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A Case Report of Two Youthful Brothers: The Spine Combines with Multiple Butterfly Vertebrae and Hemivertebrae

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Abstract

Butterfly vertebrae (BV) and Hemivertbrae (HV) are all rare congenital spinal abnormality and usually reported in the literature as an isolated finding. As far as the authors know, cases with multiple butterfly deformities and hemivertebra deformities of the spine are very rare. The condition of two brothers suffering from this disease at the same time has not been reported in the literature. We report two youthful brothers, the young brother, 14-year-old, who has butterfly vertebrae at T6-T11, L1, L3, and a hemivertebrae at L4; the old brother, 18-year-old, who has butterfly vertebrae at T5, T6, T8, T10, T11, T12, L1, L3, L4, L5, S1. They all have scoliosis and low back pain, and a 46, XY karyotype. The old brother performed TBX6 and APOB mutation.

Keywords: spine, butterfly vertebra, hemivertebra

1. Introduction

A butterfly vertebrae is a vertebrae with a midline sagittal cleft due to failure of fusion of the lateral halves of the vertebral body. This defect is considered to occur between the third and sixth week of gestation (Sonel, B., Yalçın, P., Öztürk, E. A., & Bökesoy, I., 2001). Rachael M. Hopkins et al. think that the failure of this fusion of the two halves, believed to be due to persistent remnants of the notochord (Hopkins, R. M., & Jh, A., 2015). BV most commonly occurs in the lumbar spine although it can be seen anywhere (Slouma, M., Cheour, E., Sahli, H., & Elleuch, M., 2016). BV can be seen in patients with syndromic diseases such as Pfeiffer's syndrome, Jarcho-Levins (spondylothoracic or spondylocostal dysostosis) Syndrome, Crouzon Syndrome, Alagille Syndrome (Sonel, B., Yalçın, P., Öztürk, E. A., & Bökesoy, I., 2001); or associated by additional spinal anomalies such as intervertebral bars, supernumerary lumbar vertebrae, spina bifida, diastematomyelia, kyphosis/scoliosis or kyphoscoliosis. (Kapetanakis, S., Giovannopoulou, E., Nastoulis, E., & Demetriou, T., 2016)

Hemivertebrae has been typically considered as

a failure of formation or segmentation during somitogenesis (Bao, B., Yan, H., & Tang, J., 2022). HV can combine with many other abnormalities such as Cardiac, Genitourinary abnormalities (Bao, B., Yan, H., & Tang, J., 2022).

2. Case Report

An 18-year-old male was seen in our hospital for progressive low back pain complaints. His low back pain was not observed to radiate toward lower limbs. He indicated that the pain mainly occurs after standing or exercising for a long time, which is unbearable when the pain is severe, and can be significantly relieved after lying down and resting. No history of trauma was present for this patient. Physical examination of the patients found that the lumbar flexion and extension activity was slightly limited, and the lumbar spinous process was slightly tenderness and percussion pain. The physical examination of the sensory and motor system of the lower extremities was normal, and the straight leg elevation test and reinforcement test were negative. Other systems and results of blood tests were normal.

A standing radiograph of the whole spine showed that the number of ribs was normal, the spine curve was estimated at 18° between T11 and L2 (Figure 1), and the pelvis is tilted without obvious rotation. Three-dimensional CT images demonstrated that there are 11 butterfly vertebrae at T5, T6, T8, T10, T11, T12, L1, L3, L4, L5, S1 (Figure 2). Magnetic resonance imaging confirmed the presence of this anomaly and also revealed that no disc protrusion or nerve entrapment and syringomyelia (Figure 3). The result of chromosome analysis showed a 46, XY karyotype and no obvious chromosome structure or numbers abnormality. This patient and his father performed whole exome sequencing, and showed TBX6 and APOB mutation. The location of this variation of chromosome is at chr16:30100431, and the missense variants of genomic region of the TBX6 gene is NM-004608.3:c.454c > T (p.Arg152Cys). The mutation is considered to originate from his father. However, his father has no congenital scoliosis, low back pain or butterfly vertebrae and hemivertebrae.

