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Spondyloepiphyseal dysplasia related osteoporosis in a young male patient

Ebru Aytekin, Nil Sayiner Caglar, Ertan Yüce, Yasemin Pekin Dogan, DidemİnceboyYalçın

ABSTRACT

Spondyloepiphyseal Dysplasia (SED) is a rare bone growth disorder. Clinical manifestations may include dwarfism, short trunk and neck, flat face, shortened limbs, coxa vara, genu varum/valgum, kyphoscoliosis, increased lomber lordosis, platyspondylia, barrel-shaped chest (pectus carinatum). Type II collagen is the dominant protein of cartilage in which is thought to play a major role in the etiology of the disorder. Many congenital diseases, including osteogenesis imperfecta, hypophosphatemic rickets and lysosomal storage diseases, have been associated with abnormal bone metabolism and osteoporosis. Coexistence of SED and osteoporosis rarely reported in the scientific literature. In this case report, we discussed SED related osteoporosis in a young male patient in the context of current literature.

Keywords: Osteoporosis, Spondyloepiphyseal dysplasia, Type II collagen

Ebru Aytekin[1], Nil Sayiner Caglar[1], Ertan Yüce[1], Yasemin Pekin Dogan[1], DidemİnceboyYalçın[1]

Affiliation: [1]Health and Sciences University, Physical Medicine and Rehabilitation Clinic, Istanbul Turkey.

Corresponding Author: Ebru Aytekin, Kasapİlyas Mahallesi, Org. Abdurrahman Nafiz Gürman Caddesi Fatih, İstanbul; Email: ebruaytekin@hotmail.com

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INTRODUCTION

Spondyloepiphyseal dysplasia (SED) is a rare, non-lethal genetic bone growth disorder that results in dwarfism and skeletal abnormalities. This condition affects the bones of the spine and the epiphysis of long bones in the arms and legs so as named spondylo-epiphyseal [1, 2]. Clinical manifestations may include short stature with a very short trunk and neck, flat facial features, rhizomelic shortened limbs, coxa vara, genu varum/valgum, kyphoscoliosis, increased lomber lordosis, platispondily, barrel-shaped chest. [3,4]. Early radiological features include flattened or deformed epiphyseal ossification centers of the long bones and vertebraes [5]. Three different types of SED is described. Each is inherited differently and diagnosed at different periods of the child's life: 1. SED conjenita form inherited as autosomal dominant trait. 2. SED tarda (SEDT) 3. Progressive artropathy with SEDT.Tarda forms inherited as X-linked trait. Decreased joint mobility and arthritis often develop early in life [6]. Intelligence is usually unaffected [4]. Advanced paternal age is associated with an increased risk of SED [7]. Type II collagen is the dominant protein of cartilage in which is thought to play a major role in the etiology of the disorder [5]. Type II collagen adds construction and strength to the

connective tissues. Type II collagen is found primarily in cartilage. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones. Type II collagen is also part of the center portion of the discs between the vertebrae in the spine [1, 8]. Abnormalities of type II Collagen have been found in a number of chondroplasias, including type II achondrognesis, hypochondrogenesis. These disorders have substantial phenotypic and radiologic similarities [5]. Coexistence of SED and osteoporosis rarely reported in the scientific literature. In this case report we have discussed SED releated osteoporosis in a young male patient in the context ofcurrent literature.

