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"Congenital instability of cervical spine in a pediatric patient with cleft lip and palate"

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ABSTRACT

Introduction: Cervical spine anomalies can coexist with anomalies of the head and neck. The association of cervical vertebrae anomalies (CVA) with cleft lip and palate (CLP) has been described as probably the result of a failure in normal embryological segmentation. The most frequent congenital alterations of cervical spine CLP-related are vertebral fusion (VF) and deficiency of the posterior arch (PAD).

Case Presentation: We report a case of an acute, non-traumatic onset of paraparesis in a 14 years old girl with history of CLP and bilateral conductive deafness. Magnetic resonance (MR) and Computed Tomography (CT) imaging of the cervical spine revealed C4-C5 myelopathy sign and a misunderstood C2-C3 and C5-C6 partial posterior VF. A C2 deficiency of the posterior arch was also present. Dynamic X-Rays showed a junctional instability of C4-C5 metamers. The patient was surgically treated with anterior cervical discectomy and fusion (ACDF) with immediate improvement of the symptoms.

Conclusions: The new onset of spinal cord involvement in pediatric patients with a history of head and neck developmental disorder could be ascribed to congenital cervical spine malformation and therefore should be detected by appropriated radiological imaging as early as possible to optimize surgical management and reducing the risk of neurological impairment.

1. Introduction

Congenital anomalies of the cervical spine in pediatric patients are rare conditions that should be early recognized and carefully managed to prevent neurologic impairment. In some cases, cervical spine anomalies are associated with evident organs abnormalities as manifestation of hereditary and systemic diseases. Skeletal dysplasia (i.e. Osteogenesis imperfecta, Neurofibromatosis), connective tissue disorders (i.e. Marfan syndrome), inflammatory arthritis (i.e. juvenile rheumatoid arthritis) or congenital disorders (Klippel-Feil syndrome, KFS) are the commonest and most severe pediatric disorders with involvement of the cervical spine [1,2].

Moreover, cervical anomalies with mild representation can be misdiagnosed if not framed in specific syndromes or in congenital disorders. The association of cervical vertebrae anomalies (CVA) and orofacial abnormalities such as cleft lip and palate (CLP) has been reported since 1965 [3]. Many studies focused on embryogenesis development of the neck and maxillofacial complex have contributed to understand the possible association among cervical anomalies and orofacial and acoustic disorders and to explain the etiology of CLP [3]; however, strong evidence of this association is currently insufficient [4,5].

CLP disorder can result in complications affecting feeding, speech, hearing and psychological development. During childhood, these individuals undergo several procedures to restore normal maxillofacial and functions, and generally are asymptomatic for cervical spine involvement. The cervical spine is generally not screened for associated anomalies and CVA is discovered incidentally during the second or third decade of life [6].

Prevalence of cervical vertebrae anomalies in patients with cleft lip and palate reach 20.3% [7].

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The most frequent congenital alterations of cervical spine CLP-related are vertebral fusion (VF) and deficiency of the posterior arch (PAD) [8]. Patients with VF may be predisposed to degenerative changes and hypermobility at segments adjacent to fused vertebrae in adulthood [9].[1]

We report a case of an acute, non-traumatic onset of paraparesis in a misdiagnosed congenital instability of cervical spine in a pediatric patient with cleft lip and palate. To our knowledge this is the first reported case of early occurrence of sudden cervical myelopathy in a patient affected by congenital CLP, bilateral hearing loss and cervical spine anomalies. The aim of this case presentation is to enforce the importance to investigate cervical spine in presence of multiple anomalies of the head and neck, even in non-syndromic patients.

2. Case report

A young patient of 14 years old presented two acute, non-traumatic onsets of transient para/tetraparesis initially not related to a precise diagnosis. The neurological examination documented impaired sensation below the C5 dermatome with numbness and dysesthesia of the lower limbs and increased muscle hypertonus, and presence of Babinski and Lermitte signs.

