Ocular and Imaging Findings in Bardet-Biedl Syndrome with Advanced Stage Retinal Dystrophy

İleri Evre Retina Distrofisi Olan Bardet-Biedl Sendromunda Oküler ve Görüntüleme Bulguları

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ABSTRACT

Background: To evaluate the ocular and imaging findings in cases of Bardet-Biedl syndrome (BBS) with advanced-stage retinal dystrophy.

Materials and Methods: Ophthalmic examinations of patients with clinically proven BBS reported in this retrospective observational study. Optical coherence tomography, fundus autofluorescence (AF) and optical biometry measurements were evaluated in detail.

Results: Twenty-eight eyes of 14 patients with BBS were evaluated and compared with those in the control group. The mean age of the patients was 31.7±11.2 years. The mean axial length (AL) of the eye was 22.9±0.9 mm and mean anterior chamber depth (ACD) was 3.01±0.37 mm. Cataract was observed in 17 eyes (68%). The mean central macular thickness (CMT) was 99.1±35.3 μm and the mean subfoveal choroidal thickness (SCT) was 196.1±32.3 μm. The mean AL, ACD, CMT, and SCT all were significantly lower in patients with BBS than in the control group (p<0.001). In all BBS cases, ellipsoid zone and external limiting membrane integrity were partially or completely disturbed. The retinal pigment epithelium and Bruch's membrane were observed to be thinner. In addition, eight eyes (29%) had deposit-like appearances on Bruch's membrane, six eyes (21%) had intraretinal hyper-reflective foci, ten eyes (36%) had internal limiting membrane (ILM) thickening, seven eyes (25%) had epiretinal membrane, three eyes (11%) had ILM wrinkling, three eyes (11%) had hyper-AF ring, and ten eyes (36%) had abnormally hyper-AF patterns with an irregular distribution.

Conclusion: In BBS, ocular pathologies can be seen in the outer retina, intraretinal, vitreoretinal interface, and anterior segment. This study provides insight into the ocular pathologies of BBS and may be useful to evaluate patients with BBS for treatment options.

Keywords: Fundus autofluorescence, optical biometry, optical coherence tomography, retinal dystrophy

ÖZ

Amaç: İleri evre retina distrofisi olan Bardet-Biedl sendromu (BBS) olgularında oküler ve görüntüleme bulgularını değerlendirmek.

Gereç ve Yöntemler: Bu retrospektif gözlemsel çalışmada klinik olarak BBS'si olan hastaların oftalmik muayeneleri bildirilmiştir. Optik koherens tomografi, fundus otofloresans (AF) ve optik biyometri ölçümleri detaylı olarak değerlendirildi.

Bulgular: BBS'li 14 hastanın 28 gözü değerlendirildi ve kontrol grubundakilerle karşılaştırıldı. Hastaların ortalama yaşı 31,7±11,2 yıldı. Gözün ortalama aksiyel uzunluğu (AL) 22,9±0,9 mm ve ortalama ön kamara derinliği (ACD) 3,01±0,37 mm idi. On yedi gözde (%68) katarakt görüldü. Ortalama santral makula kalınlığı (CMT) 99,1±35,3 mikron ve ortalama subfoveal koroid kalınlığı (SCT) 196,1±32,3 mikron idi. Ortalama AL, ACD, CMT ve SCT, BBS'li hastalarda kontrol grubuna göre anlamlı olarak daha düşüktü (p<0,001). Tüm BBS olgularında, elipsoid zon ve eksternal limitan membran bütünlüğü kısmen veya tamamen bozulmuştu. Retina pigment epiteli ve Bruch membranının daha ince olduğu görüldü. Ayrıca, sekiz gözde (%29) Bruch membranında depozit benzeri görünüm, altı gözde (%21) intraretinal hiper-reflektif odaklar, on gözde (%36) internal limitan membran (ILM) kalınlaşması, yedi gözde (%25) epiretinal membran, üç gözde (%11) ILM kırışıklığı, üç gözde (%11) hiper-AF halkası ve on gözde (%36) düzensiz dağılım gösteren anormal hiper-AF paternler saptandı.

