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Unusual Association of KBG Syndrome with Scheuermann's Disease

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Abstract

In this paper we discuss a novel case in which a patient had a comorbid diagnosis of KBG syndrome and Scheuermann's disease. The patient was a 14-year-old boy, referred to orthopaedics for assessment of his spinal deformity. Initial assessment revealed that he had a rib prominence on his right side, which corrected upon bending forward. SLR examination indicated significantly tight hamstrings. Plantars were upturning, but other reflexes were normal. He had kyphosis measuring up to 59.4 degrees. MRI of the spine depicted features of classic Scheuermann's disease, from D6-D10. The patient was given conservative treatment consisting of physical therapy and postural training. He remained asymptomatic during the course of a 5 year follow up period. This case is unique due to the comorbidity of Scheuermann's disease and KBG syndrome, which has never been reported in the literature. This case report suggests that routine spinal screening in cases of KBG syndrome would contribute to a better understanding of treatment and diagnosis.

Keywords: KBG syndrome, Scheuermann's disease, Case Report

Introduction

KBG syndrome (OMIM 148050) is a rare condition of autosomal dominant inheritance. The syndrome has four cardinal manifestations: macrodontia of upper central incisors, skeletal anomalies (mainly costovertebral), facial dysmorphism and developmental delay. The syndrome is caused by mutations in the ANKRD11 gene encoding ankyrin repeat domain 11 [3] or microdeletion of 16q24.3 that enclose the ANKRD11 gene [2, 4].

Scheuermann's disease is the most common cause of hyperkyphosis of the thoracic and thoracolumbar spine that affects adolescents. It is characterized by vertebral body wedging and endplate irregularity, narrowing of intervertebral disc spaces, premature disc degeneration, diminished anterior vertebral growth and Schmorl's nodes. The prevalence of this disease varies from 0.4% to 10% [5, 6]. The exact causes of the disease are still unknown. It is believed to follow an autosomal dominance pattern [7].

Here, we report a case of KBG syndrome in association with Scheuermann's disease; this combination to the best of our knowledge has not been reported earlier.

Case Presentation

A 14-year-old boy with a known diagnosis of KBG syndrome was referred for assessment of his spinal deformity. He was the first child of a non-consanguineous couple. Family history was non-contributory. Pregnancy was uneventful, and exposure to teratogens was denied. He was born of C-section secondary to breach and had delayed milestones at every age.

The boy appeared short in stature for his age and had developmental delay. Facial dysmorphism, bulbous nose and kyphosis were evident (Figure 1). He was found to have learning difficulties, macrodontia of central incisors, bilateral hearing loss and anosmia. The right shoulder was on a higher level than the left and slight rib prominence was noticed on the right side with correction on bending forward (Figure 2). He did not have any motor or sensory deficit. SLR examination was negative bilaterally and there was no hamstring tightness. Reflexes were normal, plantars downgoing, abdominal reflexes present.

X-rays revealed kyphosis measuring up to 59.4 degrees. (Figure 4 a and 4 b) MRI of his thoracic spine revealed mid dorsal kyphosis with anterior wedging shown in the D6, D7, D8, D9 and D10. Associated

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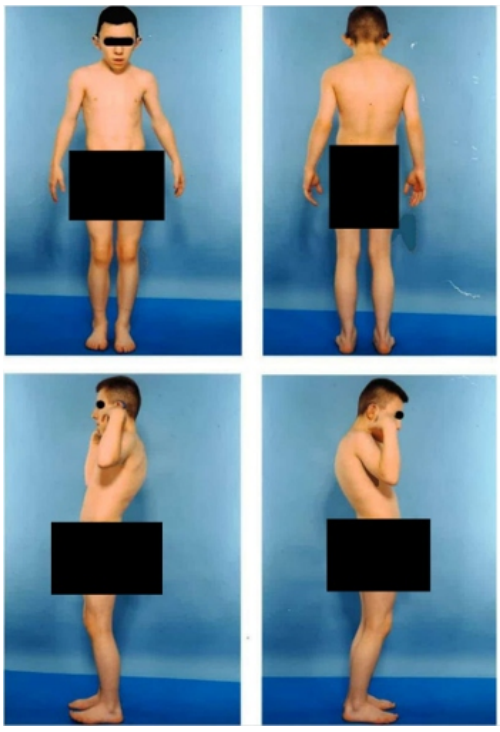

Figure 1: Showing facial dysmorphism, bulbous nose and kyphosis.

