Severe Kyphoscoliosis in a Patient with Goldenhar Syndrome: A Case Report

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Keywords
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Abstract
Introduction: Goldenhar syndrome includes a wide spectrum of congenital anomalies involving structures arising from the first and second branchial arches. It is characterized by impaired development of structures such as eyes, ears (with or without hearing loss), lip, tongue, palate, mandible, maxilla, and deformations of vertebrae. The etiology of this syndrome is unclear since it varies genetically and is linked to a plethora of reasons. Case Report: A 13-year-old male presented to the Orthopedic Department with complaints of back bulge with bilateral microtia since birth. X-ray and computed tomography revealed severe congenital kyphoscoliosis. Posterior vertebral column resection surgery with instrumentation and anterior cage support with bone graft fusion were performed. A postoperative plain X-ray film demonstrated a good correction rate. His follow-up was asymptomatic and well balanced in the sagittal and coronal planes at 6 months following operation. Systemic involvement was clinically and
Conclusion: Patients with Goldenhar syndrome due to a large variety of abnormalities and different severity of symptoms pose a challenge for clinicians. All of this necessitates an individual approach to each single patient and involvement of a team of specialists in treatment planning. Although complex scoliosis surgery could be achieved safely in Goldenhar syndrome patients, careful preoperative management is required.

Introduction

Goldenhar syndrome, also known as oculo-auriculo-vertebral dysplasia, is a developmental disorder primarily involving structures derived from the first and second pharyngeal arches during embryogenesis. The phenotype is clinically heterogeneous and is typically characterized by abnormal development of the ear, mandible anomalies, and vertebra abnormalities. It was first described by Dr. Maurice Goldenhar in 1952 [1]. In 1963, Gorlin suggested the name oculo-auriculo-vertebral dysplasia for this condition and he also included vertebral anomalies as signs of this syndrome [2]. The incidence of Goldenhar syndrome has been reported to be 1:35,000–1:56,000. The ratio of male to female was 3:2.2 [3]. Etiology of this condition is not yet fully established. Abnormalities of neural crest cells and chromosomes, and environmental factors during pregnancy like drugs (e.g., cocaine and retinoic acid) were also related to the development of the disease. In addition, intake of alcohol by the mother and maternal diabetes have also been suggested as etiologic factors [4]. Clinically, the patient may present with a variety of features ranging from facial, ear and eye abnormalities, vertebral deformities, to congenital heart problems.

There have been several reports regarding the facial and auricular anomalies in Goldenhar-related conditions [5–7], but very little information is available in the orthopedic literature regarding the associated spine problems. In this article, we report a case of Goldenhar syndrome with severe kyphoscoliosis, along with a discussion on clinical features, the importance of early diagnosis, and systemic involvement, which was assessed clinically and radiographically.

Case Report

A 13-year-old male child presented to the Orthopedic Department with complaints of back bulge with bilateral microtia since birth. The child was born by vaginal delivery at a hospital, with a birth weight of 2.7 kg. He was the first born. There was no history of trauma to the thoracolumbar region. No signs of mental retardation or impairment of cognitive function were seen during examination. There was no history of exposure to known teratogenic agents, drugs, or maternal diseases. The parents were healthy and their marriage was non-consanguineous. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

On physical examination, facial asymmetry and bilateral microtia with bilateral ear tag was observed (Fig. 1). We kept a watchful eye on appearance of the spine and trunk, including shoulders uneven, back bulge, and trunk shift to the left (Fig. 2). He has urogenital abnormalities with fine urine speed and bilateral hearing fell slightly. The diagnosis was established according to the minimal diagnostic criteria proposed by Strömland et al. [8], considering the
combination of vertebral anomaly and ear malformation. He did not suffer from ocular, cardiological or gastrointestinal abnormalities.

His radiographs of the spine demonstrated thoracolumbar kyphoscoliosis. The coronal curve is 40° and thoracolumbar kyphosis had a Cobb angle of 102° (Fig. 3). The trunk shift is 45 mm. This suggests that the patient needs surgical correction. Magnetic resonance imaging revealed no evidence of any spinal cord or canal abnormalities. Computed tomography revealed T12 semi-segment hemivertebra and failure segmentation from L3 to L5.

In June 2017, a posterior T12 vertebral column resection with instrumentation and anterior cage support with bone graft fusion were performed. The fusion level is from T8 to L3, using the premier spinal system. The total operation time was 5 h and 10 min. The total amount of blood loss was 700 mL. During the operation, the signal of this patient was normal using intraoperative spinal cord monitoring. The wake-up test was also negative. Postoperatively, there was no sign of neurological impairment. The patient has been kept on regular follow-up and observation for spine malformations.

