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(CASE REPORT)

Klippel-Feil syndrome associated with congenital megacolon: a case report

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Abstract

Klippel-Feil syndrome is a congenital abnormality characterized by the fusion of the bones of the cervical spine. Its association with a gastrointestinal abnormality is rare. We report a case of a 14-year-old girl affected by Klippel-Feil syndrome associated with congenital megacolon. Klippel-Feil syndrome should be considered as any polymalformative syndrome and its management should be multidisciplinary.

Keywords: Klippel-Feil syndrome; Congenital megacolon; Cervical spinal; Clinical case

1. Introduction

Klippel-Feil syndrome (KFS) is a complex condition presenting due to abnormal fusion of cervical vertebrae. KFS is caused by a defect in the normal segmentation of the cervical spine vertebrae during early fetal development [1]. The prevalence of KFS is evaluated at 0.0058% (1 in 172) [2].

The KFS can be associated with non-musculoskeletal abnormalities [1]. However, a gastrointestinal abnormality is rare. In this case, we report a 14-year-old girl affected by KFS associated with congenital megacolon; which we will discuss in light of the literature.

2. Patient and observation

A 14-year-old girl from a consanguineous marriage was born after a normal vaginal delivery with a good Apgar score. She is in school with good psychomotor development.

The history of the disease goes back to the age of about 5 years with intermittent constipation, treated symptomatically at home. This clinical picture has never been investigated. For the past month, she has been experiencing urinary incontinence associated with constipation for more than six days and cramping sensations in the lower limbs, which motivated the parents to consult the pediatric department. Therefore, the girl was referred to our service.

The clinical examination noticed a normal neck, hairline above the nape of the neck, and the limitation of flexion-extension in the neck's motion. She had posteriorly rotated ears. There was no motor or sensory deficit. Her ankle and knee reflexes were normal. The abdomen was not bloated, slightly painful in places. The rectal exam had not been done.

MRI of the spine revealed fusion of the cervical vertebrae at C6-C7-T1 without spinal cord abnormalities and loss of sacral kyphosis (figure 1). We also performed a full-body CT scan to seek other abnormalities. It found an abnormality of the vertebral body of L3 and congenital megacolon (figure 2).

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The treatment was mainly medical and symptomatic. She was then referred to the gastroenterology department for better management.

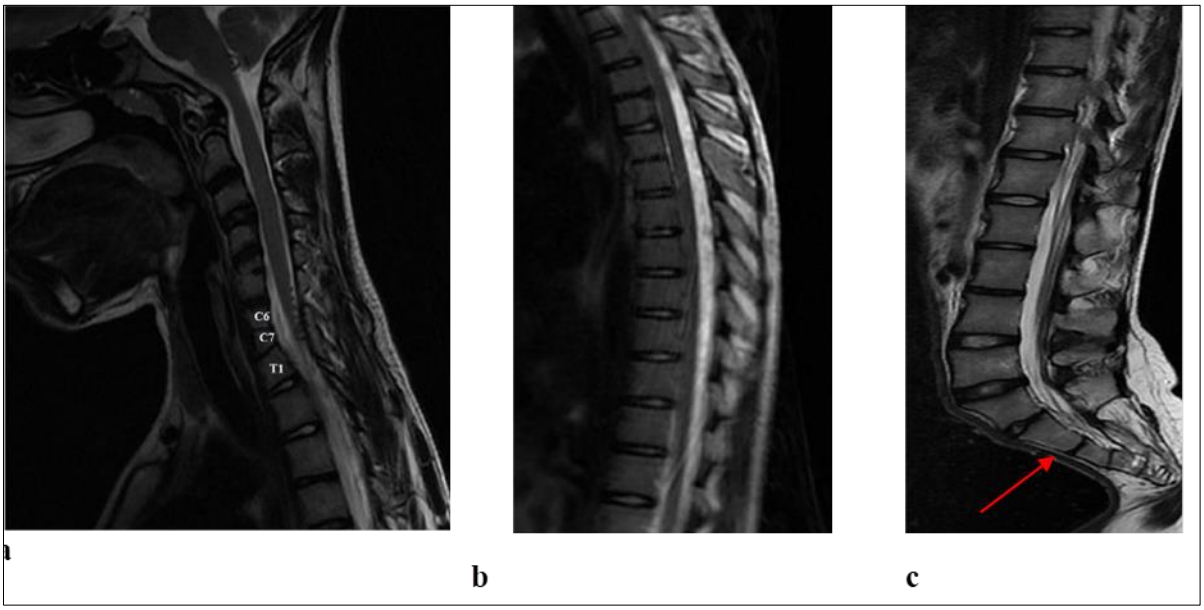

Figure 1 Fusion of the cervical vertebrae at C6-C7-T1 in MRI (a). Dorsal spinal cord (b) and cauda equina are free (c). Loss of sacral kyphosis (red) (c)

