

Available online at [www.sciencedirect.com](http://www.sciencedirect.com)

ScienceDirect

journal homepage: [www.elsevier.com/locate/radcr](http://www.elsevier.com/locate/radcr)

## Case Report

# A new lumbar vertebral anomaly in Goldenhar syndrome: A case report ☆☆☆

Mauro Costa Morais Tavares Junior, MD<sup>a,\*</sup>, Dennis J. Heaton, DO<sup>b</sup>, Brian M. Everist, MD<sup>b</sup>, Brandon B. Carlson, MD, MPH<sup>a</sup>

<sup>a</sup>Department of Orthopedic Surgery, The University of Kansas Medical Center, Kansas City, KS, USA

<sup>b</sup>Department of Diagnostic Radiology, The University of Kansas Medical Center, Kansas City, KS, USA

## ARTICLE INFO

## Article history:

Received 4 March 2022

Revised 18 March 2022

Accepted 20 March 2022

## Keywords:

Goldenhar syndrome

Nerve root anomalies

Spinal surgery

CT scan

MRI

## ABSTRACT

Goldenhar syndrome has reported incidence ranging from 1:3500 to 1:20000 live births. It consists of abnormalities involving the first and the second branchial arches and its etiology is heterogenous. A newborn with this condition can have a normal life and intelligence, so it is important to correctly diagnose and manage the various conditions associated with Goldenhar syndrome to preserve patient quality of life. This case report describes a unique vertebral abnormality in a patient with Goldenhar syndrome, where a lumbar nerve root or vessel traverses an anomalous vertically oriented osseous foramen in a lumbar spine pedicle. If this anomaly goes unidentified, pedicle screw placement may pose a significant surgical risk to the traversing nerve or vessel.

© 2022 The Authors. Published by Elsevier Inc. on behalf of University of Washington.

This is an open access article under the CC BY-NC-ND license

(<http://creativecommons.org/licenses/by-nc-nd/4.0/>)

## Introduction

Goldenhar syndrome (oculo-auriculo-vertebral spectrum) had been first described in 1952 by Maurice Goldenhar [1,2]. The syndrome is a sporadic or inherited genetic syndrome characterized by limbal dermoids, preauricular skin tags and mandibular hypoplasia [3].

Goldenhar syndrome has reported incidence ranging from 1:3500 to 1:20000 live births [3,4]. It consists of abnormalities involving the first and the second branchial arches and its etiology is heterogenous [2,3,5,6]. Clinical presentation is widely variable, with some affected individuals presenting

with mild features such as facial asymmetry or ear anomalies alone, and others with more significant presentations such as epibulbar dermoids, cardiopulmonary malformations and osseous abnormalities of the spine and extra axial bones [5,7–10].

Diagnosis is based principally on clinical aspects, which is associated with the patient's systemic conditions and radiologic findings [3,11,12]. The syndrome can also be discovered prenatally by ultrasonography [11,12].

A newborn with this condition can have a normal life and intelligence [4], so it is important to correctly diagnose and manage the various conditions associated with Goldenhar syndrome to preserve patient quality of life [1,2,13–15].

☆ Funding: This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

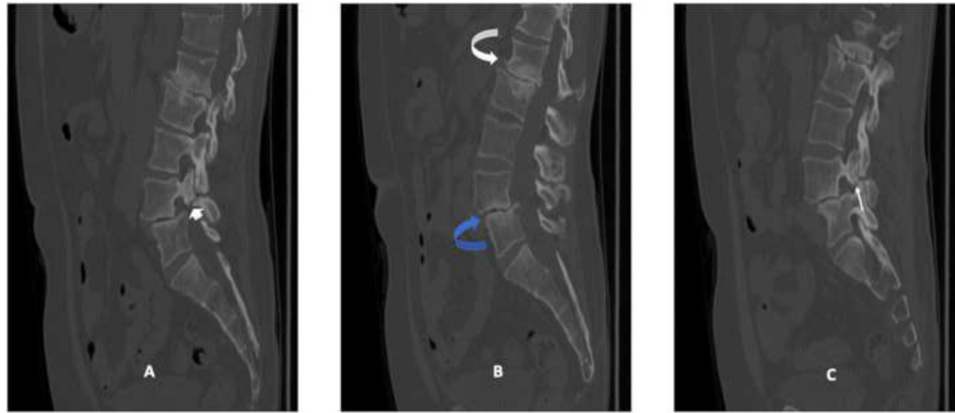
☆☆ Competing Interests: None.

