Bilateral congenital mydriasis in a child case

Dear Editor,

In this letter, a case presentation has been made to recognize congenital mydriasis, discuss the causes of mydriasis and emphasize the importance of pupil examination.

Congenital mydriasis is defined as congenital absence of iris sphincter muscle which occurs rarely and shows autosomal dominant inheritance (1-4). It was described by White and Fulton (2) in 1937 for the first time. Fixed and dilated pupil is present from the time of birth, observed more frequently in girls and typically bilateral (1-4). Iris sphincter traumas, pharmacological dilatation and acquired neurological disorders should be excluded in these patients.

A 14-year-old female patient presented to our outpatient clinic because of absent pupillary light reflex on neurological examination performed in another center. It was observed that her brain magnetic resonance imaging was reported to be normal. She had no complaints related with her eyes. She had no history of trauma, seizure, surgical intervention related with the eyes or any surgical intervention or use of systemic medication. She had no familial history of pupil anomaly. On ophthalmologic examination of the patient who was mentally normal, uncorrected visual acuity was full in both eyes. In the right eye, light reflex was absent, the pupil was fixed, moderately enlarged and had an oval appearance. In the left eye, light reflex was observed weakly only with very bright light. The pupil was moderately enlarged and had a smooth appearance (Figure 1). The pupil was 0.5 mm more dilated in the right eye compared to the left eye (4/3.5 mm). No difference was observed in the pupillary diameters in the dark and in the light. Relative afferent pupillary defect was not found. On dynamic retinoscopy, it was observed that accommodation was absent. The near point of convergence was 7 cm. The close reflex was absent. Diplopia was not described, because mydriasis was congenital.

On biomicroscopic examination, the iris and other anterior segment findings were observed to be natural. The intraocular pressures were found to be normal and fundoscopic examination was found to be normal. The eye movements were free and strabismus was not found. The laboratory tests of the patient who was evaluated in the department of pediatrics were found to be normal (complete blood count and routine biochemical tests). No additional systemic anomaly was found. Pilocarpine test (0.1% diluted pilocarpine) was performed for both eyes considering tonic pupil in both eyes. Assessment performed 45 minutes later showed that no change occurred in the pupillary diameters. The patient was started to be followed up with a prediagnosis of isolated congenital mydriasis considering congenital defect of sphincter efficiency.

Mydriatic pupil observed in the iris which appears to have a normal structure is named congenital mydriasis or familial iridoplegia. Other causes which lead to bilateral mydriasis include toxic conditions (atropine intoxication, Parkinson drugs, antidepressants, carbonmonoxide intoxication, cocaine), conditions secondary to morbidities (migraine, schizophrenia, hyperthyroidism, coma) and Parinaud oculoglandular syndrome. Adie's

Figure 1. a, b. Absent light reflex, fixed, moderately enlarged and oval right pupil. Absent light reflex, fixed, moderately enlarged and smooth left pupil

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tonic pupil, third cranial nerve palsy and pharmacological agents (unilateral use of mydriatic drop) are among the causes of unilateral mydriasis.

Lindberg and Brunvand (5) reported a 12-year old girl with bilateral congenital mydriasis and aneurismal dilatation of patent ductus arteriosus (5). In the literature, cases of congenital mydriasis accompanied by coronary artery disease, aortic aneurism, smooth muscle cell dysfunction (6), gene mutations (ACTA2) (7), Prune Belly syndrome (7), septo-optic dysplasia which may be observed in association with cerebrovascular diseases (8) and megacystic microcolon intestinal hypoperistaltism syndrome (9) have been reported. In the differential diagnosis of pupil dilatation, occurrence of tears in the iris sphincter caused by blunt eye trauma should be considered. In this case, direct and indirect light reaction is absent. Gotz-Wieckowska and Kociecki (10) reported an eight-year old female patient with bilateral Adie's pupil.

In conclusion, the diagnosis of congenital mydriasis should be kept in mind in the differential diagnosis of mydriasis. Recognition of this rare clinical picture by ophthalmologists and consultation with pediatricians in terms of screening additional systemic anomalies are important in the follow-up of the patients.

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