in AO-SLO and future research directions.	REVIEW	REVIEW	AO-SLO, AO-OCT, AO-FIO, FAF, FF-A, ICG-A, OCT,			29181321	2017	Zhang B, Li N, Kang J, He Y, Chen XM	Int J Ophthalmol.	Adaptive optics scanning laser ophthalmoscopy in fundus imaging, a review and update

Disease	Gene/mutation (if specified)	Retinal structure	Central finding	Number patients	Comments	Imaging modality	Functional testing	Treatment	PMID	Year	Author and year	Journal	Manuscript title
Methods													
Geographic Atrophy		RPE	This new method can be used to study RPE morphology in AMD and other diseases, providing a powerful tool for understanding disease pathogenesis and progression, and offering a new means to assess the efficacy of treatments designed to restore RPE health.	N = 4	method evaluation	AOSLO			24298413	2013	Rossi EA, Rangel-Fonseca P, Parkins K, Fischer W, Latchney LR, Folwell MA, Williams DR, Dubra A, Chung MM.	Biomed Opt Express	In vivo imaging of retinal pigment epithelium cells in age related macular degeneration.
Method description		Microvasculature	Images from the AO-SLO noninvasively revealed pathways with and without dark tail flow in the human parafovea.	N = 5 healthy	method evaluation	AOSLO (Canon)			24586959	2014	Arichika S, Uji A, Ooto S, Miyamoto K, Yoshimura N.	PLoS One	Adaptive optics-assisted identification of preferential erythrocyte aggregate pathways in the human retinal microvasculature.
Method description		Nerve fibres	AO-SLO revealed hyperreflective bundles and dark lines in the RNFL, believed to be retinal nerve fiber bundles and Müller cell septa. The widths of the nerve fiber bundles appear to be proportional to the RNFL thickness at equivalent distances from the optic disc.	N = 20 healthy	method evaluation	AOSLO			22427978	2012	Takayama K, Ooto S, Hangai M, Arakawa N, Oshima S, Shibata N, Hanebuchi M, Inoue T, Yoshimura N.	PLoS One	High-resolution imaging of the retinal nerve fiber layer in normal eyes using adaptive optics scanning laser ophthalmoscopy.
assessment of hemodynamics in Diabetic retinopathy		Microvasculature	Parafoveal hemodynamics, such as capillary velocity, wall shear stress, and capillary perfusion pressure can be noninvasively and reliably characterized with this method in both healthy and diabetic retinopathy patients.	N = 4 patients, N = 4 healthy	method evaluation	AO-SLO, CFP			28078170	2016	Lu Y, Bernabeu MO, Lammer J, Cai CC, Jones ML, Franco CA, Aiello LP, Sun JK.	Biomed Opt Express	Computational fluid dynamics assisted characterization of parafoveal hemodynamics in normal and diabetic eyes using adaptive optics scanning laser ophthalmoscopy
Method description		RPE	The results highlight the need for standardization of image reflectivity to facilitate quantification of en face OCT images and longitudinal analysis.	N = 4 patients, N = 3 healthy	method evaluation	OCT, AO-FIO	microperimetry		27959968	2016	Sampson DM, Alonso-Caneiro D, Chew AL, Lamey T, McLaren T, De Roach J, Chen FK.	PLoS One	Enhanced Visualization of Subtle Outer Retinal Pathology by En Face Optical Coherence Tomography and Correlation with Multi-Modal Imaging
Diabetic Retinopathy		Microvasculature	The technique was able to correct peripheral aberrations to a level that was sufficient for the enhanced visualization of microvasculatures and microaneurysms in diabetic patients.	N = 1 patient, N = 1 healthy	method evaluation	OCT-A, AO-OCT			28059209	2016	Polans J, Cunefare D, Cole E, Keller B, Mettu PS, Cousins SW, Allingham MJ, Izatt JA, Farsiu S.	Opt Lett	Enhanced visualization of peripheral retinal vasculature with wavefront sensorless adaptive optics optical coherence tomography angiography in diabetic patients