\* Corresponding author.

E-mail address: [mcosta@kumc.edu](mailto:mcosta@kumc.edu) (M.C.M. Tavares Junior).

<https://doi.org/10.1016/j.radcr.2022.03.076>

1930-0433/© 2022 The Authors. Published by Elsevier Inc. on behalf of University of Washington. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>)



**Fig. 1 – Pre-operative sagittal CT images of the lumbar spine. (A) Right spondylolysis at L4-L5 (short arrow). (B) Midline sagittal demonstrating retrolisthesis, loss of intervertebral disc height, and posterior disc osteophyte complex at L1-L2 (curved white arrow). At L4-L5 the midline sagittal image demonstrates spondylolisthesis and marked loss of intervertebral disc height (curved blue arrow). (C) Left spondylolysis at L4-L5 (long arrow). (For interpretation of the references to color in this figure, the reader is referred to the web version of this article).**

This case report describes a new and unique vertebral abnormality with an anomalous vertically oriented intrapedicular foramen transmitting either an anomalous nerve root or vessel in a patient with Goldenhar syndrome. To the best of our knowledge these findings and spinal anatomical variations have not been previously reported in a Goldenhar patient.

## Case presentation

A 62-year-old male with Goldenhar syndrome presented in May 2021 with low back pain and bilateral lumbar radiculopathy with leg pain, numbness and weakness with walking. His pain score was eight and Oswestry disability index was 36 (0-20 minimal disability, 21-40 moderate disability, 41-60 severe disability, 61-80 crippled and 81-100 bed-bound) [16,17]. Given the patient's complaints of lower extremity radiculopathy, he was referred for diagnostic selective nerve root blocks at L4 bilaterally and had temporary relief of the lower extremity pain. Based on his spondylolisthesis, foraminal stenosis and positive response to the selective nerve root blocks, surgical intervention was discussed and the patient wished to proceed.

Past medical history included acid reflux, alcoholism, arthritis, asthma, cataracts, cervical and lumbar degenerative disease, scoliosis and Goldenhar syndrome with left sided facial paralysis, left vocal cord abnormality, left eye ptosis and absent left ear with deafness on the left. The diagnosis of Goldenhar syndrome was made in childhood based on characteristic clinical aspects.

Family history included arthritis and cardiac disease (father), atrial fibrillation (brother), and dementia (mother).

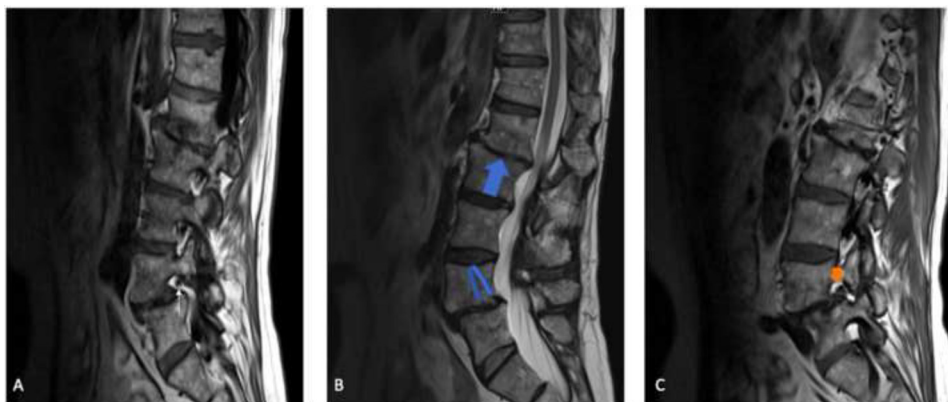
Social history included: former heavy drinker, quit in 1991 and former smoker (30 pack-year history), quit in 2014. Unaware of asbestos exposure, but more than 30 years exposure to welding fumes and particulates.

