Lumbosacral junction congenital kyphosis in an adult patient: a case report

Arvin Eslami[1], Mohammadreza Chehrassan[1], Shaya Alimoghadam[2], and Mohammadreza Shakeri[1]

[1]Iran University of Medical Sciences
[2]Shahid Beheshti University of Medical Sciences

December 14, 2022

Abstract

This is an extremely rare case of congenital lumbosacral junction kyphosis (CLSJK) in a 40-year-old woman due to S1-S2 hemivertebra. Performing surgery has helped to improve neurological disturbances but not completely after 1 year of follow-up.

Lumbosacral junction congenital kyphosis in an adult patient: a case report

Co-Authors

Arvin Eslami[1], M.D

a-eslami@alumnus.tums.ac.ir

Mohammadreza Chehrassan[1], MD

morchehrasan@yahoo.com

Shaya Alimoghadam[2], Medical Student,

shayalimoghadam@sbmu.ac.ir

Mohammadreza Shakeri[1], MD

mshakeri3116@gmail.com

[1] Bone and Joint reconstruction research center, Shafa Yahyaeian Orthopedic Hospital, Iran University of Medical Sciences, Tehran, Iran

[2] School of Medicine, Shahid Beheshti University of Medical Sciences, Tehran, Iran

Corresponding Author

Mohammadreza Shakeri[1], MD

Bone and Joint reconstruction research center, Shafa Yahyaeian Orthopedic Hospital, Iran University of Medical Sciences, Tehran, Iran

Add: Mojahesin Eslam St. Shafa Yahyaeian Orthopedic Hospital

Tel: +982133542003

Fax: +982133542004

Email: mshakeri3116@gmail.com

Data availability statement

All data are included in this published article and its additional information files.

Funding statement

No funds and resources were received in support of this work.

Conflict of interest disclosure

We have no conflicts of interest to disclose.

Ethics approval statement

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Patient consent statement

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

Key Clinical Message:

Congenital lumbosacral junction kyphosis like other congenital kyphosis types should be treated before the appearance of neurological symptoms. However, surgery in adulthood can help to improve neurological disturbances even if not completely.

Abstract

This is an extremely rare case of congenital lumbosacral junction kyphosis (CLSJK) in a 40-year-old woman due to S1-S2 hemivertebra. Performing surgery has helped to improve neurological disturbances but not completely after 1 year of follow-up.

Keywords: Hemivertebra, Sacral, neurologic bladder, Urinary retention, PSFI, MRI Spine, congenital anomalies, gait problem, lumbar, lordosis

Background

Hemivertebra(HV) is a complete failure of the formation of a vertebral body [1]. Sacral HV is a very rare congenital anomaly; prior to this, about seven cases had been reported (Table 1).[2-7] Posterior HV are wedge-shaped deformities caused by failure of anterior vertebral formation, resulting in progressive kyphosis. [8,9] The lumbosacral HV usually does not present a risk of kyphosis deformity and previously in the case of lumbosacral HV, no cases of kyphosis have been identified.[10] A sacral HV between S1-S2 is too rare and just one case had been reported previously which happened in a 10-month-old infant and no explanation has been given about its clinical symptoms in adulthood and the possible problems caused by it. [11]In this article, the condition of a patient with an S1-S2 HV that cause congenital Lumbosacral junction kyphosis (CLSJK)with a neurological bladder is explained.

Case Presentation

A 40 years old woman had mild spine deformity in the form of increased lumbar lordosis from childhood with no other symptoms. Gait disturbance started four years earlier and her trunk tilt was exacerbated in the last two years. 11 months before surgery, the patient started suffering from urinary problems in the form of increased urinary residual volume and showed symptoms such as frequency, dribbling, and suprapubic pain. Two months later, her urinary symptoms worsened and subsequently, she started having urinary retention resulting in the need to undergo frequent catheterization. Furthermore, the patient's gait aggravation, along with low back pain, disrupted her daily activities. In the physical examination, she had gait problems,

