Congenital Cervical Fusion as a Risk Factor for Development of Degenerative Cervical Myelopathy

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■ BACKGROUND: **Congenital fusion of cervical vertebrae, including Klippel-Feil syndrome (KFS), is a suspected risk factor for development of degenerative cervical myelopathy (DCM). We aimed to establish prevalence and degenerative patterns of congenital cervical fusion (CCF) among a global cohort of patients with DCM.**

■ METHODS: **Data from 3 prospective DCM studies were merged, including clinical data for 813 patients and imaging for 592 patients. CCF was diagnosed by presence of fused cervical vertebrae without signs of degenerative fusion. A wasp-waist sign was used to define a KFS subgroup. Characteristics of patients with CCF and the KFS subgroup were compared with the remainder of patients with DCM.**

■ RESULTS: **Twenty-three patients with CCF (14 KFS) were identified, indicating a prevalence of 3.9% (2.4% KFS). Patients with CCF were older ($P = 0.02$), had more operated levels ($P = 0.01$), had higher rates of ossified posterior longitudinal ligament ($P = 0.02$), and demonstrated worse degenerative changes at C3-4, including spinal cord compression ($P = 0.002$) and T_2 weighted image T2WI signal hyperintensity ($P = 0.04$). Levels adjacent to fusions showed a trend toward increased spinal cord compression ($P = 0.09$), with fusions at C3-4 or above showing cord compression below in 9 of 10 patients, fusions at C5-6 or below having cord compression above in 8 of 8 patients, and fusions at C4-5 showed cord compression above and below in 2 of 2 patients.**

■ CONCLUSIONS: **The prevalence of CCF and KFS is higher in DCM than for the general population, suggesting that these patients are predisposed to DCM development. Patients with CCF also have an altered pattern of degenerative changes, seemingly related to adjacent segment degeneration that preferentially affects midcervical levels.**

INTRODUCTION

Congenital cervical fusion (CCF), including Klippel-Feil syndrome (KFS), is relatively uncommon, and the prevalence of KFS in the general population has been estimated at 0.71%.[1] KFS arises due to failure or incomplete segmentation of somites early during gestational development of the spinal column.[2] Historically, the condition has been associated with a classic triad of physical characteristics, including a short neck, low posterior hairline, and restriction of neck motion[3,4]; however, it has been estimated that only 34%–74% of patients present with this constellation of findings.[5] KFS can manifest in a wide spectrum of phenotypes, and its impact on health and quality of life is largely dependent on the severity of the condition, accompanying deformity, and the potential development of spinal cord compression and myelopathy. Typically, cervical spine deformity increases with the number of levels involved. Additional associated congenital defects affecting visceral, musculoskeletal, otolaryngologic, and neurological systems can also present concomitantly in these patients.[4-9] Unfortunately, there has been little research addressing the long-term consequences of KFS. Previous research has suggested that patients with KFS may be predisposed to accelerated degenerative changes and hypermobility at segments adjacent to fused vertebrae.[10-13] In

Key words
- Adjacent segment degeneration
- Adjacent segment disease
- Adjacent segment pathology
- Cervical spondylotic myelopathy
- Klippel-Feil syndrome
- Magnetic resonance imaging

Abbreviations and Acronyms
CCF: Congenital cervical fusion
DCM: Degenerative cervical myelopathy
KFS: Klippel-Feil syndrome
ICF: Idiopathic cervical fusion
MRI: Magnetic resonance imaging
OPLL: Ossification of the posterior longitudinal ligament