Standing plain radiographs identified scoliosis, L1-2 disc degeneration and a Grade II (Meyerding classification) spondylolisthesis of L4 on L5 with severe disc height loss and disc collapse [18] (Fig. 1).

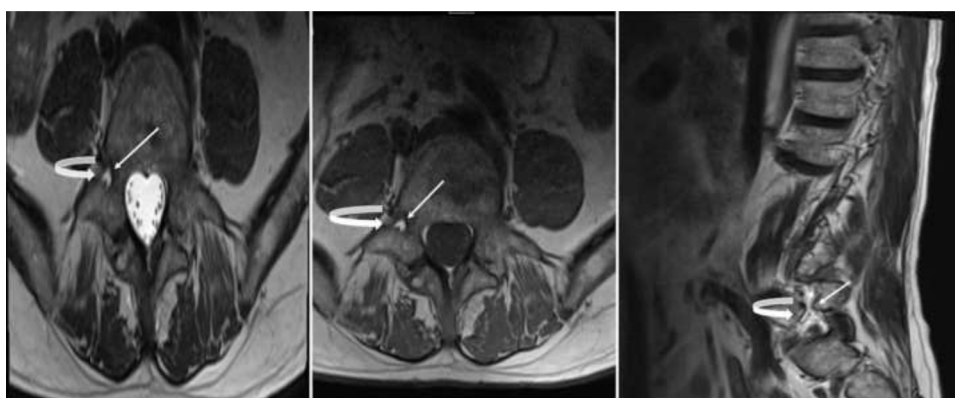
Preoperative MRI was performed in the standard recumbent position on a Siemens (Erlangen, Germany) 3.0 tesla MRI scanner. Axial T1 and T2, and sagittal T1, T2, and STIR sequences were obtained at 5mm slice thickness without post-contrast imaging. Marked lumbar degenerative changes were described at L1-L2 and L4-L5 with associated degenerative retrolisthesis and anterolisthesis, respectively. At L4-5 there was severe disc height loss due to degenerative disease which resulted in moderate right and severe left foraminal stenosis and exiting (L4) nerve root compression (Fig. 2). Computed tomography (CT) was performed as part of the preoperative evaluation using a GE MDCT scanner (General Electric Company, Boston, MA) with 0.625 mm slice thickness, multiplanar reformatted images, and dedicated soft tissue and bone kernels. The CT further delineated bilateral pars fractures at L4 with 7 mm anterolisthesis further characterizing and confirming the diagnosis of isthmic spondylolisthesis.

Within the right L5 pedicle was a vertically oriented fat containing channel with a linear T1 and T2 hypointense structure traversing the channel. The linear structure appeared contiguous with the exiting L4 nerve and likely represents a component of the lumbar plexus; possibly an extradural anastomosis to the caudal L5 nerve (Fig. 3). An anomalous vascular structure would be an additional consideration. The CT demonstrated similar findings to the MRI with a sharply margined lucent channel traversing the right L5 pedicle in a cranial-caudal direction containing fat-dense tissue and a subtle, linear, non-fat structure traversing the channel (Fig. 4).

Based on the clinical history of lumbar radiculopathy responsive to selective root blocks and associated isthmic spondylolisthesis causing foraminal stenosis at L4-5, the recommended surgical plan included a L4-5 spinal arthrodesis. The senior author (BBC) suggested an interbody fusion with titanium cage placement for restoration of disc height, reduction of the anterolisthesis and increase in the foraminal



**Fig. 2 – Multiple sagittal MR images through the lumbar spine.**(A) T1 weighted sagittal image demonstrating mild right neural foraminal stenosis at L4-L5 (white arrow). (B) T2 weighted sagittal image through the midline demonstrating marked loss of intervertebral disc height at L1-L2 (blue arrow) and L4-L5 (curved blue arrow), retrolisthesis and posterior disc osteophyte complex at L1-L2, and spondylolisthesis at L4-L5 without significant central spinal stenosis. (C) T1 weighted sagittal image demonstrating marked left neural foraminal stenosis at L4-L5 (orange arrow). (For interpretation of the references to color in this figure, the reader is referred to the web version of this article).

