JAPANESE MAKE-UP WITH HEARING LOSS

MM Patil, SS Kalyanshettar, SV Patil
Department of Paediatrics, BLDE University's Shri BM Patil Medical College Bijapur, Karnataka. India

Address for Correspondence: Dr MM Patil, Associate Professor, Department of Paediatrics, Shri BM Patil Medical College Bijapur 586103, Karnataka, India. E-mail: mmp076@gmail.com

A twelve year old male child born by non-consanguineous marriage, 3rd in birth order presented with scholastic backwardness. He had recurrent right ear discharge and mouth breathing of 1 year duration. Anthropometry revealed stunting. Sexual Maturity Rating (SMR) was stage III. He had large prominent ears, arched eyebrows with sparseness on lateral one third and long palpebral fissures

with eversion of the lateral portion of the lower eyelid (Fig 1). Oropharyngeal examination revealed adenoid hypertrophy with crowding of teeth. There was no cleft palate. Ear examination revealed healed perforation in the right tympanic membrane. Audiometric examination revealed sensory neural hearing loss (SNHL) in left ear and mixed hearing loss in the right ear. Chest x-ray was normal. Echocardiogram revealed atrial septal defect (ASD). Adenoids were surgically removed along with tonsils under general anaesthesia.

What is the diagnosis?

Kabuki Make-Up syndrome. It is a multiple congenital anomaly , mental retardation syndrome. (1) It is currently known in the literature as Kabuki syndrome: the 'make-up' portion of the original name has been discarded secondary to concern that it might cause parental confusion or offense. (1) It is characterized by peculiar facial features like elongated palpebral fissures with eversion of the lateral third of the lower eyelid, arched and broad eyebrows resembling the makeup of actors in Kabuki, the traditional Japanese theatre. (2) It is also associated with mild to moderate intellectual disability. Otitis media and conductive hearing loss are some of the otolaryngologic findings reported in the western literature. (3) Other manifestations like dysmorphic pinnae, hearing loss and airway problems are also known. (3)

In Japan where it is more frequent, it affects 1 in 32000 newborns. Molecular genetic testing for MLL 2 the only gene in which mutation are known to cause Kabuki syndrome is available. (4, 5)

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