CASE REPORT

SPONDYLOEPIPHESEAL DYSPLASIA TARDA: A CASE REPORT

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PJR April - June 2023; 33(2): 106-109

ABSTRACT

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

Key words: X-linked recessive, spondyloepiphyseal dysplasia tarda, osteochondrodysplasia.

ABREVIATIONS:

Spondyloepiphyseal dysplasia congenita (SED C)
Spondyloepiphyseal dysplasia tarda (SED T)
spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA)

Introduction

Spondyloepiphyseal dysplasia a type of bony dysplasia having three types: Spondyloepiphyseal dysplasia congenita (SED C), Spondyloepiphyseal dysplasia tarda (SED T) and spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA).[1,4] The disease majorly involves the vertebrae and the epiphysis of long bones.[6] The various modes of inheritance include X-linked disease, autosomal recessive and dominant.[5] SED T is a milder form of SED with 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 but is 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 are also abnormal showing 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 slight increase in diameter and lies close to iliac crest.Other features include dysplasia of femoral head and neck. The articular surfaces of femurs are irregular.[2,3] Pelvic bones are also seen to be involved in these patients, having a deep narrow pelvis. There is disproportion in the ilium, ischium and pubic bones. These patients often show early bone degenerative changes. Other large joints may also be involved.[2,3,13] We have presented a child with features of this rare disorder who presented to us through the outpatient

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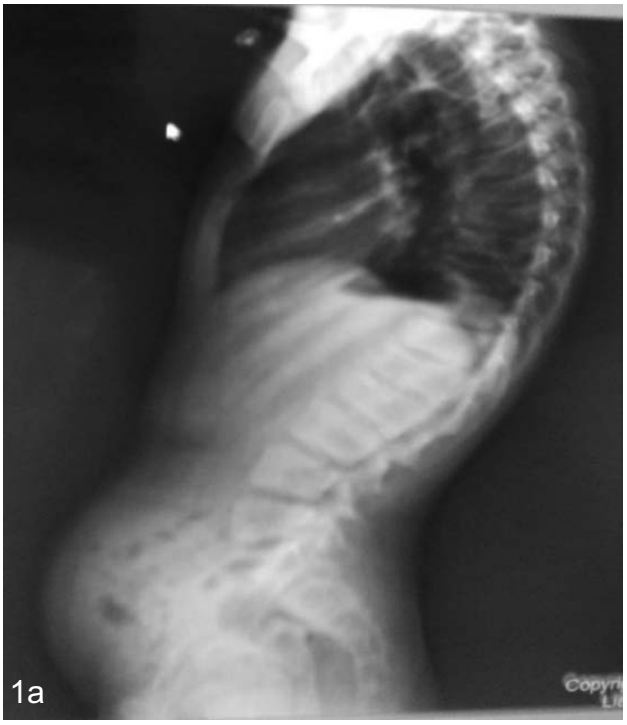
Submitted 28 May 2023, Accepted 9 July 2023

department and was diagnosed as SED T on clinical and radiological grounds.

Case Report

An eleven year old male presented to our department after 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 complains however no workup or documentary evidence of their illness was available. After 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 end plates 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.



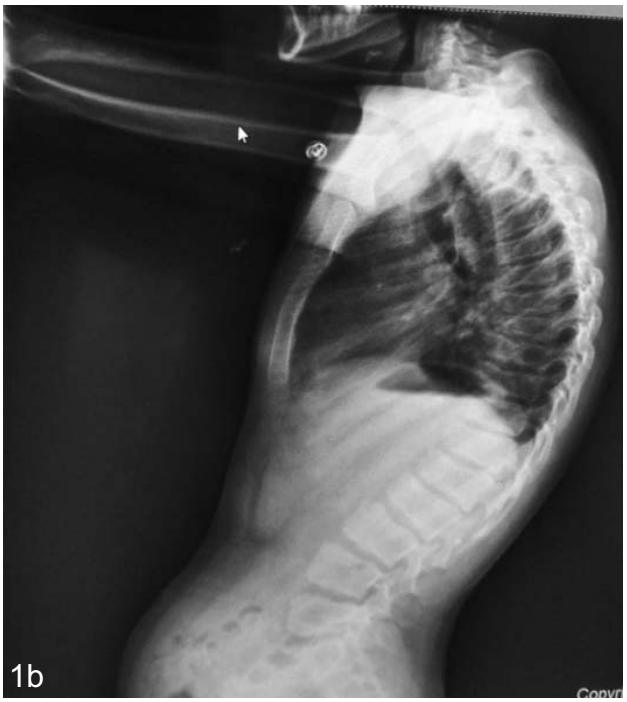


Figure 1a,b: lateral view of the dorsolumbar spine show flattening and widening of dorso-lumbar vertebral bodies along with end plate irregularity suggestive of platyspondyly. The edges of the endplates of vertebral bodies show characteristic anterior and posterior humped shape or heaped-up appearances

Discussion

Amongst the various osteochondrodystrophies, spondyloepiphyseal dysplasia tarda is one rare form that results in dwarfism primarily due to defect in the vertebrae. However as the name suggests it also involves the epiphysis of long bones.[6] This is a rare congenital disorder that has three further types as discussed previously, spondyloepiphyseal dysplasia congenita, spondy-loepiphyseal dysplasia tarda, and spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA).[5] 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 the distinctive features of other types of SED.[1-6] Later in 1911 Bannerman, Ingall and Mohn studied the familie s genetic linkage.

