VIEWERS CHOICE

Moebius Syndrome

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A full term, small for gestational age baby born of non-consanguineous marriage with birth weight of 1.6 Kg had dysmorphic facies like malformed right ear with underdeveloped external auditory meatus. There was facial asymmetry with deviation of angle of mouth to left, absent nasolabial fold and hypoplasia of tongue on right side. There was also inability to close the eye on right side. Based on these findings, a diagnosis of Moebius syndrome was made. There were no other congenital abnormalities found in the baby. CT brain and echocardiography were normal.

Figure 1: Shows assymetrical facies due to facial nerve palsy with deviation of angle of mouth to left, obliteration of nasolabial fold and inability to close right eye.

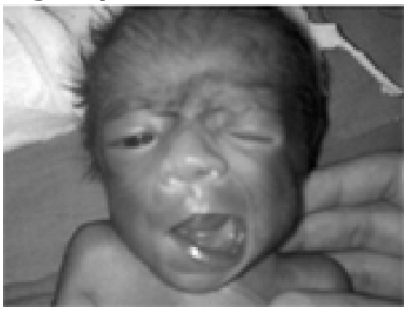
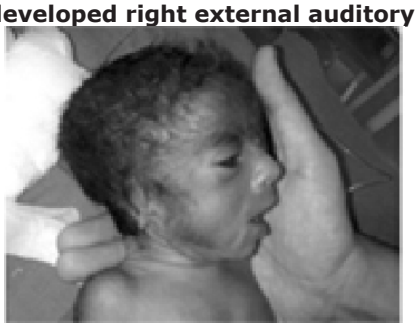


Figure 2: shows malformed right ear with underdeveloped right external auditory meatus



In 1888, Moebius first described the association of facial diplegia with other malformations like multiple cranial neuropathies, mental deficiency, endocrine disorders, musculoskeletal deformities and cardiovascular defects. Moebius syndrome is an extremely rare condition characterized by complete or partial facial nerve paralysis, bilateral or unilateral. Other cranial nerve palsies commonly seen are VI, XII but IX and X are also seen. First symptom may be inability to suck. Other symptoms involves feeding, swallowing, chocking problems; inability to smile, drooling of saliva, inability to close eye on one side, asymmetrical crying facies and mask like facies. Other associated abnormalities include orofacial malformations, ear anomalies, musculoskeletal deformities and limb malformations. Orofacial abnormalities include high or cleft palate, tongue or jaw abnormalities like fissured tongue, microglossia or micrognathia (1). Ear abnormalities include malformed external ear and external auditory meatus. Limb abnormalities include clubfoot, syndactaly, brachydactly, absent digit or talipes or radius or ulna. Aplastic or hypoplastic latissimus dorsi, serratus anterior, intercostals muscles or pectoralis major has also been reported. Ten-15% patients have mental retardation and 30-40% have autistic behavior. Cardiac anomalies include dextrocardia, dextrocardia with ventricular septal defect, transposition of great vessels, ventricular septal defect and supracardiac total anomalous pulmonary venous return (2).

Etiology of Moebius Syndrome remains unknown. Moebius syndrome with its wide spectrum of congenital malformations points to some disturbance during 4th -7th gestational week during which cranial nerve nuclei differentiation and organogenesis for other body part including heart occurs (3). Association of all these defects supports the vascular theory of embryopathogenesis. According to it cranial nuclear necrosis due to an insufficient blood supply is the cause of Moebius syndrome. The interruption of the development of subclavian artery and its tributaries like the basilar, vertebral and internal thoracic arteries at or around sixth intrauterine week causes vascular necrosis of the nerve nuclei (4). The regression of the primitive trigeminal artery supplying the hindbrain before the establishment of vertebral or basilar artery may also impair the cranial nerve nuclei development. Facial weakness with cardiovascular anomalies gives indirect evidence for the vascular pathogenesis of Moebius anomaly. Mesodermal dysplasia of the 1st and 2nd branchial arches and drugs like misoprostol, benzodiazepines and cocaine have also been implicated as causes in certain studies.

Although most of the cases of Moebius Syndrome are sporadic; autosomal recessive, autosomal dominant, and X-linked recessive inheritance pattern have also been described (5). Reciprocal translocation between chromosome 1 and 13, t (1p34; 13q13) has been described in seven members of a family over three generations with congenital facial diplegia, finger flexon deformities and mild mental retardation (6)

There is no specific course of treatment for Moebius syndrome. Treatment is supportive and in accordance with symptoms. Infants require feeding tubes or special bottles to maintain sufficient nutrition. Surgery may correct crossed eyes and improve limb and jaw deformities. Physical and speech therapy often improves motor skills and coordination, and leads to

better control of speaking and eating abilities. Plastic reconstructive surgery may be beneficial in some individuals. Nerve and muscle transfers to the corners of the mouth have been performed to provide limited ability to smile. Neuropsychological and intelligence testing should be done to detect learning difficulties, autism and visual apraxia.

References:

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