Previous surgical procedures of CLP repair (at the age of 2 years old) were reported in patient's clinical history. A bilateral hearing aid device (BiCros system: Bilateral Controlateral Routing of Signal) was applied for a transmissional hearing loss. Genetic evaluation was negative for congenital disorder.

A MRI of brain and cervical spine revealed an increased T2 signal within the cord at C4-C5 (Fig. 1). Subsequently CT scan showed a C5-C6 VF previously misdiagnosed as osseous continuities without complete separation at the intervertebral disc. A severe dynamic instability and anterolisthesis of C4, demonstrated by the flexion- extension X rays was the main cause of spinal cord compression (Fig. 1). Therefore, an anterior cervical discectomy and fusion of C4-C5 (ACDF) procedure was performed to provide immediate stability to the cervical spine (Fig. 2). Fully informed written consent was obtained regarding the risks and benefits of the procedure.

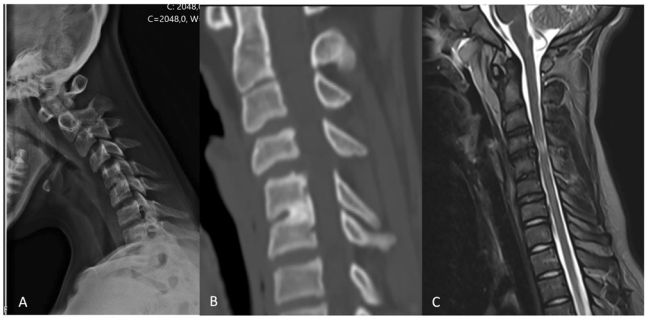

Fig. 1. Preoperative imaging. A) preoperative Flexion Cervical X-Ray showing C5-C6 posterior vertebral body fusion and C4 anterior listhesis. B) Preoperative sagittal CT-scan that confirm C5-C6 posterior vertebral body fusion and the reduction of C4 listhesis in neutral position. C) Preoperative sagittal MRI T2 weighted image showing hyperintense signal of the spinal cord in C4-C5, above the level of CV fusion.

[1] Abbreviations: Klippel-Feil syndrome (KFS), cleft lip and palate (CLP), isolated CP (ICP); unilateral CLP (UCLP) and bilateral CLP (BCLP), cervical vertebrae anomalies (CVA), Magnetic resonance (MR) and Computed Tomography (CT), vertebral fusion (VF), deficiency of the posterior arch (PAD), recurrent laryngeal nerve palsy (RLNP), sternocleidomastoid (SCM), motor and somatosensory evoked potential (SSEP and MEP), ACDF: anterior cervical discectomy and fusion

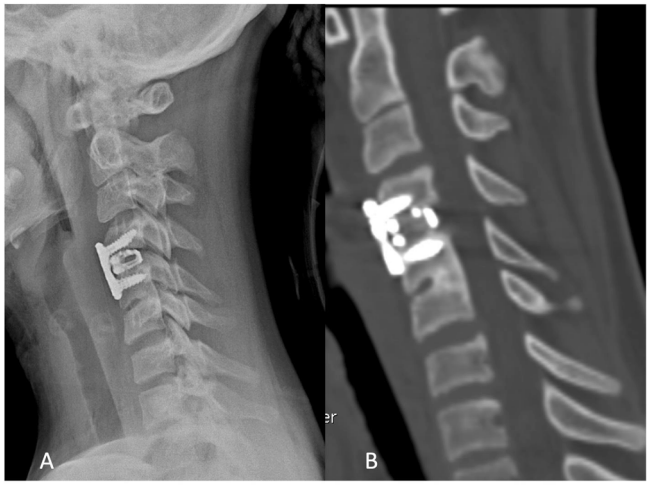

Fig. 2. Postoperative imaging. A): Postoperative lateral X-Ray (A) and sagittal CT-scan (B) showing C4-C5 interbody fusion and anterior plating.