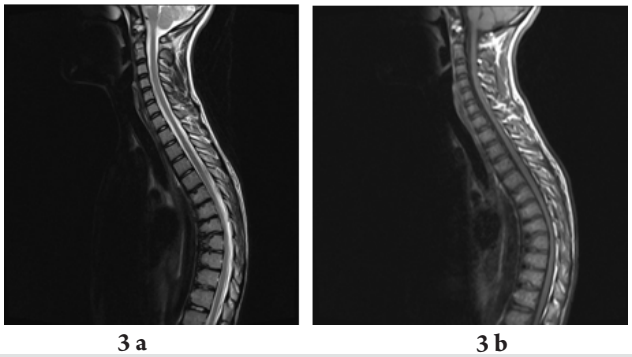

Figure 3a and 3b – MRI thoracic spine showing features of Scheuermann's disease.

disc degenerations were noted at these levels. Small Schmorl's nodes were noted in the superior endplates of the vertebral bodies D5/6/7/8 and along the inferior endplates of D9/10. These appearances indicate underlying Scheuermann's disease (Figure 3a and 3b).

Pituitary MRI done to evaluate anosmia showed no structural abnormalities and his endocrine profile was normal.

Treatment

Non-operative management was indicated as the patient was asymptomatic and the kyphosis curve was less than 60 degree. He was treated with non-operative management with Bracing physiotherapy which included postural training.

Out Come & Follow-up

Follow-up was done for a period of 5 years. At each visit, spinal range of motion was evaluated in all planes and the degree of progression of kyphosis was assessed by serial imaging. He was symptom-free throughout the follow-up period.

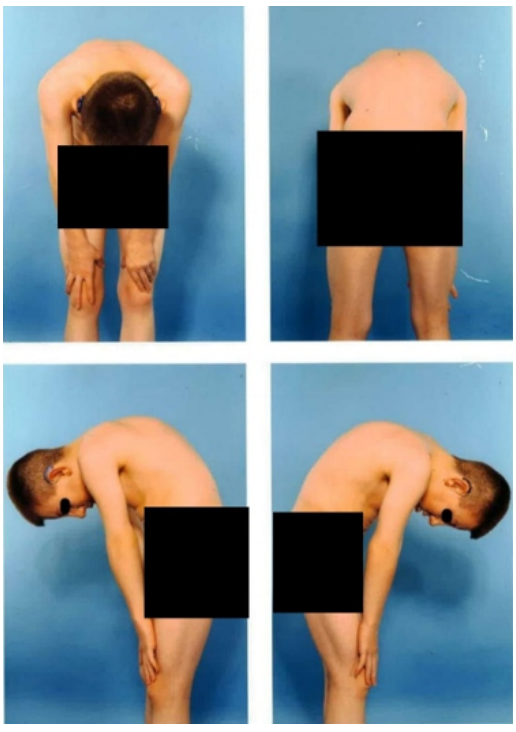

Figure 2: Showing rib prominence on the right side which corrects itself on bending forward.