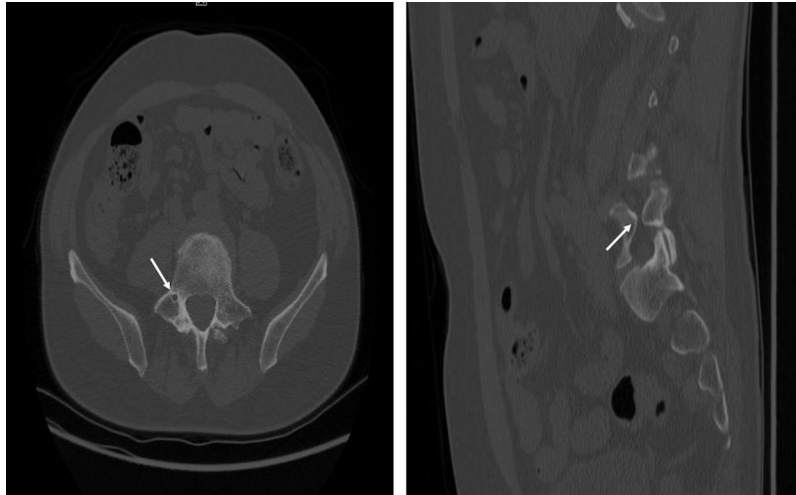


**Fig. 3 – Axial T2 (A), axial T1 (B), and sagittal T2 (C) weighted axial images through the L5 vertebral level demonstrating a fat filled channel within the L5 pedicle (straight arrow). A hypointense linear structure traverses the channel (curved arrow), possibly communicating with the L4 nerve root (not pictured).**

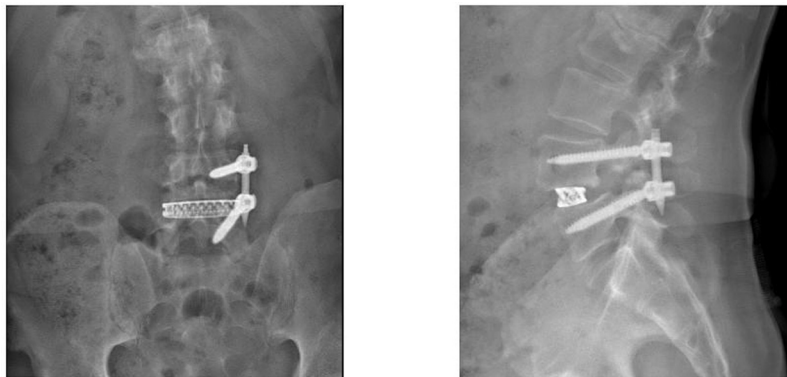
height to reduce nerve root compression. Multiple surgical approaches were discussed with the patient including transforaminal lumbar interbody fusion vs transposas lateral lumbar interbody fusion with minimally invasive percutaneous pedicle screw fixation. The treating surgeon routinely performs lateral lumbar interbody fusion with percutaneous screw fixation procedures utilizing either Navigation or Robotic Spinal Navigation technology.

The surgeon elected to utilize robotic spinal navigation for screw placement. This technology allows a surgeon to preplan pedicle screw trajectories prior to surgery by uploading the preoperative CT to the robot and using software specify screw placement position. The robotic arm then allows for accurate screw placement following the preplanned trajectory. During the preplanning process, the patient's vertebral abnormality and anomalous structure within the L5 pedicle was identified and unable to be avoided with any transpedicular trajectory. Prior to starting surgery, the surgeon

contacted the senior musculoskeletal radiologist (BE) and discussed this structure both on CT and MRI. Based on their joint evaluation, it was believed this either represented a nervous or vascular structure and should be avoided when placing spinal instrumentation to ensure patient safety. With this consensus, unilateral spinal instrumentation was planned at L4-5 on the left only with lateral lumbar interbody fusion L4-L5. The anomalous pedicle finding altered the surgical plan from bilateral instrumentation to unilateral instrumentation only. Surgery was performed using robotic navigation assistance and instrumentation was placed successfully at L4 and L5 on the left without incident and the lateral interbody fusion with cage placement was performed as planned (Fig. 5). Surgery was completed in 100min with 25cc blood loss and no complications. The patient was awakened with no neurologic deficits and radiculopathy improved. He ambulated 450ft with physical therapy on postoperative day 1 and discharged home in stable condition. He has maintained his implant position