CASE REPORT

A 33-year-old male referred to the physical medicine and rehabilitation clinic with bilateral painful and restricted shoulder, elbow and knee. The facial appearance of the patient was compatible with the phenotypic characteristics of SED that he has been diagnosed as SED at childhood. Neorological examination findings were within normal limits, intellectuel development was good as well. In his physical examination mobility of both shoulders, elbows, hips and knees were diminished and shortness in all extremities was present. In postural examination anteflexion posture, barrel chest deformity, increased lumbar lordosis, coxa vara and genu valgum deformities were observed. His height and arm span were measured as 131 cm and 94 cm respectively. Routine laboratory test results including kidney and liver functions, calcium, phosphorus, alkaline phosphatase, and parathyroid hormone were all normal. In addition, acute phase response proteins (i.e., C-reactive protein, erythrocyte sedimentation rate), rheumatic profile, thyroid functions, gonadal hormones, and tumor markers were normal. The level of 25(OH)D vitamin was 49,90 ng/mL (30–100 ng/ml). A lateral lumbar X-ray showed increased apparent radiolucency of the vertebral bodies, loss of both anterior, middle and posterior height of the vertebral body (platyspondylia) (Figure 1). In his sagittal lomber magnetic resonance imaging (MRI) there was a wedge-shaped vertebrae at the level of T12-L1 region (Figure 2). There were degenerative arthritic changes as narrowing of the joint space, increased radiolucency on both shoulder and knee plain radiographs. Also there was genu valgum and coxa vara deformities on his knee plain radiographs (Figure 3, 4). A dual energy X-ray absorptiometry (DEXA) scan revealed osteoporosis (Left femur neck T score: -2,5, Z score: -2,2, lumbar L 2-4 T score: -3,0, Z score: -3,0). The bone isotope scan confirmed degenerative changes on bothshoulders, elbows, knees and vertebraes (Figure 5.). Other causes of secondary osteoporosis(cigarette, alcohol, drug consumption, malabsorption, history of parent fracture, nutrition problems, chronic kidney insufficiency, hypogonadism, endocrine disorders, immobilization) were excluded. Alendronate 70 mg/week, 1000 mg calcium and 880 IU vitamin D3/day were prescribed for osteoporosis medical treatment. Throughout the medical treatment, the range of motion, stretching and strengthening exercises and 10 sessions of physical therapy were instructed to painful and restricted areas. There was a marked reduction in complaints at the end of three months.

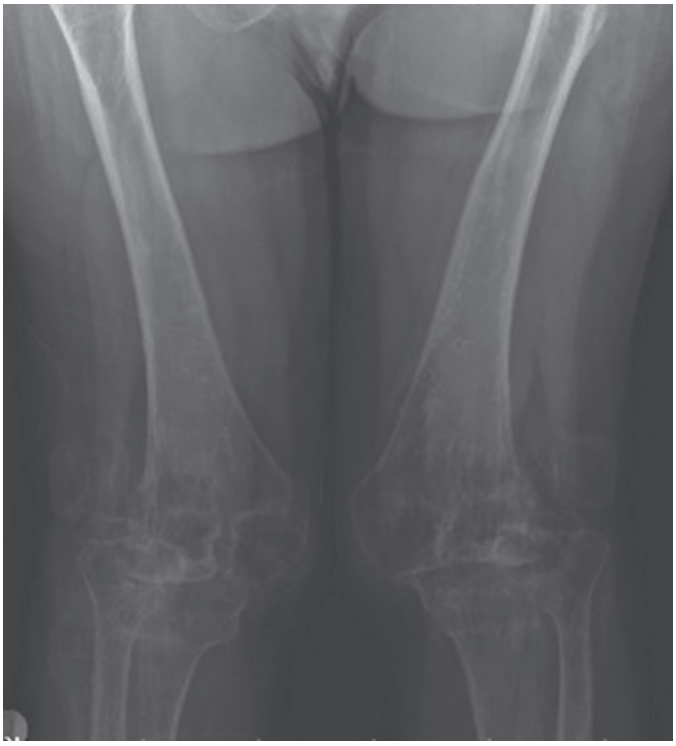

Figure 1: Lateral thoracolumbar graphy showing increased apparent radiolucency of the vertebral bodies, loss of both anterior, middle and posterior height of the vertebral body (platyspondylia).

