release of noradrenaline is involved, since adrenergic blocking drugs prevent relaxation (Bucknell and Whitney, 1965). One possibility is that noradrenaline exerts an inhibitory effect on the ganglia (Norberg, 1964 ; Jacobowitz, 1965). This idea is consistent with the presence of α-adrenergic receptor sites within neurones in the gut (Kosterlitz and Watt, 1965). Another possibility involves non-adrenergic inhibition, which has been found in animal and human intestine (Burnstock *et al.*, 1964 ; Bucknell, 1966). Some of the ganglia might take part in this inhibition and their absence would prevent the colon from relaxing.

SUMMARY

The histological distribution of adrenergic myenteric nerves has been studied by the catecholamine fluorescence technique in large bowel resected from young children, five because of Hirschsprung's disease and two for other reasons. Normal large bowel showed a dense arrangement of adrenergic nerves around myenteric ganglion cells. None of the ganglion cells contained catecholamines. Few adrenergic nerves were seen in the muscle layers. In Hirschsprung's disease the myenteric ganglion cell complex was absent from the distal segment of bowel, but increased numbers of adrenergic nerves were present in the muscle layers. The large nerve bundles which are characteristically seen in the intermuscular zone in this disease were non-fluorescent, but some medium-sized bundles were fluorescent. Transitional tissue, which shows features intermediate between this and the normal bowel, was seen in three cases. These findings indicate that inability of the affected segment to relax is not due to an absence of adrenergic nerves.

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Medical Memoranda

Goldenhar's Syndrome

Brit. med. J., 1968, **1**, 489–490

Goldenhar (1952) reviewed a symptom-complex of ocular, aural, and vertebral defects. This is usually referred to as the oculo-auriculovertebral syndrome of Goldenhar. Though this syndrome is rare, a number of cases have now been reported. Certain characteristic manifestations have been described and incomplete cases are also found, such as those without vertebral anomalies (Sugar, 1966) and those without the auricular anomalies (Weyers and Thier, 1958) ; these probably all belong to the same symptom-complex. The case reported is another fairly typical example of this syndrome.

CASE REPORT

This male infant was born at term after a normal pregnancy and labour. He weighed 7 lb. 15 oz. (3,600 g.) at birth. His parents were healthy (father aged 25 and mother aged 21). There was no history of any maternal illness during pregnancy. When first examined the face was asymmetrical ; there were multiple skin tags on the right side (Fig. 1) and two preauricular tags on the left side (Fig. 2). The lower jaw was maldeveloped and the right ramus of the mandible could not be palpated. The right external meatus was occluded by skin. There was a coloboma of the left upper eyelid at the junction of the middle and inner thirds. Both pupils were small and the corneae appeared to be large. Radiological examination confirmed the absence of the right ramus of the mandible and the lateral third of the zygomatic arch (Fig. 3). The vertebral column was normal. Examination under anaesthesia at 3 months of age confirmed that both corneae were large (11 cm.), but the

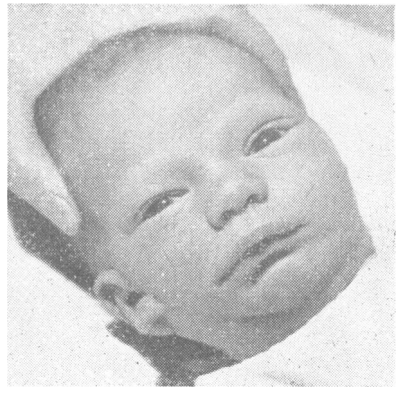

FIG 1.—Skin tags on right side of face and coloboma in left upper eyelid.

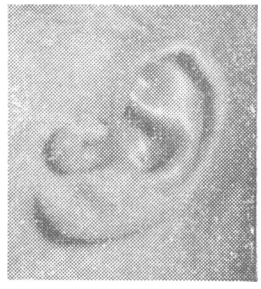

FIG. 2.—Left preauricular skin tags.

intraocular pressures and optic discs were normal. There was no coloboma of the iris or choroid. In the succeeding months the facial asymmetry became more pronounced. The preauricular and facial tags were removed on the eighth day and the coloboma will be repaired at a later date.

