



Case Report

Spondyloepipheseal dysplasia tarda

Raisa Altaf*, Rabiya Siraj, Ramsha Fatima Qureshi, Uzma Panhwer, Hina Naseer and Khansa Abro

Department of Radiology, Liaquat National Hospital Karachi, Pakistan

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***Corresponding authors:** Raisa Altaf, Department of Radiology, Liaquat National Hospital Karachi, Pakistan, Tel: 03323704910; Email: RAISAALTA88@GMAIL.COM

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Abstract

Skeletal disorders are common entities that we encounter in our daily practice. Their early diagnosis is key to proper management and genetic counselling. Spondyloepipheseal dysplasia is one such disorder. It is a genetic bone deformity that affects the spine, proximal epiphysis and pelvis. The disease is either manifested at birth or during adolescence therefore given the terms SED congenita or SED tarda. Patients with SED present with variable features including short height, short neck, club foot, cleft palate, kyphoscoliosis or lordotic abnormalities. We also present a case of an 11-year-old boy who presented to us with complaints of stunted growth and abnormal posture and underwent radiological imaging.

Abbreviations

SED C: Spondyloepipheseal Dysplasia Congenita; SED T: Spondyloepipheseal Dysplasia Tarda; SEDT-PA: Spondyloepipheseal Dysplasia Tarda with Progressive Arthropathy

Introduction

Spondyloepipheseal dysplasia is a type of bony dysplasia having three types that are Spondyloepipheseal Dysplasia Congenita (SED C), Spondyloepipheseal Dysplasia Tarda (SED T) and Spondyloepipheseal Dysplasia Tarda with Progressive Arthropathy (SEDT-PA) [1,2]. The disease majorly involves the vertebrae and the epiphysis of long bones [3]. The various modes of inheritance include X-linked disease, autosomal recessive and dominant [4]. SED T is a milder form of SED with a slightly older age of presentation and possible normal birth history. X-linked SED T has an incidence of about 1.7 per million [1]. Although rare it progresses slowly presenting between 5-10 years of age [1].

The characteristic features of SED tarda include short stature presenting between 5-10 years, this is partly due to

the spinal involvement. There is platyspondyly with a central hump in the vertebral bodies at all levels. The ring apophysis is also abnormal showing a lack of bony architecture. Scoliosis is also a feature evident in such patients. During the later stages, these patients develop early bony degenerative changes. The spinal involvement does not only result in short stature but also changes in the thoracic cavity, which shows a slight increase in diameter and lies close to the iliac crest. Other features include dysplasia of the femoral head and neck. The articular surfaces of the femurs are irregular [5,6]. Pelvic bones are also seen to be involved in these patients, having a deep narrow pelvis. There is a disproportion in the ilium, ischium and pubic bones. These patients often show early bone degenerative changes. Other large joints may also be involved [5-7].

We have presented a child with features of this rare disorder who presented to us through the Outpatient department and was diagnosed as SED T on clinical and radiological grounds.

Case report

An eleven-year-old male presented to our department after a referral from the Outpatient Department. According to the

parents he had a history of vague backache, delayed growth and abnormal posture for four years. He has a family history of three cousins with the same complaints however no workup or documentary evidence of their illness was available. After the initial lab workup, he was sent for radiological imaging. His X-ray lumbosacral spine was done initially which showed flattening and widening of dorso-lumbar vertebral bodies along with end plate irregularity suggestive of platyspondyly. The edges of the endplates of vertebral bodies showed characteristic anterior and posterior humped shape or heaped-up appearances. Bone density was mildly reduced and there was exaggerated lordosis. On the suspicion of the radiologist further, a skeletal survey was advised. His skeletal survey however showed normal intervertebral disc spaces, iliac bones and femoral heads. He was reported as a case of SED T. The patient was counselled regarding the disease and the genetic counselling of the family was also done. He was further advised genetic testing however due to affordability issues patient was lost to follow-up Figure 1.

Discussion

Amongst the various osteochondrodystrophies, spondyloepiphyseal dysplasia tarda is one rare form that results in dwarfism primarily due to defects in the vertebrae. However, as the name suggests it also involves the epiphysis of long bones [3]. This is a rare congenital disorder that has three further types as discussed previously, spondyloepiphyseal dysplasia congenita, spondyloepiphyseal dysplasia tarda, and spondyloepiphyseal dysplasia tarda with progressive arthropathy (SED T-PA) [4]. The incidence of the rare X-linked SED T is 1.7 per million. It was first described by Jacobsen in 1939 who studied an American family with this disorder and proved the X-linked mode of inheritance, he, however, did not elaborate on the distinctive features of other types of SED [1-6]. Later in 1911 Bannerman, Ingall and Mohn studied the families' genetic linkage