Sonuç: BBS'de dış retinanın yanı sıra intraretinal, vitreoretinal ara yüzey ve ön segmentte de oküler patolojiler görülebilmektedir. Bu çalışma BBS'nin oküler patolojileri hakkında fikir vermektedir ve BBS'li hastalarının oküler tedavi seçeneklerini değerlendirmek için faydalı olabilir.

Anahtar Kelimeler: Fundus otofloresansı, optik biyometri, optik koherens tomografi, retina distrofisi

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Introduction

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder that causes multiple system anomalies and displays broad clinical features. Its central clinical features include retinal dystrophy, polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction (1).

Retinal dystrophy becomes clinically evident in early childhood. Subsequent visual loss is progressive and the most severe vision loss occurs in the early adolescence period. Morphological characteristics of BBS include a bull's eye appearance of the macula, and the development of retinitis pigmentosa (RP), characterized by bone spicules, at advanced stages of BBS (2).

BBS is a ciliopathy, as the cause of retinopathy is thought to be due to the involvement of connecting cilia on the outer retinal layer. Retinal dystrophy is progressive and its severity shows variability (3).

In this study, the ocular findings of 14 BBS cases with advanced stage RP were evaluated using biometry, optical coherence tomography (OCT) and fundus autofluorescence (FAF). To the best of our knowledge, this is the first such study on the ocular and imaging findings of BBS patients in the literature, and thus represents the first quantitative survey of ocular findings in BBS patients with RP.

Material and Methods

Patients with clinically defined BBS, characterized as having retinal dystrophy, dysmorphic extremities or polydactyly, obesity, renal abnormalities, hypogonadism, mental retardation, were included in our study. Included patients were diagnosed by the internal medicine, pediatrics, urology, or nephrology clinics and were referred to our hospital's ophthalmology clinic between the dates of June 2014 and November 2016 (1,4). All included patients were Caucasian, to control for ethnic variability. This study was approved by the Local Human Research Ethics Committee, in accordance with the Declaration of Helsinki, and written informed consent was obtained from all participants (University of Health Sciences Türkiye, Şişli Hamidiye Etfal Training and Research Hospital Ethics Committee, 24.01.2017, approval number: 1377).

Examinations performed on patients included the best corrected visual acuity (BCVA) test, intraocular pressure (IOP) measurement, and anterior and posterior segment examinations. Patient family history of consanguineous unions was recorded. Additional systemic and ocular pathologies were recorded. Axial length (AL) and anterior chamber depth (ACD) measurements were performed using optical biometry (NIDEK Optic Biometry, AL-scan, Japan). Pupil dilatation was obtained using 1% tropicamide and 10% phenylephrine. Spectral domain OCT (3DOCT-2000; Topcon Inc., Tokyo, Japan) was used to evaluate a 6×6 mm macular area, a 9 mm choroidal thickness in the horizontal plane, and FAF images. The values of central macula thickness (CMT) and subfoveal choroidal thickness (SCT) were recorded between 12.00 p.m. midday and 2.00 p.m. in the afternoon. The SCT was measured manually at a 500 microns interval from the fovea, so that the inner border would be the sclera and the outer border would be retinal pigment epithelium (RPE). The same observer carried out all the measurements.

Retinal nerve fiber layer thickness and visual field could not be assessed in the patients due to nystagmus, the inability to focus because of cataracts, or the poor quality of shooting. Full-field standard electroretinograms (ERGs) were obtained in accordance with the protocols of the International Society for Clinical Electrophysiology of Vision (5). However ERGs of patients eyes, which were non-recordable for the patient under all standard stimuli and recording conditions due to advanced-stage retinal dystrophy.

Values that could be measured with biometry and OCT were taken into account. AL, ACD, CMT and SCT values were compared with an age-matched control group. Healthy cases that applied to our clinic for routine ophthalmologic examination were selected as the control group. The control group included cases with ±0.50 diopters, BCVA of 20/20, IOP was below 20 mm Hg, and there was no pathology in the ocular examinations.

Statistical Analysis

SPSS 15.0 for Windows program was used for statistical analysis. Descriptive statistics were given as number and percentage for categorical variables and as mean, standard deviation, minimum, maximum, and median for numerical variables. When the numerical variables met the conditions of the normal distribution, the Student's t-test was used to compare the two independent groups and the Mann-Whitney U test was used when the normal distribution conditions were not met. Since the relations between numerical variables did not meet the condition of a parametric test, they were analyzed using Spearman correlation analysis. Statistical significance level of alpha was accepted as $p<0.05$.