Retinitis Pigmentosa (RP) and Stargardt (ST)		Cones	The developed and presented algorithms do not require spatial regularity in cone packing and are, therefore, useful for counting cones in diseased retinas, as demonstrated for eyes with Stargardt's macular dystrophy and retinitis pigmentosa.	N = 1 RP-patient, N = 1 ST-patient, N = 3 healthy	Testing of cone counting algorithm	AO flood-illum			17429482	2007	Xue B, Choi SS, Doble N, Werner JS.	J Opt Soc Am A Opt Image Sci Vis	Photoreceptor counting and montaging of en-face retinal images from an adaptive optics fundus camera
Macular dystrophy, Retinitis Pigmentosa (RP) and Acute zonal occult outer retinopathy (AZOOR)		Photoreceptors	The presented algorithm is more stable than conventional methods in cases of non-periodical photoreceptor structures such as the affected retinal area.	N = 15 healthy, N = 3 (macular dystrophy, RP, AZOOR)	Testing of cone counting algorithm	AO-SLO (Topcon Corporation)			28479850	2017	Miyagawa S, Fukuyama H, Hirota M, Yamaguchi T, Kitamura K, Endo T, Kanda H, Morimoto T, Fujikado T	Clin Ophthalmol	Automated measurements of human cone photoreceptor density in healthy and degenerative retina by region-based segmentation
Method description		Microvasculature	The magnitude of these proportional changes implies that the capillary beds themselves play an important role in the retinal response to changes in carbon dioxide levels.		effects of altered gas-breathing conditions	AO-SLO			28522835	2017	Duan A, Bedggood PA, Metha AB, Bui BV	Sci Rep	Reactivity in the human retinal microvasculature measured during acute gas breathing provocations
Maculopathies		Cones	Outer retinal reflectivity on en-face optical coherence tomography correlates well with photoreceptor density. This cone density estimation method based on retinal reflectivity could have interesting applications in the exploration and management of maculopathies.	N = 9 eyes of 6 patients	Testing of cone counting algorithm for OCT	rtx-1, SD-OCT			28791546	2017	Saleh M, Flores M, Gauthier AS, Elphege E, Delbosc B	Graefes Arch Clin Exp Ophthalmol.	Quantitative analysis of photoreceptor layer reflectivity on en-face optical coherence tomography as an estimator of cone density
Stargardt disease (STGD) and retinitis pigmentosa GTPase regulator (RPGR)-associated retinopathy		Cones	Split-detector AO-SLO greatly improved the reliability and repeatability of cone density measurements in both disorders and will be valuable for natural history studies and clinical trials using AO-SLO. However, it appears that these indices may be disease dependent.	N = 12 STGD patients, N = 8 RPGR patients	Evaluation of imaging method and cone counts	AO-SLO			28738413	2017	Tanna P, Kasilian M, Strauss R, Tee J, Kalitzeos A, Tarima S3, Visotcky A, Dubra A, Carroll J, Michaelides M	Invest Ophthalmol Vis Sci.	Reliability and Repeatability of Cone Density Measurements in Patients With Stargardt Disease and RPGR-Associated Retinopathy
Method description		Retinal vascular networks	In cases requiring accurate and detailed retinal vasculature observation, AO-SLO might be useful for evaluating retinal vascular lesions as a supportive imaging method of OCTA.	N = 16 healthy	Method testing	AO-SLO, OCT-A			28875064	2017	Kaizu Y, Nakao S, Wada I, Yamaguchi M, Fujiwara K, Yoshida S, Hisatomi T, Ikeda Y, Hayami T, Ishibashi T, Sonoda KH	Transl Vis Sci Technol.	Imaging of Retinal Vascular Layers: Adaptive Optics Scanning Laser Ophthalmoscopy Versus Optical Coherence Tomography Angiography