Congenital Mydriasis Associated with Absence of Accommodation: Case Report

Akomodasyon Kaybıyla Seyreden Konjenital Midriyazis

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ABSTRACT A 14-year-old patient presented to our hospital with dilated pupils and decreased near vision. Complete ocular examination was done and pupil responses to pharmacological agents were evaluated. The patient had bilateral congenital mydriasis. The pupils were mydriatic. At pupil margins there were pigmented rudimentary nipple-like structures and remnants of persistent pupillary membrane. Pupils did not respond to pharmaceutics. The patient was not able to accommodate. Ultrasound biomicroscopy demonstrated hypoplasia of ciliary processes. Congenital mydriasis may be associated with absence of accommodation. Patients with that condition may need near vision correction first.

Key Words: Accommodation, ocular; congenital; mydriasis

ÖZET On dört yaşında kadın hasta kliniğimize pupillalarda dilatasyon ve yakın görmede bozukluk yakınmalarıyla başvurdu. Tam göz muayenesi yapıldı ve farmakolojik ajanlara pupil yanıtları değerlendirildi. Hastada bilateral konjenital midriyazis mevcuttu. Pupillalar midriyatikti. Pupil kenarlarında pigmente rudimenter meme başı benzeri yapılar ve pupil membranı kalıntıları vardı. Pupillalar farmasötik ajanlara yanıtsızdı. Hasta akomodasyon yapamıyordu. Ultrasonografik biyomikroskopide siliyer proseslerde hipoplazi olduğu gözlendi. Konjenital midriyazise akomodasyon yokluğu eşlik edebilmektedir. Bu durumdaki hastalarda öncelikle yakın görmenin düzeltilmesi gerekli olabilmektedir.

Anahtar Kelimeler: Uyum, oküler; konjenital; midriyaz

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Congenital mydriasis is a rare ocular abnormality. This condition is defined as the presence of fixed and dilated pupils, which are present at birth and occur in otherwise normal eyes with normal sight.[1] It may be associated with failure of accommodation.[2] In this case report, we described a 14-year-old patient who had congenital mydriasis with absence of accommodation.

CASE REPORT

A 14-year-old female patient presented with photophobia. General physical examination was unremarkable and mental development was normal. There was no consanguinity of her parents. Ocular examination showed bi-

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lateral fixed, dilated pupils observed by parents since birth. Neither direct nor indirect reaction to light could be detected bilaterally. Near reflexes were absent. There was no reaction of pupils with cover test. Best spectacle corrected visual acuities were 20/20 with-0.50 dioptry (D) correction in the right eye and-0.25 D correction in the left eye. Near vision was at the level of J1 with +2.5 D correction. Pupil diameters were 7.5 mm and 8 mm in the right and left eyes, respectively. Both pupils were round and regular. The irises were brown and lacked stromal furrows. In both eyes, there was a reasonable rim of iris with many, nipple-like pigmented structures at pupil margin. The stroma was hypoplastic, without crypts (Figure 1). The irises did not show transillumination defect. Grid filiform tissue at the pupillary margin indicated persistence of the pupillary membrane. Iridocorneal angle was open but some pigmented filiform attachments arising from iris tissue and reaching to pigmented trabeculum were detected by gonioscopy. Ocular tensions were 15 and 17 mm Hg in the right and left eyes, respectively. The cornea, lens, vitreus, optic nerve, macula, and peripheral retina were normal but increased retinal arterial tortuosity was observed by ophthalmoscopy.

Phenylephrine hydrochloride 2.5% eye drops were instilled to stimulate the dilator muscles. After 30 minutes, there was no change in the pupil size in either eye. Cyclopentolate 1% was instilled to paralyze the sphincter muscle and show unopposed dilator activity; however, it produced no change in pupil sizes or refractions. Cycloplegic refractions were detected as -0.50(-0.50α128) D and -0.25 (-0.25α25) D in the right and left eyes, respectively. After a washout period, pilocarpine 2% was instilled on both eyes to demonstrate sphincter activity by direct stimulation and 1 hour later the pupil diameters and refractions did not change.

Axial lengths, anterior chamber depths and lens thicknesses were 21.82 mm, 2.85 mm, and 3.72 mm in the right eye and 21.24 mm, 2.59, and 3.74 mm in the left eye, respectively.

Ultrasound biomicroscopy revealed slightly narrow anterior chambers, dilated and hypoplastic irises. The number and length of ciliary processes were normal but there was diffuse hyporeflective appearance around ciliary body, which was thought to be a reflection of hypoplasia (Figure 2).

Similar iris changes with loss of accommodation were observed in the father of the patient (Figure 3). The ocular examination of the mother was entirely normal.