Results

Twenty-eight eyes of 14 BBS patients with advanced stage retinal dystrophy and 28 eyes of 14 people in the control group were evaluated. Gender, age, consanguineous

marriage, BCVA, and clinical phenotypic characteristics of the patients are shown in Table 1. In both the case and control groups, four patients were female (29%) and ten patients were male (71%). The mean age of the case group was 31.7±11.2 (16-58) years, and the mean age of control group was 31.7±11.2 (16-57) years. The parental consanguinity rate among cases was 86%. The BCVA of case patients was determined as presence of only light perception in 11eyes (39%), presence of only hand motion in 13 eyes (47%), and counting fingers at 1 meter in 4 eyes (14%). The strabismus examination, presence of nystagmus, anterior segment examination, IOP, AL, and ACD values of the cases are shown in Table 2. Exotropia was present in 12 cases (86%), nystagmus was present in 11 cases (76%), and 2 cases were orthotrophic (14%). In the anterior segment examination, no pathology was detected in eight eyes (29%). The posterior subcapsular cataract (PSC) was observed in 17 eyes (61%), nuclear and PSC was observed in two eyes (7%), and one eye (3%) was pseudophakic. The mean IOP was 14.3±2.4 (10-19) mm Hg. The mean AL was 22.9±0.9 (20.8-24.9) mm, and the mean ACD was 3.01±0.37 (1.95-3.5) mm. In the control group, the mean IOP was 14.8±2.6 (11-19) mm Hg, the mean AL was 23.7±0.6 (22.9-25) mm, and the

mean ACD was 3.45±0.39 (2.88-4.09) mm. Mean values for AL and ACD were statistically significantly lower (p<0.001) in the BBS group compared to the control group. The means of IOP were not significantly difference between the case and control groups (p=0.465).

The OCT and FAF imaging features are shown in detail in Table 3. The mean values of CMT and SCT in the OCT were 99.1±35.3 (42-171) and 196.1±32.3 (127-257) μm in the BBS group, and 213.9±22.4 (171-246) for mean CMT and 289.1±48.1 (198-365) μm for mean SCT in the control group, respectively. The values of CMT and SCT were statistically significantly lower for the BBS group than the control group (p<0.001). The values of CMT and SCT were also statistically significant correlated in the BBS group (p=0.023, rho=0.428), but no correlation was detected in the control group (p=0.442). In all cases, complete or partial disruption of the ellipsoid zone (EZ), disruption of the external limiting membrane (ELM) line integrity, and thinning of RPE/Bruch's membrane were observed. Intraretinal hyper-reflective foci were detected in a total of six eyes (21%). Among these six cases, four eyes had BCVA limited to hand motion and two eyes had BCVA limited to light detection. Deposit-like hyper-reflective foci were observed in the Bruch's

Table 1. Gender, age, consanguineous marriage, visual acuity and clinical phenotypic characteristics of the patients with Bardet-Biedl syndrome										
Case	Gender	Age	Consanguineous marriage	Visual acuity (right/left)	Polydactilia	Obesity	Learning difficulty	Hypogonadism	Kidney anomaly	Other
1	M	42	-	lp/lp	+	+	+	-	-	
2	F	58	-	lp/lp	+	+	+	-	-	DM
3	M	43	+	lp/lp	+	+	+	+	-	
4	M	31	+	lp/lp	+	+	+	-	+	DM
5	M	31	+	lp/lp	+	+	-	-	+	DM
6	M	20	+	lp/hm	+	+	+	+	+	DM
7	F	25	+	hm/hm	+	+	+	-	-	DM
8	M	41	+	hm/hm	Bradydactilia	+	+	+	-	HT, depression
9	M	29	+	hm/hm	+	Overweight	+	+	-	
10	M	31	+	hm/hm	+	+	-	+	+	DM
11	F	16	+	hm/hm	+	+	+	-	-	DM, hypothyroidism
12	F	29	+	hm/hm	+	+	-	+	+	
13	M	29	+	1 mcf/1 mcf	+	+	+	-	-	Developmental retardation, speaking D/O
14	M	19	+	1 mcf/1 mcf	+	+	+	-	-	DM, clotting D/O
lp: Light perception, hm: Hand motion, 1 mcf: Counting fingers at 1 meter, DM: Diabetes mellitus, HT: Hypertension, D/O: Disorder										

