Unilateral Retinal Pigment Epithelium Dysgenesis - The First Case in Pakistan

Hina Qazi and M. A. Rehman Siddiqui

Department of Ophthalmology and Visual Sciences, The Aga Khan University Hospital, Karachi, Pakistan

ABSTRACT

Unilateral retinal pigment epithelium dysgenesis (URPED) is one of the exceedingly rare conditions of the retina. Herein, the first case of URPED in Pakistan is reported in a 33-year male who presented with a uniocular decrease in vision in his right eye. His general, ophthalmic, and family history were unremarkable. His right eye fundus examination revealed a pale centrally atrophic area bounded by scalloped fringe-like margins in the peripapillary region. The lesion showed hypo-autofluorescence on fundus autofluorescence. Optical coherence tomography (OCT) scan showed central atrophy, thinned-out ellipsoid, and interdigitation zone with hyperplastic changes in the retinal pigment epithelium (RPE). The left eye revealed no abnormal findings either on clinical examination or on imaging modalities. He was diagnosed with URPED with preserved vision. No treatment was required. The patient was counselled about the nature of his disease, associated complications, and the need for its long-term follow-up.

Key Words: *Retina, Vision, Unilateral retinal pigment epithelium dysgenesis.*

How to cite this article: Qazi H, Siddiqui MAR. Unilateral Retinal Pigment Epithelium Dysgenesis - The First Case in Pakistan. *JCPSP Case Rep* 2023; **1**:102-104.

INTRODUCTION

Unilateral retinal pigment epithelium dysgenesis (URPED) was initially reported by Cohen *et al*. as unilateral, leopard-spot lesions in the retinal pigment epithelium (RPE).[1] The characteristic clinical features of the disease are well-defined, scalloped margins of RPE, hyperplasia, and fibrosis, and the remarkable inversion of fundus autofluorescence (FAF) with hyperfluorescence observed on fundus fluorescence angiography (FFA).[2] These characteristic findings were recently reported in two Asian patients by Zhu *et al*.[3] With only 24 reported cases worldwide, and only a few from Asia, this is the first case of URPED in Pakistan to the best of authors' knowledge.

CASE REPORT

A 33-year Afghan male presented with blurred vision in his right eye for the last few months. His ophthalmic history was unremarkable as was his past medical or drug history. His family history was negative for any familial disorders. Any ocular trauma or an inflammatory episode was denied. On examination, his best corrected visual acuity was 20/30 in the right eye and 20/25 in the left eye.

Correspondence to: Dr. Rehman Siddiqui, Department of Ophthalmology and Visual Sciences, The Aga Khan University Hospital, Karachi, Pakistan
E-mail: rehman.siddiqui@gmail.com

Received: July 08, 2023; Revised: September 17, 2023; Accepted: September 22, 2023
DOI: https://doi.org/10.29271/jcpspcr.2023.102

Examination revealed normal anterior segments of both eyes with intraocular pressures in the normal range. The posterior pole of his right eye showed a large flat solitary lesion, yellowish pale in colour, extending from the optic disc to the inferior half of the macula bordered by fringe-like margins. The central area of the lesion showed RPE atrophy, with fibrotic hyperplastic RPE changes in the scalloped borders (Figure 1A). A greyish zone of hypopigmentation was seen in the mid-periphery. The vascular arcades appeared normal. The posterior segment in the left eye was unremarkable.

The FAF of the right eye revealed a markedly hypo-autoflourescent lesion on the macula with scalloped fringe-like margins. The inferior mid-periphery of the fundus showed a triangular hypo-autoflourescent area surrounded by a zone of hyper-autoflourescence indicating a gravitational tract (Figure 1B).

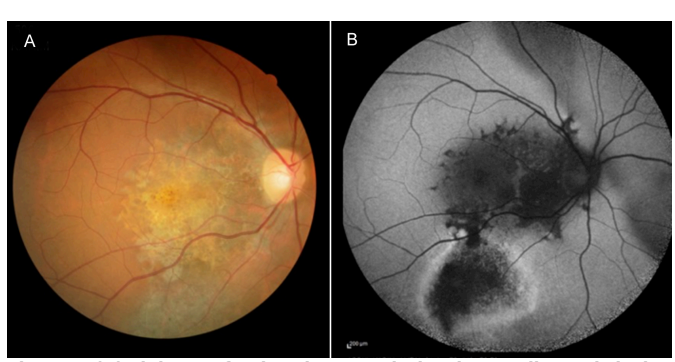

Figure 1: (A) Right-eye fundus photograph showing a yellow pale lesion with scalloped fringe-like margins. (B) Fundus autofluorescence (FAF) with marked hypo-autoflourescent area correlating with fundus photo. Gravitational tract inferiorly (arrow).