low-grade pelvic tilt, and severe lumbar lordosis. All lower limb forces, sensory examination, Upper motor reflexes, and anus sphincter tone were normal. Radiologically severe CLSJK due to the existence of S1-S2 HV was identified on Plain x-ray, CT-scan and magnetic resonance imaging features. (Figure 1,2,3) The patient was operated in prone position and S1-S2 HV resection was performed through the posterior approach. Subsequently, pedicle screws were applied from L5 to S2. Lumbarized S1 was reduced partially on S2 and posterior spinal fusion was done using allograft bone chips mixed with harvested bone from corpectomy. The neuromonitoring control was performed through the surgery and remained as the records just before the surgical incision (Fig.4). one year of follow-up shows that the patient's gait has improved and her low back pain has eliminated. The patient can perform daily activities without restriction. Her urinary retention problem has not yet been completely eliminated but has improved as compared to before.

Discussion

Sacral HV and CLSJK is a very rare abnormality and the natural progress is unclear.[12] Daher et al., reported a 10 month old infant with anorectal malformation who underwent rectal surgery, which in further examinations, a HV at S1-S2 level was found but they did not mention the details of the spine problem and did not give an explanation about its follow-up [6]. Karaeminogullari et al., reported an 11-month-old boy who was treated surgically for congenital heart disease with a sacral HV at S2-S3 level with no thoracic or lumbar scoliosis. They followed the patient for seven years and no symptoms such as lower back pain in either standing or seating positions were found. Also the gait, muscle power, sensation and reflexes for the lumbar and sacral nerves were all normal [7]. Whereas Ansari et al., reported two cases with L5-S1 HV who both had functional kyphosis at the lumbosacral junction. Both walked with a waddling gait and flexed hips and had bowel and bladder difficulties with abnormal urodynamic testing. [13]. It is notable that patients showed similar symptoms and clinical course to our report. The difference is that our patient had a HV at S1-S2 level.

Winter et al. described 41 degree of deformity advancement following dorsal HV in a sample of 130 patients over 6 years.[14]Also, Nazareth et al have pointed out that that CLSJK like other congenital kyphosis types should be treated before the appearance of neurological symptoms. [9] In this regard, like other congenital kyphosis types, CLSJK needs to be treated before neurological symptoms present. However, even if adult surgery doesn't totally resolve neurological problems, it can still contribute to improvement.

Conclusion

CLSJK following HV at S1-S2 level is a very rare condition and no symptomatic adult patients had been reported for previously. CLSJK like other congenital kyphosis types should be treated before the appearance of neurological symptoms. However, performing surgery in adulthood can help to improve neurological disturbances even if not completely.

ACKNOWLEDGEMENTS

None.

AUTHOR CONTRIBUTION

First author 1 (A.E): wrote and edited the manuscript

Second author 2 (M.C): Performed the surgery

Third author 3 (S.A): wrote the manuscript

Fourth author 4 (M.S): Performed the surgery and editing the manuscript

Data availability statement

All data are included in this published article and its additional information files.

Funding statement

No funds and resources were received in support of this work.

Ethics approval statement

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Consent for publication

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

Conflict of interest disclosure

We have no conflicts of interest to disclose.

Figure legends:

Figure .1: A mild lumbar scoliosis, tiled pelvic (a), a severe CLSJK (b), and the S1-S2 HV (c) are seen in X-rays.

Figure .2: HV between S1-S2 are seen in coronal view (a) and sagittal view of CT scan (b).

Figure .3: The S1-S2 HV with compressive effect on the roots and dura is seen in MRI.

Figure .4: HV between S1-S2 resected and PSFI was done.