Results

A postoperative plain X-ray film demonstrated Cobb angles of the thoracolumbar scoliosis correction from 40 to 20° (correction rate 50%) and the Cobb angles of the thoracolumbar kyphosis were between 102 and 16° (correction rate 84%) (Fig. 4). His follow-up was asymptomatic and well balanced in the sagittal and coronal planes at 6 months following operation (Fig. 5). Both the patient and his parents were satisfied with the results of the surgery.

Discussion

Spine deformities occurring during the mesenchymal stage may be due to either a unilateral defect of formation or segmentation of the primitive vertebrae. It can result in a unilateral imbalance in the growth of the spine, which produces congenital scoliosis. On rare occasions, these vertebral abnormalities may be associated with a unilateral failure of formation of the face and ear. This combination of anomalies is also described as hemifacial microsomia. The exact origin of Goldenhar syndrome is unknown. The most widely accepted theory is that hemifacial microsomia is the result of a disturbance in the embryological development of the first and second branchial arches during the first 6 weeks of gestation [9]. During these first 6 weeks of embryological development, both the skull and spine are formed. Therefore, a common pathogenic mechanism is likely to be the basis of both craniofacial and vertebral malformations in patients with hemifacial microsomia [10].

The nature and extent of vertebral anomalies in Goldenhar syndrome patients were reported in previous studies. Tsirikos and McMaster [10] reviewed 668 consecutive patients with congenital scoliosis. They found that the prevalence of hemifacial microsomia was 2%. A thoracic scoliosis was the most common deformity. Of these patients, 8 had an isolated hemivertebra, and the remaining 2 had a unilateral unsegmented bar with contralateral hemivertebra at the same level. A thoracolumbar kyphoscoliosis occurred in only 1 patient and was caused by a posterolateral quadrant vertebra. They considered hemifacial microsomia as a baseline deformity in patients with Goldenhar syndrome. However, torticollis and cervicothoracic scoliosis were the major deformities in another study. Thoracolumbar scoliosis and kyphoscoliosis were of lesser occurrence [11]. Anderson and David [12] reported a wide range...
of spinal and rib anomalies in cases of Goldenhar syndrome. The anomalies occurred with almost equal incidence in the cervical and thoracic spine of those who had whole spine assessment. The lumbar spine was less commonly affected. In our study, posterolateral semi-segment hemivertebra in thoracolumbar region were the critical factor of severe kyphoscoliosis.

Treatment of angular and rigid kyphosis is a challenge for spine surgeons. Osteotomies were needed in this severe kyphoscoliosis along with a high risk of neurologic deficit. In patients with more severe and complex deformities needing posterior vertebral column resection surgery, neurologic complications were reported to be as high as 17.1%, with 3.3% permanent neurologic deficits [13]. In the present study, the case was examined by an experienced surgeon. Fortunately, there was no sign of neurological impairment after the surgery.

**Conclusion**

Patients with Goldenhar syndrome, due to a large variety of abnormalities and different severity of symptoms, pose a challenge for clinicians. All of this necessitates an individual approach to each single patient and involvement of a team of specialists in treatment planning. Although complex scoliosis surgery could be achieved safely in Goldenhar syndrome patients, careful preoperative management is required. When performing surgery on scoliosis patients with Goldenhar syndrome, surgeons and anesthesiologists should take into account the associated airway management.

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**Statement of Ethics**

The authors have no ethical conflicts to disclose.

**Disclosure Statement**

The authors have no conflicts of interest to declare.

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**Author Contributions**

X.X. analyzed and collected the patient imaging data, and was a major contributor in writing the manuscript. S.Z. performed the revision surgery. The authors read and approved the final manuscript.
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Fig. 1. Images showing facial features of the Goldenhar syndrome: facial asymmetry and bilateral microtia with left ear tag.

Fig. 2. Appearance of the Goldenhar syndrome before the operation: shoulders uneven, back bulge, and trunk shift to the left.
Fig. 3. Standing anteroposterior and lateral radiographs and computed tomography scan image before the operation.

Fig. 4. Standing anteroposterior and lateral radiographs initially following the operation (a, b) and 3 months following the operation (c, d).
Fig. 5. Appearance of the Goldenhar syndrome 6 months after surgery: good balance in the coronal and sagittal view.