CASE REPORT

More Than Just a Torticollis: A Klippel-Feil Syndrome Case Report

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Abstract

Although often benign, torticollis may be the manifestation of a serious disease, such as Klippel-Feil syndrome. We report a pediatric clinical case of an isolated form of Klippel-Feil syndrome. A 6-month-old male infant, whose previous history is restricted to a sacrococcygeal dimple, developed limitation of cervical mobility, presenting with a short neck, low implementation of the posterior hairline and scoliosis, with normal psychomotor development. There was no improvement after conservative treatment. The medullary magnetic resonance revealed cervical scoliosis, rudimentary disc spaces, fusion of the posterior elements, and hydromyelic dilatation of the ependymal canal. No other changes were found in the multidisciplinary evaluation and no disease was found in the parents. Klippel-Feil is rare. It can manifest either isolated or associated with other malformation, such as skeletal, cardiac, or nephrourological. If this syndrome is suspected, both the patient and his family should be evaluated by a multidisciplinary team.

Keywords: Infant; Klippel-Feil syndrome/diagnostic imaging; Klippel-Feil Syndrome/complications; Torticollis/etiology

Introduction

Klippel-Feil syndrome is a rare entity in the pediatric age group with an estimate prevalence of 1:40,000-42,000. This congenital malformation can occur as a *de novo* event or be inherited as an autosomal dominant (caused by *GDF3* and *GDF6* genes) or autosomal recessive (caused by *MEOX1* and *MYO18B* genes) pattern. Klippel-Feil syndrome is characterized by the presence of congenital synostosis of some or all cervical vertebra developed between the third and eighth week of gestation. The classical triad consists of limited neck mobility, short neck, and low posterior hairline, and it is present in 50% of the cases. Cervical mobility impairment is the most common feature, especially in patients with extensive vertebral fusion.[1]

The classification proposed by Feil in 1919 distinguishes three types of the Klippel-Feil syndrome - I, II, and III - depending on whether there is only the fusion of one segment of vertebra, multiple noncontiguous segments, or contiguous multiple segments, respectively. More recently, there was a better understanding of the dynamics and complexities of Klippel-Feil syndrome, particularly in pediatric patients, which seem to have a type II predominance, although, overall, in male sex type III predominates.[2] A different classification (types 1-4) based on patterns of inheritance, associated anomalies, and the axial level of the most anterior fusion was proposed in 1995.[3]

In addition to vertebral fusion malformation, Klippel-Feil syndrome may be associated with other musculoskeletal malformations, such as scoliosis. Cardiac,[4,5] nephro-urological,[6] ophthalmologic,[7] and audiological abnormalities are also described.[8-10] Regarding treatment, a surgical approach is rarely indicated, except in cases of the instability of the cervical spine or in myelopathy and radiculopathy that are refractory to conservative treatment. The later depends on the other anomalies.

We report a pediatric clinical case of an isolated form of Klippel-Feil syndrome, discussing its etiology, therapeutic approach as well as the most probable differential diagnoses.

Case Report

A 3-year-old male, son of a non-consanguineous healthy couple, with irrelevant antenatal history was detected in the neonatal period with a simple sacrococcygeal dimple was detected. The evaluation by ultrasonography

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revealed a fistulous pathway of the spinal canal to the skin lining without cutaneous fistula. There was no further investigation at this point.

At 6 months, the cervical position in left antero-lateral flexion with mobility limitation was noticed, with no other alterations to physical examination. His neurodevelopment and growth were adequate. Cervical ultrasound and radiography showed no relevant alterations. Conservative treatment with physical therapy was initiated, without clinical improvement.

At 14 months, there was evidence of a short neck, a cervical tilt to the left, with a low posterior hairline implantation and sinister-convex scoliosis (Fig. 1).

For etiological clarification, a cervical and lumbosacral magnetic resonance was performed, confirming sinister-convex cervical scoliosis with rudimentary disc spaces (Fig. 2) and the fusion of posterior elements between cervical vertebra C2-C3 and C3-C4. Hydromyelia (D5 - medullary cone) and moderate coccygeal hypoplasia were also identified. There was no imaging suggesting closed spinal dysraphism.

Evaluations by pediatric cardiology, ophthalmology, otorhinolaryngology, and nephrology did not detect any associated alterations. The patient's parents were consulted by a clinical geneticist and, after analysis of the cervical radiographs, it was concluded that they are not affected.

