CASE REPORT

Spondyloepiphysial Dysplasia

Dawood Sadik. Al-Obidi* ,Qasim Mohammed. Shakir**

ABSTRACT :

BACKGROUND:

A 13 years old girl from north of Iraq, presented with gross deformity of her back & difficulty in walking& then progressive difficulty in breathing. A thoracic scoliotic deformity with rib hump obvious on her back, it is a case report of spondyloepiphyseal dysplasis(SED) with primary involvement of the vertebrae & epiphyseal centers all over the body.

AIM OF THE STUDY:

To present a very rare case of (SED), including the deformities& the complications that can be presented with it.

METHOD:

Examining & survying a 13 years old Iraqi female from north of Iraq.

RESULTS:

A thoracic scoliotic deformity was found& became more obvious on bending foreword, also rib hump on the right side.Both hips in fixed flexion deformity & limitation of abduction.

DISCUSSION:

In SEDT the vertebral bodies are malformed& flattened, 1st described by Nilssone (1924).Wynne-Davies& Gormley(1985) estimated the prevalence to be 1 per 100.000 in a Scottish population.

KEY WORDS: Spondylopepiphyseal dysplasia Tarda (SEDT),Spondyloepiphyseal dysplasia congenita (SEDC).

INTRODUCTION:

A 13-years old female from north of Iraq, she was presented with gross deformity of her back & difficulty in walking, the story of this patient started since early childhood when there was a delay in walking, at the age of 11-year, her mother noted that there was a deformity of the back which became more progressive & she started to complain from difficulty in breathing

*Iraqi Board for Medical Specializations
**Surgical Specialities Teaching Hospital

Past history:.

There is no past history of trauma, chronic infection or other chronic diseases. She is a product of normal vaginal delivery of full term pregnancy. Her father & mother are relatives.

On\Examination:

When patient stands right, thoracic scoliotic deformity was obvious & became more obvious in bending foreword. Rib hump was obvious to the right side.

Lateral bending test was positive; the skin was normal, no pigmentation, no hair tuff, no signs of spina bifida.

Both hip joints on fixed flexion deformity & limitation of abduction.

Back ground:

Spondyloepiphyseal dysplasia (SED) is a descriptive term for a group of disorders with primary involvement of the vertebrae & epiphyseal centers resulting in a short-trunk disproportionate dwarfism(1).SED- congenital is caused by a mutation of the gene coding for collagen TypeII (COL2A1) found on chromosome 12(2).Most cases of this dysplasia are due to spontaneous mutations (3).

Types:

Two major types of SED are recognized namely: SED Congenita& SED Tarda.

Frequency:

The prevalence is approximately 3.4 per one million populations (Wynne_ Davies, 1982).

Sex:

SED congenita is autosomal dominant, SED tarda is X-Linked recessive, hence,in SEDT only males are affected.

Deformities:

- Thoracolumbar scoliosis (50%).
- Kyphosis & Kyphoscoliosis are common in these patients.
- These deformities occur much earlier, progress more quickly & are more rigid than those in idiopathic conditions.
- Some patients may have valgus deformities of the knee in addition to coax vara of the hip.

- Thoracic scoliosis or kyphosis may become evident in adolescence, single & double curve patterns have been noted.
- Thoracic kyphosis may be severe measuring up to 130°; kyphosis or kyphoscoliosis can be rigid & severe.

Differential diagnosis:

1. Achondroplasia: (This has characteristic facial changes).
2. Diastrophic dysplasia: (deformities of hands, club feet, joint contractures and cauliflower ears).
3. Morquio disease.
4. Pseudochondroplasia: (short limb dwarfism, hands& feet affected).
5. Perthe´s disease: (asymmetrical stages, evidence of healing).
6. Kniest disease: (characteristic facies, absent platyspondyly).
7. Metatrophic dwarfism: (paper-thin vertebrae, dumb-bell shaped metaphyseal flaring, battle-axe like pelvic configuration).

Complications: common conditions associated with SED include the following:

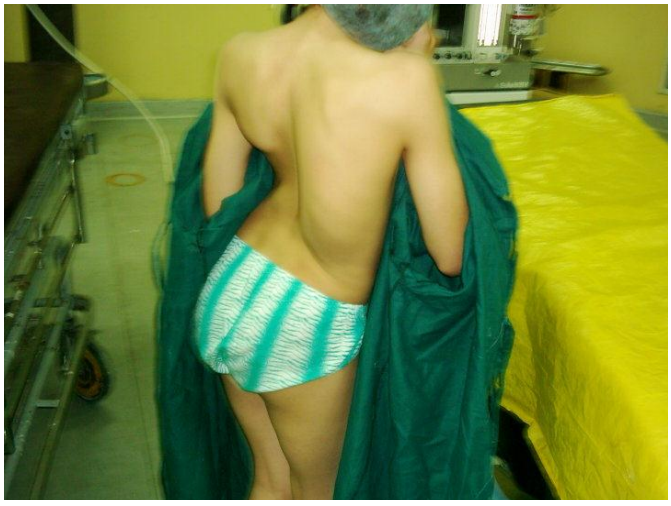
- Neck instability.
- Spinal deformities such as scoliosis, kyphosis, or lordosis.
- Ocular abnormalities such as myopia or retinal detachment.
- Hearing deficits.
- Hip deformities including coax vara, capital femoral epiphyseal involvement.
- Genu valgum.
- Equinovarus feet.
- Degenerative joint disease of the hips, knees, or shoulders.

