

Table S1. Clinical data of patients diagnosed with STGD or with the differential diagnosis of STGD.

P a t i e n t I D	Diag nosis	Age onset	Severit y	Visual acuity details	Best-corrected Visual Acuity (LogMAR)		Tension	Fundus autofluoresce nce (FAF)	Autofluorescenc e and deposits (Severity)	optical coherence tomography (OCT)	central macular thicknes s	macular volume	Electroretinogram m (ERG)	Fluorescei n angiograph y	Color vision	Goldmann Visual	Others
					OD	OS											
0 0 8	CRD	45	severe	progressive visual impairment OU (blurry vision) over years, central vision distortion including metamorphopsi a OU, night vision problems, reduced contrast sensitivity, glare, no color vision problems besides inherited protanopic color vision deficiency	0.1	0.1	16mmHg/16 mmHg	bilateral finely speckled hypoautofluore scent spots, adjacent hyperautofluor escent rim, (para-)central retinal atrophy, OS foveal sparing	deposits none, atrophy medium	bilateral (para-)central lesions, cRORA, OD ORTs, OS foveal sparing, iRORA and retinal thinning up to the vascular arcades	177µm/23 1µm	6.21mm³/6.40m m³	ffERG: scotopic and photopic amplitudes slightly reduced below normal values (combined rod cone dystrophy)	N/A	mainly protanopic confusions	OD>OS concentric scotoma	N/A
0 1 7	RP	childhood	severe	Initial photopsia, glare, night vision problems, reduced contrast sensitivity, variable visual impairment OU (blurry vision)	0.1	0.1	14 mmHg/15 mmHg	bilateral hypoautofluore scent spots, slight adjacent diffuse hyperautofluor escence	deposits none, atrophy medium, (small lesions, great distribution)	bilateral indistinct borders of atrophy, mainly iRORA, foveal sparing, cystoid macular edema	286µm/27 0µm	7.55mm³/7.31m m³	ffERG: reduced scotopic and photopic amplitudes, but near to normal	N/A	inconspicuo us	regular borders of the visual field	N/A
0 1 9	STG D	20	N/A	Initial symptom: Central	0.8	1.2	N/A	OD: Mild perimacular	N/A	OD: Hyperreflective and irregular photoreceptor	205µm/22 1µm	8.42mm³ 8.89mm³	/N/A	OD: Mild perimacular staining, no	N/A	N/A	N/A

				Scotoma right eye in 2016				autofluorescence with flecks		inner/outer segment junction				leakage, no dark choroid			
								OS: Inconspicuous		OS: Dry neurosensory retinal layers				OS: Inconspicuous			
020	MD	51	N/A	No visual field defects in 2012	Date: Value 08/16: 0.05 05/16: 0.32 04/16: 0.4 02/16: 0.4 01/16: 0.5 12/15: 0.5 08/15: 0.5 05/15: 0.7 03/14: 0.8 03/11: 1.0	Date: Value 08/16: 0.3 05/16: 0.32 04/16: 0.5 02/16: 0.4 01/16: 0.5 12/15: 0.5 08/15: 0.63 05/15: 0.7 03/14: 0.8 03/11: 1.0	Date: Value 05/15: 18/19 mmHg 12/15: 20/20 mmHg 01/16: 22/22 mmHg 04/16: 16/16 mmHg	Central autofluorescence due to multiple Drusen (Drusen maculopathy)	N/A	OD: Numerous drusen especially centrally. Dry neurosensory retina OS: Drusenoid pigment epithelial detachment	Date: Value 08/16: 170 um/489 um 06/15: 516 um/504 um	Date: Value 08/16: 8.63 mm³/10.65mm³ 06/15: 10.68 mm³/10.69mm³	N/A	OD: Intense staining, no leakage OS: Intense staining, no leakage	N/A	N/A	Alopecia, Fibromyalgia. Does not look like classical Stargardt but something like Drusenoid maculopathy
021	STG D	69	N/A	Left Eye: Eccentric vision, subjective visual impairment left eye when presenting in Nov. 2015	Hand movement	Finger-counting	N/A	Perifoveolar hyper-auto-fluorescence with flecks OS>OD, centrally no evaluation possible due to retinal pigment epithelial atrophy	N/A	Bilateral: Central pigment epithelial and central retinal atrophy	236um/227um	5.77 mm³/6.23mm³	N/A	OD>OS dark choroid, centrally large RPE defects. Late staining, no leakage	N/A	N/A	Developed Myelodysplastic syndrome 04/2017. Arterial hypertension, Cardiac arrhythmias, post stent operation, fibromyalgia