Table I: Characteristics of published cases of unilateral retinal pigment epithelium dysgenesis worldwide.

S.No	Year	Origin	Author	Journal	Laterality	Age	Gender	Presenting vision	SD=OCT	Complications/ Findings	Treatment	Final vision
1	2002	White	Cohen *et al.*	Arch Ophthalmol	OS	34	M	20/20	-	CNV on FFA	Krypton laser	20/20
2		White			OD	27	M	20/25	Central atrophy, thickened sensory retina, cystic changes, ERM	ERM/ vascular tortuosity/ Progression over 7 years		20/40
3		White			OD	16	F	20/20	-	-	-	20/20
4		White			OD	27	M	20/128	-	CNV- FFA	Krypton laser	20/126
5	2008	-	Berthout *et al.*	J Fr Ophthalmol		36	F	-	-	Retinal folds	No follow-up	N/A
6	2009	Asian	Cohen *et al.*	Am J Ophthalmol	OD	19	M	20/20	Central RPE atrophy/ transmission defects	-	-	20/20
7		White			OD	36	F	20/32	Central atrophy, ERM	ERM, retinal folds	-	20/32
8		White			OS	18	M	20/40	Central RPE atrophy/ transmission defects	Torturous vessels	-	20/40
9		White			OD	42		20/400	Central atrophy, transmission defects, cystic irregular retinal surface, localised detachment			20/400
10		White			OD	16	F	20/25	Central RPE atrophy/ transmission defects	Vascular tortuosity, retinal folds		20/25
11	2012	-	Renz *et al.*	Arch Ophthalmol	OD/OS	35	F	20/25	Outer retinal thinning, attenuated IS/OS	-	Lost to follow up	N/A
12	2014	-	Shioyama *et al*.	Case Rep Ophthalmol	OS	8	M	20/20	Subfoveal CNV	Progression/ treatment resistantCNVM	SITA /Bevacizumab	20/50
13	2017	-	Yamasaki *et al*.	Retina Cases Brief Rep	OS	8	M	20/20	Attenuated IS/OS junction, choroidal thinning	Mild progression of atrophic lesion over 2 years	-	20/20
14	2018	-	Krohn *et al*.	Acta Ophthalmol	OS	21	M	20/20	Disorganized retinal architecture, Foveal RPE atrophy	Progressive RPE atrophy over 10 years	-	20/100
15	2019	African-American	Florakis *et al*.	Retina Cases Brief Rep	OS	47	M	20/20	RPE hyperplasia, shallow RPE detachment, thinned outer retina.	-	-	20/20
16	2019	White	Gar-Or *et al*.	Retina Cases Brief Rep	OS	30	F	20/25	Hyporeflective subretinal mass originating from RPE with subretinal fluid	Presumed RPE tumour	Ranibizumab + aflibercept intravitreally lead to reduction in subfoveal/ subretinal fluid	20/20
17	2019	-	Preziosa *et al*.	Retina Cases Brief Rep	OS	51	F	20/200	RPE hyperplasia with atrophy and subfoveal PED	CNVM	2 injections of bevacizumab	20/50
18	2020	-	Riga *et al*.	Ocul Oncol Pathol	OS	52	M	20/40	ERM, retinal tortuosity and folds, RPE atrophy	Progressive atrophy over 8 years	-	20/200
19	2020	Chinese	Ding *et al*.	BMC Ophthalmol	OS	10	F	20/25	Thinned out ellipsoid zone, irregular RPE-Bruch's complex. Inhomogeneous signals of outer segments of photoreceptors at the fovea.	-	-	20/25
20	2020	-	Sekeryapan Gediz *et al*.	Turk J Ophthalmol	OD	32	M	20/32	Retinal folds, CNV with SRF	Retinal folds, CNV	Bevacizumab	20/20
21	2020	-	Diafas *et al*.	Am J Ophthalmol Case Rep		16	F	Counting Fingers at 1 meter	Localized tractional detachment with macular hole	TRD with FTMH	PPV with ERM-ILM peel	20/32
22	2021	-	Handa *et al*.	Can J Ophthalmol	OS	16	F	20/30	RPE loss with hypertransmission+ RPE thickening	-	-	N/A
23	2022	Chinese	Zhu *et al*.	BMC Ophthalmol	OD	51	M	14/20	Cystic changes, disorganised architecture, ERM, CNVM	CNV, ERM	-	N/A
24					OD	39	M	20/25	Abnormal RPE, cystic changes, subretinal hyperreflectivity	-	-	N/A

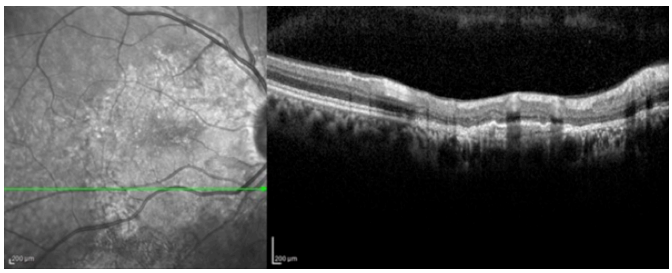

Figure 2: Spectral-domain optical coherence tomography (SD-OCT): Retinal pigment epithelium (RPE) hyperplasia with choroidal hyper-reflectivity secondary to hyper-transmission effect.