The patient was moved on the surgical table, and immobilized with cervical collar. A general anesthesia was performed with endoscopic endonasal intubation to avoid extreme extension of the neck. Patient was then placed in a supine position. Preoperative radiological imaging excluded other anatomical anomalies of the neck and therefore a standard pre-sternocleidomastoid (SCM) muscles transvers skin incision was performed. Intraoperative neuromonitoring was obtained with combined motor and somatosensory evoked potential (SSEP and MEP). The anterior cervical spine was reached from the left side to reduce the risk of recurrent laryngeal nerve palsy (RLNP), as the nerve is more reliably protected within the tracheoesophageal interval on the left side. The treatment for cervical instability aims to obtain spinal cord decompression, correction, and fusion. Once the C4-C5 disc space was exposed, anterior complete discectomy was completed. An intersomatic titanium cage filled by heterologous bone was implanted to provide intersomatic fusion and anterior listhesis reduction. The cage was fixed with an anterior plate to improve primary stability. At the end of surgery, the C4 listhesis was completely reduced, without neurological or surgical problems.

Postoperative clinical follow-up after 6 and 12 months showed great improvement of myelopathy and absence of any neurological signs (normal muscles tone), and complete recovery of symptoms as numbness and dysesthesia of the lower limbs. The 6 months MRI showed great improvement of radiological signs of spinal cord signal (Fig. 3) and the correct alignment of the cervical spine.

3. Discussion

Nontraumatic congenital cervical spine instability is caused by rare conditions, which sometimes are difficult to diagnose during childhood [2]. Our young patient presented a dynamic cervical instability above a VF. According to the literature, the instability can be triggered by the mechanical stress, which is a consequence of the vertebral fusion. The presence of a rigid segment, associated with ligamentous laxity and immature neck musculature can modify the physiological range of motion of the spinal segment (including 2 vertebral bodies, the intervertebral disc, joints, and ligaments), resulting in instability [10,11]. In our case, a nontraumatic cervical spine instability triggered also neurological impairment. Therefore, a surgical treatment was required to decompress the spinal cord, realign, and fuse the segment of motion. An anterior approach was chosen to achieve direct spinal cord decompression, and consequent progressive neurological improvement. A posterior cervical decompression or fusion in this case was not

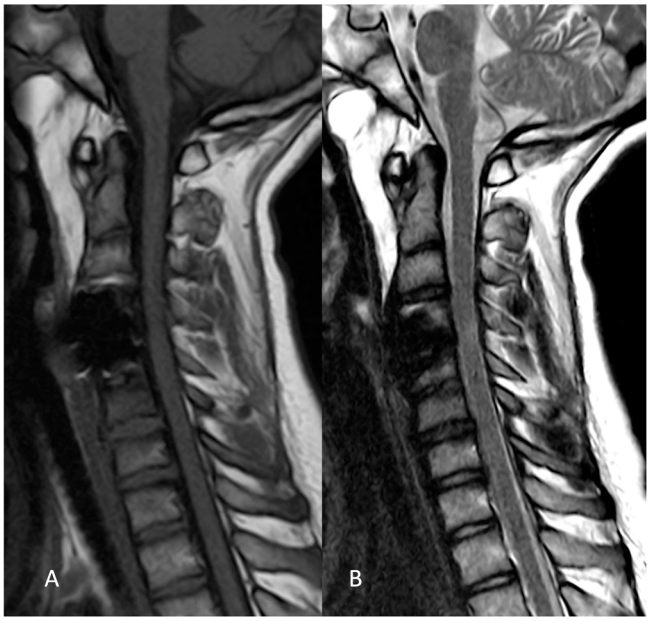

Fig. 3. Follow-up MRI A-B) Six-months follow-up MRI in T1 (A) and T2 (B) weighted image showing the radiological improvement of preoperative sign of myelopathy.

performed because the anterior approach allowed for the optimal listhesis reduction; moreover, the posterior approach may have led to posterior muscles impairment.

