Spondylocarpotarsal synostosis syndrome. A rare case of short stature and congenital scoliosis

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Summary

Spondylocarpotarsal synostosis is a very rare skeletal disorder characterized by vertebral malsegmentation defects. Apart from severe vertebral defects, the disease is associated with carpal and tarsal synostosis which is quite characteristic for the disease. We report a case of young child who presented with short stature and congenital scoliosis. The radiological and clinical findings were compatible with the above diagnosis. Apart from the classical findings, the patient had evidence of odontoid aplasia which has not earlier been described in association with this disorder. We report this case for rarity of this disorder and the associated novel finding.

KEY WORDS: spondylocarpotarsal synostosis; carpal fusion; odontoid aplasia; tarsal fusion; skeletal dysplasia.

Introduction

Vertebra malsegmentation can give rise to variable degree of spinal deformity including congenital scoliosis. These individuals are brought to clinical attention either for evaluation of skeletal dysplasia or associated short stature. Spondylocarpotarsal synostosis (SCT) is a very rare form of vertebral segmentation defect characterized by abnormal fusion of vertebra and fusion of carpal and tarsal bones (1, 2). We report a case of SCT in a female child who presented for evaluation of severe short stature and congenital scoliosis.

Case report

A 2 year 6 months female child was brought by her parents for evaluation of poor gain in height and skeletal deformity. The child was born out of non consanguineous marriage delivered at term by normal vaginal delivery. There was history of two first trimester abortions in mother prior to delivery of index case. There was delay in both motor and social developmental milestones of child as per her age. There was presence of severe joint contractures of bilateral elbow and knee joint. There was some improvement in mobility of joints from baseline after patient underwent rehabilitative orthopedic physiotherapy. She had severe short stature (height SDS -4.94) and low weight (weight SDS -1.5). There was presence of severe degree of congenital scoliosis. There was evidence of disproportionate short stature with short trunk, thoracic scoliosis and lordosis. Clinical evaluation showed presence of wide nasal bridge, prominent forehead, anteverted nose and low set ears. There was presence of bilateral pes planus. Neurological and ophthalmological evaluation did not reveal any gross abnormality. Skeletal imaging revealed presence of fusion of posterior elements of all levels of vertebral column with intermittent paucity or absence of posterior elements (Figure 1). There was presence of gross kyphoscoliosis of vertebral column (Figure 1). Computed tomography (CT) of spine also confirmed these findings (Figure 2). Along with fusion of anterior segment of C1 and C2 vertebra, there was presence of odontoid aplasia (Figure 3). X-ray of bilateral hands and feet revealed presence of capito-hamate and cuboid-cuneiform fusion (Figure 4). Magnetic resonance imaging of spine (T2 w sagittal image) revealed extensive fusion of posterior spinal arch at multiple levels of cervical and thoracic vertebrae along with fusion of anterior arch of atlas with body of axis vertebra without any gross cord compression (Figure 5). Liver function tests, renal function tests and serum electrolytes were normal. Ultrasonography of abdomen and pelvis did not reveal any abnormality. Biochemical evaluation for metabolic bone disease including serum calcium, phosphorous, alkaline phosphatase, vitamin D and serum parathyroid hormone were normal. Based on above findings a diagnosis of spondylocarpotarsal synostosis syndrome (SCT) was made. The parents were counseled regarding the genetic nature of disease and future prognosis. The patient was referred to department of orthopedics and spine surgery for further management of scoliosis.

Discussion

Spondylocarpotarsal synostosis syndrome (SCT) (OMIM # 272460) is an extremely rare genetic disorder. The term was coined by Langer et al. who reported the classical clinical and radiological findings for the first time in 1994 (2). Till 2008, only 26 cases were described in medical literature and less than a dozen more have appeared since then (3). The cardinal features of SCT include multiple abnormal segmentation of spine and vertebral fusion, scoliosis, lordosis, carpal and tarsal synostosis, disproportionate short stature with...
short trunk, short neck, pes planus, dental enamel hypoplasia, hearing loss and mild facial dysmorphism (2, 3). Other reported anomalies include elbow joint mobility limitation, brachydactyly, club foot, cleft palate, hearing loss, high arched palate, kidney anomalies, inguinal hernia and dentition defect (4). The main differential of diagnosis of SCT include Spondylocostal dysplasia (STD), Ischiovertebral dysplasias (IVD), Cerebro facio thoracic dysplasia (CFTD), Robinow syndrome (RS) and other rare syndromes (4, 5). These group of disorders usually present with variable vertebral anomalies (block vertebrae, hemi vertebrae, butterfly vertebrae) and severe rib changes (missing, bifid, fused) (5). The presence of carpotarsal synostosis clinches the diagnosis in favor of SCT. Hence a simple inexpensive skeletal survey is crucial for diagnosis. The most common isolated carpal fusion include capitate/hamate and lunate/triquetrum (5). Similarly, the most common tarsal fusion include talocalcaneal and calcaneo-navicular (5, 6). Hence it is prudent to include radiographic evaluation of hands and feet in patients presenting with congenital progressive scoliosis due to
The scoliosis of SCT appears early and is of severe degree as compared to scoliosis of the spondylothoracic dysplasia. The first breakthrough in deciphering the genetic basis of disease came when Steiner et al. reported a locus for SCT at chromosome 3p14 in 2004 (7). Krakow et al. reported for the first time that homozygosity and compound heterozygosity for nonsense mutation in the filamin B (FLNB) gene on chromosome 3p14.3 resulted in SCT (8). The disease is inherited as an autosomal recessive disorder. FLNB is expressed throughout the cartilage growth plate as well as in the cartilaginous condensations of developing vertebrae (8). Biallelic loss of function mutations leading to loss of FLNB cause SCT, while heterozygosity for missense mutations in FLNB produces a spectrum of autosomal dominant skeletal disorders including boomerang dysplasia, Larsen syndrome and atelosteogenesis I and III (9).

Zieba et al. reported that FLNB is involved in attenuation of TGF beta/BMP signaling and influence annulus fibrous cell fate, thereby causing intervertebral disc disruptions in case of loss of function mutation in FLNB (9). Further breakthrough in understanding molecular basis of SCT came when an animal model of SCT that is FLNB knockout mice was discovered (10). It was initially hypothesized that SCT vertebral fusions resulted from failure to segment the vertebrae properly (2, 11), but analysis of the FLNB knockout mouse model demonstrated that the vertebrae form normally but subsequently fuse (10). It is worth mentioning that our patient had evidence of fusion of anterior arch of atlas with malformed vertebrae (5).
body of axis vertebra and aplasia of odontoid process. Complete agenesis of odontoid is extremely rare and usually occurs in the context of collagenopathy syndromes such as spondyloepiphyseal and spondylometaphyseal dysplasias (12). Cervical spine instability and findings like atlanto-axial rotatory fixation have been very rarely reported in SCT (11, 13). These could predispose towards possible cervical spine instability and atlanto axial rotatory fixation. Odontoid hypoplasia is also a rare reported finding in SCT (2, 11) and to best of our knowledge odontoid aplasia has not been reported earlier in SCT. Hence periodic neuroimaging may be required for early identification and management of potential adverse events.

Conclusion
Hence SCT should be an important differential in any child presenting with short stature and severe congenital scoliosis. Identification of carpo-tarsal synostosis and absence of rib abnormalities establishes the clinical diagnosis. The finding of odontoid aplasia in our patient expands the spectrum of craniovertebral junction anomalies associated with SCT. Knowledge about this rare disease entity among physicians is essential for early identification and management for optimal outcome.

References