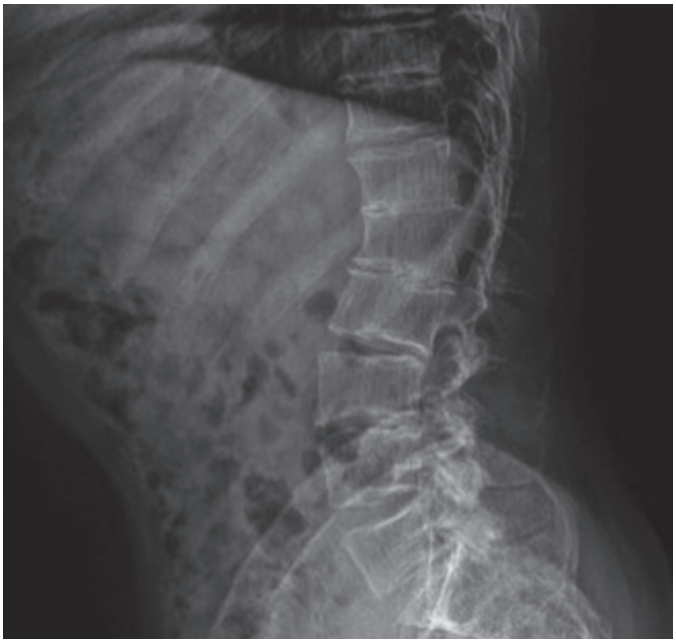

Figure 2: A vertebrae wedge-shaped at the level of T12-L1 region on sagittal T2 weighted lomber MRI.

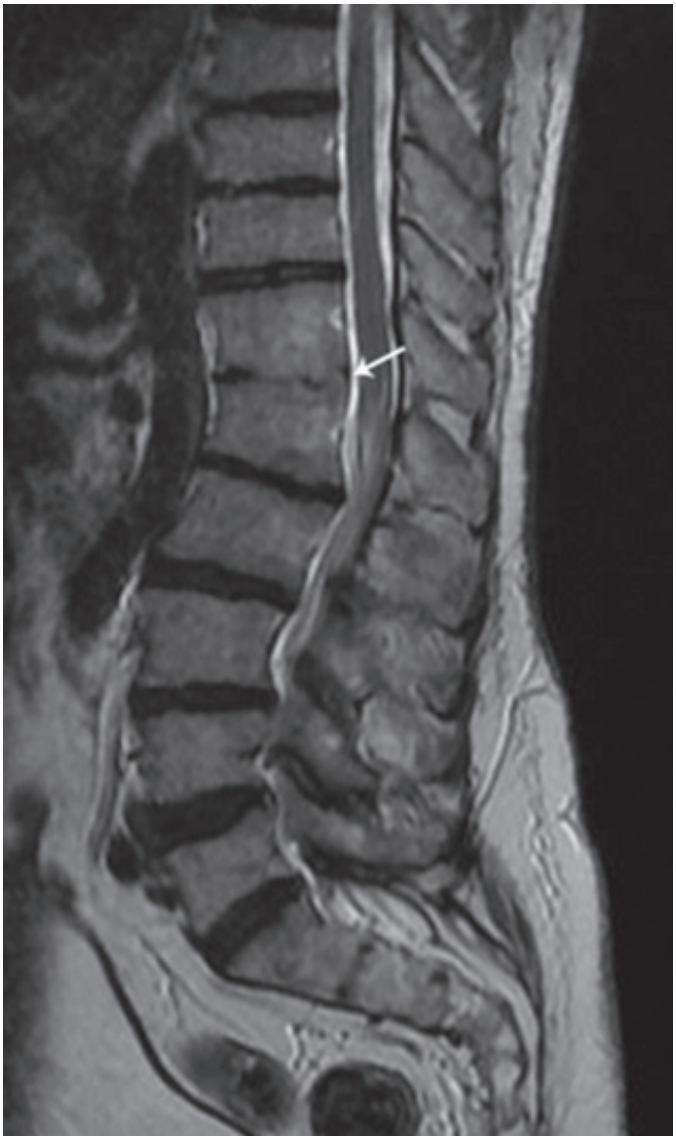

Figure 3: Degenerative arthritic changes, narrowing of the joint space, increased radiolucency on both shoulder plain radiographs.