The importance of MRI imaging in the early stage of symptoms onset is crucial. In absence of clear syndromic diseases, other bony pathologies of the spine, such as eosinophilic granuloma or benign osteoblastoma of the spine should be ruled out [12].

Primitive tumors of the spine have usually an history of increasing local pain and progressive neurological impairment. Tumors can lead to vertebral instability mainly due to the osteolitic effect of the tumor cells [12].

The CVA can coexist with development anomalies of the head and neck. According to Samartzis classification [2], the congenital cervical VF can be classified in three types based on level of fusion. Single congenitally fused cervical segment characterizes the type I; multiple noncontiguous congenitally fused segments characterize the type II, whilst Type III is represented by multiple contiguous congenitally fused cervical segments.

Young subjects with congenitally fused cervical vertebrae present an increased risk of degenerative changes of the contiguous metamers that can lead to myelopathy in adulthood [9].

Several mechanisms of spinal cord injuries have been proposed, such as the coexistence of spinal cord anomalies, or congenital canal narrowing, vertebral instability and vascular dysfunctions [11].

In adult patients, the fused vertebra may contribute to alter the stress forces and/or a degenerative cervical process, thus causing instability [9,10].

In pediatric population, neurological involvement of the cervical spine is described in patients affected by specific syndromes such the KFS, which is characterized by short and stiff neck, abnormal fusion of at least two vertebrae and low hairline [10]. In these patients neurological changes slowly progress or they can be worsened by minor traumas, especially in presence of occipitocervical abnormalities [11]. Embryogenetic disorder may explain the association of non-syndromic cervical spine and head/neck anomalies [3].

The type of CVA (VF, PAD, vertebral artery canal, anomalies of the anterior arch in C1, odontoid process abnormality), the level (upper or subaxial cervical spine) and the number of involved segments may be associated with different kinds of cleft lip and palate anomalies: isolated CP (ICP); unilateral CLP (UCLP) and bilateral CLP (BCLP). According to the literature, the prevalence of CVA is highest in the UCLP (52.8% of PAD, 33.9% of fusions) and BCLP groups (56.0% of PAD; 32.0% of fusions) [3]. However, no strong relations with types of cleft lip and palate and cervical anomalies have been observed [3]. Moreover, no significant difference in prevalence of CVA between males and females was found [3].

The diagnosis of non-syndromic CLP during childhood is frequently associated with transmissional hearing loss. Despite the higher prevalence of CVA in CLP patients [3], cervical anomalies are not routinely investigated in those subjects, thus increasing the risk of neurological damages.

In this case report (type II of Samartzis classification and bilateral CLP, BCLP) the two noncontiguous C2-C3 and C5-C6 fusions produced a pivot on the C4-C5 “healthy” disc, leading to segmental instability and then to spinal cord progressive injury.

Hence, interbody fusion was necessary to obtain anterior column support and stability. At the last follow-up complete neurological improvement has been observed, probably thank to the young age of our patient.

The study has some limitations; as we could not establish the effective cause of the cervical instability, that could be only postulated according to the literature. Moreover, we did not investigated the interbody fusion by CT scan after surgery, to avoid an excessive radiation exposure for our young patient.

4. Conclusions

A strong association among cervical anomalies and cleft patients is well known. Therefore, a cervical spine screening by MRI of CLP patients should be advocated to avoid sudden and early spinal cord injury in those subjects. A new onset of neurological symptoms in pediatric patients with a story of CLP, should be detected by radiographic imaging (MRI and cervical X-Rays) as early as possible to optimize management and to reduce the risk of further neurologic impairments.

CRediT authorship contribution statement

Carlotta Morselli: Writing – original draft, Writing – review & editing. **Patrizia Mancini:** Writing – review & editing. **Agostino Cirullo:** Writing – review & editing. **Laura Mangiavini:** Writing – review & editing. **Roberto Bassani:** Conceptualization, Supervision, Writing – review & editing.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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