Spine Deformity
www.spine-deformity.org

Spine Deformity 6 (2018) 448–453

The Prevalence of Klippel-Feil Syndrome: A Computed Tomography–Based Analysis of 2,917 Patients

Jillian Gruber, MD[a], Ahmed Saleh, MD[b], Wajeeh Bakhsh, MD[a], Paul T. Rubery, MD[a], Addisu Mesfin, MD[a,*]

[a]*Department of Orthopaedic Surgery, University of Rochester School of Medicine and Dentistry, 601 Elmwood Ave, Box 665, Rochester, NY 14642, USA*
[b]*Department of Orthopaedic Surgery, Maimonides Medical Center, 4802 10th Ave, Brooklyn, NY 11219, USA*

Received 4 August 2017; revised 28 November 2017; accepted 2 December 2017

Abstract

Study Design: Cross-sectional study.
Objective: To determine the prevalence of KFS in asymptomatic patients in New York State.
Summary of Background Data: Klippel-Feil syndrome (KFS) is characterized by congenitally fused cervical vertebrae and may not be diagnosed clinically because most patients do not have the classic triad of short neck, low posterior hairline, and decreased neck range of motion. KFS may be associated with abnormalities such as congenital scoliosis and deafness, and patients are at higher risk for neurologic injury following cervical spine trauma. The prevalence of KFS has not been evaluated in a large series but is estimated to occur every 40,000 births.
Methods: A total of 3,534 cervical computed tomography (CT) scans at the emergency department of a level I trauma center were obtained during a one-year period. Duplicate scans and outside hospital imaging were excluded, resulting in 2,917 cervical CT scans for review. Demographic information was collected, and if KFS was present, level(s) fused, Samartzis classification type, and presence of cervical scoliosis and cervical spine fractures were recorded.
Results: The prevalence of KFS was 0.0058% (1 in 172). Of the 17 subjects with KFS, 8 were female and 9 were male. The most commonly fused levels were C5–C6 and C2–C3. All 17 subjects were classified as Samartzis type I, with a single congenitally fused cervical segment. None of the subjects had cervical scoliosis or cervical spine fractures.
Conclusions: The prevalence of KFS in our series is much higher than previously described. Because clinical diagnosis may not be reliable, it is likely that this condition is underreported and may only be found incidentally on imaging.
Level of Evidence: Level III.

Keywords: Klippel-Feil syndrome; Prevalence; CT; Congenital cervical fusion; Population study; New York state

Background

Klippel-Feil syndrome (KFS) is characterized by congenital synostosis of the cervical vertebrae because of a segmentation or formation defect. Although once defined as a triad of short neck, low posterior hairline, and decreased neck range of motion, its phenotype has evolved to encompass a wide range of associated congenital anomalies, including as scoliosis, deafness, and cardiovascular abnormalities [1]. Pain, neurologic symptoms, and decreased cervical range of motion are the most common symptoms that lead to a diagnosis of this syndrome [2]. In patients with KFS, trauma may result in disastrous consequences such as spinal cord injury or facet dislocations. KFS also predisposes to accelerated degenerative change of the segment adjacent to the fused segments [3]. Magnetic resonance imaging of myelopathic patients with KFS demonstrate increased degenerative changes as compared to patients without KFS [3].

Author disclosures: JG (none); AS (none); WB (none); PTR (grants from AOSpine, during the conduct of the study); AM (grants from Corelink, AOSpine, OMEGA Foundation, Globus, and LES Society, outside the submitted work; and Axiomed, equity).

*Corresponding author. Orthopaedic Surgery and Neurosurgery, University of Rochester School of Medicine and Dentistry, 601 Elmwood Ave, Box 665, Rochester, NY 14642, USA. Tel.: (585) 275-5196; fax: (585)756-4726.

E-mail address: amesfin@gmail.com (A. Mesfin).

2212-134X/$ - see front matter
https://doi.org/10.1016/j.jspd.2017.12.002

The true prevalence of KFS has not been reported because of a lack of population screening studies, but it has been estimated to be 1 in 40,000 with a slight female predominance [2]. In a prospective series of 131 surgical patients with cervical spondylotic myelopathy (CSM), 5 (3.82%) patients had KFS [3]. The objective of this study was to assess the prevalence of KFS in the general population as determined by modern, high-resolution computed tomography (CT) scans.