**Fig. 4 – Axial and sagittal CT on bone window through the L5 vertebral level demonstrating an abnormal sharply margined channel traversing the right L5 pedicle (arrow). A subtle linear structure is present centrally within the channel and is better depicted on MR images.**



**Fig. 5 – Frontal and lateral post-operative computed radiographs of the lumbar spine demonstrating left posterior spinal fusion and placement of an intervertebral cage at L4-L5.**

without cage subsidence, screw pullout or loss of reduction at 3 months after surgery (Fig. 6).

## Discussion

Goldenhar syndrome is a developmental anomaly of the maxillofacial skeleton and vertebrae [2]. Gorlin et al were the first to recognize that this syndrome could also be associated with congenital anomalies of the vertebrae [1,2].

Orthopedic findings previously reported include spinal anomalies, clubfoot, congenital dislocation of the hip, Sprengel's deformity, and radial limb defects [9,13,19]. In most studies, the reported prevalence of vertebral anomalies varied from 19% to 79% [7,13,14,19].

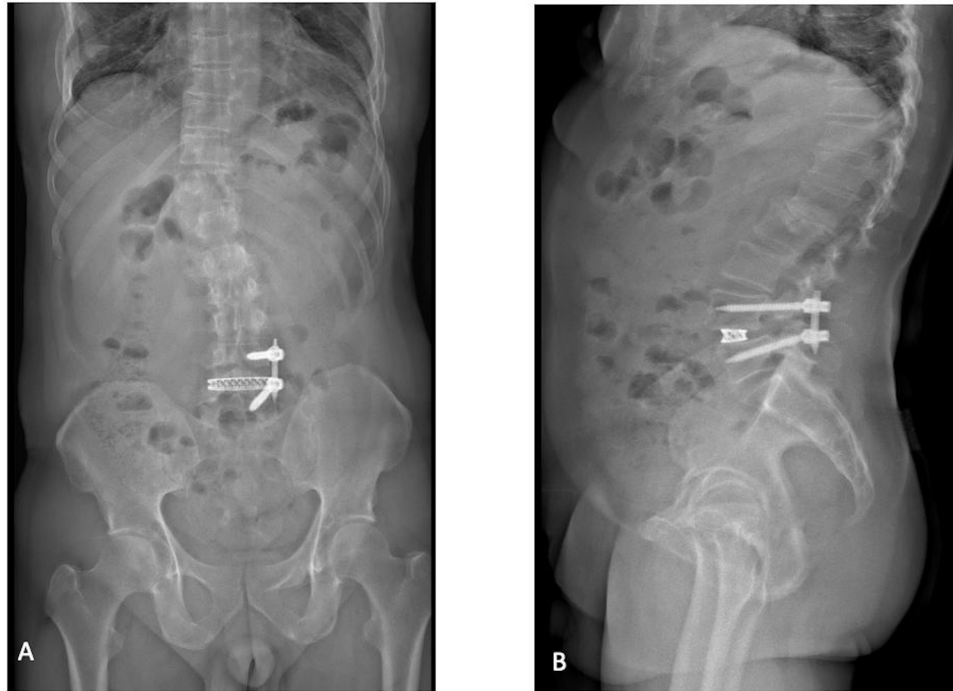
Reported vertebral anomalies without specification of the location were hemivertebra, scoliosis and spina bifida occulta [7,13,19]. The cervicothoracic spine is the most prevalent region affected [13]. In the lumbar region, vertebral anomalies

were seen less frequently and when present, hemivertebra, block vertebra, and/or scoliosis were most often described [13].

