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Case Report

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

Kabuki syndrome is a rare syndrome and is composed of multiple congenital anomalies and mental retardation [1-6]. The frequency of Kabuki syndrome is estimated to be approximately 1/32,000 in Japan [5]. Five cardinal diagnostic criteria are postnatal short stature, mental retardation, skeletal anomalies, dermatoglyphic anomalies, and characteristic facial dysmorphism such as long palpebral fissures, large ears and a depressed nasal tip. In addition, other clinical features, including cardiac and renal malformations, deafness, ophthalmologic anomalies, hyperlaxity (including hip dislocation), missing teeth, frequent infection, feeding difficulties, intestinal malrotation, anorectal anomalies, seizures, and endocrine anomalies, have been reported [1,5]. However, there have been few prior, detailed descriptions of strabismus or stereopsis in Kabuki syndrome patients. There has been only one case report in Korea of Kabuki syndrome that described its ophthalmologic features [6]. More information about the ophthalmologic findings as well as more typical clinical features of Kabuki syndrome might allow easy recognition of this clinical entity. Here, we report a more typical patient of Kabuki syndrome who showed a long eye fissure and eversion of the lateral third of the lower eyelid as well as the strabismus.

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Case Report

A 9-year-old girl with developmental delay was born at 37 weeks gestation and had a birth weight of 2.84 kg. Her presenting height, weight and head circumference were 135.3 cm (10th percentile), 38 kg (50th percentile) and 53 cm (50th percentile), respectively. Past medical history, perinatal history and family history were unremarkable. The patient's intelligence quotient was measured to be 60. Her language was at the level of a 6 year old. Chromosome analysis showed a 46, XX karyotype with a breakage at 3p14.

Ophthalmologic examination revealed an esotropia of 8 prism diopters (PD) at distance and near in the primary position. Ductions and versions showed a mild limitation of abduction in both eyes (Fig. 1). The pupils were isocoric and reactive to light, and the fundoscopic examination was normal. Cycloplegic refraction revealed a hyperopic astigmatism of +1.75 Dsph -0.50 Dcyl × 180A in both eyes. The corrected visual acuity was 20/30 in both eyes. With correction, there were 4 PD of intermittent exotropia at distance and 2 PD of intermittent exotropia for near vision. The pa-

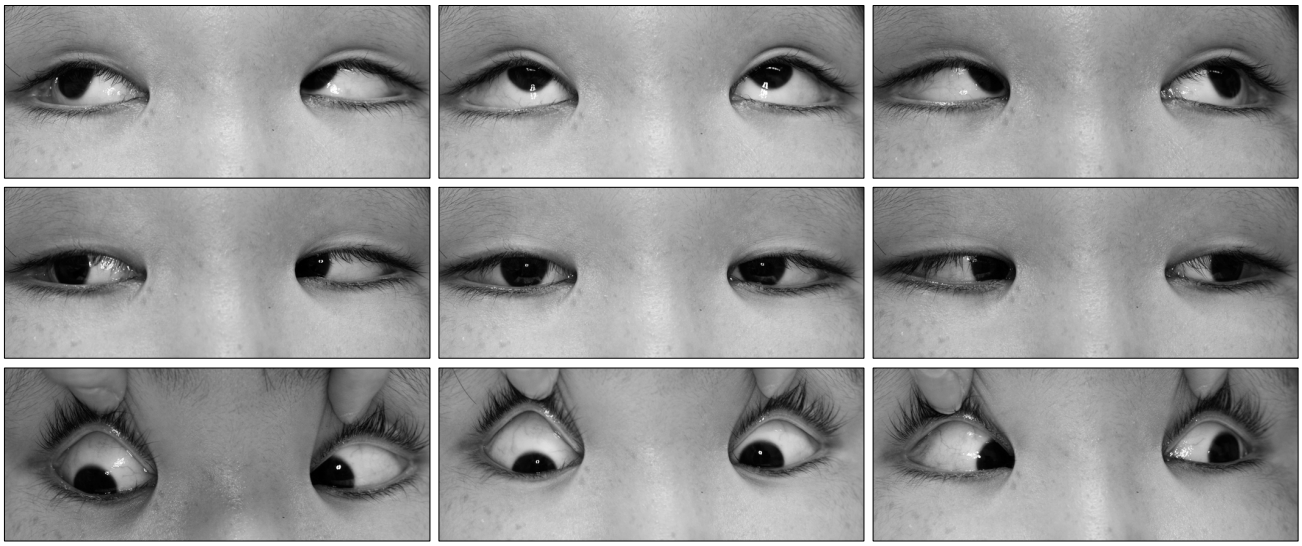

Fig. 1. Photograph of the patient with Kabuki syndrome shows a small-angle esotropia in the primary position. Mild limitations of abduction in both eyes were noted.

tient 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 f issures 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 pigmentation/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 of exotropia after a full correction of hyperopic astigmatism.

The external eye and eyebrow findings are important diagnostic criteria of Kabuki syndrome [5]. The most consistent finding was reported to be a long palpebral fissure, which was most significant at younger ages [7]. Our patient also showed the characteristic long palpebral fissure. Refractive errors such hyperopia, myopia, and astigmatism were reported; therefore, the importance of a full ophthalmologic examination (including refraction) should be stressed, especially for patients who may not communicate their visual difficulties. Any potentially reversible causes of reduced vision should be identified and corrected in this group of patients with intellectual difficulties. Nocturnal lagophthalmos (sleeping with eyes open) was reported to be common in patients with Kabuki syndrome; therefore, the possibility of complications such as keratitis or conjunctivitis should be investigated.

Our patient had a mild hyperopic astigmatism and a very small-angle esotropia that switched to a small-angle exotropia with full correction of the hyperopic astigmatism. She also had 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 stereoacuity; however, she showed a fusion response with the Worth four-dot test. The patient's low intelligence may have contributed to the poor stereoacuity, 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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