Case 18275

Eurorad

Klippel-Feil spectrum

Published on 24.08.2023

DOI: 10.35100/eurorad/case.18275
ISSN: 1563-4086
Section: Musculoskeletal system
Area of Interest: Musculoskeletal spine Neuroradiology spine
Imaging Technique: CT
Imaging Technique: MR
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 45 years, female

Clinical History:

A 45-year-old female patient presents with neck pain lasting over a year. She has a history of congenital scoliosis and underwent cardiac surgery for atrial septal defect 20 years ago. During the physical examination, she demonstrates severe muscle stiffness in the cervico-occipital region that restricts her range of motion.

Imaging Findings:

MR and CT of the cervical segment of the vertebral column revealed partial fusion of the posterior and lateral aspect of C4-C6 associated with narrowing of anteroposterior diameter of the vertebral bodies, as well as a decrease in the development of the corresponding intervertebral discs.

Signs of lateral hemivertebra or wedged vertebra at D3 and fusion of D2-D3-D4 vertebral bodies with partial development of the intervertebral disc D2-D3 and absence of a disc at the D3-D4 level, resulting in left convex dorsal scoliosis.

Discussion:

Background

Klippel-Feil spectrum (KFS) is a congenital disorder where two or more bones in the cervical spine fuse abnormally. Its prevalence is around 1 in 40,000 newborns, with females accounting for 60% of cases. The exact cause is unclear, but it may result from vascular disruptions, fetal insults, neural tube complications, or gene disruptions during embryonic development. While mostly sporadic, KFS can also be inherited in an autosomal recessive or dominant pattern. [1,2,3]

Clinical Perspective

Although the classic clinical triad includes short neck, low posterior hairline, and limited neck movement, it's important to note that less than 50% of patients exhibit all three simultaneously. These abnormalities can cause chronic headaches, neck muscle pain, and contribute to cervical spinal deformity. Associated anomalies include scoliosis (60%), atlantoaxial instability, spina bifida (45%), renal agenesis (35-55%), deafness (30-40%), rib deformity (20-30%), and vascular/cardiac abnormalities (8-14%) [1,2]. KFS is typically diagnosed in childhood, but asymptomatic patients may remain unaware until adulthood, increasing their risk of congenital spinal stenosis. [1,2]

The fusion of affected vertebral segments restricts their mobility, leading to increased mobility in adjacent segments. This overload can cause spondylotic changes in interapophyseal joints and discs, potentially resulting in canal or foraminal stenosis.Certain patterns, such as C2-C3 block with atlas occipitalization, two separate cervical blocks with an unfused segment between them or long blocks involving multiple segments, carry a higher risk of spinal cord injury. [1,5,8].

Imaging Perspective

Anteroposterior and lateral cervical radiographs depict the vertebral fusion of the body, facets and spinous process. We must include thoracic and lumbar spine radiographs that may reveal scoliosis or hemivertebrae. [1,4] Anterior-posterior narrowing ("wasp-waist" sign) can also be present. Functional radiographs can be utilized to assess spinal stability and movement. [1] C2-C3 and C5-C6 are the most commonly fused vertebral levels. [8]

CT demonstrates other accompanying abnormalities: absent or hypoplastic intervertebral spaces, loss of vertebral body height, narrow foramina and cervical stenosis. [5]

MRI is indicated in cases of suspected neurological injury in assessing the integrity of the spinal cord, disc space, nerve roots, ligaments and soft tissue structures. [1,5] It also demonstrates increased degenerative changes. [8]

There are three morphological subtypes: Type I involves a massive single fused cervical segment; Type II includes multiple noncontiguous fused segments and other anomalies such as hemivertebrae or occipito-atlantal fusion; and Type III comprises multiple fused cervical, thoracic or lumbar segments. [2,5,6,8]

Other associated disorders may require additional tests such as an echocardiogram, abdominal ultrasonography and intravenous pyelogram. [1,4]

Outcome

In the majority of cases, non-operative management is sufficient, involving activity modification; unless there are acute neurological deficits, cervical instability or risk of chronic neurological issues, extensive spinal surgeries may be necessary in such cases. Considering the poly-syndromic nature, coordination among various specialists is vital, with pediatricians playing a crucial role for younger patients [1,2].