The case presented here is a case of isolated Klippel-Feil Syndrome, probably sporadic or *de novo* event. The patient was re-evaluated in consultation by a multidisciplinary team. In Fig. 4, there is a photograph of the child at 3 years old, maintaining the cervical tilt to the left.

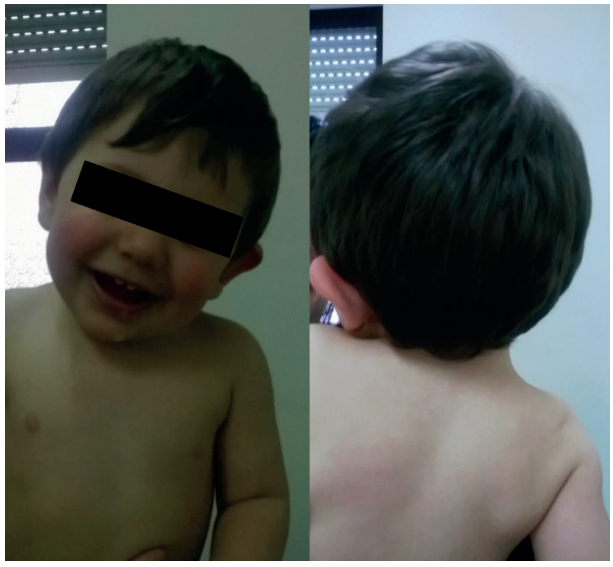

Figure 1. The child at 14 months with evidence of a short neck, a cervical tilt to the left, with a low posterior hairline implantation and sinister-convex scoliosis.

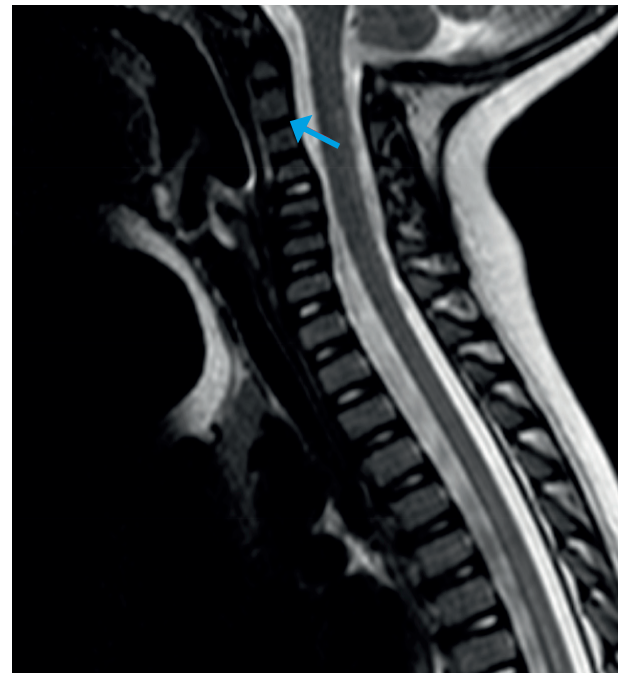

Figure 2. Magnetic resonance image confirming sinister-convex cervical scoliosis with rudimentary disc spaces.

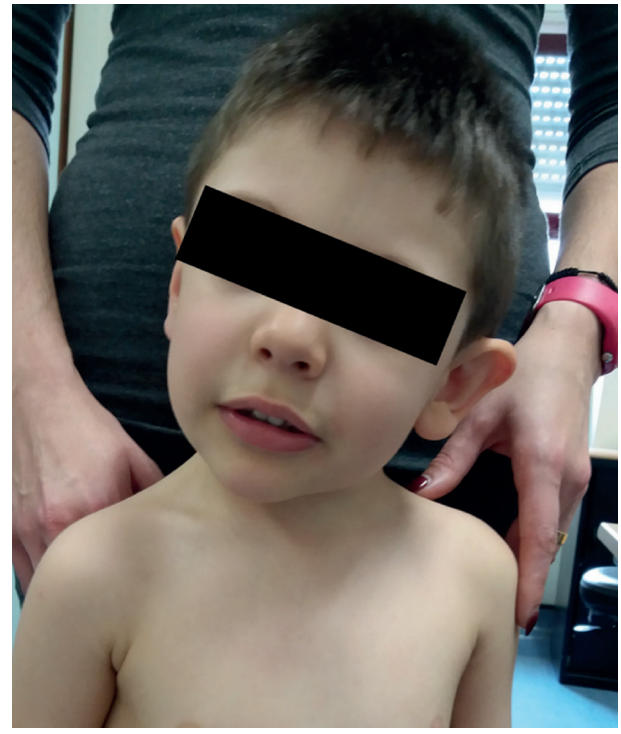

Figure 3. The child at 3 years old, maintaining the cervical tilt to the left.

