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Lumbar Spondylolysis and Spondylolisthesis in Down Syndrome: A Cross-sectional Study at One Institution

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Abstract

Study Design: Retrospective cross-sectional study.
Objectives: To describe lumbar spondylolysis and spondylolisthesis and establish their prevalence in individuals with Down syndrome.
Summary of Background Data: Orthopedic problems in Down syndrome are variable and numerous. Lumbar spondylolysis and spondylolisthesis may be common conditions in Down syndrome. However, there has been a paucity of data on the association of these conditions in the published literature.
Methods: A retrospective review of 110 patients with Down syndrome seen at a single institution from 2000 through 2012 was performed. Medical records, X-rays, and physician dictations were carefully reviewed to establish a detailed database of the study population.
Results: Of the 110 patients in the study, 20 exhibited spondylolysis (unilateral, n = 11; bilateral, n = 9), whereas 38 had lumbar spondylolisthesis (isthmic, n = 9; dysplastic, n = 2; degenerative, n = 27). No gender difference was noted ($p \geqslant .7732$). Fifteen patients reported low back pain (LBP) and/or leg pain. There was no significant association between LBP, leg pain, and spondylolysis ($p = .9232$). Both of these symptoms were highly predictive of lumbar spondylolisthesis, however ($p = .0006$). No significant findings were noted in pelvic parameters (pelvic incidence, sacral slope, pelvic tilt, or lumbar lordosis) in this study.
Conclusions: The prevalence of spondylolysis and spondylolisthesis in individuals with Down syndrome may be as high as 18.7% and 32.7%, respectively, significantly higher than in the non-Downs population. Etiopathogenesis of these conditions in Down syndrome does not appear to be related to pelvic parameters. Low back pain and leg pain may be more predictive of spondylolisthesis in Down syndrome than in the general population. Therefore, it is recommended that individuals with Down syndrome and LBP and/or leg pain be evaluated for lumbar spondylolisthesis.

Keywords: Spondylolysis; Spondylolisthesis; Down syndrome; Low back pain

Introduction

Down syndrome (DS) may manifest itself in a variety of orthopedic disorders, abnormalities, and pathologies. It has been widely hypothesized that orthopedic problems in DS result from a combination of ligamentous laxity and hypotonia. This synergism is believed to contribute to a diverse nature of orthopedic problems such as joint instability, dislocation, and recurrent fracture [1].

Much research and attention has been devoted to describing upper cervical, hip, and knee abnormalities in DS. However, there is a paucity of data documenting the prevalence of lumbar spondylolysis and spondylolisthesis and describing them in DS.

Spondylolysis is defined as a defect in the pars interarticularis of the vertebral arch [2-4]. It is typically reported in 3% to 6% of the Caucasian population [4-6]. One extensive study reported the prevalence in the community-based population to be 4.2% in childhood and 6% in adulthood [4], with slight ethnic variations noted in other studies [3,7]. With the use of computed tomography (CT), the prevalence

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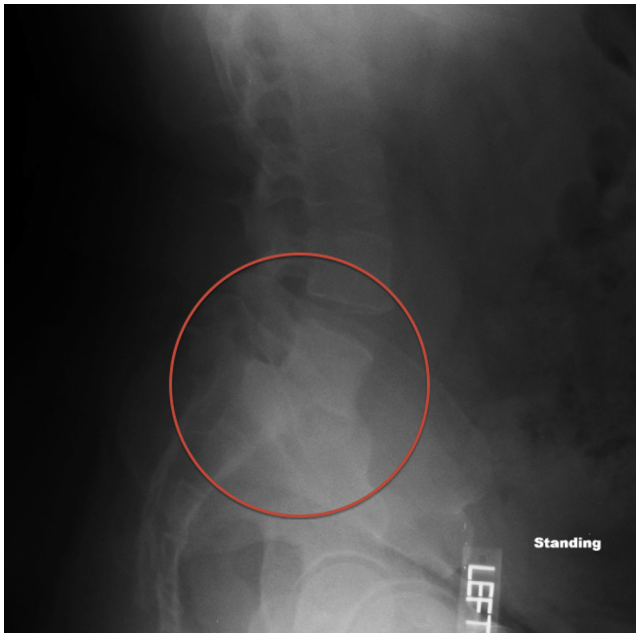

Fig. Grade 2 isthmic spondylolisthesis at L5–S1 in an individual with Down syndrome (circled).

has been reported to be as high as 11.5% in the general population [8].