The symptoms and clinical presentation are unlikely before 11 to 13 years as this is the time when the element of dwarfism becomes evident [3]. Most of the patients present with backache and curvature deformity [1-6] as seen in our case a male with backache and delayed growth presented to our department after a referral from the OPD. The family history is positive in

the majority of the cases [1-6]. Our patient also had a positive family history although no proper documentation was available to delineate the pedigree of the patient. The typical features of the disease include flattening of vertebrae, and a humped-shaped appearance mostly central and posteriorly along the body of the vertebrae [3,8,9]. These changes involve the spine diffusely from the 2nd cervical vertebrae up to the lumbar region. Scoliosis is also one of the features of this disorder. Later the patients develop reduced disc space with degeneration and vacuum phenomenon. The diffuse involvement and shortening of the spine results in deformity of the thoracic cavity. The lower ribs lie in close proximity to the iliac bones. Whereas the iliac bones are also small as compared to the ischium and pubis resulting in a narrow pelvis. Femoral head degeneration with deep acetabula and degenerative changes involving the two. Shortening of the femoral neck is also a feature of the disease [3,6,8-13]. Despite the extensive bony involvement the patients have a normal intellect and do not suffer from any systemic complications. The bony pathology may become severe in adult age and result in severe degenerative changes [1-6]. The phenotypic characteristics of individuals with SED are thought to be caused by aberrant production of type II collagen, which inhibits bone formation [9,10]. Although our patient did not have the extra spinal features however the spinal involvement showed the typical picture of SED T with the supporting history of familial involvement. Conventional imaging can easily diagnose the radiological features of the disease however genetic testing is important for the final diagnosis. Our patient also was diagnosed on X-ray imaging however due to affordability issues patient could not afford any further expenses.

A frequent, dangerous consequence in individuals with SED is spinal cord compression brought on by atlantoaxial instability. The lack of spinal cord symptoms does not diminish the necessity for fusion and decompression procedures because, from our perspective, surgery's benefits are mostly preventative [11]. Since there is no cure for this illness, only symptomatic alleviation is frequently possible with rehabilitation and surgical procedures. Osteotomies or arthroplasty may be recommended for some individuals with SED T-PA [11,12]. Non-Steroidal Anti-Inflammatory Medicines (NSAIDs) or even necessary opiates may be necessary for some people. Exercise and physical therapy are also taken into consideration [9-12]. Management of complications includes spine surgery to repair scoliosis or kyphoscoliosis. Osteoarthritis pain treatment as needed is a necessary joint replacement (hip, knee, shoulder). Monitoring is done to check for clinically severe odontoid hypoplasia, cervical spine films should be taken before the patient reaches school age and before any surgery requiring general anaesthesia. Follow-up exams every year to monitor joint discomfort and scoliosis. Situations to avoid are neck flexion and extension at extreme angles in those with odontoid hypoplasia. Unnecessary stress on the spine and weight-bearing joints from certain activities and employment. Assessment of at-risk relatives needed for presymptomatic assessment in males at risk may save needless diagnostic testing for alternative reasons of short stature and/or osteoarthritis [12-14]. The best course of action for SED T

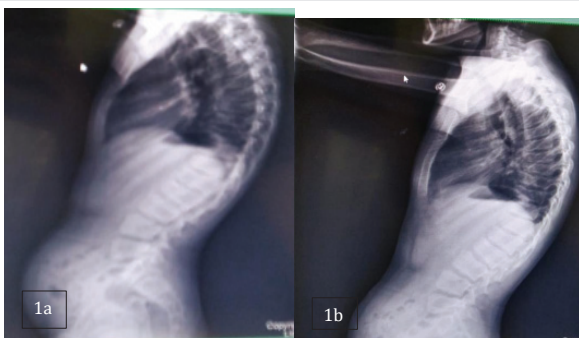


Figure 1a,b: Lateral view of the dorsolumbar spine show flattening and widening of dorso-lumbar vertebral bodies along with end plate irregularity suggestive of platyspondyly. The edges of the endplates of vertebral bodies show characteristic anterior and posterior humped shape or heaped-up appearances.



is to spot individuals who are experiencing progressively worsening neurological and joint mobility issues and undertake the necessary surgical procedures. Neurological performance and general well-being of life can be improved via surgery. Surgery does not, however, stop the illness from progressing as palliative care does [9,10,14,15]. Genetic counselling plays a crucial role in the further management of the patients and intrauterine workup is also necessary.

Conclusion

SED T is a rare congenital osteochondrodystrophy resulting in dwarfism and it has a strong family lineage. This disease has interesting radiological features including flattening of vertebrae, humped-shaped appearance, Scoliosis, Femoral head degeneration and Shortening of the femoral necks which should be considered and known to the radiologists at the time of reporting to help establish the diagnosis and differentiate this disorder from other dystrophies. It is also important for the radiologist to give particular look at the family history of the patient which can also help in making the diagnosis.

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