Discussion

Torticollis is not a diagnosis *per se* and may be a manifestation of an underlying pathology. In the presented case, the differential diagnoses to be considered are

congenital anomalies that present as neck asymmetry and a limitation of cervical mobility such as congenital muscular torticollis, the third most common congenital malformation, and some acquired alterations, such as inflammation in infections, joint injuries, or tumors. Klippel-Feil syndrome may be a feature of another condition as part of other syndromes.[11] During follow-up, patients may present with neurological complications, such as spinal cord compression, especially at a cervical level, with complaints of the loss of function of the extremities (myelopathy) or pain and muscle weakness (radiculopathy).

Klippel-Feil syndrome may be caused by mutations in the *GDF6* or *GDF3* gene and inherited in an autosomal dominant manner. It may also be caused by mutations in the *MEOX1* or *MYO18B* genes and inherited in an autosomal recessive manner. According to some authors, Klippel-Feil syndrome 1 has a recessive inheritance pattern, Klippel-Feil syndrome 2 has an autosomal dominant pattern, and in Klippel-Feil syndrome 3, there is a recessive pattern or reduced penetrance. Klippel-Feil syndrome includes cases of Wildervank and Duane syndrome.[3]

Considering the findings in the study by medullary magnetic resonance imaging, the absence of other malformations, such as spinal dysraphism and the absence of pathological findings in the parents, it was interpreted that the most likely hypothesis was Klippel-Feil syndrome as an isolated feature. For this reason, the recurrence risk for this couple in future gestation is low and sporadic. In turn, the child himself may have a risk of 50% of having an affected offspring. Consequently, genetic testing is required when intending to start a family.

Therefore, even in the absence of the classic triad of Klippel-Feil syndrome, if there is a high level of clinical suspicion, it would be advisable for the patient and family to be evaluated by a multidisciplinary team, given the features that may involve various organs and systems. This is particularly true when clinical findings are associated with other spinal changes, such as scoliosis, and in this age group, since the complete triad is only present in 50% of cases.[1]

WHAT THIS CASE REPORT ADDS

- Torticollis is often benign and no further studies are necessary.
- Clinicians should be aware of the clinical manifestations of Klippel-Feil syndrome, even in the absence of the clinical triad.
- A multidisciplinary approach is mandatory.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Consent for publication

Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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Mais do que Apenas um Torcicolo: Um Caso Clínico de Síndrome de Klippel-Feil

Resumo:
Apesar de habitualmente benigno, o torcicolo pode ser a manifestação de uma doença grave, como é o caso da síndrome de Klipple-Feil. Os autores apresentam um caso clínico pediátrico de uma forma isolada de síndrome de Klipple-Feil. Lactente de 6 meses, sexo masculino, cujos antecedentes relevantes se limitam a uma fosseta sacrococcígea, desenvolveu limitação da mobilidade cervical, objetivando-se pescoço curto, baixa implantação da linha posterior do cabelo e escoliose, com desenvolvimento psicomotor normal. Não houve melhoria com tratamento conservador. A ressonância magnética medular revelou escoliose cervical, espaços discais rudimentares, com fusão dos elementos posteriores e dilatação hidromiélica do canal ependimário. A restante avaliação multidisciplinar foi irrelevante e não foi diagnosticada qualquer patologia nos pais. A síndrome de Klippel-Feil é rara. Pode-se manifestar de forma isolada ou estar associada a outras malformações, como as esqueléticas, cardíacas ou nefrourológicas. Se existe uma suspeita clínica, o doente e a família deve ser avaliada por uma equipa multidisciplinar.

Palavras-Chave: Lactente; Síndrome de Klippel-Feil/complicações; Síndrome de Klippel-Feil/diagnóstico por imagem; Torcicolo/etiologia