Table 1

Accompanying symptoms	**Level of sacral hemivertebra**	**Age**	Article
VACTERL	Not mentioned	child	Faivre et al.,[2]
bilateral severe hypoplasia of the femora, hypoplastic and vertical pelvic bones	Not mentioned	fetus	Darouich et al.,[3]
cleft palate, atrial septal defect (ASD), bicuspid aortic valve (BAV), Anal atresia partial synostosis of C3-C4, C6-C7, T3-T4, lumbar scoliosis, unilateral postaxial polydactyly of the right hand	Not mentioned	6 years old	Dentici et al.,[4]
bilateral hydronephrosis, two pelvic cystic masses behind the bladder, an abnormal rectum	S3-S4	fetus	Fayard et al.,[5]
Anorectal malformation	S1-S2	10 months old	Daher et al.,[6]
congenital heart disease	S2-S3	11 months old	Karaeminogullari et al.,[7]

Table 1: Including previous case reports related to sacral hemivertebra and associated symptoms in reported

patients.

Reference:

1. McMaster MJ, Ohtsuka K. The natural history of congenital scoliosis. A study of two hundred and fifty-one patients. *J Bone Joint Surg Am* . Oct 1982;64(8):1128-47.

2. Faivre L, Portnoï MF, Pals G, et al. Should chromosome breakage studies be performed in patients with VACTERL association? *Am J Med Genet A* . Aug 15 2005;137(1):55-8. doi:10.1002/ajmg.a.30853

3. Darouich S, Amraoui J, Amraoui N. Femoral-facial syndrome: Report of 2 fetal cases. *Radiol Case Rep* . Oct 2019;14(10):1276-1282. doi:10.1016/j.radcr.2019.08.001

4. Dentici ML, Barresi S, Niceta M, et al. Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. *Clin Genet* . Feb 2018;93(2):401-407. doi:10.1111/cge.13029

5. Fayard C, Blondiaux E, Grigorescu R, Garel C. AIRP best cases in radiologic-pathologic correlation: prenatal and postmortem imaging of a complex cloacal malformation. *Radiographics* . Nov-Dec 2014;34(7):2056-63. doi:10.1148/rg.347140018

6. Daher P, Daher R, Riachy E, Zeidan S. Do low-type anorectal malformations have a better prognosis than the intermediate and high types? A preliminary report using the Krickenbeck score. *Eur J Pediatr Surg* . Oct 2007;17(5):340-3. doi:10.1055/s-2007-965462

7. Karaeminoğullari O, Sahin O, Akgun RC, Karaman A, Atabey M. Sacral hemivertebra: a case report. *European Journal of Orthopaedic Surgery & Traumatology* . 2005;15:316-318.

8. Winter RB, Moe JH, Wang JF. Congenital kyphosis. Its natural history and treatment as observed in a study of one hundred and thirty patients.*J Bone Joint Surg Am* . Mar 1973;55(2):223-56.

9. Nazareth A, Andras LM, Krieger MD, Skaggs DL. Bilateral Congenital Posterior Hemivertebrae and Lumbar Spinal Stenosis Treated With Posterior Spinal Fusion and Instrumentation. *J Am Acad Orthop Surg Glob Res Rev* . Oct 2019;3(10)doi:10.5435/JAAOSGlobal-D-19-00054

10. Bollini G, Docquier PL, Viehweger E, Launay F, Jouve JL. Lumbosacral hemivertebrae resection by combined approach: medium- and long-term follow-up. *Spine (Phila Pa 1976)* . May 15 2006;31(11):1232-9. doi:10.1097/01.brs.0000217616.17692.a0

11. Acharya S, Palukuri N, Gupta P, Kohli M. Transcranial Motor Evoked Potentials during Spinal Deformity Corrections-Safety, Efficacy, Limitations, and the Role of a Checklist. *Front Surg* . 2017;4:8. doi:10.3389/fsurg.2017.00008

12. Goldstein I, Makhoul IR, Weissman A, Drugan A. Hemivertebra: prenatal diagnosis, incidence and characteristics. *Fetal Diagn Ther* . Mar-Apr 2005;20(2):121-6. doi:10.1159/000082435

13. Ansari SF, Rodgers RB, Fulkerson DH. Dorsal midline hemivertebra at the lumbosacral junction: report of 2 cases. *J Neurosurg Spine* . Jan 2015;22(1):84-9. doi:10.3171/2014.9.Spine1411

14. Winter RB. Congenital kyphosis. *Clin Orthop Relat Res* . Oct 1977;(128):26-32.