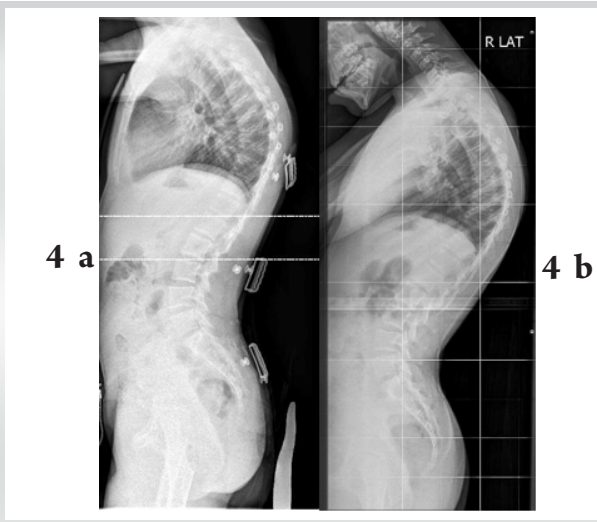

Figure 4 a On first presentation; **4 b** Radiograph in follow up appointment

Discussion

The first case of KBG syndrome was reported in 1975 by Herman et.al [1]. In their paper, they discussed 7 affected individuals from 3 unrelated families. The KBG syndrome was named after the surname initials K, B, and G of the three original families. The diagnosis of KBG syndrome is usually based on clinical findings. Brancati et al. described diagnostic criteria, inclusive of 7 major and 7 minor features [8].

For a definitive diagnosis of KBG syndrome, the patients must have at least 4 of the 7 major findings. Our patient exhibited 5 major features namely, facial dysmorphism, macrodontia, kyphoscoliosis,

psychomotor delay, short stature and additionally hearing loss, thus satisfying the clinical diagnosis of KBG syndrome. Apart from these, our patient also had anosmia, which is inconsistent with the syndrome. His MRI spine report revealed Scheuermann's disease, which is also not a typical feature of the syndrome.

The cause of Scheuermann's disease is unknown. In this condition, there is increased curvature of spine in posterior plane than the anterior. Also, uneven growth of vertebrae in saggital plane could be seen. This is a self-limiting disorder, found mostly commonly between ages 12 and 15 [9]. Scheuermann's disease is classified into two types - Type I (Classic), where there is involvement of thoracic spine only, with the apex of curve T7-T9 and Type II, with thoracic and lumbar involvement, with the apex of curve T10-T12 [17]. Our patient has the classic variant of Scheuermann's disease. There are many treatment methods and options available that aim to correct the kyphosis while the spine is still growing, and especially aim to prevent it from worsening. Adolescents whose kyphosis remains<60 are usually treated only by exercises with or without partial time bracing and are periodically followed up by X-rays [10]. Surgery is usually considered in adolescents with Scheuermann's disease only when the deformity is severe (>80^0) and cannot be controlled with brace treatment [10, 11]. There are two primary surgical techniques to correct kyphosis: posterior-only fusion and anterior/posterior fusion. Recently several published studies suggest treatment trends are favoring posterior-only fusion [12, 13, 14]. However, surgery is indicated only for cosmetic purposes as some of the symptoms of spinal deformity cannot be totally corrected [15,16].

The association of Scheuermann's disease with KBG syndrome in our patient warrants a routine screening of the spine including an MRI should be included in the work-up of KBG patients.

Major features
Facial dysmorphisms*
Macrodontia*
Skeletal abnormalities
Abnormal ribs/vertebra*
Delayed bone age
Short hand tubular bones
Brachy-clinodactylous 5th finger
Abnormal spine curvature*
Short femoral necks/hip dysplasia
Sternum abnormalities
Wormian bones in skull
Cognitive deficits/psychomotor delay*
Short stature*
Abnormal EEG with seizures
Abnormal hair implantation
Minor features
Cutaneous syndactyly, toes II/III
Webbed/short neck
Cryptorchidism
Hearing loss*
Palatal defects (including uvula)
Strabismus
Congenital heart defects
*Features found in our patient

Table 1. Clinical features of KBG syndrome [8]

Conclusion

The association of Scheuermann's disease with KBG syndrom is rare. This patient was managed with conservative management with routine screening of the spine including an MRI. Physicians should be observant for characteristic manifestations of KBG syndrome for accurate diagnosis and should carefully watch for any progression of kyphosis.

Declaration of patient consent : The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his/her consent for his/her images and other clinical information to be reported in the Journal. The patient understands that his/her name and initials will not be published, and due efforts will be made to conceal his/her identity, but anonymity cannot be guaranteed.

Conflict of Interest: None; **Source of Support:** None

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