The treatment objective is to prevent cervical instability and degenerative joint disease. [5,7] Thorough evaluation and proper referral are important to assess the presence of other abnormalities. Prompt and appropriate treatment yields a favorable prognosis in most cases, emphasising the importance of avoiding activities that may cause neck injury [2,5].

All patient data have been completely anonymised throughout the entire manuscript and related files.

Differential Diagnosis List: Ankylosing spondylitis, VACTERL syndrome, Klippel-Feil spectrum, Chronic sequelae of discitis or healing osteomyelitis, Juvenile idiopathic arthritis, Surgical intervertebral fusion

Final Diagnosis: Klippel-Feil spectrum

References:

[1] Menger RP, Rayi A, Notarianni C. (2022) Klippel Feil Syndrome. StatPearls Publishing; 2023 Jan (PMID: 29630209)

[2] Frikha R. (2020) Klippel-Feil syndrome: a review of the literature. Clinical Dysmorphology, 29:35–37 (PMID: 31577545)
[3] ?ota S, Žagar I, Delimar V, Pap M, Peri? D, Peri? P. (2019) Klippel-Feil syndrome misdiagnosed as spondyloarthropathy: case-based review. Rheumatology International. 39:1655-1660.(PMID:31214770)
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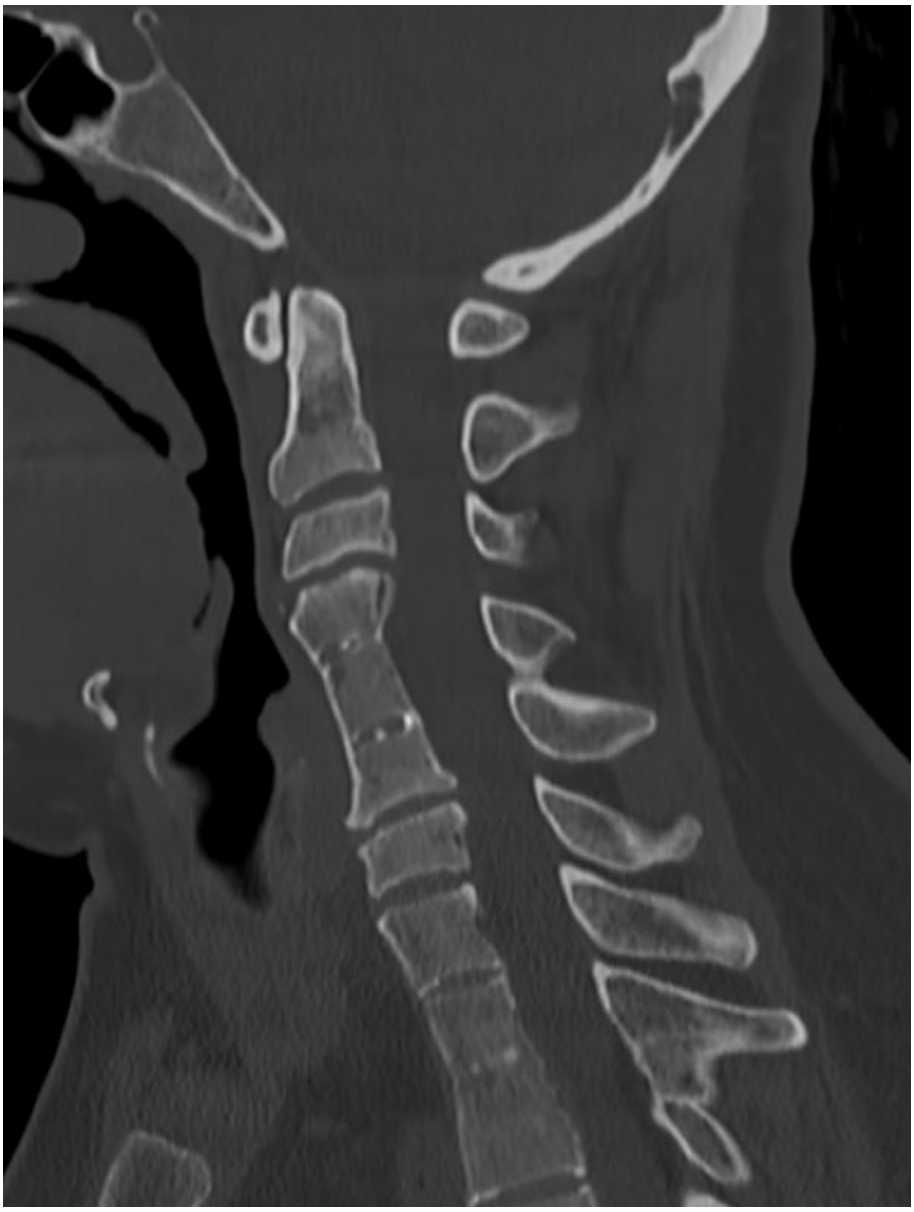
[5] Palacios Martínez D, Díaz Alonso RA, Gutiérrez López M, Gordillo López FJ. (2013) Síndrome de Klippel-Feil, una cervicalgia poco frecuente. Semergen 39:e63-67. (PMID: 24315079)
[6] Huke?ová Z, Krošláková M. (2021) Klippel-Feil syndrome cases from Slovakia. International Journal of Paleopathology; 33:188-195. (PMID: 33971397)
[7] Verla T, Prablek M, Ropper AE, Xu DS, Raber M. (2020) Congenital Fusion of Dens to T3 Vertebra in Klippel-Feil Syndrome. World Neurosurgery. 143:18-22. (PMID: 32652274)
[8] Gruber J, Saleh A, Bakhsh W, Rubery PT, Mesfin A. (2018) The Prevalence of Klippel-Feil Syndrome: A Computed Tomography-Based Analysis of 2,917 Patients. Spine Deformity. 6:448-453. (PMID:29886918)

