Case Series

<u>Case series - Slipped capital femoral</u> <u>epiphysis in three members of same family</u> <u>as an unusual presentation</u>

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Abstract

Slipped capital femoral epiphysis is a disorder of the hip that affects children in late childhood and early adolescence, characterized by medial and posterior displacement of the proximal femoral epiphysis on the metaphysis. Although the diagnosis and treatment of slipped capital femoral epiphysis have been well described, the search for its cause and a method of early identification continues. We have reported here an interesting case of a bilateral slipped capital femoral epiphysis as an unusual presentation in one family - father and two siblings, had no record of any hormonal imbalances or endocrine abnormalities; had good nutrition; and presented with atypical characteristics of slipped capital femoral epiphysis.

Key words

Slipped capital femoral epiphysis, X-ray, Diagnosis of slipped epiphysis.

Introduction

Slipped capital femoral epiphysis is a disorder of the hip that affects children in late childhood and early adolescence. It is characterized by medial and posterior displacement of the proximal femoral epiphysis on the metaphysis. This results in the classic varus appearing hip on X-ray. Slipped capital femoral epiphysis (SCFE) is most common in the adolescent period (i.e. boys aged 10-16 years and girls aged 12-14 years). Males have 2.4 times the risk compared with females. The left hip is affected more commonly than the right [1, 2].



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Some hips may slip in a postero-lateral displacement, producing an apparent valgus deformity and a so-called valgus SCFE. This occurs when shearing forces applied to the femoral head exceed the strength of the capital femoral physis. The exact etiology is unknown, but is believed to be secondary to multiple factors, including obesity which increases mechanical strain on the physis. The periosteal thinning and widening of the physis seen during rapid adolescent growth acceleration may also be a predisposing factor for the weakening of the physis. Only small percentage of SCFE (5.2-6.9%) is associated with endocrinopathies such as hypothyroidism or growth hormone deficiency. Short stature, early age at presentation, and the atypical appearance of a valgus SCFE have all been suggested as indicators for endocrinopathy screening [3].

Case Series

We have presented the 3 cases of slipped capital femoral epiphysis in one family: father and two siblings - daughter and son.

Patient - 1

The first child (7 years old girl) presented with a history of limping gait and difficulty in walking. At presentation, she was diagnosed with a stable, mild left hip slipped capital femoral epiphysis. Her medical history was remarkable excepting the above mentioned complaints in her gait. Patient was investigated with X-rays of both lower limbs. (Photo – 1)

Patient - 2

The second child (6 years old boy) presented with similar history as his sister i.e., limping gait while walking. The patient was evaluated with normal birth history and no developmental anomaly excepting the difficulty in gait since the time she learned started walking. Patient came in our department with above complaints and was investigated with X-ray of both lower limbs. (Photo – 2)

<u>Photo – 1</u>: X-ray findings suggestive of bilateral coxa vera deformity with slipped epiphysis.



<u>Photo – 2</u>: X-ray findings suggestive of slipped epiphysis with coxa vera deformity.



Patient - 3

The third patient was father of both children (39 years old) complaint of difficulty in walking and valgus deformity in both knees. (Photo -3)

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<u>Photo – 3</u>: X-ray findings suggestive of slipped epiphysis with coxa vera deformity.



Discussion

Our case series is unique given the atypical presentation characteristics: the patients were non-obese, with bilateral involvement, and no history of endocrine problems. According to Kay [4], after a diagnosis of slipped capital femoral epiphysis has been made in a family member, a second family member has been reported to be affected in 3% to 7% of reported cases. There are few reports of slipped capital femoral epiphysis occurring in siblings.

Several studies have described the genetic characteristics of children presenting with slipped capital femoral epiphysis. In the past several decades, human leukocyte antigen testing has allowed for inferences about possible genetic causes of slipped capital femoral epiphysis, but such studies differ by region and do not identify a distinct genetic marker [5, 6]. The inheritance pattern seen in familial occurrences of slipped capital femoral epiphysis is thought to be autosomal dominant with variable penetrance [4, 7].

Although the cause of slipped capital femoral epiphysis is still debated, most sources agree with regard to its epidemiology and demographics [1, 4]. Most patients with slipped capital femoral epiphysis are obese, with weights above the 95th percentile according to age at the time of diagnosis [3]. The racial predilection of slipped capital femoral epiphysis is 1.0 for Caucasians, 4.5 for Pacific Islanders, 3.9 for African Americans, and 2.5 for Hispanics [1, 8]. Several case series support these demographic representations in their descriptions of the occurrence of more than one case of slipped capital femoral epiphysis in the same family [7, 9, 10, 11].

Our series, however, is unique in the consistent atypical presentation of baseline characteristics. The patients were siblings, both sister and brother and not obese. These atypical characteristics are important indicators that if there is a hereditary or genetic link with slipped capital femoral epiphysis, the mechanism may transcend the physical characteristics of patients. The patients in our series did not want to pursue genetic testing.

Loder, et al. [12] found that slipped capital femoral epiphysis occurs at a higher incidence in children with endocrine disorders, specifically hypothyroidism. Thyroid dysfunction was present in another familial case series of slipped capital femoral epiphysis in Australia [11]. After we noticed the familial trend in our patients, endocrine studies, including thyroid tests, were performed on the second and third patients. Their thyroglobulin, thyroid-stimulating hormone, and T3 levels were normal; the only notable deviation was a decreased serum creatinine level in both patients.

Loder, et al. [12] also noted that previous use of growth hormone supplementation was an associated factor with slipped capital femoral

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epiphysis [1]. None of our patients were on gonadotropin-releasing hormone supplements or had previously taken these substances. Similarly, their testosterone and estrogen levels were normal. Systemic diseases, such as renal osteodystrophy, have commonly been associated with patients with slipped capital femoral epiphysis who have heights less than the 10th percentile for age; however, none of our patients had any signs of systemic diseases or chondrodysplasias [4]. Finally, none of our patients had undergone previous radiation therapy, which also has been implicated in slipped capital femoral epiphysis [4].

The cause of slipped capital femoral epiphysis most likely involves biochemical and biomechanical factors that combine to weaken the physis [1]. All 3 of our patients participated in similar athletic endeavors such as field hockey, volleyball, softball, soccer, and skiing, and all described themselves as being "very active" in these sports. The sisters also reported consuming well-balanced diets. They ate many of the same foods and took multivitamins most days of each week. We have previously reported on an association between slipped capital femoral epiphysis and vitamin D deficiency [13]. It was interesting to note, however, there is no previous research reporting the combination of cardiac abnormalities and slipped capital femoral epiphysis.

Conclusion

While familial slipped capital femoral epiphysis appears to be associated with mainly autosomal dominant mode of inheritance with variable penetrance in certain populations, we believe that in most other populations, a multi-factorial form of inheritance is more likely, in this disease. More studies need to be done to understand the underlying genetics of this disease, with the recognition that local environmental conditions can affect the different incidence of slipped capital femoral epiphysis across populations. However, currently available studies suggest that first degree relatives of patients with slipped capital femoral epiphysis are at an increased risk for the disease, and the affected families should be educated accordingly, and screened when indicated.

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