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Short Trunk Dwarfism and Kyphoscoliosis are the First Alarming Signs in Children with Kozlowski Type of Spondylometaphyseal Dysplasia

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ABSTRACT

Background: In early childhood moderate short trunk dwarfism associated with spine misalignment can occur in a number of heritable bone disorders. Later on, scoliosis or kyphoscoliosis associated with curved limbs associated with waddling gait evolved. Stubby hands and feet with stiff joint movements associated with gait disturbance are additional abnormalities which manifest itself during the course of the disease. Clinical and radiological phenotypic characterizations are the baseline tools to delineate and diagnose the different types of skeletal dysplasia.

Patient and Methods: Short trunk dwarfism, progressive kyphoscoliosis in a group of children with pre and postnatal growth retardation (two boys and two girls and one adult unrelated parents (aged 6 -13 and a-33-year-old woman have been enrolled in this study). Family history and radiographic documentation revealed two parents of the two unrelated children are manifesting typical phenotype akin to their children. Progressive kyphoscoliosis deformity in a-6-year-old-boy (kyphosis 30°Cobb´s angle and scoliosis 20° and a 13-year-old girl kyphosis - 70◦ Cobb's angle and scoliosis - 55◦ Cobb´s angle) were the main deformity. Clinical and radiological phenotypic characterizations were the baseline tool for management.

Results: The distinctive clinical features in these children and adults were severe short trunk dwarfism, prominent forehead/frontal bossing, hyper lordosis and forearm deformity. Waddling gait, restricted joint mobility and progressive kyphoscoliosis were evident. Radiological phenotype revealed flattening of the vertebrae with rounding of the anterior ends and posterior wedging. The pelvis showed squared iliac wings, narrow sciatic notches, flat trident acetabular roofs, short femoral necks resulted in coxa vara. The overall clinical and radiological phenotype were compatible with the diagnosis of Spondylometaphyseal dysplasia –Kozlowski type (SMD). Correction of kyphoscoliosis deformity has been successfully performed in the 13-year-old-girl - «growing rod» construction. After 5 years the patient performed staged operations. Final correction kyphoscoliosis deformity - final posterior fusion Th2-L5 (posterior spondilodesis)

Conclusion: Three major skeletal problems have been seriously correlated to the SMD Kozlowski type in our group of children and adults. Firstly, the early onset progressive kyphoscoliosis, secondly odontoid hypoplasia with subsequent development of os odontoideum with subsequent development of atlantoaxial instability. Thirdly, the development of coxa vara which is correlated to progressive rarefaction of the bone matrix. Furthermore, the lack of the normal cohesiveness of the femoral necks in connection with metaphyseal dysplasia play a negative role in the biomechanics of the locomotor system. In this study we discussed the management of the musculoskeletal abnormalities in patients with SMD Kozlowski type.