The symptoms and clinical presentation is unlikely before 11 to 13 years as this is time when the element of dwarfism becomes evident.[6] 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 referral from the OPD. The family history is positive in 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, humped shaped appearance mostly central and posteriorly along the body of the vertebrae.[6-8] These changes involve the spine diffusely from the 2nd cervical vertebrae up to the lumber 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 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 ischium and pubis resulting in a narrow pelvis. Femoral head degeneration with deep acetabula and degenerative changes involving the two. Shortening of the femoral necks is also a feature of the disease.[7-13] 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 an aberrant production of type II collagen, which inhibits bone formation.[8-9] 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 expense.

A frequent, dangerous consequence in individuals with SED is spinal cord compression brought on by atlanto-axial instability. The lack of spinal cord symptoms does not diminish the necessity for fusion and decompression procedures because, in our perspective, surgery's benefits are mostly preventative.[10] 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 SEDT-PA.[10-11] 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.[8-11] Management of complications includes spine surgery to repair scoliosis or kyphoscoliosis. Osteoarthritis pain treatment as needed is necessary joint replacement (hip, knee, shoulder). Monitoring 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.[11-12,14] The best course of action for SEDT 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.[8,9,14,15] 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 also important for the radiologist to give particular

look to the family history of the patient which can also help in making the diagnosis.

Conflict of Interest: None

References

1. MacKenzie, J. J., Fitzpatrick, J., Babyn, P., Ferrero, G. B., Ballabio, A., Billingsley & Costa, T. X linked spondyloepiphyseal dysplasia: a clinical, radiological, and molecular study of a large kindred. Journal of medical genetics 1996; **33(10):** 823-28.

2. Harper, P. S., Jenkins, P., & Laurence, K. M. Spondyloepiphyseal dysplasia tarda: a report of four cases in two families. The British Journal of Radiology 1993; **46(549):** 676-84.

3. Bannerman, R. M., Ingall, G. B., &Mohn, J. F. X-linked spondyloepiphyseal dysplasia tarda: clinical and linkage data. Journal of medical genetics1972; **8(3):** 291-94.

4. Kocyigit, H., Arkun, R., Ozkinay, F., Cogulu, O., Hizli, N., &Memis, A. Spondyloepiphyseal dysplasia tarda with progressive arthropathy. Clinical Rheumatology 2000; **19(3):** 222-35.

5. Iceton, J. A., & Horne, G.E.O.F.F.R.E.Y. Spondyloepiphyseal dysplasia tarda. The X-linked variety in three brothers. The Journal of Bone and Joint Surgery. British volume 1996; **68(4):** 616-19.

6. Whyte, M. P., Gottesman, G. S., Eddy, M. C., & McAlister, W. H. X-Linked Recessive Spondyloepiphyseal Dysplasia Tarda Clinical and Radiographic Evolution in a 6-Generation Kindred and Review of the Literature. Medicine 1996; 78(1): 9-25.

7. Gedeon, A. K., Tiller, G. E., Le Merrer, M., Heuertz, S., Tranebjaerg, L., Chitayat & Mulley, J. C. The molecular basis of X-linked spondyloepiphyseal dysplasia tarda. The American Journal of Human Genetics 2001; **68(6):** 1386-97.

8. Cao, L. A., & Bennett, J. T. Spondyloepiphyseal Dysplasia. In Orthopaedics for the Newborn and Young Child: A Practical Clinical Guide 2023; (pp. 241-245). Cham: Springer International Publishing.

9. Veeravagu, A., Lad, S. P., Camara-Quintana, J. Q., Jiang, B., &Shuer, L. Neurosurgical interventions for spondyloepiphyseal dysplasia congenita: clinical presentation and assessment of the literature. World Neurosurgery 2013; **80(3-4):** 437-e1.

10. Gembun, Y., Nakayama, Y., Shirai, Y., Miyamoto, M., Sawaizumi, T., & Kitamura, S.. A case report of spondyloepiphyseal dysplasia congenita. Journal of Nippon Medical School 2001; **68(2):** 186-89.

11. Bal, S., Kocyigit, H., Turan, Y., Gurgan, A., Bayram, K. B., G venc, A., ... &Dirim, B. Spondyloepiphyseal dysplasia tarda: four cases from two families. Rheumatology international 2009; **29:** 699-702.

12. Tiller, G. E. (2020). X-linked spondyloepiphyseal dysplasia tarda. GeneReviewsfi[Internet].

13. Tabban, H., Salem, T. A., & Salem, K. Y. Skeletal Dysplasia: Approach to Simplify Diagnosis, Looking for Radiographic Clue Signs. Asian Journal of Medicine and Health 2002; **20(12):** 65-76.

14. Ayaz, S. B., Rauf, S., & Rahman, F. Spondyloepiphyseal dysplasia congenita: report of a case and review of the literature. Journal of Postgraduate Medical Institute 2019; **33(1):** 213-16.

15. Chen, Z., Zhang, Z., Ye, F., Lei, F., & Feng, D. Multiple disc herniation in spondyloepiphyseal dysplasia tarda: A rare case report and review of the literature. BMC Musculoskeletal Disorders2016; **23(1):** 1-8.