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Case Report

Severe scoliosis and restrictive lung disease associated with Goldenhar syndrome: A case report

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ABSTRACT

Goldenhar syndrome is a rare congenital condition associated with a wide range of phenotypic variations resulting structural abnormalities in the face, eyes, ears and various organs. Primary findings are vertebral abnormalities which are mainly localized in cervical and thoracic regions. Scoliosis may develop as a result of vertebral abnormalities and can cause important clinical outcomes and disability. Diagnosis is usually made by physical examination and radiological imaging; however, genetic consultation may be required to confirm the diagnosis. Treatment protocol varies according to age and the severity of clinical manifestations.

Keywords: Congenital, Goldenhar syndrome, restrictive lung disease, scoliosis.

Goldenhar syndrome (GS) (MIM 164210), also known as oculo-auriculo-vertebral spectrum or hemifacial microsomia, is characterized by facial asymmetry and accompanying structural anomalies of varying severities in the ears, eyes, heart, central nervous system and urogenital system.[1] Its prevalence is estimated to be 1 in 3,500 to 5,600 live births, with a male predominance. Unilateral manifestation is most commonly observed on the right side and typically causes dysmorphic facial features appearance.[2,3] Although pathogenesis is still unclear, a multifactorial etiology, including genetic and environmental risk factors, is to be blamed.[4] Gestational diabetes mellitus, alcohol consumption, and teratogenic drugs are listed among maternal causes of the GS. Nevertheless, impairments in neural crest migration and developmental defects of the first and second branchial arches that emerge in the fourth to eighth weeks of pregnancy are most frequently considered etiologies.[5] Hypoplasia in mandibular, zygomatic, temporal bones, cleft lip and palate in the face, auditory canal atresia and microtia in the ear, epibulbar dermoid, iris/ chorioretinal coloboma in the eye, congenital vertebral abnormalities, scoliosis in the spine, tetralogy of Fallot, atrial and ventricular septal defects in the heart, horseshoe kidney and agenesia in the urogenital system are common findings of this syndrome. Although there are no definitive diagnostic criteria, it has been postulated that ear anomalies and facial asymmetry are the cardinal signs for diagnosis. The main goal of the treatment protocol is to correct functional disorders due to craniofacial anomalies, as well as to improve aesthetic appearance.[6]

Early detection of spinal abnormalities decreases the risk of progression and related complications, since early treatment and frequent follow-up can be offered. While there are numerous reports about craniofacial anomalies in the literature, the widespread presentation of vertebral abnormalities and resulting scoliosis are rarely mentioned.[6-8] In this report, we present a case of severe scoliosis and restrictive lung disease associated with GS and emphasize that vertebral abnormalities may lead to severe scoliosis and cause disability.

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CASE REPORT

A 51-year-old male patient was admitted to our outpatient clinic complaining of a progressive back deformity, limited mobility, and shortness of breath with exertion. He was the first child in a non-consanguineous family. His medical history revealed that he underwent scoliosis surgery at the age of 17, but the instrumentation was removed 20 days later due to surgical failure. Since the patient was born and raised in a rural area with limited access to healthcare services, he remained undiagnosed despite having multiple congenital anomalies. A comprehensive evaluation was never performed during childhood.

Physical examination revealed the absence of the right external auditory canal and auricle, along with periauricular skin tags, a limbal dermoid, facial asymmetry, and micrognathia. Although painless, a severe restriction of movement was noted in the cervical, thoracic, and lumbar regions. The finger-to-floor distance was 44 cm. The Adams forward bend test result was positive in both the thoracic and lumbar regions, with the thoracic region displaying a more prominent hump (Figure 1). Using a Bunnell scoliometer, the angle of trunk rotation was 28 degrees in the thoracic region and 16 degrees in the lumbar region. The patient exhibits a coronal imbalance with a lateral deviation of 35.85 mm to the right. The extremely rigid main curve was localized