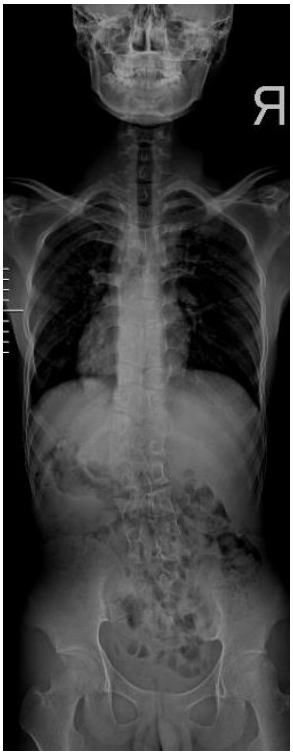

Figure 1.

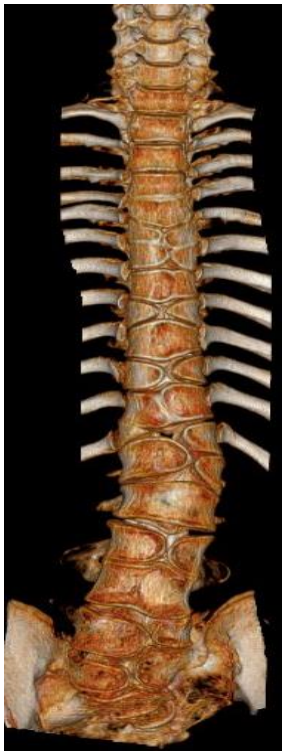

Figure 2.

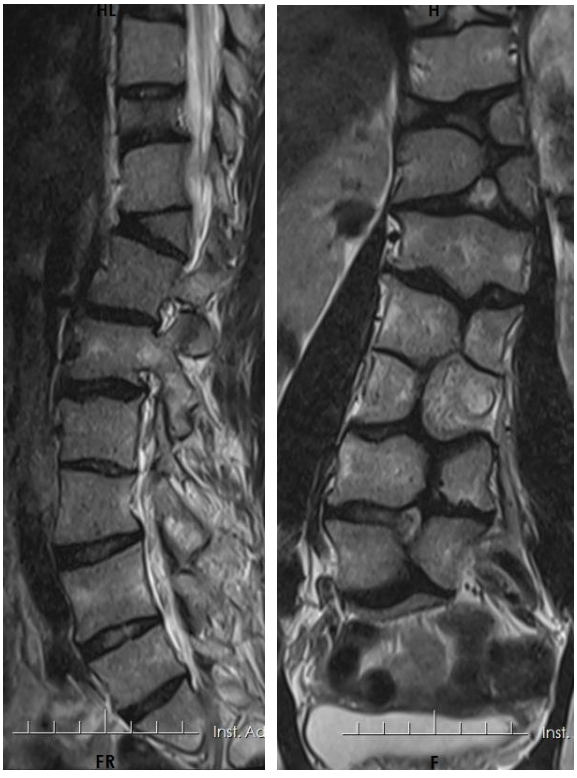

Figure 3.

His 14-year-old brother was operated on in our hospital three years ago because of scoliosis with obvious low back pain. After the operation, scoliosis was well controlled and low back pain

was significantly improved. The results of his brother's physical examination and blood test were similar to his, and there was no obvious abnormality. The standing radiograph of the whole spine showed that he had 13 pairs of ribs, but numbers of the vertebra was normal, because one of the ribs was at C7. The Cobb angle of scoliosis between T-12 and L-5 was 58° (Figure 4). Three-dimensional CT images demonstrated that there are 8 butterfly vertebrae at T6-T11, L1, L3, and a hemivertebrae at the right side of L4 (Figure 5). Magnetic resonance imaging indicated a narrowed disc space between multiple thoracic or lumbar vertebrae (Figure 6). Finally, he underwent the hemivertebrae resection.