Keywords: Spondylometaphyseal, Dysplasia, Childhood, Adulthood

1. Introduction

Spondylometaphyseal dysplasia, Kozlowski type (SMD-Kozlowski) (OMIM 184252) is characterized by a constellation of abnormalities noted in early childhood such as moderate dwarfism, most marked in trunk region, short neck, scoliosis/kyphoscoliosis, mildly curved limbs, short and stubby hands and feet. The earliest sign of the disease is often growth retardation with spinal kyphosis appearing around the age of 18 -24 months of age. Though several reports showed that scoliosis of progressive nature was noted at birth[1-3]. Musculoskeletal features of limitations in joint mobility associated with gait disturbance and in some patient's genu valgum. Progressive kyphoscoliosis, either manifested in early childhood or late in adulthood[4]. The radiological phenotype is characterized by generalized severe platyspondyly with increased height of the intervertebral disc space, open staircase vertebral bodies, medially placed pedicles, scoliosis/kyphoscoliosis and early osteoarthritic spine changes are common manifestations of the disease. Broad short basilar portion of the iliac bones, with broad horizontal acetabular bones and horizontal acetabular roofs (these are mostly noted in children). Progressive metaphyseal widening, sclerosis and irregularity causing effectively the development of coxa vara. Coxa vara is a common finding which is correlated to the irregular metaphyseal ossification in the tubular bones (of maximal intensity in the proximal femora). The metaphyses are characterized by widening, sclerosis and irregularity which effectively leads to the development of coxa vara.)[5-8]. Congenital coxa vara is commonly associated with a significant number of diverse forms of skeletal dysplasia and in some can lead to limb-length discrepancy. An association between coxa vara and abnormalities of development of the femur such as congenital short femur, congenital bowed femur and proximal focal femoral deficiency has long been recognised[1-8]. The autosomal dominant TRPV4 disorders are a group of skeletal dysplasia's encompassing spondylometaphyseal dysplasia Kozlowski type, spondyloepiphyseal dysplasia Maroteaux type, Metatropic dysplasia and other disorders of neuromuscular origin. All these disorders are caused by mutation in TRPV4 gene (OMIM 605427). Differentiation and diagnosis can only made via clinical and radiographic phenotypic characterizations[1-3, 9-11].

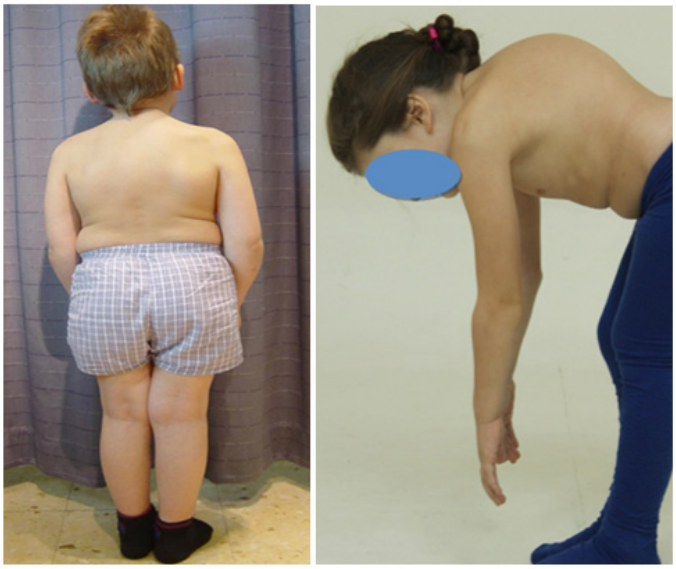
2. Patients and Methods

The study protocol was approved by Ethics Committee of the National Medical Research Center for Traumatology and Orthopedics, (number 4(50) issued on 13.12.2016) Kurgan, Russia. Informed consents were obtained from the patient's Guardians to publish. Six patients (girls and 4 boys with age average of 3 years). We fully documented these children through detailed clinical and radiological phenotypic characterizations at the axial skeleton and neurosurgery department, Ilizarov Center, Russia. At birth babies showed pre and postnatal growth deficiency and later on in infancy children with SMD type Kozlowski syndrome manifested short trunk dwarfism. The clinical phenotype of the backbone in a-6-year-old-boy who showed short trunk dwarfism with no associated rhizomelia. Similarly, a-13-year-old –girl showed apparent trunk dwarfism associated with progressive kyphosis, though with normal length of the limbs (Figure 1a and b). Their subsequent course of development has been described as being retarded in acquiring motor skills. Spinal kyphosis/kyphoscoliosis appeared around the age of 18 to 24 months which is characterized as being of progressive nature. All our patients were products of uncomplicated gestation. All born full term, at birth the length and weight were around the ١٠th Percentile and OFC around the 25th percentile. Walking has been achieved around the ages of ١٥ to 18 months albeit with difficulties. Gait abnormalities predominate the motor activity followed later on by the development of progressive limp. At time of presentation all manifested proximal varus deformity with an angle of 80°. Limb length inequality was around the average of 2.5 cm to 3 cm. No particular cranio-facial dysmorphic has been noted. Musculoskeletal examination showed a positive Trendelenburg sign, abduction limited to 30° and internal rotation limited to 10° were present mostly on the left side. The right hip could be abducted 60° and internally rotated 35°. No ligamentous hyperlaxity but apparent joint stiffness has been noted. Upper limbs showed no abnormalities (except for stubby hands and feet). Neurological examination was normal. Hearing, vision and intelligence were normal. Examination of hair, teeth, nails and skin were normal. Laboratory studies showed normal white and red blood cell and platelet counts, normal serum calcium, phosphorus and alkaline phosphatase levels. Screening of urine for amino acids and mucopolysaccharides were negative and all manifested normal karyotype. Fluorescence In Situ Hybridization (FISH) tests showed normal results.