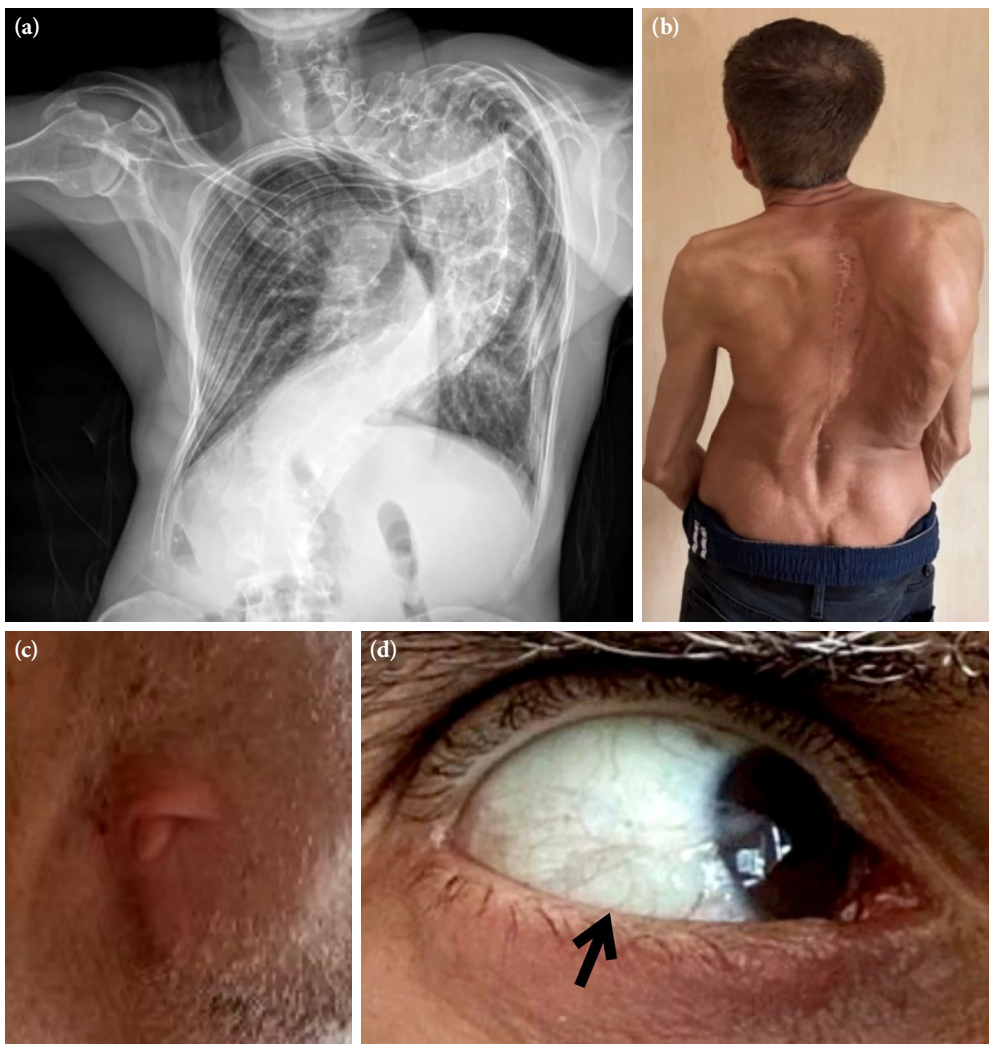

Figure 1. **(a)** X-ray image of the thoracic and lumbar spine showing severe scoliosis. **(b)** Clinical photograph of the patient demonstrating a severe spinal deformity. **(c)** Absence of the right external auditory canal and auricle, along with periauricular skin tags. **(d)** Limbal dermoid (ocular mass typical of Goldenhar syndrome).

in the right thoracic spine with compensatory left lumbar scoliosis. The thoracic Cobb angle was approximately 104 degrees, and the lumbar Cobb angle was 67 degrees. While bilateral patellar and Achilles reflexes were slightly increased, there were no sensory or motor deficits.

Pulmonary function test results were as follows: forced vital capacity (FVC): 30%, forced expiratory volume in 1 sec (FEV1): 34%, and FEV1/FVC: 95%, resulting in a diagnosis of severe restrictive lung disease. The echocardiography demonstrated ascending aortic dilation, bicuspid aortic valve, and mild aortic and tricuspid valve insufficiencies. Other organs and system evaluations or radiological imaging revealed no additional pathologies.

The patient was clinically diagnosed with GS, and considering his age, genetic testing was not performed; however, genetic counseling for his children and future progeny was recommended. Consultations with various specialists, including ophthalmologists, otolaryngologists, cardiologists, pulmonologists, and spine surgeons, were performed to evaluate the systemic manifestations. The patient was found to be unsuitable for a brace due to the extreme and rigid spinal curvature. Reoperation was not recommended due to the complexity of the case, high surgical risks, and previous failed instrumentation.

The patient underwent an exercise program for his restrictive lung disease, including diaphragmatic breathing, segmental breathing exercises, and Triflo training to improve his pulmonary function and capacity. With the aim of improving the overall physical condition, quality of life, and functional independence, the treatment plan included mobilization exercises to improve joint and spinal flexibility, upper and lower extremity strengthening exercises, and balance-coordination training. The patient was informed about the importance of regular cardiology and pulmonology follow-ups. Written informed consent was obtained from the patient.