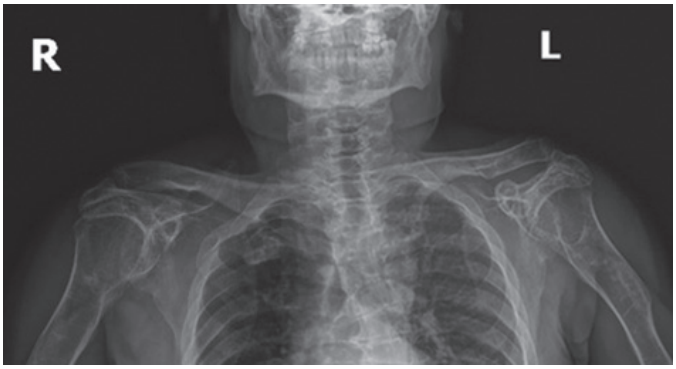

Figure 4: Degenerative arthritic changes, narrowing of the joint space, increased radiolucency, coxa vara, genu valgum deformity on both knees plain radiographs.

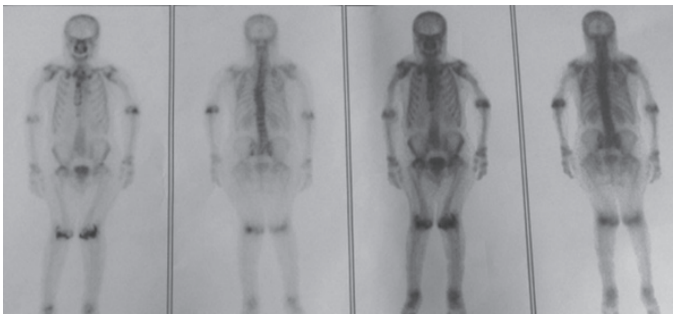

Figure 5: Degenerative changes on both shoulders, elbows, knees and vertebraes on bone isotope scan.

DISCUSSION

There are several congenital diseases with abnormal bone metabolism and osteoporosis, including osteogenesis imperfecta, hypophospatemic rickets and lysosome storage diseases [9]. However, SED have been rarely described as osteoporotic in the literature and coexistence of SED and osteoporosis has rarely been reported in the scientific literature.

Osteochondrodysplasias are shorter than normal population. The mean height of men was measured 118-145 cm and woman was 112-136 cm and arm span is almost the same as their height [5,10]. The height of our patient was consistent with this range but his arm span was shorter than his height. It may be linked to severe contractures in upper limbs.

Three forms of SED has been defined. The third form SED tarda with progressive arthropathy usually occurs at 3-8 years. Especially the clinic of SEDT with progressive arthropathy consists of pain, stiffness and contracture development in hands and other joints [11]. Individuals with SEDT with progressive arthropathy are more likely to develop pain, inflammation and damage in affected joints at an early age (early-onset osteoarthritis) [12] like in our case. Clinical manifestations of SEDT with progressive arthropathy resembles rheumated arhritis but laboratory findings and rheumatic profile must be in the normal limits. In addition, the onset of the disease at an early age should be considered in the differential diagnosis of spondyloarthropathies because of the similarity of symptoms (chronic back and buttock pain, swelling of the joints, morning stiffness) and radiological findings. Also the risk of osteopenia or osteoporosis is higher in this form [4]. Joint abnormalities may lead to development of hip deformity in which the thigh bone is angled towards the center of the body (coxa vara) and/or knee deformities, including bow legs (genu varum) and 'knock knees' (genu valgum) [12]. We thought our case to be compatible with SEDT with progressive arthropathy as a diagnosis. Also the fact that our patient is male and that there is not a history of kinship between the parents supported an X-linked inheritance. In addition to physical therapy including analgesic currents, for existing joint contractures; strengthening, joint range of motion,

stretching exercises were also instructed to him. And also strengthening exercises for the low back and abdominal muscles were prescribed.