DISCUSSION:

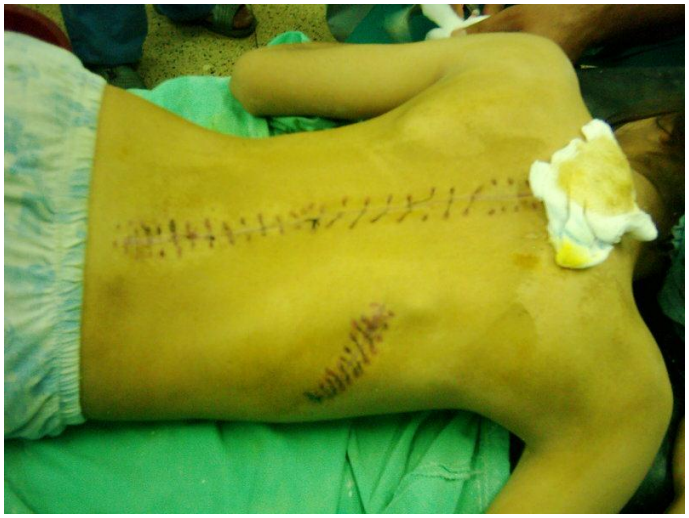
In SEDT the vertebral bodies are malformed & flattened and most of the dense area is part of the vertebral plate. This disorder was probably first described by Nilsonne (1927) [8].

Follow up by Tacobsen's family was provided by Bannerman et al.(1971) [9]. Bannerman (1981) reviewed his material & concluded that heterozygotes show no abnormality such as as short stature. Wynne-Davies and Gormely(1985), estimated the prevelance to be 1 per 100.000 in a Scottish population.[4]

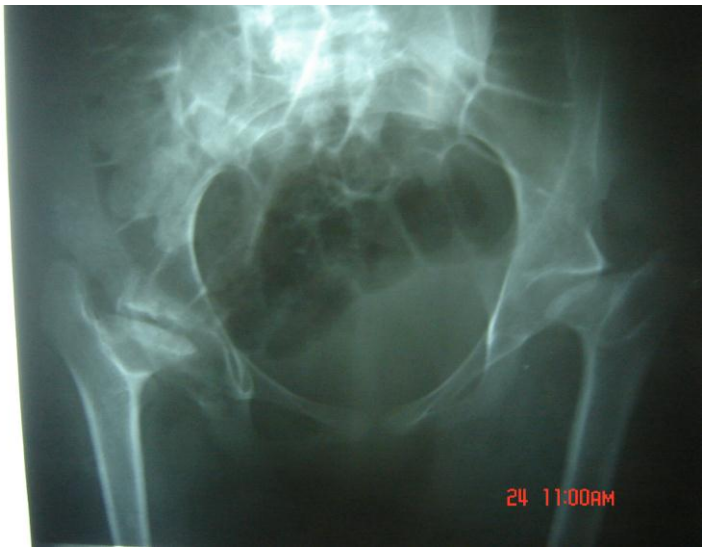
Whyte et al.(1999) described the clinical & radiographic evalution of a second large American kindred with X-linked recessive SEDT, the first such family being the classic family reported by Jacobson(1939). [10]



Sondyloepiphyseal Dysplasia (Spinal deformity)



(SED) After correction of scoliosis



(SED) Coxa Vara

REFERENCES:

1. Bailey J.A. Disproportionate short stature: Diagnosis& management.Philadelphia, WB Saunders. 1973.
2. Jones, Kenneth L.Recognizable patterns of Human Malformation. Philadelphia,PA: Elsevier Saunders.2006.6ed.Pp426.
3. Scott,Charles I, Dwarfism.Clinical Symposium, 1988; 40,17-18.
4. Wynne-Davies,R.Gormley,J.:The prevalence of skeletal dysplasia;an estimate of their minimum frequency and the number of patients requiring orthopedic care,J. Bone Joint surg.1985;67B,133-137.
5. Whyte,M.P.:gottesman,G.S.:Eddy,M.C:Mc Alister,W.H:X-Linked recessive Spondyloepiphyseal Dysplasia Tarda:clinical & radiographic evolution in a 6-generation kindred and review of the literature Medicine 1999;78, 9-25.
6. Kopits,Steven E. Orthopedic Complication of Dwarfism.Clinical Orthopedics and related Research. 1976;144,153-179.
7. Weinfeld A, Ross MW, Sara Sohn SH. Spondyloepiphyseal dysplasid; a cause of premature osteoarthritis AJR 1967;101,851.

8. Nilsonne, H:Eigent ue mliche wirbelkorper-veraenderungen mit familiaerem auftereten. Aeta chir. Scand. 1927;62,550-554.

9. Bannerman,R.M.: Ingall,G.B: Mohn,J.F.X-Linked Spondyloepiphseal dysplasia tarda:clinical & linkage data.J.Med.Genet. 1971;8,291-301.

10. Taybi, Hooshang.Lachman,Ralph S. Radiology of Syndromes, Metabolic Disorders, and Skeletal Dysplasias. St. Louis,MO: Mosby-Year book,Inc. 1996.