3. Results

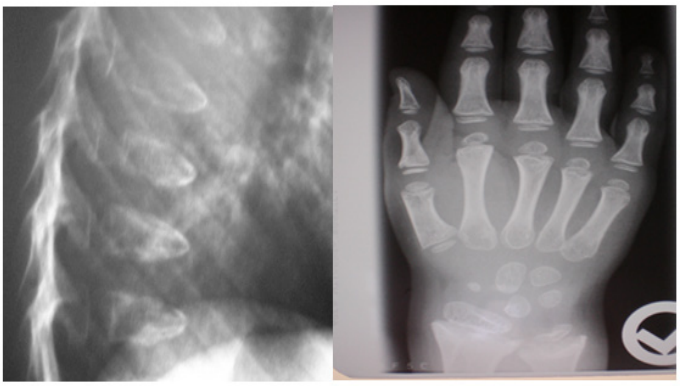
In accordance with the aforementioned clinical features, the radiological phenotype has been performed to further interpret the constellation of the skeletal abnormalities. Lateral thoracic spine radiograph of a-six-year-old-boy with SMD (Kozlowski type) showed generalized platyspondyly (flat vertebral bodies) (a). AP hand radiograph of a-12-year-old-boy with SMD (Kozlowski type) showed small carpal bones, retardation of the carpal ossification's centres with minimal metaphyseal

changes of the short tubular bones which look short and broad. Note marked metaphyseal dysplasia of the inferior ends of the radius and ulna (Figure 2a and b). Lateral spine radiograph of a -13-year-old-girl with the diagnosis of SMD type Kozlowski showed evident generalized platyspondyly with anterior wedging (a). Note, severe kyphoscoliosis deformity. (kyphosis - 70◦ Cobb and scoliosis - 55◦ Cobb) (Preoperative radiographs) (Figure 3a and b). Postoperative spine radiographs showed correction kyphoscoliotic deformity - «growing rod» construction. For 5 years the patient performed staged operations. final correction kyphoscoliotic deformity - final posterior fusion Th2-L5. Posterior spondilodesis (Figure 4a and b).



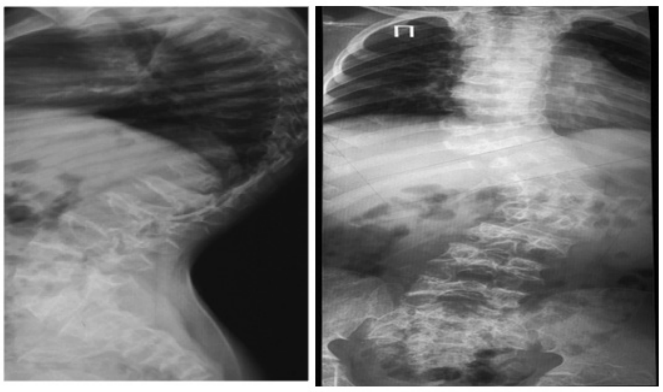
(a) (b)

Figure 1a): A-6-year-old-boy showed short trunk dwarfism with no associated rhizomelia. **b):** A-13-year-old –girl showed marked short trunk dwarfism associated with progressive kyphosis with normal limb lengths.



