Strabismus and Poor Stereoacuity Associated with Kabuki Syndrome

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Kabuki syndrome is characterized by long palpebral fissures, large ears, a depressed nasal tip, and skeletal anomalies associated with postnatal dwarfism and mental retardation. There have been few prior detailed descriptions of strabismus or stereopsis in these patients. We report a patient with Kabuki syndrome who showed small-angle strabismus and poor stereopsis. This case illustrates the need for patients with a diagnosis of Kabuki syndrome to have an ophthalmologic evaluation. Strabismus associated with Kabuki syndrome may have a small angle that can be easily overlooked.

Key Words: Kabuki syndrome, Stereoacuity, Strabismus
Fig. 1. Photograph of the patient with Kabuki syndrome shows a small-angle esotropia in the primary position. Mild limitations of abduction in both eyes were noted.

The patient recognized nothing with the Randot stereotest. She fused 4 dots at a distance and near with the Worth four-dot test.

The patient had sparse arched eyebrows, long palpebral fissures and eversion of the lateral third of the lower eyelids (Fig. 1), a broad and depressed nasal tip, cleft palate, gynecomastia, short, incurved fifth fingers, epicanthal folds, retrognathia, a small mouth, a tented upper lip, a short nasal septum, and prominent ears. A single flexion crease was observed on each of the fourth and fifth fingers. These features are characteristic of Kabuki syndrome.

Discussion

The reported ocular findings associated with Kabuki syndrome include amblyopia, refractive errors, ptosis, strabismus, nystagmus, ophthalmoplegia, sixth cranial nerve palsy, microphthalmia, microcornea, megalocornea, corneal opacities, Peter’s anomaly, blue sclerae, cataracts, retinal pigmentary hypopigmentation, abnormal electroretinogram and visual evoked potential, obstructed nasolacrimal ducts, tilted discs, and colobomas of the iris and retina [1-6]. Forty-three cases of strabismus have been reported out of 200 documented patients with Kabuki syndrome [1]. However, few prior, detailed descriptions of the associated strabismus or stereopsis exist. One patient with a description of the pattern of strabismus showed a small angle of exotropia of 12 to 16 PD. Our patient showed a small angle of esotropia, which converted to a small-angle exotropia with full correction of the hyperopic astigmatism. She also showed a mild limitation of abduction in both eyes, which was too mild to meet criteria for paralysis of the sixth cranial nerve reported in Kabuki syndrome. She did not show any evidence of stereaoeuity; however, she showed a fusion response with the Worth four-dot test. The patient’s low intelligence may have contributed to the poor stereoaucuity, but the possibility of the effect of longstanding esotropia on stereopsis could not be denied. This case illustrates the need for patients with a diagnosis of Kabuki syndrome to have an ophthalmologic evaluation. Strabismus associated with Kabuki syndrome may have a small angle that can be easily overlooked.

Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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