022	STG D	30	N/A	N/A	2015: 1/35 (Visual acuity chart) 2011: 0.05	2015: 1/35 2011: 0.05	18mmHg/19mmHg	Bilateral: Diffuse paracentral and midperipheral autofluorescence, central large RPE defects	N/A	Bilateral: Central pigment epithelial and central retinal atrophy	132µm/70µm	4.39mm³/4.48mm³	N/A	Bilateral: Dark choroid Late-phase: Perimacular staining, no leakage	N/A	N/A	Hypertension, Diabetes II,
023	STG D	50	Mild	metamorphopsia OD>OS, glare, progressive visual impairment OU (blurry vision), reduced contrast sensitivity	0.22	0.22	12mmHg/12mmHg	bilateral alterations of the retinal pigment epithelium (RPE) and circumscribed areas of paracentral retinal atrophy (foveal sparing), surrounding hypo- and hyperautofluorescent drusenoid spots	deposits low atrophy medium	circumscribed paracentral complete retinal pigment epithelium and outer retinal atrophy (cRORA), adjacent incomplete retinal pigment epithelium and outer retinal atrophy (iRORA), foveal sparing of RORA, degenerative intraretinal fluid	266µm/304µm	7.95mm³/8.08mm³	mfERG: OD>OS reduced amplitudes in the central area, some artefacts	N/A	inconspicuous (Panel D15)	regular borders of the visual field, OS (para-)central scotoma	N/A
024	STG D	50	Mild	progressive paracentral scotoma OU, progressive visual impairment OU (blurry vision), metamorphopsia	0.1	0.22	13mmHg/14mmHg	bilateral hyperautofluorescent macular and peripapillary spots, few spots of paracentral retinal atrophy (foveal sparing)	deposits medium atrophy low	hyperreflective spots, partially invading into the outer retina, partly confluent lesions of cRORA, foveal sparing, degenerative intraretinal fluid	328µm/322µm	9.31mm³/9.00mm³	mfERG: bilateral amplitudes in normal range, slight relative reduction in the paracentral areas	FA: bilateral dark choroid, mild paracentral dye pooling in the late phase	Inconspicuous	N/A	Clinical examination: pattern-like distribution of the lesions
025	STG D	49	N/A	N/A	0.05	0.3	N/A	N/A	N/A	OD: Foveolar retinal thinning with intraretinal cysts and questionable retinal pigment epithelial atrophy	216µm/110µm	6.79mm³ 6.99mm³	/N/A	N/A	N/A	N/A	Decrease of VA at the age of 40. Sister has M. Stargardt (020 II.3)

										OS: Mild central retinal atrophy with reduced foveolar depression, signs of retinal pigment epithelial atrophy							
02511.3	STG D	23	N/A	N/A	Date: Value 02/87: 0.4 08/15: 0.05	Date: Value 02/87: 0.2 08/15: 0.05	Date: Value 02/87: 13/15 mmHg 08/15: 16/16 mmHg	OD: Central retinal pigment epithelial atrophy, perimacular hyperautofluorescent flecks, dark choroid OS: Central retinal pigment epithelial atrophy, perimacular hyperautofluorescent flecks, dark choroid	N/A	OD/OS: Central retinal pigment epithelial and outer retinal atrophy	257µm / 192µm	7.12mm³ / 6.96mm³	N/A	OD/OS: Central retinal pigment epithelial defect, macular hyperfluorescent flecks, late staining, no leakage	N/A	N/A	N/A
026	STG D	16	severe	Subjective visual impairment since 2013 visual field: Bilateral central scotoma in year 2016	Date: Value 3/16: 0.16 1/16: 0.2p	Date: Value 03/16: 0.2p 1/16: 0.2p	14 mmHg/14 mmHg	OD/OS: Perimacular autofluorescent spots	N/A	OD/OS: Central foveal retinal pigment epithelial and retinal atrophy	20 µm / 61 µm	7.33mm³ / 7.15mm³	OD/OS: Dark choroid, foveolar hyperfluorescence with central late staining, no leakage	N/A	N/A	Central scotoma	N/A

N/A: Not available.