Table 2. Strabismus, nystagmus, anterior segment examination, intraocular pressures, optical biometry axial length and anterior chamber depth values of the patients with Bardet Biedl syndrome

Case	Strabismus	Nystagmus	Anterior segment examination (right/left)	Intraocular pressure (right/left) mmHg	Axial length (right/left) mm	Anterior chamber depth (right/left) mm
1	Exotropia	+	Normal/normal	14/16	23.28/22.88	2.91/2.89
2	Exotropia	+	PSC+NC/PSC+NC	15/12	23.00/21.50	NA/NA
3	Exotropia	+	PSC/pseudophacia	18/17	22.73/22.48	NA/NA
4	Exotropia	+	PSC/PSC	12/16	20.80/20.95	2.27/1.95
5	Exotropia	-	PSC/PSC	12/13	22.92/22.52	3.11/3.09
6	Exotropia	+	PSC/PSC	14/14	22.07/21.97	NA/NA
7	Orthotrophic	+	PSC/PSC	13/12	23.63/23.34	2.93/2.82
8	Orthotrophic	-	Normal/normal	15/15	22.77/22.61	3.14/3.08
9	Exotropia	+	PSC/PSC	13/11	23.40/23.34	3.05/3.01
10	Exotropia	+	PSC/PSC	18/17	23.26/23.71	NA/NA
11	Exotropia	+	Normal/normal	14/17	23.49/23.63	2.83/2.95
12	Exotropia	+	PSC/PSC	19/16	23.43/23.23	3.27/3.43
13	Exotropia	-	Normal/normal	19/19	22.43/22.64	3.21/3.25
14	Exotropia	+	PSC/PSC	14/13	24.70/24.92	3.50/3.48

PSC: Posterior subcapsular cataract, NC: Nuclear cataract, NA: Not available

Table 3. Optical coherence tomography and fundus autofluorescence imaging features of the patients with Bardet-Biedl syndrome

Case	OCT imaging					FAF imaging	
	Central macular thickness (right/left)	Subfoveal choroidal thickness (right/left)	Intraretinal hyperreflective foci (right/left)	Hyperreflective foci on Bruch's membrane (right/left)	Vitreoretinal interface pathologies (right/left)	Macular area (right/left)	Perimacular area (right/left)
1	54/45	127/134	-/-	-/-	-/-	-/d	-/-
2	92/85	163/178	-/-	-/-	-/-	-/-	-/-
3	68/85	175/170	+/+	+/+	-/a	-/-	-/-
4	98/42	196/213	-/-	-/-	b/b	-/d	-/-
5	102/114	212/204	-/-	-/-	a/-	d/-	-/-
6	90/123	257/240	-/-	-/-	b/b	-/-	-/-
7	143/171	255/186	-/-	-/+	-/b	d/d	+/+
8	55/78	188/196	-/-	-/-	-/-	-/d	-/-
9	117/137	196/206	+/-	+/-	a/a	d/-	-/-
10	122/124	208/197	+/-	+/-	a/a*	c/-	+/-
11	75/59	215/222	+/+	+/+	a/a	-/-	-/-
12	88/78	186/194	-/-	-/-	-/-	d/-	+/+
13	167/156	232/227	-/-	-/-	b*/b*	c/c	-/-
14	106/101	171/143	-/-	+/-	a/a	d/d	-/-

OCT: Optical coherence tomography, ILM: Internal limiting membrane, ERM: Epiretinal membran, FAF: Fundus autofluorescence, a: ILM thickening, b: ERM presence, *: ILM wrinkling, c: Increased FAF ring in macula, d: Abnormally increased FAF patterns which are distributed irregularly in the macula