Materials and Methods

This retrospective review was approved by the university institutional review board. CT scans of the cervical spine obtained in the emergency department of our level I trauma center were reviewed for the presence of KFS over a one-year period, from January 2014 to December 2014. Inclusion criteria included a CT scan of the cervical spine in which all of the cervical spine vertebrae were visualized. Exclusion criteria included CT of the head or spine that only extended to the C2 level or poor visibility of vertebral bodies on imaging.

Investigators all trained in identifying Klippel-Feil syndrome on imaging reviewed CT scans. None of the investigators were involved in the direct care of the patients. KFS was diagnosed when the wasp-waist sign was present as well as bony fusion of adjacent vertebrae [4]. All potential candidates were reviewed by attending orthopaedic spine surgeons who made the final decision about the classification. If KFS was present, the vertebrae involved and Samartzis classification were recorded. According to the Samartzis classification, type I denotes a single fused cervical segment, type II multiple noncontiguous fused segments, and type III multiple contiguous fused segments [5]. Subject age, gender, race, ethnicity, cervical scoliosis, and presence of concurrent cervical spine fractures were also recorded. The medical records of patients with a KFS diagnosis were reviewed for the presence of prior diagnosis of KFS as well as the mechanism of injury prompting their visit to the emergency room. The prevalence of KFS was quantified in the overall patient population.

Statistical analysis

Continuous variables were presented as a mean and associated range, and categorical variables were presented as fractions of the overall population. Independent *t* tests were used to compare continuous variables, and the Fisher exact test was used to compare categorical variables.

Results

A total of 3,534 cervical CT scans were obtained. Duplicate scans and outside hospital imaging were excluded, resulting in 2,917 unique cervical CT scans of adequate quality for review. There were no significant differences in demographic data between patients with KFS and patients without KFS (Table 1).

Table 1
Demographics.

Parameter	Non-KFS subjects (n = 2,900)	KFS subjects (n = 17)	p value
Age, years, mean (range)	47.8 (1–101)	52.6 (16–85)	.41
BMI, mean (range)	27.7 (10.8–83.7)	26.6 (8.2–41.6)	.52
Sex, n (%)			
Men	1,339 (46.2)	9 (52.9)	.58
Women	1,561 (53.8)	8 (47.1)	
Race, n (%)			
White	2,208 (76.1)	15 (88.2)	.32
Black or African American	567 (19.6)	1 (5.9)	
Asian	26 (0.9)	1 (5.9)	
American Indian or Alaskan Native	7 (0.2)	0 (0.0)	
Native Hawaiian or other Pacific Islander	1 (0.0)	0 (0.0)	
Other	87 (3.0)	0 (0.0)	
Not reported	4 (0.1)	0 (0.0)	
Ethnicity, n (%)			
Hispanic or Latino	191 (6.6)	1 (5.9)	.91
Not Hispanic or Latino	2,706 (93.3)	16 (94.1)	
Not reported	3 (0.1)	0 (0.0)	

KFS, Klippel-Feil syndrome.

Table 2
KFS level and classification (N = 17).

Variable	Number (%)
Level of fusion	
C2–C3	6 (35.3)
C3–C4	1 (5.9)
C4–C5	3 (17.6)
C5–C6	6 (35.3)
C6–C7	1 (5.9)
Samartzis classification	
Type I	17 (100)

Seventeen patients had KFS for a total prevalence of 0.0058% (1 in 172). The group was composed of 8 females and 9 males with an average age of 52.6 years (range 16–85). The most commonly fused segments were C2–C3 and C5–C6, and all 17 had a type I KFS classification with a single congenitally fused cervical segment (Table 2). None of the patients had cervical scoliosis or a cervical spine fracture. Review of the 17 patients' medical records revealed only 1 patient with documentation of prior KFS diagnosis. Ten patients presented to the emergency room following a fall or syncopal episode, 6 patients presented following a motor vehicle collision and 1 patient following an assault. None of the patients required surgical intervention for their cervical spine. Four patients were deceased at latest follow-up from non–KFS-related etiologies.

Discussion

This is the first study and largest series reported to date evaluating the prevalence of KFS with CT scans. We found