

## 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 microphthalmia. 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



**Figure 1** Accessory tragi on left side of face.

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.

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.<sup>1</sup> It occurs in 1 per 5800 births with male:female ratio being 3:2.<sup>1</sup> 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.<sup>2</sup> Most of the cases are sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested.<sup>3</sup> 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 microphthalmia, epibulbar dermoids and coloboma; aural features such as preauricular tragi, hearing loss and microtia; and vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion.<sup>4</sup> 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.<sup>5</sup> 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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## Breast carcinoma presenting as carcinoma en cuirasse

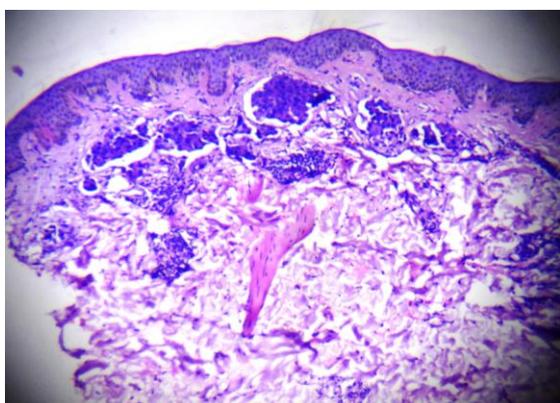
Sir, carcinoma en cuirasse is a form of cutaneous metastasis. Although this condition is rare, it is most commonly associated with breast carcinoma with local recurrence after mastectomy. It presents most commonly a few months or years after the surgical operation or rarely, the condition presents as the presenting manifestation of the disease. It is of diagnostic importance because it may be the first manifestation of hitherto undiscovered internal



**Figure 1** Erythematous indurated plaques on neck and breast with diffuse edema of left hand.



**Figure 2** Indurated plaques on neck and retroauricular areas.



**Figure 3** Groups of malignant cells suggestive of cutaneous deposits of ductal carcinoma.

**Table 1** Cutaneous manifestations of breast carcinoma described in the literature.

<i>Clinical morphology</i>	<i>N (%)</i>
Nodular carcinoma	141 (46.8)
Alopecia neoplastica	36 (12)
Telangiectatic carcinoma	25 (8)
Malignant melanoma-like metastasis	19 (6.3)
Carcinoma ersipelatoides	19 (6.3)
Subungual metastasis	14 (4.6)
Carcinoma <i>en curasse</i>	13 (4)
Zosteriform metastases	11 (3.6)
Metastasis to eyelids	7 (2.3)
Paget-like metastases	4 (1.3)
Dermatomyositis	3 (0.9)
Granuloma pyogenic-like metastases	2 (0.6)
Multicentric reticulohistiocytosis	1 (0.3)
Sister Mary Joseph's nodule	1 (0.3)
Metastatic squamous cell carcinoma	1 (0.3)
Occult metastasis	1 (0.3)
Infection-like acral cutaneous metastasis	1 (0.3)
Clown nose	1 (0.3)
Targetoid cutaneous metastases	1 (0.3)
Nodule on benign intradermal nevus	1 (0.3)
Erythema annulare centrifugum-like	1 (0.3)
Dermatitis-like metastases	1 (0.3)
Calciophylaxis	1 (0.3)

malignancy or the first indication of an inadequately treated malignancy.

We report a case of a 35-year-old female patient who presented with indurated skin lesions over the breast, the axilla and the chest wall. She presented with 6 months history of multiple skin lesions over the left breast, left shoulder and the left side of the upper back, extending to involve the retroauricular region and the neck. She also had swelling of the left upper limb for duration of one month. The lesions initially started as asymptomatic papules and plaques on the neck and retroauricular areas which increased in number and size to involve chest and back of the same side. She also gave history of difficulty in having full range of motion of the left shoulder joint. There was no history of anorexia or weight loss. She denied any dermatological diseases and her personal history was unremarkable.

On examination, multiple indurated nodules and plaques were present over the left breast,

the left retroauricular area, the scapular region and the neck (**Figure 1, 2**). Multiple grouped vesicles on an erythematous base were seen over the left breast and the neck. Breast examination revealed a hard non-tender mobile swelling of 3-4 cm over the left breast. Posterior axillary lymph nodes were significantly enlarged and there was diffuse swelling of the left upper limb. On the basis of cutaneous and breast examination findings, a provisional diagnosis of carcinoma en cuirasse was made and diagnosis was confirmed with skin biopsy which showed groups of malignant cells suggestive of cutaneous deposits of ductal carcinoma (**Figure 3**).

Cutaneous metastasis is a phenomenon that results from a tumor spreading via lymphatic or vascular embolization, direct implant during surgery or skin involvement by contiguity. The primary malignant tumor that most commonly metastasizes to the skin in women is breast cancer, which can be manifested through wide range of clinical features (**Table 1**).<sup>1,2</sup>

The incidence of cutaneous metastasis varies from 6% to 10%. Carcinoma en cuirasse is a form of metastatic cutaneous carcinoma. It is usually seen in patients with carcinoma breast who have undergone mastectomy, but rarely this can be the primary presentation of carcinoma breast (similar to our case).<sup>3</sup>

*It was* first described by Velpeau in 1838, a description chosen because of its resemblance to the metal breastplate of a cuirassier. It has also been called scirrhous carcinoma, pachydermia and *Acarcine eburnee* by various authors.<sup>4</sup>

In our case the patient only had skin lesions and was unaware of underlying malignancy. She sought the medical help only after lesions

progressed to entire half of the trunk and caused restriction of shoulder movement. Cutaneous metastasis always indicates a grave prognosis and early diagnosis can make a big difference in the management and subsequently in prognosis. Carcinoma en cuirasse may be the first clinical manifestation of internal malignancy especially carcinoma breast. So it is the responsibility of the dermatologist to have high index of suspicion in patients presenting with cutaneous features suggestive of metastasis and manage accordingly.

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