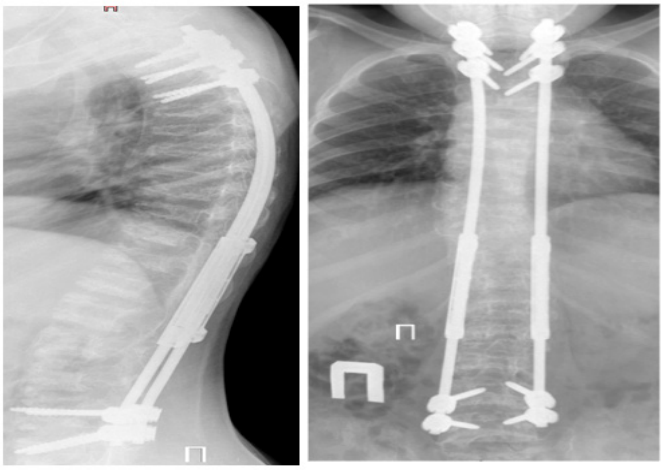
(a) (b)

Figure 2: a) Lateral thoracic spine radiograph of a-six-year-old-boy with SMD (Kozlowski type) showed generalized platyspondyly (flat vertebral bodies). **b)** AP hand radiograph of a-12-year-old-boy with SMD (Kozlowski type) showed small carpal bones, retardation of the carpal ossification's centres with minimal metaphyseal changes of the short tubular bones which look short and broad. Note marked metaphyseal dysplasia of the inferior ends of the radius and ulna.



(a) (b)

Figure 3: a) Lateral spine radiograph of a -13-year-old-girl with the diagnosis of SMD type Kozlowski showed evident generalized platyspondyly with anterior wedging. **b)** Note, severe kyphoscoliosis deformity. (kyphosis - 70◦ Cobb and scoliosis - 55◦ Cobb)(Preoperative radiographs).



(a) (b)

Figure 4: a) Postoperative spine radiographs showed correction kyphoscoliotic deformity - «growing rod» construction. **b)** For 5 years the patient performed staged operations. final correction kyphoscoliotic deformity - posterior fusion Th2-L5. Posterior spondilodesis.

4. Discussion

Spondylometaphyseal dysplasia is a heterogeneous group of disorders characterised by platyspondyly and metaphyseal changes not unlike those of Schmid's metaphyseal chondrodysplasia[12]. Spondylometaphyseal dysplasia has been discussed by various authors by whose names are well known such as Kozlowski[1], Sutcliffe[13], Murdoch and Walker[14], Borochowitz[15], Wiedemann and Spranger[16]. Kozlowski type of spondylometaphyseal dysplasia can manifest itself early on in life or later in life. Spondylometaphyseal dysplasia Kozlowski type has been published by various authors. A series of further cases was described by Krakow et al.[17]. Four of patients had typical findings that included scoliosis, a waddling gait and radiologically, platyspondyly, over faced vertebral pedicles, flared iliac wings, a mildly flattened acetabular roof and irregular capital femoral metaphysis and delayed carpal ossification. Some had findings akin to non-lethal metatropic dysplasia. Heterozygous mutations in TRPV4 (as in metatropic dysplasia and mild autosomal dominant brachyolmia) were found in some patients. The differential diagnosis of patients with

SMD-Kozlowski type has to be made with metatropic dysplasia (which differs by the presence of dumb-shaped long tubular bones and more severe epiphyseal dysplasia as well as crescent shaped iliac wings[18,19]. Kozlowski type SMD should also be differentiated from Spondylo-chondrodysplasia[20]. Dyggve-Melchior-Clausen syndrome (Smith-McCart dysplasia), which does not include mental retardation, should be included in the differential diagnosis[21]. Spondylometaphyseal dysplasia cone – rod dystrophy is another entity akin to SMD Kozlowski type c but in the former lateral views of the spine showed ovoid shaped vertebral bodies rather than flat as seen in the latter[22]. Spondyloepimetaphyseal dysplasia type Maroteaux shows varying degrees of platyspondyly with less metaphyseal and more epiphyseal involvement[23].

