Short Communication

Goldenhar syndrome: a report of rare case with subtle clinical features

Sir, a 10-year-old girl presented to us with painless nodule on the cheek since birth. Though the lesion was static and asymptomatic, she wanted it to be removed as it posed a significant cosmetic concern. There was no history suggestive of deafness, blurred vision, convulsions or scholastic backwardness. She was born to second-degree consanguineous marriage and her mother had no perinatal or antenatal complications. There was no maternal illness or drug intake during pregnancy. All other family members including two siblings were normal. On examination patient was found to have few skin coloured papules on both preauricular areas and soft nodule of size 1cm x 1cm on left cheek (**Figure 1**). These lesions were identified as accessory tragi especially the one located on left cheek as the structure suggestive of cartilage could be felt on palpating with the pulp of the fingers. She also had low set ears (appreciated more on the right side), mild submucous cleft in hard palate which was appreciated on palpation as a groove. Ocular examination revealed yellowish soft noninflamed mass in the right eye identified as epibulbar dermoid at the inferotemporal limbus. There was no redness or discharge from the eyes and ocular motility was normal bilaterally. Her visual acuity was normal 6/6 and there was no evidence of coloboma or microopthalmia. Examination of respiratory, cardiovascular, central nervous system revealed no abnormality. The patient's hemogram and blood investigations were normal. Radiological investigations in the form of X-rays of chest and of lumbosacral spine and ultrasonography of abdomen and pelvis revealed no abnormality. Pure tone audiometry did not show any evidence of deafness. ECG and 2D echo of heart were normal. The diagnosis of Goldenhar syndrome was made on the basis of epibulbar dermoid, accessory tragi and the presence of cleft palate. Accessory tragi were surgically removed under the guidance of paediatric surgeon. Cleft palate was left untreated as patient had no history of snoring, difficulty in breathing or swallowing. Currently she is following up with ophthalmologist for the treatment of epibulbar dermoid.

Figure 1 Accessory tragi on left side of face.

Goldenhar syndrome (also known as oculoauriculo vertebral dysplasia and hemifacial microsomia) is a rare congenital disorder which manifests as ocular, auricular, dental, vertebral and systemic abnormalities.[1] It occurs in 1 per 5800 births with male: female ratio being 3:2.[1] Numerous hypotheses have been invoked to explain the etiopathogenesis of this syndrome. The reported cases illustrate causal heterogeneity of the Goldenhar complex. Experimental observations have shown that destruction of differentiating tissue in the region of ear and jaw by an expanding hematoma will produce brachial arch dysplasia. Severity of the dysplasia is related to the degree of local destruction which would help explain the syndrome's variability of expression.[2] Most of the cases are sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested.[3] The

aetiology of Goldenhar syndrome is not well established. It is thought to be due to exposure of various viruses (rubella and influenza) and chemicals (thalidomide, retinoic acid, tamoxifen and cocaine) during pregnancy. Some researchers also suggested diabetes mellitus as one of the cause. However, in our patient there was no history of maternal drug intake, febrile illness or diabetes during pregnancy. Goldenhar syndrome is associated with wide range of features like macrostomia, micrognathia, cleft palate, bifid tongue and malocclusion, vertebral abnormalities, facial muscle hypoplasia, neurological, visual, cardiac and genitourinary abnormalities. However, the classic features of this syndrome include ocular changes such as micropthalmia, epibulbar dermoids and coloboma; aural features such as preauricular tragi, hearing loss and microtia; and vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion.[4] Our patient had accessory tragi as a presenting feature which can also be found in various other syndromes like Trecher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome, Townes-Brocks Syndrome and Delleman syndrome.[5] However, our patient also had classical limbal dermoid and mild cleft palate which confirmed the diagnosis of Goldenhar syndrome. Successful treatment requires a multidisciplinary approach involving otolaryngologist, ophthalmologist, paediatrician, dermatologist and orthopedician. The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. Prognosis of the disease is good in otherwise uncomplicated cases without any systemic associations. To conclude, extreme variability is a characteristic feature of this disease and patients presenting with subtle clinical features might remain without correct diagnosis and proper management for a very long period. The present case is being reported to create awareness on various clinical features of this entity and increase the chances of early diagnosis.

References

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