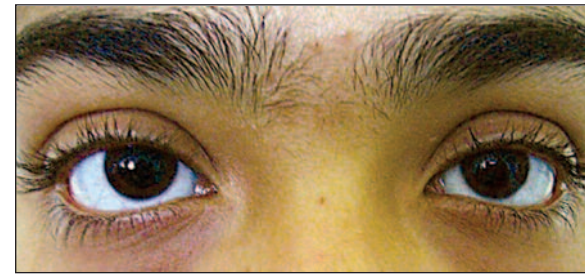

FIGURE 1: Photography of the patient showing fixed, dilated pupils.
(See for colored form http://tipbilimleri.turkiyeklinikleri.com/)

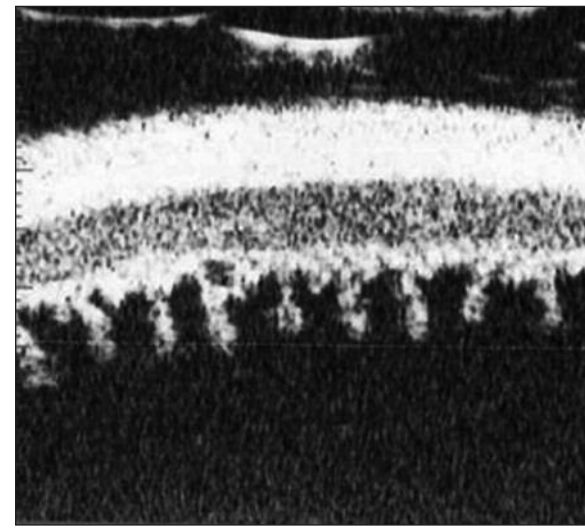

FIGURE 2: Ultrasound biomicroscopy of the ciliary processes.

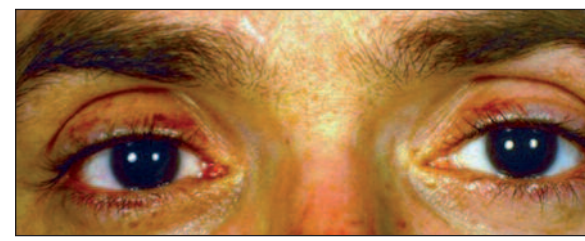

FIGURE 3: Photography of the father.
(See for colored form http://tipbilimleri.turkiyeklinikleri.com/)

An informed consent was obtained from the patient and her father.

DISCUSSION

A few cases of bilateral congenital mydriasis have been documented previously in the literature. The first cases-monozygotic twins-described by White and Fulton, had large irregular pupils, which reacted only slightly to light. The authors suggested that the abnormality was due to congenital absence of the sphincter iris.[1]

Congenital mydriasis is an extremely rare ophthalmic condition. Fixed and dilated pupils are present at birth and occur in normal seeing eyes. It is interesting that females are affected more frequently than males, typically bilaterally.[1-4] For congenital mydriasis, autosomal dominant mode of inheritance and a possible X-linked mode with nonviability of males were suggested.[1-5] Our patient was female and the presence of the affected father suggests the autosomal dominant mode of inheritance.

The iris is derived from the neuroectoderm and the surrounding mesoderm. Sphincter (at 12 weeks of gestation) and dilator muscles (at 24 weeks of gestation) arise from the neuroepithelial layer of the optic cup. The iris stroma develops from the mesoderm. It consists of two layers. The superficial layer forms the pupillary membrane. The deeper layer ends at the pupil. By the 32nd week of gestation, the pupillary membrane begins to resorb.[6] In our patient, there was hypoplasia of the iris stroma. Normal architecture of the iris disappeared with the absence of branching bands or trabeculae giving a velvety appearance to the iris. Fibrillar tissue representing pupillary membrane remnants were present at pupillary margin. Neither the annular band of muscle at the pupillary margin representing the sphincter, nor the concentric circular folds in the peripheral iris representing the dilator muscle were present. The lack of response to pilocarpine in our patient confirmed the absence of sphincter activity. The absence of a change in the diameter of pupils after instillation of cyclopentolate showed that there was neither dilator activity nor sphincter activity to be blocked. Absence of sympathetic dilator activity was confirmed with lack of response to a phenylephrine drop. In our patient, the slit-lamp appearance of the irises and the absence of response to pharmacologic agents demonstrated functional absence of sphincter and dilator muscles.

Previously, association of congenital mydriasis with lack of accommodation was reported.[3] Absence of a change in refraction after instillation of pilocarpine hydrochloride revealed that accommodation of our patient was also affected. This finding led us to conclude that the ciliary body might be hypoplastic and nonfunctional. Ultrasound biomicroscopy confirmed this.

Since our patient did not have any other ocular abnormalities, we think that the iris changes are primary abnormality not related to the other anterior segment syndromes, like aniridia, circumpupillary aplasia or anterior chamber cleavage syndromes. Iris changes in congenital mydriasis may also be observed in Gillespie's syndrome but cerebellar ataxia and mental subnormality were not present in our patient, so we concluded that the iris anomaly of our patient was an isolated condition.[7]

The ophthalmologic findings of our patient represents a rare, nonprogressive condition called congenital mydriasis which can be recognised by clinical observation, pharmacologic testings and sophisticated anterior segment examination like ultrasound biomicroscopy. This condition can be associated with hypoplasia of ciliary processes with loss of accommodation. Patients with that condition may immediately need near vision correction firstly.

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