A closely related condition, spondylolisthesis, is defined as a forward displacement of the vertebral body in relation to the level below (Fig.) [2]. Five types of spondylolisthesis have been described: dysplastic, isthmic (lytic), degenerative, traumatic, and pathological [9]. Isthmic spondylolisthesis occurs in the presence of a bilateral pars defect with reduction of the stabilizing ability of the posterior elements of the spine [10]. This pathogenesis highlights the fact that the 2 conditions are interrelated. The precise natural history of spondylolysis progressing to spondylolisthesis is not fully understood; however, genetic factors are highly suspected [3]. Spondylolisthesis may also affect males and females at unequal rates. One study reported a lower rate of spondylolisthesis in males compared with females, at 2.7% and 8.4%, respectively [11]. Furthermore, many orthopedic surgeons may attribute higher occurrence of olisthesis to increased body mass index (BMI).

Both conditions are common radiographic findings and may be symptomatic or entirely asymptomatic. When symptomatic, the most common patient problems are location-specific back pain (low back pain [LBP]) and/or lower limb radiculopathy [2,12]. Some studies have concluded that the association of LBP with spondylolysis and spondylolisthesis is not strong and that the vast majority will present asymptomatically [4,5,8].

As mentioned previously, the accepted epidemiology reports that 3% to 6% of the general adult population is affected by spondylolysis and 2.7% to 8.4% is affected by lumbar olisthesis. These frequencies may be much higher in DS. This retrospective cross-sectional study was developed to establish a relationship between the 2 orthopedic pathologies under study and DS.

Table 1
Description of patient population.

Variable	n (%)
Total subjects	110
Male	64 (58.2%)
Female	46 (41.8%)
Mean age ± standard deviation, years	40.6±16.9
Spondylolysis	20 (18.2%)
Spondylolisthesis	38 (34.6%)
Low back pain	14 (12.7%)
Leg pain	8 (7.3%)

Table 2
Comparison of prevalence of spondylolysis and spondylolisthesis, by sex.

Condition	Males (n=64), n (%)	Females (n=48), n (%)	Total population (n=110), n (%)	Gender difference? (χ^2 test)
Spondylolysis	11 (9.6%)	9 (7.8%)	20 (18.2%)	(p=.8301) No
Spondylolisthesis	21 (18%)	17 (14.8%)	38 (34.6%)	(p=.7732) No

Materials and Methods

Patient population

After institutional review board approval, a retrospective review of 110 orthopedic patients with DS was performed. These patients were referred to 1 orthopedic spinal specialist from 2000 to 2012. The patients were referred for orthopedic consultation by their primary care physicians, most of whom are DS specialists at a single institution. Eleven patients meeting the above criteria were excluded from the study because they lacked reviewable X-rays.

Patients presented to the surgeon with a variety of orthopedic and neurological problems including pain and suspected cervical instability, and for routine evaluation, among many other reasons.

Patient evaluation

Patient medical charts, dictations, and X-rays from 2000 to 2012 were reviewed to build a detailed database of the study population. Throughout this process, special attention was paid to characteristic symptoms of LBP and leg

Table 3
Distribution of lumbar spondylolisthesis, by level.

Level	Slips, n (N=43)
L1–L2	2
L2–L3	2
L3–L4	3
L4–L5	5
L5–S1	31
Even distribution?	No (p = .0)