Kabuki Syndrome: A Rare Clinical Presentation

Sir,
Kabuki makeup syndrome was described in Japanese children by Niikawa et al. based on the characteristic faces resembling kabuki play artist. Diagnosis is based on the criteria laid down by International Consensus diagnostic criteria by Adam et al. MLL-2 and KDM6A gene mutations have been identified in 70% of the patients. Few case reports are available from India.

Sixteen-year-old male presented to the Department of Neurology with seizures for 2 years. The patient is a firstborn child born of a nonconsanguineous marriage. Perinatal and neonatal period was uneventful. No history of recurrent infections in childhood. Seizures were right upper limb focal with secondary generalization and are well controlled with carbamazepine for 6 months. No other significant past or personal history. The patient was poorly built with short stature. The patient was intact mentation. Bilateral lower motor neuron facial weakness with mild quadripareisis was noted. He had generalized hypotonia with no long tract signs. Other clinical features on general examination included long palpebral fissure with arched eyebrows, mild ptosis, and long eyelashes. He had prominent ears with depressed nasal tip and preauricular pits (Figures 1a and 1b). Oral examination showed him to have a high arched palate, abnormal dentition, and cleft palate (Figure 1c). The fifth digit was short (Figure 1d).

His magnetic resonance imaging (MRI) of the brain and electroencephalogram (EEG) were normal while nerve conduction studies did not show any abnormality. Echocardiography showed him to have a small atrial septal defect (ASD).

Kabuki makeup syndrome (KMS) was described in Japanese children by Kuroki, Niikawa et al. in 1988. The name comes from the makeup of actors in Japanese traditional play “Kabuki” to which the facial features resemble. A definitive diagnosis of KMS was made in the present case based on criteria laid down by International consensus diagnostic criteria by Adam et al. The differential diagnosis in the present case includes Ehlers–Danlos syndrome, IRF6-Related disorders, and Hardikar syndrome. The prevalence of the syndrome in India is not known. However, numerous case reports are available from India about varied manifestations. Schrander-Stumpel et al. reported 29 caucasian patients and reviewed 60 Japanese and 29 non-Japanese patients. The study group reported similar clinical presentation in both groups. Neurological
manifestations were present in over 80% of the non-Japanese patients. Congenital cardiac defects are present in 58% of the patients. The present patient presented with seizures which is a rare clinical presentation (17%). The identification of MLL-2 and KDM6A gene mutation emphasized the role of genetic studies in a suspected case of KMS. However, in 30% of the cases, no gene mutation was identified.

The present case, to our knowledge, is the first case reported from India presenting primarily with seizures and perhaps, the first case from central India.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

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Submitted: 23-Oct-2019 Revised: 27-Nov-2019 Accepted: 15-Dec-2019 Published: 05-Jun-2020

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DOI: 10.4103/aian.AIAN_541_19