This syndrome was first reported in an age where MRI or high resolution CT scans did not exist [20], and many of the recent studies using the new technologies that emerged in the past few decades, has provided high-resolution images suitable for the evaluation in vivo [20]. This fact provided innumerable opportunities to discover new rare findings associated with Goldenhar Syndrome spectrum [5,21,22].

In our study, we report an abnormal L5 pedicle and an anomalous nerve root branch contained within the midportion of the bony pedicle. Identification of this variant preoperatively altered the surgical plan to avoid possible risks to the patient.

In the general population, nerve root anomalies (NRAs) most frequently involve the L5 and S1 nerve roots, routinely accounting for 50% to 70% of reported nerve root anomalies and either L4 or S2 have reported to be the next most commonly involved nerve root. NRAs have been detected in patients between the ages of 15 and 73 and the prevalence rates



**Fig. 6 – Frontal and lateral scoliosis EOS post-operative computed radiographs demonstrating unchanged position of the left posterior spinal fusion implants, and unchanged alignment of the L4-L5 vertebrae.**

of nerve anomalies intraoperatively discovered range from 0.32% to 5.8%. For comparison, reported rates of NRAs found in MRI studies ranging from 0.25% to 6.7%. Because of the great variability in the nerve root anomalies, there are several classification systems. [23] When searching the literature about similar findings, even in non-syndromic patients, we could not retrieve any similar finding.

To the best of our knowledge, this is the first report of anomalous lumbar spine nerve anatomy within a lumbar pedicle in a patient with Goldenhar syndrome. This case underscores the importance scrutinizing preoperative imaging and utilizing both MRI and/or CT when planning instrumentation placement in patients with Goldenhar syndrome. The collaboration between surgeons and radiologists when questionable anatomy is encountered is critical for safe surgical planning in patients with rare syndromes such as Goldenhar syndrome [4,8,14].

## Conclusion

Goldenhar syndrome has variable anatomical and radiologic findings related to the spine. This report is the first finding of an anomalous nerve or vessel traversing a lumbar pedicle in a Goldenhar patient and was identified when planning for spinal instrumentation placement. Based on this case, surgeons and radiologists should be keenly aware of possible abnormal neurovascular anatomy in the lumbar spine in Goldenhar patients, and use of advanced imaging is warranted to fully evaluate these patients prior to surgical interventions.

## Patient consent

Patient consent has been obtained.

## Acknowledgments

None

## REFERENCES

- [1] Bogusiak K, Puch A, Arkuszewski P. Goldenhar syndrome: current perspectives. *World J Pediatr* 2017;13(5):405–15. doi:10.1007/s12519-017-0048-z.
- [2] Al Kaissi A, Ben Chehida F, Ganger R, Klaushofer K, Grill F. Distinctive spine abnormalities in patients with Goldenhar syndrome: tomographic assessment. *Eur Spine J* 2015;24(3):594–9. doi:10.1007/s00586-014-3204-3.
- [3] Almeida LM, Diniz Mdos S, Diniz Ldos S. Do you know this syndrome? *An Bras Dermatol* 2012;87(3):495–7. doi:10.1590/s0365-05962012000300029.
- [4] Kurniawan R, Suarca IK, Suryawan IWB. Goldenhar syndrome: a case report. *Open Access Maced J Med Sci* 2019;7(8):1342–5. doi:10.3889/oamjms.2019.281.
- [5] Bedi M, Jain RK, Barala VK, Singh A, Jha H. A constellation of rare findings in a case of Goldenhar syndrome. *Case Rep Pediatr* 2017;2017:3529093. doi:10.1155/2017/3529093.
- [6] Cullins SL, Pridjian G, Sutherland CM. Goldenhar's syndrome associated with tamoxifen given to the mother during gestation. *JAMA* 1994;271(24):1905–6.