membrane in a total of eight eyes (29%). Their BCVA was only light perception in two eyes, hand motion in five eyes, and counting fingers at 1 meter in one eye (Figure 1a). When the macular region was evaluated in terms of vitreoretinal interface pathologies, internal limiting membrane (ILM) wrinkling was observed in three eyes (11%), ILM thickening was observed in ten eyes (36%), and epiretinal membrane (ERM) was observed in seven eyes (25%) (Figure 1b). Cystoid macular edema, micropseudocysts, subretinal fluid, macular or lamellar holes, choroidal neovascularization were not observed in any of the cases. In the FAF examination, a perifoveal increased hyper-autofluorescence (AF) ring was detected in three eyes (11%) (Figure 2a). In two of these cases, BCVA was counting fingers at 1 meter, while in one patient, it was at the level of hand motion. Abnormally hyper-AF patterns with irregular distribution were detected in the macula of ten eyes (36%). In FAF imaging, there were

patchy areas of AF in the perimacular area in five eyes (18%) with BCVA of hand motion. Perimacular AF loss was observed in the remaining 23 eyes (82%) (Figure 2b).

Discussion

BBS is a very rare disease characterized by progressive retinal dystrophy. To the best of our knowledge, our study presents the ophthalmic examination, optical biometry, OCT and FAF findings for the largest number of patients with BBS. In all cases, partial or complete disruption of the EZ and ELM line integrity, and a thinning of the RPE/Bruch's membrane were observed. In addition to outer retinal pathology, commonly detected pathologies in patients included diffuse retinal and choroidal atrophy, intraretinal hyper-reflective foci, vitreoretinal interface pathologies, deposit-like accumulations on the Bruch's membrane, hyper-AF rings, abnormally hyper-AF patterns with irregular distribution and cataracts, especially PSC.

Previous work has shown that patients with BBS have not only total loss of the retinal photoreceptor layer, but also loss of the inner retinal layers, including degeneration, loss of the inner-outer nuclear layer, and decrease in ganglion cell count and glial proliferation (6). In a postmortem histopathological study of the eye of a 60-year-old patient, the choroid layer was fibrotic in appearance (2). RPE cells were observed to have migrated to the inner retinal layers and accumulated on the walls of retinal blood vessels. In another study of BBS, the accumulation of lipid, calcium, and PAS-positive abnormal material, including iron, was shown between RPE and Bruch's membrane (7). In our study, there was a loss of central function in all cases. In the fundus examination, all retinal areas had diffuse atrophy of RPE, and a patchy appearance somewhat similar to bone spiculations.
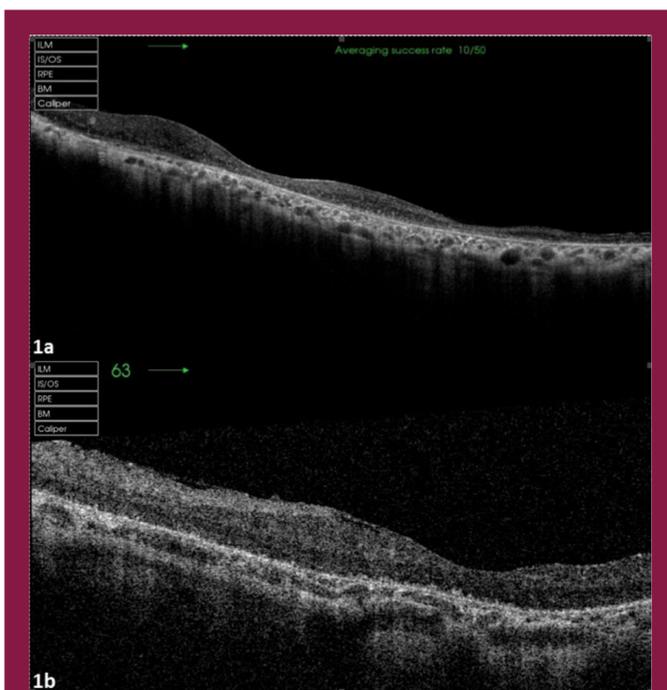

Figure 1. Presence of intraretinal hyper-reflective foci in 9 mm line macula OCT scan of a 43-year-old male patient with vision of only light perception. The patient also had thickening of ILM, complete disruption of EZ and ELM line integrity, thinning of RPE/Bruch's membrane, hyper-reflective deposits on Bruch's membrane, thinning of choroid layer thickness (1a). Epiretinal membrane at the retinal surface in the left eye, 6 mm horizontal macular OCT scan of a 25-year-old female patient with visual acuity of hand motion. The patient also had hyper-reflective deposits on the Bruch's membrane, diffuse thinning of the choroid layer, complete disruption of the EZ and ELM line integrity, and thinning of the RPE/Bruch's membrane (1b)