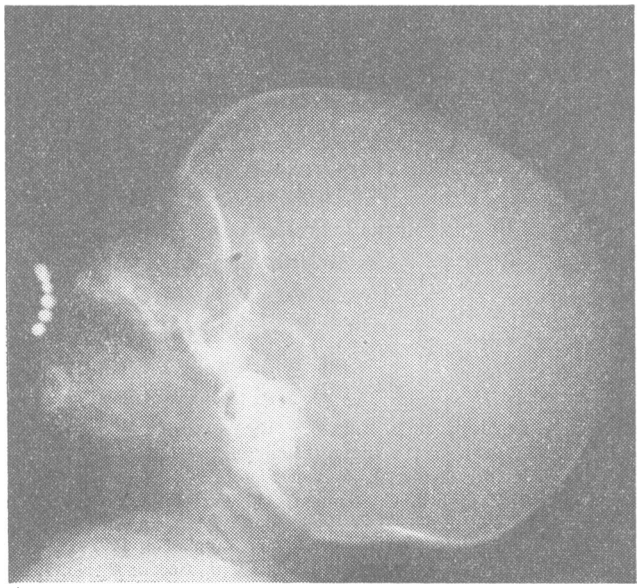

FIG. 3.—Radiograph showing cystic appearance of the left mandible.

DISCUSSION

In Goldenhar's original cases the ocular signs included dermoids and lipodermoids. Sugar (1966) described these as the most important of the ocular features, being bilateral in two-thirds of the cases. The dermoid is usually found in the lower outer quadrant and the lipodermoid in the upper outer quadrant. The next commonest ocular abnormality is unilateral coloboma of the upper lid ; this was present in our patient, at the characteristic site of the junction of the inner and middle thirds. Bilateral colobomas are rare, and the coloboma in the upper lid is an important feature in differentiating this syndrome from mandibulofacial dysostosis, when the coloboma is usually in the lower lid (Gorlin and Pindborg, 1964). Other ocular anomalies are rare and include microphthalmos, microcornea, anophthalmos, and colobomas of the iris and choroid.

Our patient showed the characteristic finding of bilateral preauricular appendages. These are usually multiple and situated in front of the tragus. Blind fistulas are occasionally found in the same situation. Microtia and deafness, with or without disturbances of the external auditory meatus, have also been described. Vertebral anomalies are usually manifest only by *x*-ray examination and were not present in our case ; numerous anomalies have been described, including cuneiform vertebrae, occipitalization of the atlas, supernumerary thoracic or lumbar vertebrae, and anomalous ribs.

The facial appearance is often striking owing to the presence of hemifacial microsomia. This was present to a marked degree in our case, which also showed absence of the right ramus of the mandible and recession of the chin. Other rare facial abnormalities, such as cleft palate, bifid tongue, and cleft lip, have also been reported (Gorlin and Pindborg, 1964 ; Sugar, 1966). In some of these children a mild degree of mental retardation has been described, though most of them are of normal intelligence. It has not yet been possible to assess the mental development pattern of our patient accurately.

No hereditary pattern has been described and the chromosomes are normal (Gorlin *et al.*, 1963 ; Sugar, 1966).

Treatment of these cases consists mainly of cosmetic surgery. The dermoids and lipodermoids can be removed, eyelid colobomas repaired, and the preauricular appendages removed. Lip and palatal deformities can be repaired. Differential diagnosis is mainly from hemifacial microsomia and mandibulofacial dysostosis. These conditions do overlap and hemifacial microsomia often occurs in cases of Goldenhar's syndrome. Paufique *et al.* (1952) have stated that all these syndromes are similar and are due to a gene undergoing mutation. It has been suggested that dermoids and auricular appendages may be part of the first arch syndrome (McKenzie, 1958 ; Schultz *et al.*, 1967). Chromosomes, however, have been normal, unlike the first arch anomalies seen in the trisomies. The position of the coloboma in the upper lid helps to differentiate it from mandibulofacial dysostosis.

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