A 4 years 6 months old male child born of non-consanguineous marriage presented with dysmorphic features excessive tearing from eyes. He had been operated for cleft palate. His development and birth history were normal. On examination, he had alopecia, epiphora, increased salivation, hypertelorism, low set ears, loss of eye lashes, bald tongue, oral leukoplakia, dystrophic nails, dental caries, loss of teeth, hypoplastic nipples, kyphosis and telangiectatic erythematos.

What is the diagnosis?

Dyskeratosis congenita. It is an inherited multisystem disorder characterized by mucocutaneous abnormalities, bone marrow failure and predisposition to cancer and myelodysplastic syndrome. The diagnostic mucocutaneous (ectodermal) triad is reticulate skin pigmentation of upper body, mucosal leukoplakia and nail dystrophy. About 73 percent patients are males compatible with X-linked recessive inheritance. The remainder has either an autosomal dominant or autosomal recessive mode of inheritance. (1) The X-linked recessive form maps to Xq28, and many mutations have been identified in the DKC1 gene, which codes for the nuclear protein dyskerin. Because of impaired telomere maintenance in all 3 inherited forms, short telomeres are demonstrated in the peripheral blood cells of all patients and are a cardinal marker for marrow failure. Androgens combined with low-dose prednisone, can induce improvement of marrow function in approximately 50 percent of patients. Allogeneic hemopoetic stem cell transplantation corrects marrow failure, but with only a 50 percent survival rate. (1) 

References


E-published: January 2013.

JAPANESE MAKE-UP WITH HEARING LOSS
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A twelve year old male child born by non-consanguinous 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)

REFERENCES