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Case Report

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İsolated Foveal Hypoplasia

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Introduction

Foveal hypoplasia (FH) is characterized by the lack of foveal depression and persistence of all neurosensory retinal layers in the supposed foveal region [1-3]. Foveal hypoplasia may be associated with albinism, microphthalmia, aniridia, achromatosis, and retinopathy of prematurity [4]. The disease can also be seen as an isolated form rarely [5]. Optical coherence tomography (OCT) is a quick and useful technique to make the suspected diagnosis and grading of the disease [1,3]. In this report, we present a case of foveal hypoplasia who had a preserved vision and spectral domain optical coherence tomography (SD-OCT) was performed to make the diagnosis and explain the structural characteristics.

Case Report

A 49-year-old female patient applied to our clinic for the routine eye examination, without any complaints. She did not have any systemic diseases except for Diabetes mellitus. The full ophthalmic examination was performed. Her visual acuity was 9/10 in both eyes. Refractive errors were -0,50 and 2,50-0,50 axe160 and intraocular pressures were 18 and 17 mmHg in the right and left eyes respectively. Bio-microscopic anterior segment examination was normal; she did not have aniridia, cataract or translumination defect. Bilateral foveal reflexes were absent but retinal pigmentation was normal on fundus examination. She did not have nystagmus and strabismus. Spectral domain optical coherence tomography (RTVue-100 OCT, Optovue, Inc., Fremont, CA) was performed and multiple retinal sections were obtained from the foveal region. In the OCT scans foveal pits are absent, inner retinal layers were continued in the fovea-like extra foveal region and the whole retina was thickened in both eyes central foveal thicknesses were 282 and 300 µm in right and left eyes respectively. Subfoveal choroidal thicknesses are 247 and 251 µm in right and left eyes respectively. The external limiting membrane and IS/OS bands were intact in both eyes. With these findings, the patient was diagnosed with isolated foveal hypoplasia.

Discussion

In FH there is a wide variability of clinical manifestations such as fundal aspect and visual acuities but there is a common finding for all cases, that is SD-OCT pattern with reduced or absent foveal pit and continuity of inner retinal layers [1-3].

Fundus appearance may vary from unremarkable except for absent of foveal reflexes to slightly mottled pigment disorganizations in foveal hypoplasia [2], as we found the first one in our case. In recent years, studies have shown that OCT can be used as a diagnostic and prognostic indicator for foveal hypoplasia [1,4,6]. Various OCT findings may be encountered in foveal hypoplasia cases. These findings are

the shallow or absent foveal pit, increased outer nuclear layer width, increased photoreceptor outer segment length, and thickening of the whole retina [1-3, 8-10]. Foveal hypoplasia is graded by optical coherence tomography according to the presence or absence of findings such as extrusion of plexiform layers in the fovea, the absence of foveal pit, the extension of photoreceptor outer segment, and expansion of outer nuclear layer. 5 grades are defined [4,11]:

Grade 1: There are shallow foveal depression; photoreceptor outer segment prolongation and outer nuclear layer expansion but no extrusion of plexiform layers.

Grade 2: There are photoreceptor outer segment prolongation and outer nuclear layer expansion but no extrusion of plexiform layers and foveal depression.

Grade 3: There is outer nuclear layer expansion but no extrusion of plexiform layers, foveal depression, and photoreceptor outer segment prolongation.

Grade 4: There are no extrusion of plexiform layers, foveal depression, photoreceptor outer segment prolongation and outer nuclear layer expansion.

Atypical: There is shallow foveal depression, but no extrusion of plexiform layers. The IS/OS band is degraded and interrupted.

In our case, we confirmed the diagnosis by OCT and graded by this classification system as grade 2. It was reported that foveal hypoplasia is associated with a broad range of visual acuities from 20/20 to 20/400 in the literature [7]. In our case, visual acuity was 9/10 in both eyes. OCT may also be useful for giving an idea about the estimated visual acuity. Some of the ophthalmologists tried to figure out a relationship between visual acuity and foveal morphology, but the foveal pit was not accepted as a prognostic factor [7]. The foveal avascular zone and the pit are not critical for the postnatal lengthening of cones or spatial packing which have a role to make the higher visual resolution. Although foveal cone specialization may be protected anatomically and functionally during the absence of a foveal depression. Fovea plana is a term recently suggested to define only the anatomic absence of a foveal pit with no functional effects [7,12]. This explains why some patients may have good visual acuity despite the absence of normal foveal depression, as found in our patient. The grading system was based on the stage at which foveal development was stopped and may be helpful to provide a prognostic indicator for visual acuity [11]. The normal thickness of the photoreceptor layer found in our patient at the presumed foveal center is also present in healthy eyes [2]. This may be a sign of lesser anatomic alteration in a subgroup of patients with foveal hypoplasia and may be the explanation of the preserved visual acuity in these patients.

In our patient, there were no signs of associated features such as regressed ROP, myopia, cataract, corneal pannus, ocular albinism or incontinent pigments which were reported previously. For this reason, our case diagnosed as an isolated form of foveal hypoplasia. This is a rare form of the disease. This report highlights the role of OCT in the diagnosis of this condition, by the demonstration of the preservation of the inner retinal layers in the fovea. SD-OCT is a noninvasive, simple and rapid procedure for the detection of foveal hypoplasia, which can be difficult to detect in the clinical and ophthalmologic examination in cases with unexplained visual decrease. FH must be kept in mind in patients who had a good but not complete visual acuity and OCT must be performed for the diagnosis.

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