- [7] Anderson PJ, David DJ. Spinal anomalies in Goldenhar syndrome. *Cleft Palate Craniofac J*. 2005;42(5):477–80. doi:10.1597/04-142051r.1.
- [8] Engiz O, Balci S, Unsal M, Ozer S, Oguz KK, Aktas D. 31 cases with oculoauriculovertebral dysplasia (Goldenhar syndrome): clinical, neuroradiologic, audiological and cytogenetic findings. *Genet Couns* 2007;18(3):277–88.
- [9] Avon SW, Shively JL. Orthopaedic manifestations of Goldenhar syndrome. *J Pediatr Orthop* 1988;8(6):683–6. doi:10.1097/01241398-198811000-00010.
- [10] Johnson KA, Fairhurst J, Clarke NM. Oculoauriculovertebral spectrum: new manifestations. *Pediatr Radiol* 1995;25(6):446–8. doi:10.1007/BF02019062.
- [11] De Catte L, Laubach M, Legein J, Goossens A. Early prenatal diagnosis of oculoauriculovertebral dysplasia or the Goldenhar syndrome. *Ultrasound Obstet Gynecol* 1996;8(6):422–4. doi:10.1046/j.1469-0705.1997.08060422.x.
- [12] Guzelmansur I, Ceylaner G, Ceylaner S, Ceylan N, Daplan T. Prenatal diagnosis of Goldenhar syndrome with unusual features by 3D ultrasonography. *Genet Couns* 2013;24(3):319–25.
- [13] Renkema RW, Caron CJJM, Mathijssen IMJ, Wolvius EB, Dunaway DJ, Forrest CR, et al. Vertebral anomalies in craniofacial microsomia: a systematic review. *Int J Oral Maxillofac Surg* 2017;46(10):1319–29. doi:10.1016/j.ijom.2017.04.025.
- [14] Uehara M, Kuraishi S, Ikegami S, Oba H, Takizawa T, Munakata R, et al. Scoliosis in Goldenhar syndrome with curve reversal during brace treatment: a case report. *BMC Musculoskelet Disord* 2020;21(1):685. doi:10.1186/s12891-020-03719-y.
- [15] Belenchia P, McCardle P. Goldenhar's syndrome: a case study. *J Commun Disord*. 1985;18(5):383–92. doi:10.1016/0021-9924(85)90028-0.
- [16] Fairbank JC, Pynsent PB. The Oswestry disability index. *Spine (Phila Pa 1976)* 2000;25(22):2940–52 discussion 2952. doi:10.1097/00007632-200011150-00017.
- [17] Fairbank JC, Couper J, Davies JB, O'Brien JP. The Oswestry low back pain disability questionnaire. *Physiotherapy* 1980;66(8):271–3.
- [18] Koslosky E, Gendelberg D. Classification in brief: the meyerding classification system of spondylolisthesis. *Clin Orthop Relat Res* 2020;478(5):1125–30. doi:10.1097/CORR.0000000000001153.
- [19] Gibson JN, Sillence DO, Taylor TK. Abnormalities of the spine in Goldenhar's syndrome. *J Pediatr Orthop* 1996;16(3):344–9. doi:10.1097/00004694-199605000-00010.
- [20] Manara R, Brotto D, Ghiselli S, Mardari R, Toldo I, Schifano G, et al. Cranial nerve abnormalities in oculo-auriculo-vertebral spectrum. *AJNR Am J Neuroradiol* 2015;36(7):1375–80. doi:10.3174/ajnr.A4273.
- [21] Konas E, Canter HI, Mavili ME. Goldenhar complex with atypical associated anomalies: is the spectrum still widening? *J Craniofac Surg* 2006;17(4):669–72. doi:10.1097/00001665-200607000-00011.
- [22] Zelante L, Gasparini P, Castriota Scanderbeg A, Dimitri L, Criconia M, Gorlin RJ. Goldenhar complex: a further case with uncommon associated anomalies. *Am J Med Genet* 1997;69(4):418–21.
- [23] Schmidt CK, Rustagi T, Alonso F, Loukas M, Chapman JR, Oskouian RJ, et al. Nerve root anomalies: making sense of a complicated literature. *Childs Nerv Syst* 2017;33(8):1261–73. doi:10.1007/s00381-017-3457-3.