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# GOLDENHAR SYNDROME WITH MULTIPLE RENAL ANOMALIES—A CASE REPORT

By

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SUMMARY A two year old girl presented with dribbling of urine dating from birth. The clinical picture was that of Goldenhar syndrome. Multiple renal abnormalities which have not been described previously were detected.

## INTRODUCTION

Oculoauriculovertebral dysplasia is a syndrome affecting the eyes and eye lids, ears, facial bones, and vertebrae. Goldenhar[2] in 1952 reviewed the clinical features in detail. The principal features of this syndrome consists of epibulbar dermoids and or lipodermoids, auricular appendices and pretragal blind-ended fistulae, and vertebral anomalies. The epidermal dermoids are usually seen at the limbus or corneal margin in the lower outer quadrant of the eye. Multiple renal anomalies have not been described previously.

## CASE REPORT

S. L., a two year old girl was admitted to General Hospital, Badulla in 1977 with a history of dribbling of urine dating from birth but there was no history of fever. Her motor milestones and speech were delayed. The birth and neonatal history were normal, and there was no significant family history.

The skull circumference was 41.9 cm, and she had facial asymmetry with hypoplasia of the right side. An epibulbar dermoid was seen on the temporal aspect of the right eye. She had marked scoliosis of the dorsolumbar spine. The neck was short with a low hairline. The cardiovascular and respiratory systems were clinically normal. She had continuous dribbling of urine from the urethra, but not from the vagina.

Examination of the urine showed a mild albuminuria, with 2—3 pus cells. The blood urea was 5 m mol/l (30 mg/dl.) The roentgenograms showed a mandibular hypoplasia of the right side and multiple hemivertebrae in the cervical and dorsal spines. Occipitalisation of the atlas was seen with multiple fused vertebrae of the short cervical spine. This gave an appearance of Klippel Feil syndrome. A cervical rib was observed on the left side, and the fourth and fifth ribs were fused posteriorly on the right side. A hemivertebra was seen between D12 and L1 vertebrae. The fourth and fifth lumbar vertebrae were fused, and the alae of the sacrum were poorly developed.

An intravenous urogram showed a large kindney, 9 cm in length, on the left side, and a stretched pelvicalyceal system with hydronephrosis on that side. There was no radiological evidence of a renal shadow on the right side. A micturating cystourethrogram showed third degree vesicoureteric reflux on the left side. The left ureter opened into the vesico-urethral junction.

## DISCUSSION

This syndrome is a symptom complex involving principally the first and second branchial arches. McKenzie[4] (1958) relates the mandibulo-facial malformations to faulty development of the blood supply during critical periods of facial development, with inadequate oxygenation of the embryonic facial tissues, resulting in abnormal bone formation. There is no hereditary pattern.

It is important to differentiate Goldenhar syndrome from Treacher Collins syndrome and hemifacial microsomia. An epibulbar dermoid which was found in this child, and/or lipodermoids are a constant feature of Goldenhar syndrome, while it is not seen in mandibulofacial dysostosis (Treacher Collins) and hemifacial microsomia. Vertebral anomalies almost always occur in Goldenhar syndrome while these are are usually absent in mandibulofacial dysostosis and hemifacial microsomia[3]. More recently, bony anomalies have been reported in hemifacial microsomia.[5]

Darling *et al.* (1968)[1] in their review on the roentgenological aspects of Goldenhar syndrome have described all the bony abnormalities seen in this child, which include unilateral underdevelopment of the face, occipitalisation of the atlas, fused vertebrae, hemivertebrae, aplasia of the sacrum, and anomalous ribs. Mental retardation has been observed in a few patients[3].

Gorlin[3] in his review mentions the occurrence of an anomalous renal artery in one patient, which could be a normal variant. Apart from this, renal abnormalities have not been reported before . This patient's hydronephrosis on the left side is very likely to be due to the third degree vesicoureteric reflux. The ectopic ureteric opening near the bladder neck explains the dribbling of urine.