Kyphosis presenting at birth or before adolescence is almost always due to congenital anomaly of vertebral development. The causation and pathogenesis of congenital kyphosis/ scoliosis and or kyphoscoliosis are arise from either a dysplastic process or from vertebral malsegmentation. Scoliosis and kyphoscoliosis are a symptom complex in more than 742 syndromic entity[24].

Coxa vara is defined as a decrease in the angle between the head or neck and shaft of the femur, often in presence of a metaphyseal defect. In other words, the neck-shaft angle is less than 110°. It is characterised by a decrease in the neck-shaft angle and clinically by a waddling gait or limb length discrepancy. Symptoms do not develop until the child starts to walk and often not before the age of 3 or 4 years. Coxa vara is a constant finding in many types of skeletal dysplasia as part of the manifestations of a generalized growth disturbance. Symptoms of coxa vara do not develop until the child starts to walk and often not before the age of 3-4years. There is a typical limp or a waddling gait resembling that of dysplastic developmental dislocation of the hip. Abduction of the hip is limited; there is a positive Trendelenburg's sign on standing on the affected side and the greater trochanter is elevated. It has been suggested that abnormal development of the proximal femoral cartilaginous physis and defective ossification of the adjacent metaphysis are responsible for the progressive decrease of the neck shaft angle[25,26]. Oh, et al.[27] studied 46 patients with coxa vara. Spondyloepiphyseal dysplasia congenita or spondyloepimetaphyseal dysplasias were the forms of osteochondrodysplasias reported in connection with congenita coxa vara. They concluded that the lack of the epiphyseal ossification was the most challenging element. Further studies indicated that histological investigations revealed abnormalities in the proximal femoral physeal chondrocyte maturation, with disruption of the normal columnar architecture and abnormal calcification of the cartilaginous matrix. This abnormal endochondral ossification resulted in decreased production of the metaphyseal bone, leading to relative osteoporosis and subsequent weakness in this area. Patients with coxa vara and a Hilgenreiner angle greater than 60 degrees are candidates for a surgical intervention, as are patients with a Hilgenreiner angle greater than 45 degrees who are symptomatic presenting with limping or showing progression. Surgical correction is performed by a valgus osteotomy according to Pauwels (VY osteotomy with plate fixation). Malalignment of the leg can be treated by gradual correction using 8-plates, as long as the epiphseal growth plates are open. The surgical correction of limb length inequality by lengthening procedures is risky. Prerequisite for indication should be absolute stability of hip and knee joints[28,29].

5. Conclusion

Congenital kyphosis/kyphoscoliosis and Coxa vara are almost always a symptom complex rather than a separate diagnostic entity until proven otherwise. Kyphosis present at birth or in pre-adolescence is almost always due to a congenital anomaly in correlation with vertebral mal-development (genetically programmed). The deformity may be purely kyphotic or, more commonly, combines kyphosis and scoliosis. The vertebral bodies mal-development in patients with skeletal dysplasia is part of the entire growth disturbance of the skeletal system, which is totally different from other forms of vertebral abnormalities as seen in dozens of syndromic entities in which vertebral dyssygmentations (failure of segmentation) as in block vertebrae is part of the symptom complex in. Coxa vara can be clinically classified as developmental, congenital, dysplastic or traumatic and may occur at the physis or in the trochanteric or subtrochanteric area. Skeletal dysplasia, in particular, can manifest a wide range of variable and confusing phenotypic features, varying from profound dwarfism, lethal in utero, to phenotypically normal individuals but with a genetically programmed disorder of unpredictable onset and form. Clinico-radiographic documentation is the base line for proper management. Our paper might indicate an area of concern to generate further research that it could lead to elaborate knowledge.

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