Figure 1

a



Description: CT sagittal and coronal shows partial fusion of the posterior and lateral aspect of C4-C5-C6 associated with a decrease in the anteroposterior diameter of the vertebral bodies revealing the classic "wasp-waist" sign, as well as a decrease in the development of the corresponding intervertebral discs **Origin:** Departamento de Radiología del Centro de Diagnóstico por Imágenes de la Clínica Internacional Lima, Perú, 2023

b

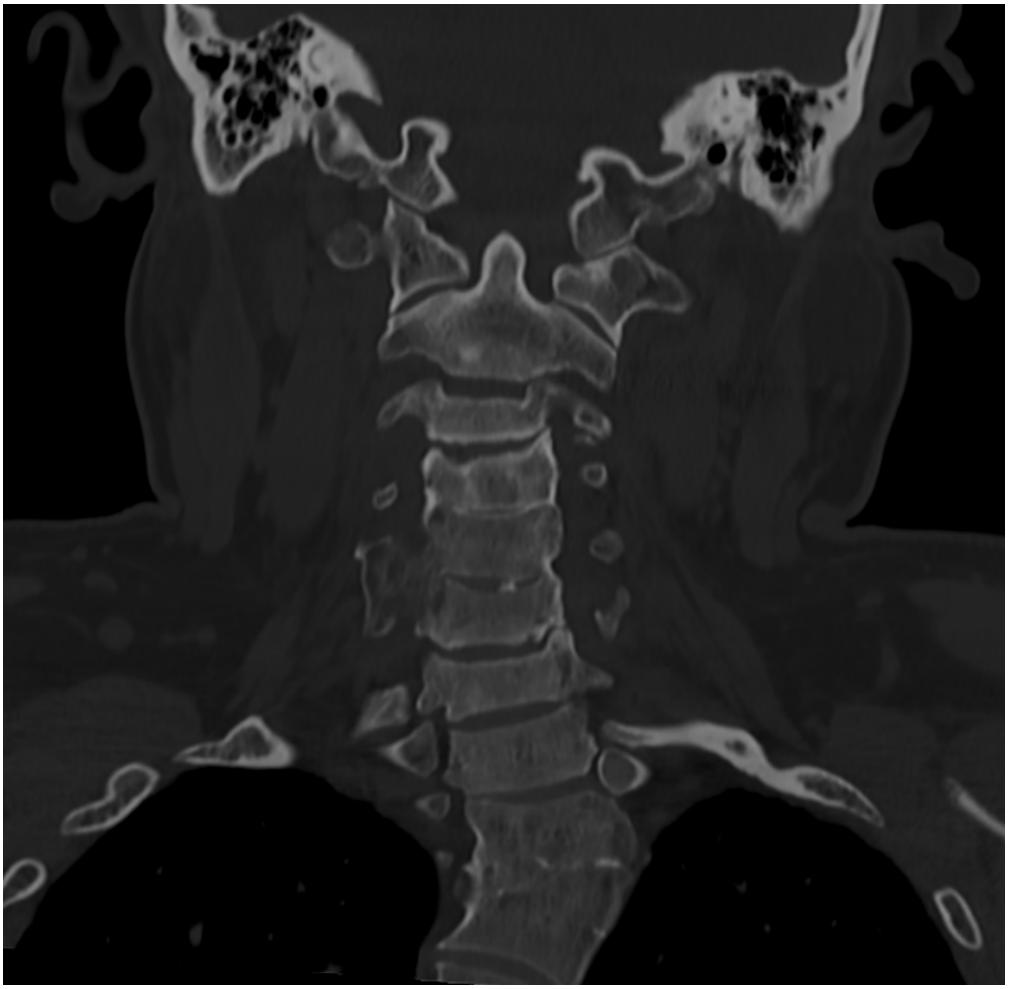


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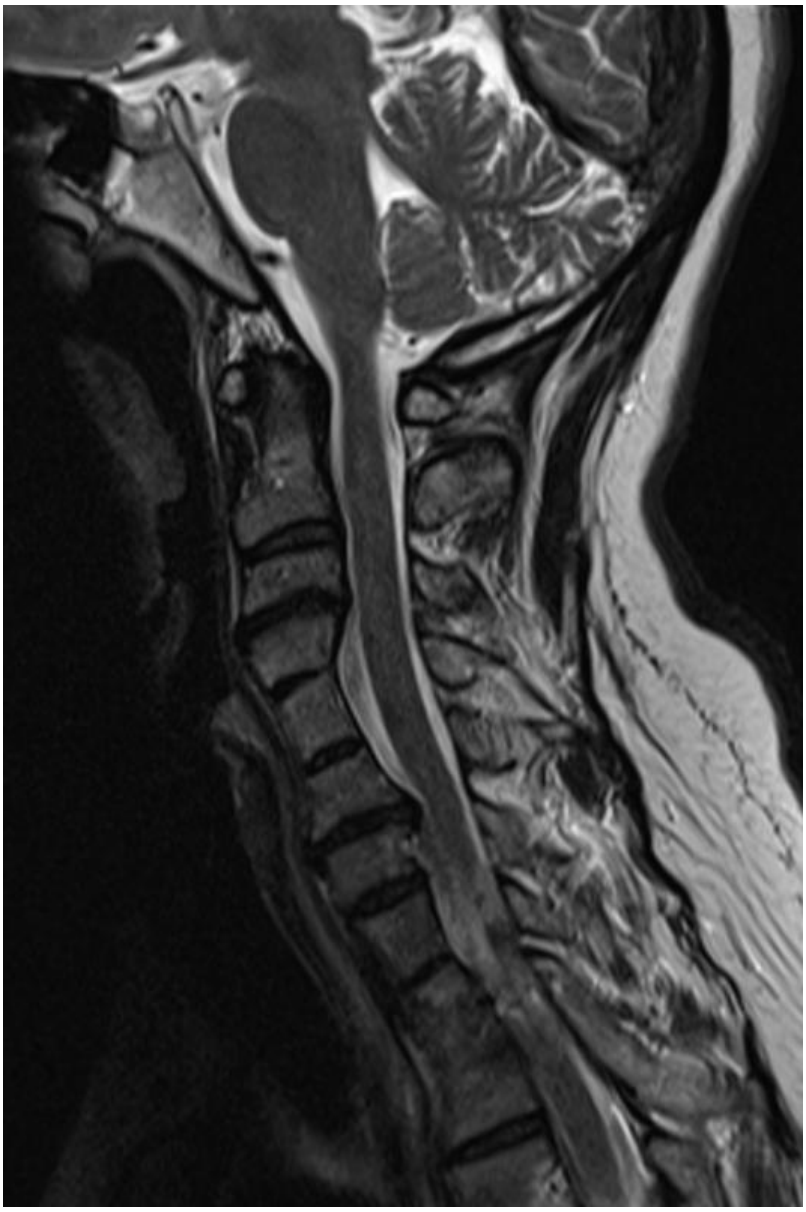
Figure 2