OCT: Optical coherence tomography, ILM: Internal limiting membrane, ERM: Epiretinal membran, RPE: Retinal pigment epithelium, EZ: Ellipsoid zone, ELM: External limiting membrane
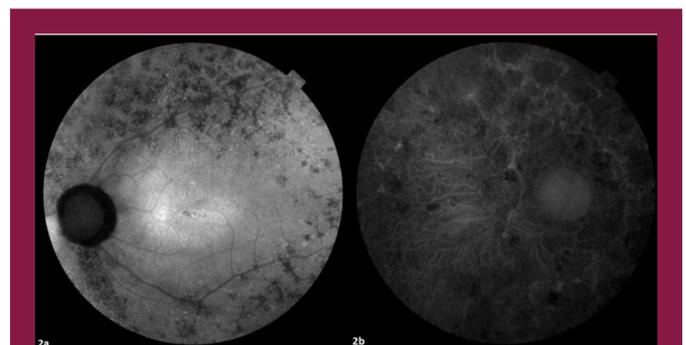

Figure 2. Appearance of perifoveal hyper-autofluorescence ring and patchy perimacular areas of fluorescence in the left eye fundus autofluorescence imaging of a 29-year-old male patient whose visual acuity was counting fingers at 1 meter (2a). Diffuse loss of autofluorescence loss in the macular and perimacular area in the right eye fundus autofluorescence imaging of a 41-year-old male patient whose visual acuity was hand motions (2b)

There was also slight or severe optic disc pallor and evident visibility of the choroid layer.

OCT is frequently used to evaluate retinal structures and layers in the eyes of the patients with retinal pathologies. It was indicated that there was a correlation between OCT and histological retinal evaluation. OCT provides a detailed non-invasive evaluation of retinal layers in patients with BBS (8). In our BBS cases, the retinal layers could not be separately examined when carrying out central macular area examinations of the OCT image due to severe diffuse atrophy in the retina. In all of our cases, partial or complete disruption of the EZ and ELM line integrity, and a thinning of the RPE/Bruch's membrane were observed, as well as thinning in the choroid layer. Furthermore, we detected a correlation between SCT and CMT. Therefore, we believe that thinning of the choroid layer thickness develops secondary to retinal dystrophy in BBS patients with advanced stage retinal dystrophy.

We evaluated intraretinal hyper-reflective foci in patients with BBS. In the OCT of patients with RP, hyper-reflective foci were observed in both the inner and outer nuclear layers. Reactive gliosis, migration of active microglia, expansion of the synaptic processes of photoreceptor cells, and synaptic remodeling of amacrine and horizontal cells by the Müller cells in the inner layers have been previously demonstrated in both human and animal studies (9). In the OCT, intraretinal hyper-reflective foci were detected in six eyes (21%) in our study. However, because of the presence of widespread retinal atrophy, we could not determine which layer was affected by these foci. Like Gerth et al. (8) we think that this condition may be related to retinal re-organization.

We examined deposit-like accumulations on the Bruch's membrane in patients with BBS. Deposit accumulation on the Bruch's membrane was shown in siblings with advanced stage RP. This accumulation was considered to be a finding of advanced stage retinopathy or a factor causing faster degeneration of retinal layers (7). In our study, eight eyes (29%) of BBS patients had a hyper-reflective deposit-like appearance on Bruch's membrane, and VA for these eyes ranged from counting fingers to light perception. We consider that the deposit on Bruch's membrane is a finding of advanced-stage retinopathy, it may be the residual RPE aggregation or remnant of RPE. Also, we observed that, in most of these cases, the deposit-like appearance that gives hyper-reflectance on Bruch's membrane and the abnormally increased FAF patterns that were distributed irregularly in the macula were localized.