The spine is mostly affected in SED. Platispondly with cone-shaped flattening of the dorsal aspects with narrowing of the interpedicular distances of the lower thoracic and lumbar vertebral bodies develops. This at last leads to wedge-shaped thoracic vertebrae and hence to severe kyphoscoliosis. Thoracolumbar kyphosis seen in patients with SED depends on the presence of one or two wedge shaped vertebra corpus. Involvement between T11 and L2 vertebral region is the most prevalent known reason of kyphosis [5]. In our case, there was a wedge shaped vertebrae due to hypoplasia of the T12-L1 disc, consistent with the literature. Some individuals may have instability of the spine in the neck region, which can increase the risk of spinal injury and eventually cervical myelopathy [13].

Back pain in SED patients has been generally considered to be associated with increased lomber lordosis and spinal canal stenosis rather than osteoporosis. Despite that increased bone turnover and low BMD may cause vertebral fragility fractures and back pain in these patients [5].

CONCLUSION

As a result in patients with SED, osteoporosis-related symptoms should be monitored and examined carefully and must be treated if osteoporosis is present. Regular exercises and proper diets should be recommended and be encouraged to practice their daily routines. These approaches may prevent future fragility fractures due to osteoporosis. Also we have to be careful about cervical and lumbar spinal stenosis releated symptoms in patients with SED.

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Author's Contributions

Ebru Aytekin – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Nil Sayiner Caglar – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Ertan Yüce – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Yasemin Pekin Dogan – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
DidemİnceboyYalçın – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor of Submission

The corresponding author is the guarantor of submission.

Source of Support
None

Consent Statement
Written informed consent was obtained from the patient for publication of this case report.

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Written informed consent was obtained from the patient for publication of this case report.

Conflict of Interest
Authors declare no conflict of interest.

ABOUT THE AUTHORS

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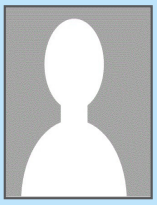
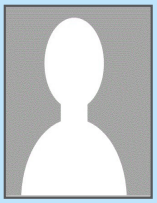
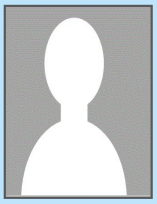


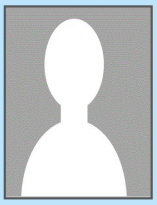
Ebru Aytekin is Associate Professor at Istanbul Training and Research Hospital, Physical Medicine and Rehabilitation Clinic, Health and Sciences University, Istanbul, Turkey.
Email: ebruaytekin@hotmail.com



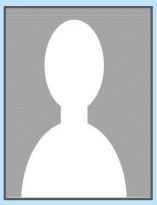
Nil Sayiner Caglar is Associate Professor at Istanbul Training and Research Hospital, Physical Medicine and Rehabilitation Clinic, Health and Sciences University, Istanbul, Turkey.
Email: nilcag@yahoo.com



Ertan Yuce is Assistant Doctor at Istanbul Training and Research Hospital, Physical Medicine and Rehabilitation Clinic, Health and Sciences University, Istanbul, Turkey.
Email: yuceertan@hotmail.com



Yasemin Pekin Dogan is Specialist Doctor at Istanbul Training and Research Hospital, Physical Medicine and Rehabilitation Clinic, Health and Sciences University, Istanbul, Turkey.
Email: yaspekin@yahoo.com



Didem Inceboy Yalcin is Assistant doctor at Istanbul Training and Research Hospital, Physical Medicine and Rehabilitation Clinic, Health and Sciences University, Istanbul, Turkey.
Email: dideminceboy@hotmail.com