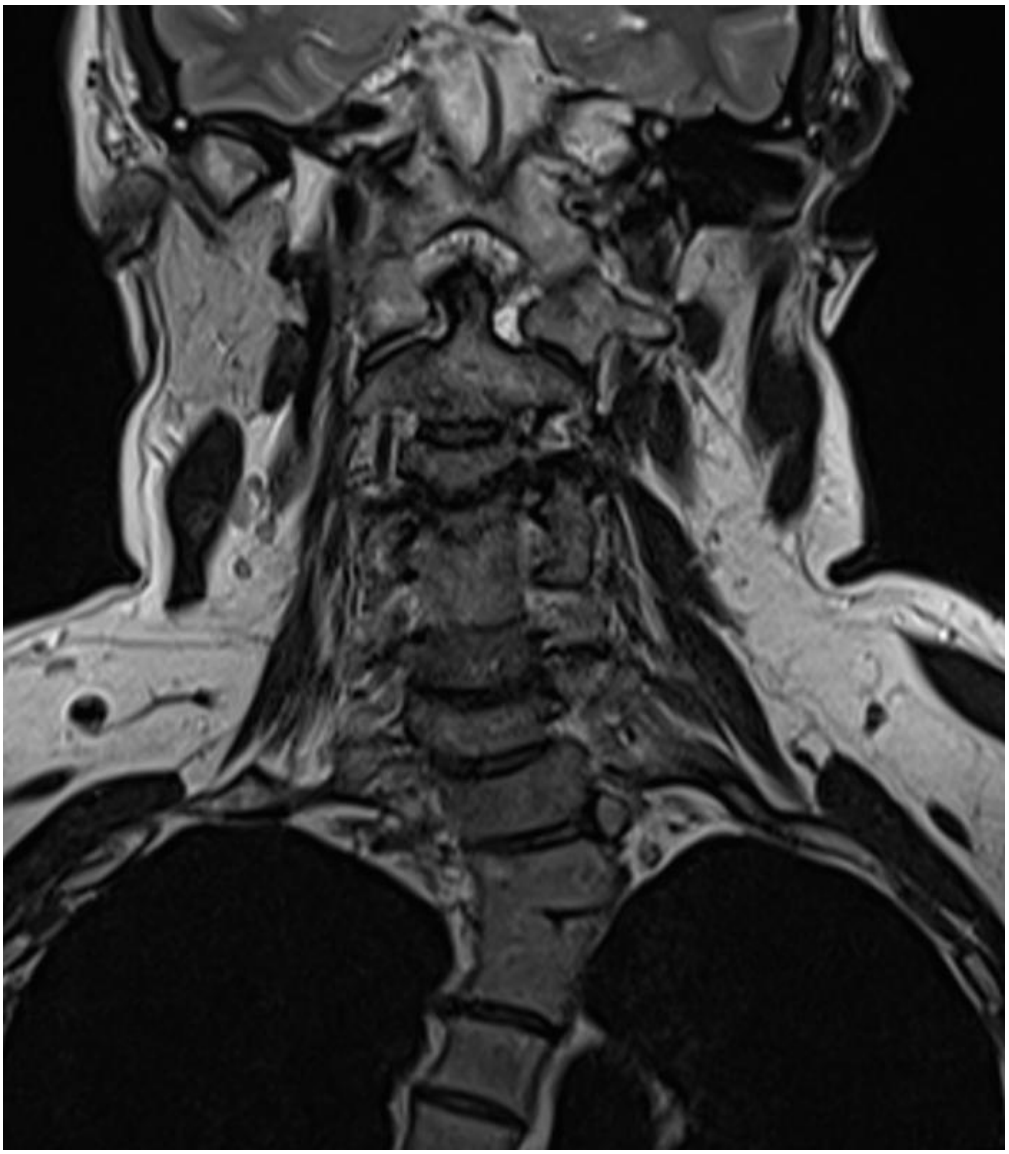
a



Description: Sagittal T1w MR sequence shows partial fusion of C4-C5-C6 associated with narrowing of the anteroposterior diameter of the vertebral bodies, as well as a decrease in the development of the corresponding intervertebral discs. Fusion of D2-D3-D4 vertebral bodies with partial development of the intervertebral disc D2-D3 and absence of a disc at the D3-D4 level **Origin:** Departamento de Radiología del Centro de Diagnóstico por Imágenes de la Clínica Internacional Lima, Perú, 2023