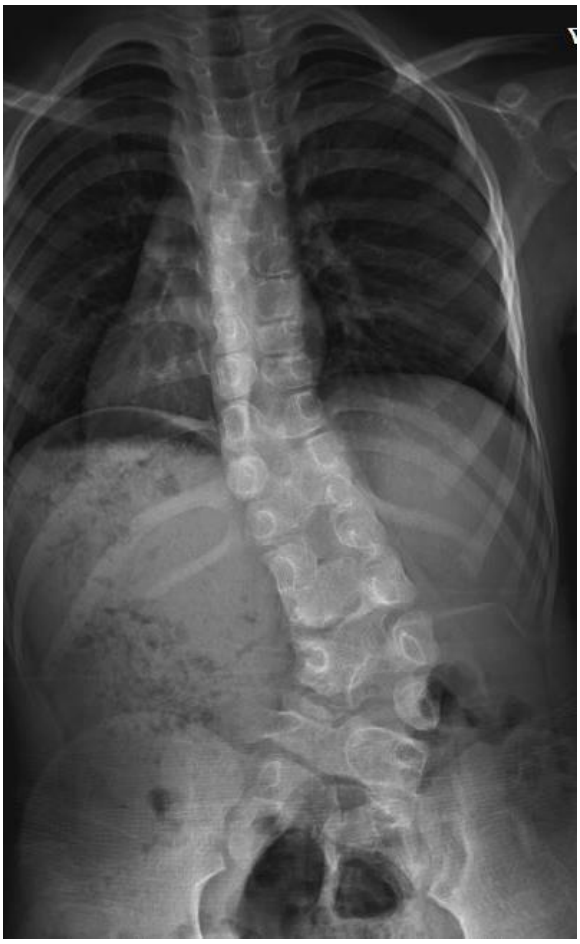

Figure 4.

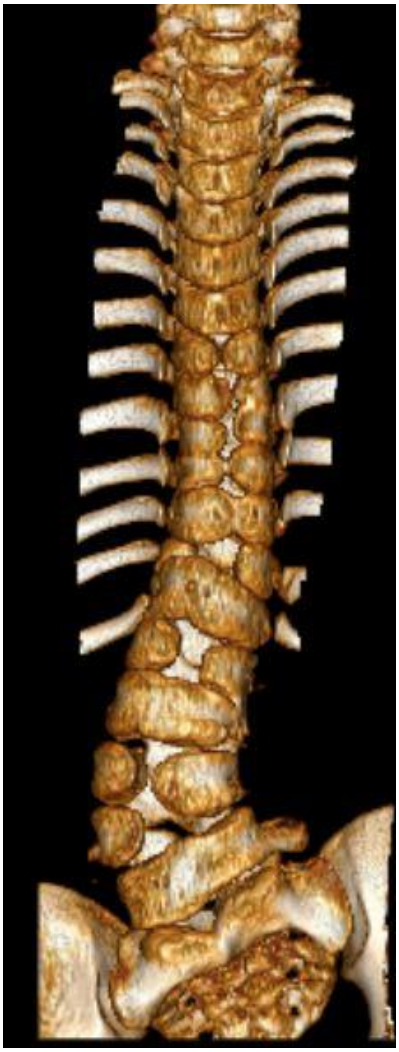

Figure 5.

