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Case Report

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A Kabuki syndrome presenting with strabismus: case report

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ABSTRACT

Kabuki syndrome affects many systems and is characterized by a typical facial appearance and is a genetically inherited disease. Ophthalmological involvement is quite common and can be diagnostic. In this article, we aimed to present the ophthalmological findings of our patient with Kabuki syndrome, who was diagnosed and treated based on ophthalmological findings.

Keywords: Kabuki syndrome, strabismus, KMT2D gene defect

INTRODUCTION

Kabuki syndrome was first described in Japan and is a genetically inherited syndrome, also known as Kabuki make-up syndrome or Niikawa-Kuroki syndrome, characterized by a typical facial appearance.[1] Although its inheritance pattern and etiology have not been elucidated yet, it is thought to be inherited in an autosomal dominant manner. Mutations are frequently found in the KMT2D, KDM6A, and MLL2 genes. In addition to the typical dysmorphic facial appearance, bone and skeletal deformities, cardiac anomalies, urinary system anomalies, gastrointestinal system anomalies, growth retardation, and psychomotor retardation may be observed. Ophthalmologic involvement is observed quite frequently and may be diagnostic.[2,3]

In this article, we aim to present the ophthalmologic findings of our patient with Kabuki syndrome, who was diagnosed with ophthalmologic findings and treated.

CASE

A 13-year-old male patient was admitted to our outpatient clinic with ocular misalignment. The best corrected visual acuity (BCVA) was 0.8 (1.50 axis 135) in the right eye and 0.9 (+0.50-1.25 axis 30) in the left eye using Snellen's threshold. Stereopsis was evaluated as an 800-second arc. Distance and near shift were evaluated as 40 prism diopters (PD) esotropia with an alternating on-off test. There was a-1 restriction on outward gaze in bilateral eyes. The patient also had bilateral ptosis. The right eyelid gap was 8 mm, the left eyelid gap was 8 mm, and the and the margin-reflex distance (MRD) was 1 mm right and 2 mm left. Levator function was measured at 11 mm bilaterally. There was a long palpebral fissure, and the laterals of the lower eyelids were turned outward. Lateral parts of the eyebrows were sparse. When we evaluated the anterior segment, the corneas were transparent, with a horizontal diameter of 10 mm and a vertical diameter of 9.5 mm bilaterally. The lens was clear, and no pathology was found in the fundus examination. The axial length measurements of the patient were 22.31 mm in the right eye and 22.28 mm in the left eye.

The patient had strabismus since the age of 2 years, but this condition was initially evaluated as pseudoesotropia, and no treatment was recommended. She used to wear glasses sometimes. The degree of strabismus has increased over the years.

Attentional hyperactivity and mild mental retardation were present. The patient with dysmorphic facial appearance, large prominent ears, and a single kidney was referred to the pediatrics and genetics unit for a multisystemic evaluation. Genetic analysis revealed a pathogenic KMT2D gene defect.

After consecutive follow-up with the patient, a 6 mm retraction operation was performed on the bilateral medial rectus due to esotropia. During the operation, gray-black pigmentation was observed in the sclera, 8mm behind the insertion in the medial rectus trace. The patient's shifts were orthophoric at the 2nd week and 3rd month postoperatively. No intervention was performed for ptosis or valve anomalies (Figure).

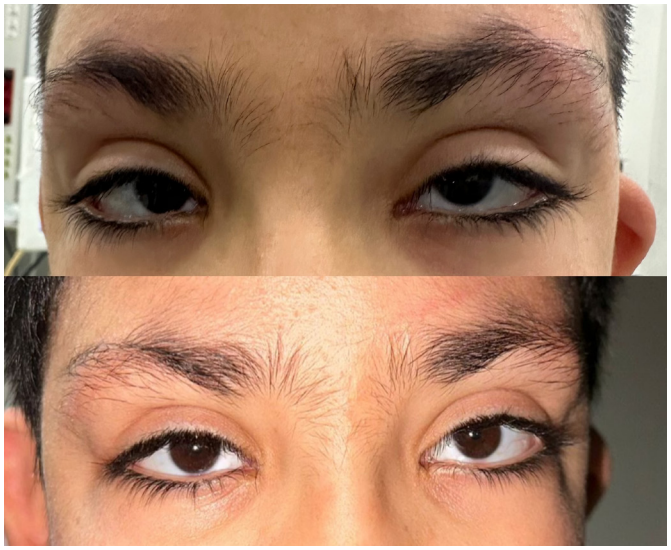

Figure. Picture of the patient before and after the operation

DISCUSSION

Kabuki syndrome is a multi-systemic, genetic disease characterized by mental retardation, developmental delay, musculoskeletal anomalies, and a typical facial appearance. The incidence in the community is estimated to be 1 in 32 thousand.[1] The characteristic facial appearance observed in Kabuki syndrome has been defined as long palpebral fissures, eversion of the lower eyelids, prominent eyelashes, a more prominent brow arch, sparseness of the eyebrows, flattening of the nasal tip, and remarkably large ears.[4] In ophthalmologic involvement, ptosis, esotropia, microcornea, corneal opacity, cataract, nystagmus, blue sclera, iris, retina, choroid, and optic disc coloboma, retinal telangiectasia, and pigmentation may be observed.[5] Other findings include cardiovascular system anomalies, gastrointestinal system and renal anomalies, autistic findings, and behavioral disorders.[6] Although the first striking feature in the diagnosis is the characteristic facial appearance, not all patients with this facial appearance are diagnosed with Kabuki syndrome. An important part of the components constituting this facial appearance is ophthalmologic. Şenel et al.[7] reported a case in which the diagnosis was based on nystagmus, coloboma, and microphthalmia findings. In our case, strabismus, microcornea, long palpebral fissures, ptosis, and eyelid disorders were present.

De novo or inherited pathogenic variants in the KMT2D gene are the most common cause of Kabuki syndrome, accounting for approximately 75% of patients. Mutations in the KDM6A gene account for approximately 5% of patients. Kabuki syndrome is very rare in Turkiye, and in a recent study by Usluer et al.,[8] a KMT2D mutation was found in 21 of 32 patients (65%) who were diagnosed with Kabuki syndrome.[2,3] Our patient's typical facial appearance, ophthalmologic findings, existing renal anomaly, and KMT2D gene mutation found in genetic analysis supported the diagnosis of Kabuki syndrome.

Strabismus is the most common ophthalmologic finding in children with Kabuki syndrome, and its frequency was found to be approximately 20% in a study by E. Ming et al.[9] in our patient, esotropia and limitations of outward gaze movement in both eyes were observed on examination at presentation. It was thought that the limitation in outward gaze was due to esotropia, which was present for a long time.

CONCLUSION

After strabismus surgery due to esotropia, we achieved an orthophoric appearance in the eyes, although we could not achieve a significant improvement in stereopsis due to the advanced age of the patient. In conclusion, ophthalmologists may assume the initiating role in the diagnosis of Kabuki syndrome, and the ophthalmologist's treatment approach to the existing findings plays an important role in improving the quality of life of patients with Kabuki syndrome. However, patients with typical examination findings should be referred to other related branches, and a multidisciplinary approach should be organized.

ETHICAL DECLARATIONS

Informed Consent

The patient's parents signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

The authors declared that this study has received no financial support.

Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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