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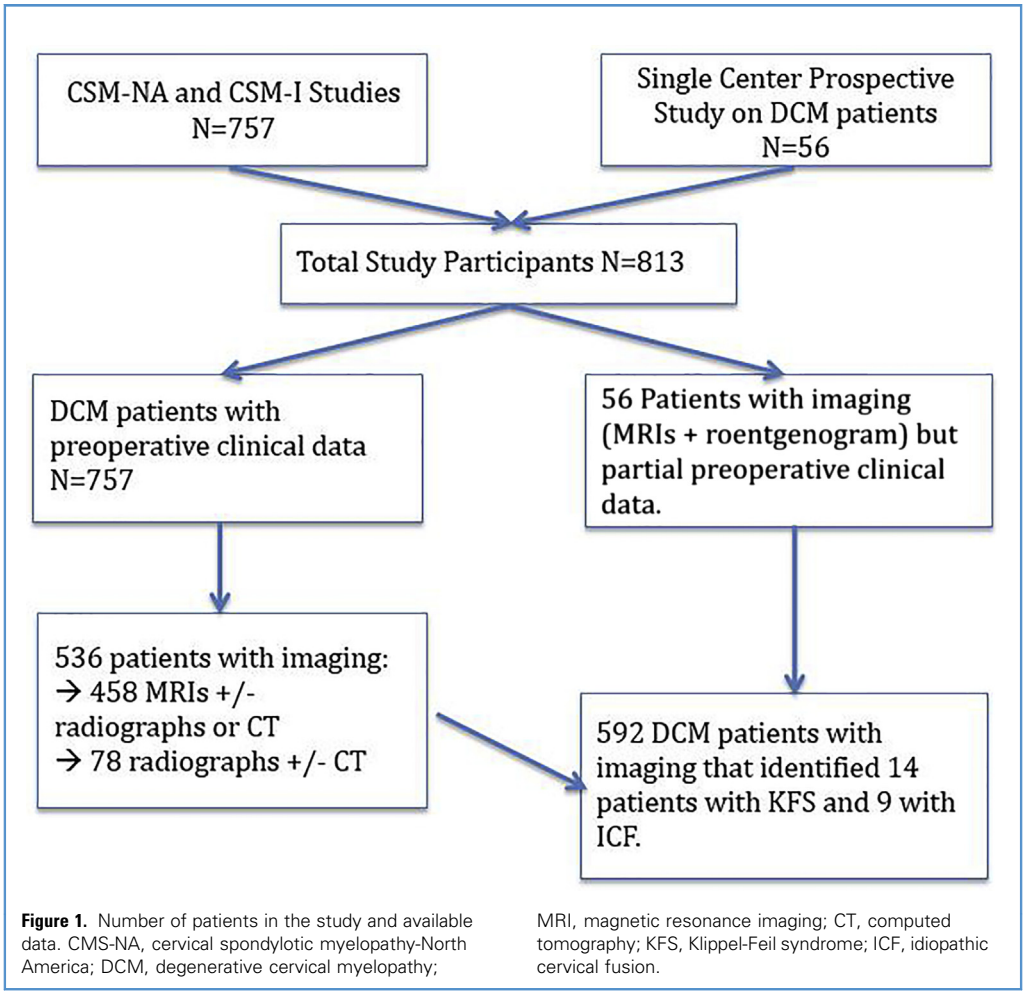

Figure 1. Number of patients in the study and available data. CMS-NA, cervical spondylotic myelopathy-North America; DCM, degenerative cervical myelopathy; MRI, magnetic resonance imaging; CT, computed tomography; KFS, Klippel-Feil syndrome; ICF, idiopathic cervical fusion.

our recent analysis of the AOSpine North America data on patients undergoing treatment for degenerative cervical myelopathy (DCM), the prevalence of KFS was 3.8% with patients tending to present with more pronounced degenerative changes on magnetic resonance imaging (MRI).[14] However, the study was limited by a relatively small number of patients with KFS. In addition, idiopathic cervical fusion (ICF), referring to autofusions that do not show evidence of degenerative fusion (e.g., osteophytes), but are strongly suspected to be congenital, was not previously reported. To investigate the impact of KFS and ICF further, the present study evaluated this broader population of patients with CCF by merging clinical and imaging data from 3 prospective studies that collectively comprise an international cohort of patients.

METHODS

Study Data

The data in the present study were derived from 3 cohorts: 1) the AOSpine North America prospective multicenter study, 2) the AOSpine International prospective multicenter study, and 3) a prospective single center study at our institution. The primary study details and results of the AOSpine studies have been previously published,[15,16] and the third study remains ongoing, but patient enrollment has stopped. All 3 studies included patients aged 18 years and older with clinically confirmed myelopathy. Research ethics approval was obtained from each study site. Details of the clinical and imaging data available for the present analysis are described in Figure 1. There were a total of 757 patients with preoperative clinical data and 592 patients with preoperative imaging.

Clinical Assessments

Clinical status was assessed before surgery and at 6, 12, and 24 months after surgery. Assessments included modified Japanese Orthopedic Association score,[17] Nurick,[18] neck disability index,[19] and physical and mental component of the 36-item short-form health survey.[20] The group of patients with CCF (including KFS and ICF) and the KFS subgroup were compared against the remainder of patients with DCM. Postoperative outcomes were analyzed based on follow-up data at 24 months or, if not available, the last completed follow-up.