Spectral-domain optical coherence tomography (SD-OCT) revealed distorted outer retinal layers. The ellipsoid and interdigitation zones were thinned out. The RPE-Bruch's membrane complex showed hyperplastic changes throughout the lesion giving hypertransmission on OCT (Figure 2). There was no fluid, blood, or exudates seen in the scans.

The FAF and OCT scans of the fellow-eye were unremarkable. The patient was diagnosed as having right-eye URPED with good vision. He was counselled about the nature of his disease and the need for its long-term follow-up.

DISCUSSION

The pale appearance surrounded by fringe-like margins, marked hypo-autofluorescence on FAF along with thinned-out interdigitation and ellipsoid zones on OCT in this patient were consistent with the findings of Cohen *et al.*[1] The triangular area of a gravitational tract on FAF suggested a now resolved accumulation of fluid (Figure 1). Whether this was a separate event or was secondary to the URPED lesion remained a query.

The disease does not show a preference towards gender. Literature showed middle-aged people being affected more than young (Table I). The aetiology of this condition remains unidentified. It was hypothesised by Renz *et al.* that an infection, inflammation, or an autoimmune condition could play a role. With the word dysgenesis, they suggested that it should be a bilateral disease and reported a bilateral variant of URPED in 2012.[4] This was one of its kind and no other bilateral cases have been reported since then.

Despite its distinctive features, it is important to consider other differentials. Acute zonal occult outer retinopathy (AZOOR) may appear as a whitish discoid lesion but the fringe-like margins of URPED can differentiate the two. Combined hamartoma of the retina and RPE (CHRRPE) is another entity of which URPED is a forme fruste variant. It can present as a raised lesion with retinal disorganisation. Furthermore, clinical manifestations, age at presentation, and FFA features may differ from URPED. Choroidal osteoma, a benign ossifying intraocular tumour, has smooth delineating margins as opposed to URPED along with a prominent elevated mass, which is not seen in URPED.

The prognosis of the disease seems good unless complications such as choroidal neovascular or epiretinal membranes, retinal detachment, and foveal atrophy develop.[2] Gal-Or *et al.* reported a presumed intraocular tumour from an existing URPED lesion.[5]

However, there are certain limitations to this case. There was a lack of the fundus fluorescein angiography (FFA) information as the patient could not financially afford further tests. Further long-term follow-up was also needed as the disease is reported to progress slowly and gradually.

PATIENT'S CONSENT:
A written informed consent was taken from the patient.

COMPETING INTEREST:
The authors declared no competing interest.

AUTHORS' CONTRIBUTION:
HQ: Prepared the draft.
RS: Revised the draft and provided critical feedback.
Both authors approved the final version.

REFERENCES

1. Cohen SY, Massin P, Quentel G. Clinicopathologic reports case reports, and small case series: Unilateral, idiopathic leopard-spot lesion of the retinal pigment epithelium. *Arch Ophthalmol* 2002; **120(4)**:512-6.
2. Cohen SY, Fung AE, Tadayoni R, Massin P, Barbazetto I, Berthout A, *et al*. Unilateral retinal pigment epithelium dysgenesis. *Am J Ophthalmol* 2009; **148(6)**:914-9.e2. doi: 10.1016/j.ajo.2009.06.033.
3. Zhu Z, Xiao J, Luo L, Yang B, Zou H, Zhang C. Common clinical features of unilateral retinal pigment epithelium dysgenesis and combined hamartoma of the retina and retinal pigment epithelium. *BMC Ophthalmology* 2022; **22(1)**:24. doi: 10.1186/s12886-022-02244-x.
4. Renz J, Fein JG, Vora R, Woodcome H, Reichel E, Duker J. Unilateral retinal pigment epithelium dysgenesis may be a bilateral disease. *Arch Ophthalmol* 2012; **130(10)**:1340-2. doi: 10.1001/archophthalmol.2012.845.
5. Gal-Or O, Finger PT, Fisher YL, Yannuzzi LA, Freund KB. Presumed retinal pigment epithelium tumor originating from unilateral retinal pigment epithelium dysgenesis. *Retin Cases Brief Rep* 2019; **13(2)**:121-6. doi: 10.1097/ICB.0000000000000568.