DISCUSSION

In this case report, we emphasize that vertebral abnormalities may lead to severe scoliosis and cause disability. This case is an outstanding example of how devastating the consequences can be, if a concomitant spinal abnormality is overlooked. Our patient had three main features of GS: facial asymmetry because of mandibular hypoplasia, oculo-auricular, and spinal deformities. Additionally, congenital cardiac defects and restrictive lung disease, caused by severe scoliosis in the thoracic region were identified.

In the literature, the prevalence of spinal deformities has been reported between 8% and 79%.[8] Caron et al.[9] found that vertebral malformations were more common in the cases of mandibular hypoplasia and bilateral anomalies. In a study, thoracic scoliosis was determined in 10 of 14 patients with craniofacial microsomia, and the most common vertebral abnormalities are hemivertebra and unilateral unsegmented bar with contralateral hemivertebra. Eight of these patients were operated on to stabilize the spine and partially correct the deformity.[5] A systematic review reported that the most frequent skeletal anomalies occur in the cervical spine, thoracic spine, and ribs, while lumbar anomalies are rarer.[8] The most reported vertebral deformities include hemivertebra, block vertebra, scoliosis/kyphoscoliosis, and spina bifida. In the cervical region, occipitalization of the atlas and cervical ribs are frequently observed. Renkema et al.[10] reported that vertebral abnormalities were the most common extra-craniofascial malformation in their study. The majority of anomalies were located in the thoracal region, and 44% of patients had symptoms such as torticollis, neck and back pain, and limited range of motion in the cervical. Although the patient did not report pain, his cervical, thoracic, and lumbar spinal mobility was severely restricted, significantly impacting daily activities. Thoracic scoliosis with severe rotation was identified as the major curve.

Vertebral abnormalities in GS are understudied compared to craniofacial anomalies.[11] This case emphasizes the importance of early intervention to prevent scoliosis-related complications by illustrating a rare, severe form of GS. It contributes to the limited research on vertebral abnormalities in this condition. In this patient, the main concern was the severe restrictive lung disease due to marked scoliosis. Congenital scoliosis from vertebral abnormalities is typically rigid and resistant to correction, with progression rates varying: 25% remain stable, 50% progress slowly, and 25% rapidly. While scoliosis progression in GS is recognized, precise data on its rate remain unclear.[12]

Treatment of GS with multiple abnormalities is symptomatic and may include a combination

of supportive, medical, and surgical therapies. Particularly in patients with congenital spinal deformities, early surgical intervention is often essential to ensure balanced spinal growth and prevent the progression of deformity.[13] Non-surgical options include bracing and physical therapy.[14] A few studies have suggested that bracing may have a short- to medium-term positive impact on pain and functionality in adults with progressive primary degenerative or idiopathic scoliosis. These studies, predominantly of low quality, involved mostly women with thoracolumbar and lumbar curves.[15] However, this patient, presenting with an extremely severe and rigid curvature, was not a suitable candidate for brace treatment. While the majority of scoliosis associated with GS is of a congenital nature, idiopathic-pattern scoliosis may also be present. A reported case of a four-year-old patient developed scoliosis at 11 without congenital anomalies, and bracing effectively managed the curve, highlighting the need for regular follow-ups.[1] In this patient, surgical intervention was performed during late adolescence; however, early complications necessitated the removal of the instrumentation. If this complication had not occurred, the current severity of restrictive lung disease could have been prevented. In clinical practice, GS can be diagnosed during the intrauterine period. Therefore, pediatricians should take a proactive role in managing affected children, ensuring timely referrals to relevant specialists for appropriate treatment and follow-up of existing anomalies.

The main limitation of our approach to this case was the inability to identify potential vertebral abnormalities due to both poor X-ray quality and severe vertebral rotation. Furthermore, due to the thoracic hump, the patient was unable to lie supine, which made performing imaging procedures such as magnetic resonance imaging or computed tomography scans impossible. Nevertheless, the strength of this case report strength is its emphasis on the identification of scoliosis in individuals with craniofacial abnormalities. It also underscores the essential requirement for early detection and monitoring of spinal deformities to prevent potential disabilities in adulthood.

In conclusion, it is of utmost importance to recognize GS in children with vertebral deformities and craniofacial abnormalities. Preventing the progression of deformities and associated complications requires early diagnosis, regular monitoring of spinal deformities, and thorough evaluations. A multidisciplinary approach along with tailored rehabilitation programs is crucial for preserving functional abilities and enhancing quality of life.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

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