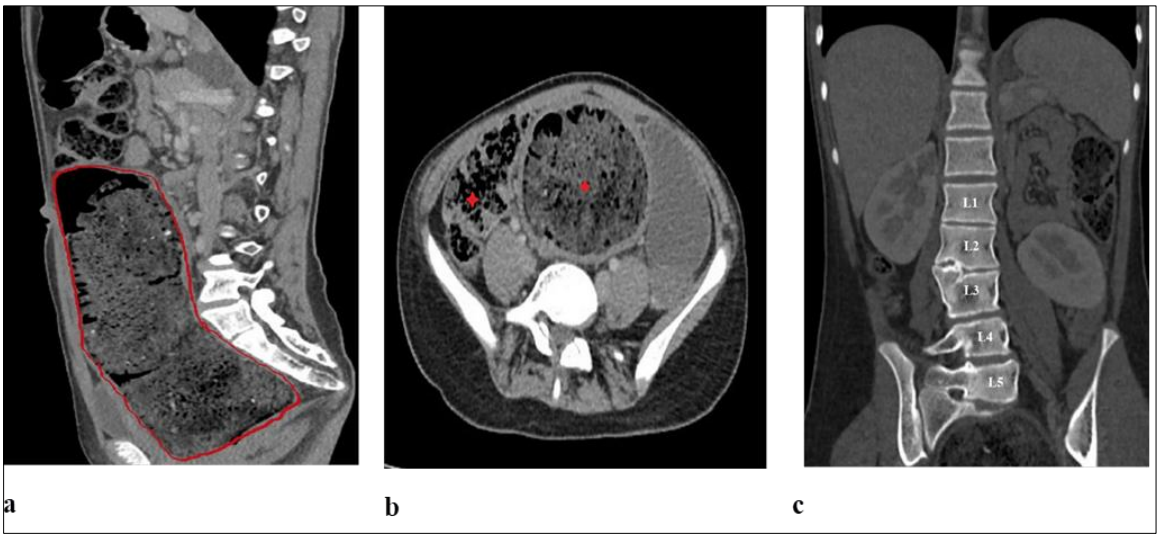

Figure 2 Congenital megacolon surrounded in red color in sagittal CT (a) and red stars in axial CT (b). Congenital abnormality of the vertebral body of L3 (c)

3. Discussion

The association of KFS and gastrointestinal abnormality is a rare pathological entity. Despite a relatively low incidence of 2%, as reported by Nouri et al for KFS [3], the most common associated abnormalities affect the musculoskeletal system, central nervous system, cardiovascular system, urogenital system, eyes, ears, and respiratory system [4].

KFS is characterized by a clinical triad short neck with reduced mobility and a low posterior hairline [5]. This manifestation occurs only in 40–50% of patients [4, 5]. In our case, this clinical triad was not complete. Hence the interest in complementary imaging examinations, which allow to highlight vertebral fusion. Three morphological subtypes of this abnormality were described according to radiographic features [4]. In our case, it is KFS type I.

Congenital megacolon and KFS were reported by Kopysc and Borowicz in 1957 [6]. Its clinical manifestation is common to Hirschsprung's disease and idiopathic megacolon [7, 8]. The main symptom is constipation. Diagnosis of congenital megacolon is easy thanks to new imaging technologies, such as CT and MRI.

The etiology of KFS is not well known. Several studies have assumed hypotheses such as vascular disruption, primary neural tube complications, or related genetic factors in the development of KFS [9].

The challenge of the management of KFS is to recognize the concomitant anomalies and to perform the appropriate workup [4]. In our case, the symptomatology relative to a suffering of the abdominal viscera was in the first place, simulating signs of spinal cord compression. Thus, abnormalities associated with KFS may delay diagnosis. The management of KFS should be multidisciplinary. In our case, the girl was referred to the gastroenterology department, with regular follow-up in the neurosurgery department.

4. Conclusion

The association of KFS with congenital megacolon is rare. KFS should be considered as any polymalformative syndrome requiring musculoskeletal and non-musculoskeletal investigations. The management of KFS should be multidisciplinary.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare that they have no conflicts of interest.

Statement of informed consent

Informed consent and verbal permission were obtained from the patient and her family before the submission of this article. In addition, this article follows both the Consensus-based Clinical Case Reporting Guideline and the Recommendations for the Conducting, Reporting, Editing, and Publication of Scholarly Work in Medical Journals.

Disclosure

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