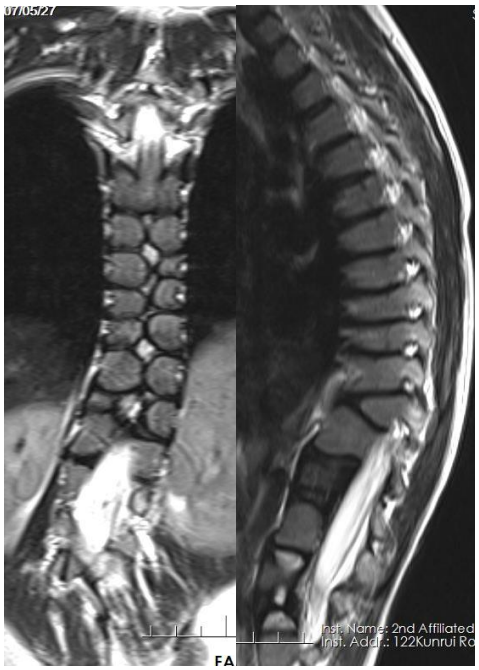

Figure 6.

The result of the two brothers' chromosome analysis showed a 46, XY karyotype and no obvious chromosome structure or numbers abnormality, and their cervical curvature was straight.

3. Discussion

Butterfly vertebrae are an uncommon congenital anomaly of the spine, which is generally benign. Butterfly vertebrae were first described by Rokitansky in 1844 (Müller, F., O'Rahilly, R., & Benson, D. R., 1986), and are known variously as: cleft vertebrae, sagittal cleft vertebrae, which occurs usually at the thoracolumbar spine, Butterfly vertebrae can be mistaken with wedge vertebral fracture. Birkan Sonela et al. reported that BV might increase the incidence of disc herniation, because there is absence of a normal disc between the two vertebrae (Sonel, B., Yalçın, P., Öztürk, E. A., & Bökesoy, I., 2001). BV also can lead neurological impairment. Yoshihiro Katsuura reported the presence of multiple butterfly vertebrae (>1) was associated with the presence of a syndrome, while single butterfly vertebrae were more likely to occur alone (Katsuura, Y., & Kim, H. J., 2019). Kazuki Takeda et al previously examined TBX6 in Japanese congenital scoliosis patients and revealed that approximately 10% of congenital scoliosis was caused by TBX6 mutations (Takeda, K., Kou, I., Mizumoto, S., Yamada, S., Kawakami, N., Nakajima, M., ... & Ikegawa, S., 2018).

The first case report on a hemivertebrae resection was from Royle in 1928. A hemivertebrae has no potential of deterioration (Halm, H., 2011). Hemivertebrae are the most frequent cause of congenital scoliosis. Different locations of hemivertebrae can lead to different scoliosis or kyphosis or lordosis. Hemivertebrae can be described as fully-segmented, semi-segmented or unsegmented. Kiyoshi Imaizumi et al. report that hemivertebrae associated with de novo balanced reciprocal translocation, t (13;17) (q34;p11.2) (Imaizumi, K., Masuno, M., Ishii, T., Kuroki, Y., Okuzumi, N., & Nakamura, Y., 1997). LFNG (a gene) mutations can lead multiple vertebral malformations including hemivertebrae, butterfly vertebrae (Slouma, M., Cheour, E., Sahli, H., & Elleuch, M., 2016).

Butterfly vertebrae and Hemivertbrae are all rare congenital spinal abnormality and usually reported in the literature as an isolated finding or coexist with one sydrome. However, in our case report, although they all have multiple butterfly vertebrae, they have no Cardiac, Genitourinary or other system abnormalities, and it has not been reported that two brothers in the same family suffer from this type of disease. Through magnetic resonance imaging and physical examination, we found no pathognomonic signs and symptoms of spinal cord injuries, disc herniation or nerve entrapment. Therefore, we can't find the cause of the low back pain. The unique abnormality is the result of whole exome sequencing, the mutation of TBX6 can lead Spondylocostal Dysostosis. An autosomal dominant or recessive inheritances is believed of this disease from a genetics perspective. But, there is no article reported how the mutation of TBX6 lead congenital scoliosis or multiple butterfly vertebrae. There is also no literature sharing the treatment of these patients whose spine combines with multiple butterfly vertebrae and hemivertebrae.

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