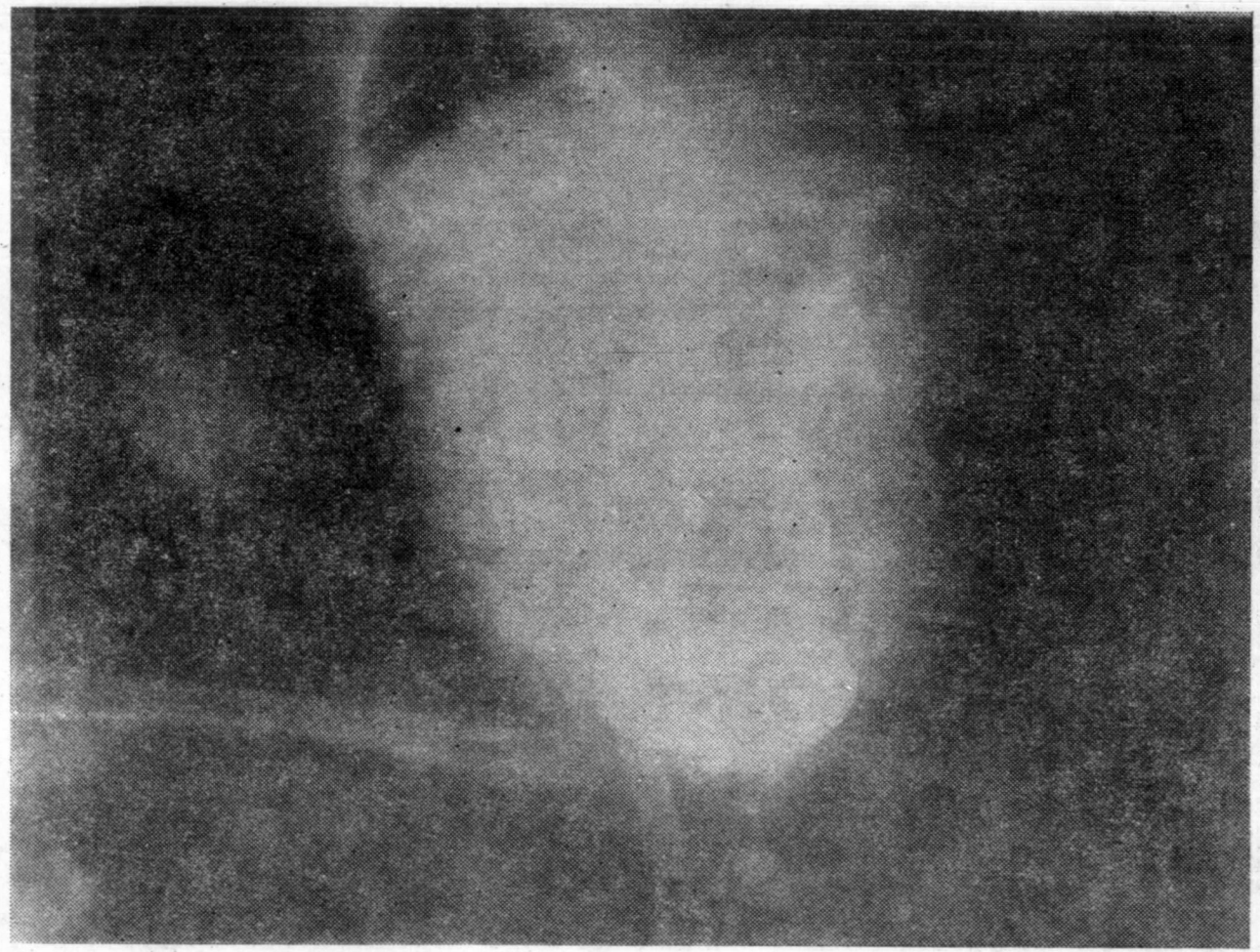

Fig. 1. Cystourethrogram showing third degree vesicoureteric reflux on the left side.

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