A 12-year-old boy with a previous medical history of tetralogy of Fallot, bilateral hearing loss and small stature, presented with limitation of neck movement after a minor neck trauma.

On clinical examination, mild dextroconvex thoracolumbar scoliosis was seen, and therefore radiographs of the full spine and the cervical spine were requested.

Imaging Findings:

Conventional radiography of the cervical spine (Fig. 1-2) showed a congenital bony fusion of four vertebrae with a wasp-waist sign (C4-C7). Complete fusion of the facet joints at the level C4-C5, C5-C6 and partial fusion of the facet joints at the level C6-C7. In addition, there is a fusion of the lamina and spinous processes of C4-C7. The disk space C4-C5, C5-C6 and C6-C7 is rudimentary with a compensatory broad disk space C2-C3 and C3-C4.

Plain films of the full spine (Fig. 3) demonstrated an S-shaped scoliosis of the thoracic and lumbar spine. Furthermore, there are postoperative findings after cardiac surgery consistent with the history of tetralogy of Fallot.

Discussion:

Klippel-Feil syndrome is defined by congenital fusion of at least two cervical vertebrae with an absence of the intervertebral discs. The syndrome was first described by Maurice Klippel and Andre Feil in 1912. It is a result of failure of normal segmentation and fusion processes of the cervical somites, which occur between the third and seventh week of embryogenic development [2]. The incidence of Klippel-Feil syndrome is approximately 1:42000. The syndrome is slightly more common in women (60%) than in men [2].

The classical triad consists of a short neck, limitation of head and neck movements and a low posterior hairline. This triad is seen in 50% of patients [3]. Klippel-Feil can be associated with several organ anomalies such as congenital heart defects (most often ventricular septal defect), deafness, learning disabilities and renal anomalies [2]. Other skeletal deformities such as scoliosis, malformed ribs and a Sprengel deformity (rotated and elevated scapula) are also common findings [3].

Klippel-Feil syndrome is classified into three categories. In type I, there are numerous fused cervical vertebrae and upper thoracic vertebrae. In type II, the patient presents with fusion of one or two vertebrae, most often C2-C3 or C5-C6. In type III, fusion of both cervical and lower thoracic or lumbar vertebrae occur. This type is often associated with the above mentioned systemic anomalies [3].

Most of the patients are asymptomatic. However, there is an increased risk of developing spontaneous or post-traumatic neurological injury. Patients may present with radiculopathy, myelopathy or quadriplegia caused by
spondylolysis or instability of vertebrae adjacent to the fused segment [1]. Furthermore, there is a risk for early
development of degenerative arthritis due to increased mobility at the normal segments adjacent to the congenital
fused segment [4].
When the diagnosis of Klippel-Feil is suspected, one can perform conventional radiographs of the cervical spine for
detection of fusion of the vertebral bodies. Other radiographic findings include fusion of the facet joints and spinous
processes. Imaging may sometimes show the wasp-waist sign (Fig. 4) The wasp-waist sign refers to the vertebral
body narrowing and associated concavity of the anterior and posterior surfaces of the vertebrae at the level of the
fused segment [5]. Plain radiography of the full spine is useful for evaluation and follow-up of an associated
scoliosis. MRI may be helpful to evaluate degenerative disk disease, narrowing of the spinal canal and myelopathy
when a patient presents with neurological symptoms [6].

**Differential Diagnosis List:** Klippel-Feil syndrome with several associated anomalies., History of vertebral fusion
surgery, Ankylosing spondylitis

**Final Diagnosis:** Klippel-Feil syndrome with several associated anomalies.

**References:**

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Figure 1

**Description:** AP view of the full spine. Congenital S-shaped scoliosis. **Origin:** Department of Radiology, Antwerp University Hospital, Edegem, Belgium
Figure 2

Description: Wasp-waist sign. Vertebral narrowing and concave anterior and posterior cortex at the level of the affected disc spaces (black arrows). Origin: courtesy of Kevin Van Looveren
Figure 3

**Description:** AP view of the cervical spine. White arrows indicate fusion of the vertebral bodies. Postoperative findings after cardiac surgery. **Origin:** Department of Radiology, Antwerp University Hospital, Edegem, Belgium
Figure 4

Description: Lateral view of the cervical spine demonstrates the wasp-waist sign (arrowhead). Fusion of the posterior elements: facet joints (dashed arrow) and spinous processes (white arrow). Origin: Department of Radiology, Antwerp University Hospital, Edegem, Belgium