Figure 3

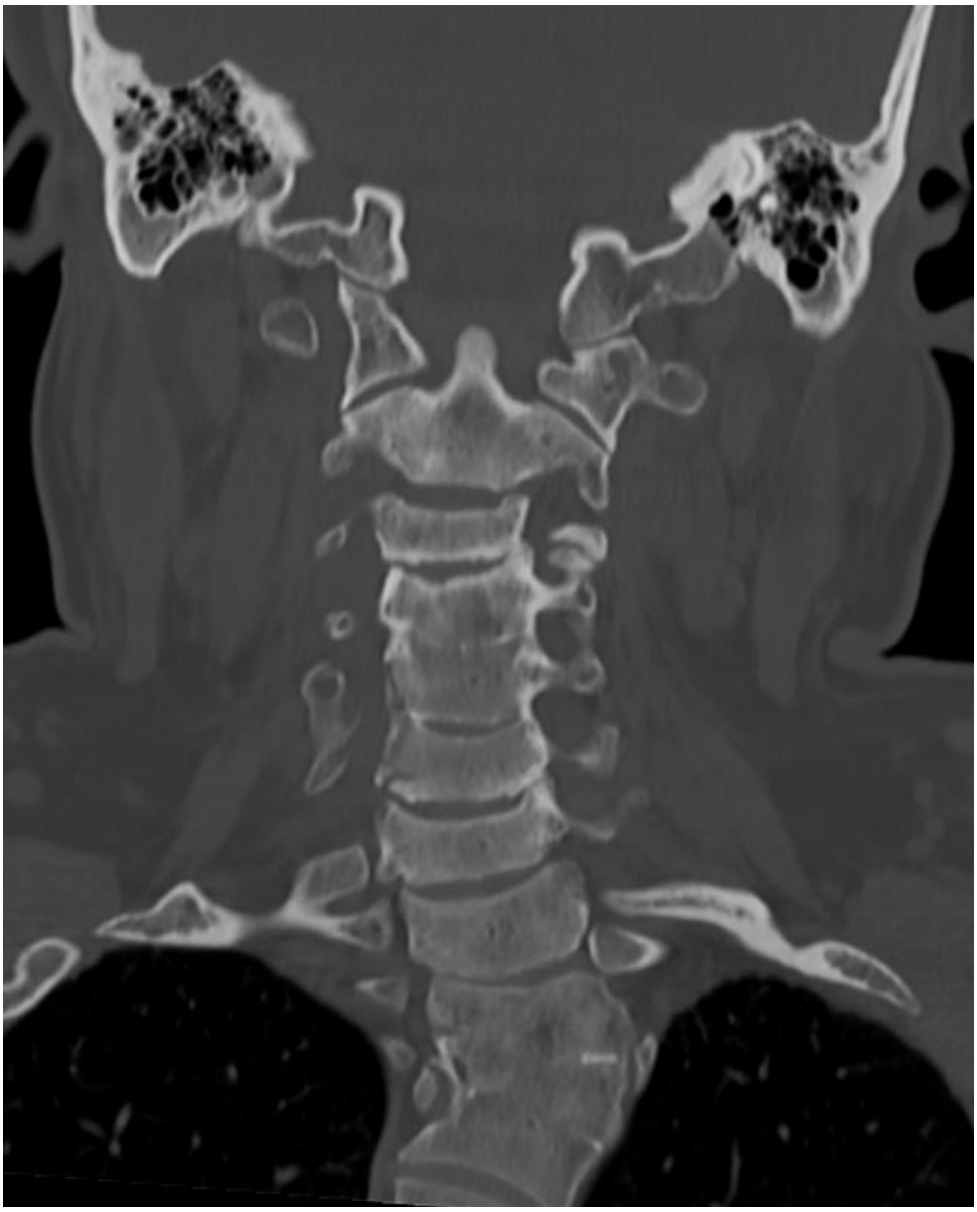
a



Description: Coronal T1w MR sequence shows signs of lateral hemivertebra or wedged vertebra at D3 leading to left convex dorsal scoliosis **Origin:** Departamento de Radiología del Centro de Diagnóstico por Imágenes de la Clínica Internacional Lima, Perú, 2023

Figure 4

a



Description: CT coronal demonstrates the presence of a right cervical rib fused with the first dorsal rib
Origin: Departamento de Radiología del Centro de Diagnóstico por Imágenes de la Clínica Internacional Lima, Perú, 2023