Vitreoretinal interface pathologies can also be seen in patients with BBS. Gerth et al. (8) found ILM wrinkling in three of eight patients with BBS. They suggested that advanced stage retinal dystrophy may result in ILM wrinkling due to the thinning or collapse of retinal layers. In our study, ILM wrinkling was seen in only 11% of the patients with advanced stage retinal dystrophy. Most of our cases with advanced stage retinal dystrophy did not demonstrate ILM wrinkling, indicating that ILM wrinkling is not always associated with the thinning or collapse of the retinal layers. For this reason, we suggest that ILM wrinkling is not exclusively caused by collapse due to thinning in the retina, but that there are other unknown causal mechanisms. In our study, the rate of ILM thickening or ERM was observed to be as high as 61%. We think that this thickening may be due to reactive gliosis due to Müller cells and the migration of active microglia.

There are few studies available in the literature on FAF imaging in patients with BBS. In studies by Billingsley et al. (10) and Cox et al. (11), perifoveal hyper-AF rings are most commonly detected in the FAF imaging of BBS patients. In a literature review by Mitamura et al. (12), diagnostic imaging findings of patients with RP were evaluated, and they stated that abnormal hyper-AF in FAF was the result of increased turnover of the photoreceptor outer segment, impaired phagocytosis, or an intrinsic defect in the RPE cells that enable recycling of phagosomes. However, as the disease progresses, AF is either not present or reduced as a consequence of RPE atrophy or loss of photoreceptor cells. They also showed that the hyper-AF ring indicated the border between the functional and non-functional retina, and that it was strongly correlated with retinal function. However, they stated that hyper-AF ring could sometimes be the result of increased photoreceptor degeneration at an abnormally high rate. Wakabayashi et al. (13) suggested that hyper-AF ring in BBS patients with RP was the result of active photoreceptor degeneration or increased phagocytosis of external segments by RPE. They stated that when the accumulation of lipofuscin in the RPE cells reaches a critical level, AF signals would reach the maximum level, but it would cause AF loss as a result of RPE atrophy and photoreceptor cell death. In our study, the perifoveal hyper-AF ring was detected in only three eyes (11%) and irregularly distributed macular hyper-AF patterns were detected in ten eyes (36%). Most of these cases were eyes with a VA that was at the level of counting fingers at 1 meter or hand motion. We think that abnormally hyper-AF patterns, which were irregularly distributed in the macula, may be the result of surviving RPE aggregation, or they could be due to the window effect caused by the outer plexiform layer thinning rather than lipofuscin accumulation. We suggest that the reason for having different FAF imaging results compared to these other studies was because the retinal dystrophies of our patients were at more advanced stages.

Our results showed ACD and AL, and the incidence of cataracts of patients with BBS. In the literature, there are no studies evaluating the AL and ACD with biometry in cases with BBS. In our study, we detected a significant narrowing in the values of AL and ACD in patients with BBS. We think that this situation may develop as a result of the influenced neurotransmitters that play a role in ocular growth in BBS patients. The incidence of cataracts in BBS is much higher than that in the normal population. Although the mean age of our patients was 31.7 years, 68% of the eyes had cataracts (especially posterior subcapsular cataracts), and 3% of the eyes had cataract surgery. We think that this condition develops secondary to inflammation.

Study Limitations

One limitation of this study is that gene analysis of our patients was not performed. In future studies, we will consider if there is a correlation between gene analysis and OCT, and FAF findings.

Conclusion

In BBS patients, changes occur not only in the outer layers of retina but also in the inner layers of retina and in the vitreoretinal interface. A significant thinning is seen in the CMT and SCT. In addition to retinal pathology of the eye, there is also marked narrowing in the values of AL and ACD, and high incidence of cataracts. Biometry, OCT, and FAF provide important quantitative data in the documentation of ocular examinations of BBS patients. This data is useful to evaluate candidate cases in groups of patients with BBS for such treatment options as stem cell transplantation, which is used in visual rehabilitation, or retinal prosthesis implantation in those patients who are cognitively suitable.

Information: This study was accepted as an oral presentation at 32. TOD Summer Symposiums 2019, İzmir.

Ethics

Ethics Committee Approval: This study was approved by the Local Human Research Ethics Committee, in accordance with the Declaration of Helsinki, (University of Health Sciences Türkiye, Şişli Hamidiye Etfal Training and Research Hospital Ethics Committee, 24.01.2017, approval number: 1377).

Informed Consent: Written informed consent was obtained from all participants.

Peer-review: Internally and externally peer-reviewed.

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